CURRICULUM VITAE

INFORMAZIONI PERSONALI

Cognome e Nome	**GASPARINI PAOLO**
Qualifica	**ABILITATO ALLA PROFESSIONE DI MEDICO-CHIRURGO, PROFESSORE ORDINARIO DI GENETICA MEDICA**
Incarico attuale	**PROFESSORE ORDINARIO, DIRETTORE DI DIPARTIMENTO (DIPARTIMENTO DEI SERVIZI E DIAGNOSTICA AVANZATA), DIRETTORE DI STRUTTURA COMPLESSA (GENETICA MEDICA)**
Indirizzo e-mail	**paolo.gasparini@burlo.trieste.it**

TITOLI DI STUDIO E PROFESSIONALI ED ESPERIENZE LAVORATIVE

• Titolo di studio

- **Maturita' Classica** conseguita a Cagliari nel **1979** riportando la votazione di 60/60

- **Laurea in Medicina e Chirurgia** conseguita nel **1985**, presso l'Universita' di Torino, con la votazione di 108/110 e l'assegnazione della dignita' di stampa discutendo una tesi su: "Meccanismo di leucemogenesi nell'uomo: traslocazione dell'oncogene 5' myc all'enhancer di una catena immunoglobulinica pesante nel DNA tumorale di un paziente affetto da leucemia linfatica acuta (L3)"

- **Abilitato** alla professione di medico-chirurgo nella sessione autunnale degli Esami di Stato (Torino) per l'anno **1985**

- **Specialita' in "Ematologia Generale (Clinica e di Laboratorio)"** conseguita nel **1988** presso l'Universita' di Verona, con una tesi su "Il riarrangiamento genico bcr-abl in corso di Leucemia Mieloide Cronica" ottenendo i pieni voti e la lode

- **Specialita' in "Genetica Medica"** conseguita nel **1992** presso l'Universita' di Verona, con una tesi su "Prevenzione della Fibrosi Cistica in Italia" ottenendo i pieni voti e la lode

• Altri titoli di studio e professionali

- **Vincitore** per l'aa. **86-87** di una borsa di studio in tema "Emopatie Maligne" bandita dal Comitato Gigi Ghirotti, presso la **Clinica Medica A** dell'**Universita' di Torino** (Diretta dal Prof. Felice Gavosto)

- **Assegnatario** per gli anni **1987, 1988, 1989** di una borsa di studio dell'ULSS 25 offerta dal "Centro Regionale Veneto per lo Studio della Fibrosi Cistica", presso l'**Istituto di**

Scienze Biologiche dell'**Universita' di Verona** (diretto dal Prof. Pier Franco Pignatti)

- **Vincitore** del **"Premio Socrea-Sirtori 1988**" per il migliore comunicazione scientifica nel campo della Genetica Medica a tema: "Diagnosi prenatale di fibrosi cistica in 24 ore mediante l'uso della Polymerase Chain Reaction" tenuta al 3°Congresso Nazionale F.I.S.M.E.

- **Vincitore** del **"Premio A.I.R.H. Junior 1991"** per il contributo dato allo sviluppo della ricerca in Genetica Medica Molecolare ed in particolare nel campo delle malattie ereditarie ad insorgenza infantile come la Fibrosi Cistica

- **Vincitore** del **"Premio SIBioC '94"** per la comunicazione "Rivelazione di mutazioni mediante RNA single strand conformation polymorphism (rSSCP)

- **Vincitore** del **"Premio Augustale 99"** per la ricerca scientifica

- **Vincitore** nel 2000 del **"International Journal of Molecular Medicine Award"** perla ricerca scientifica

- **Vincitore** nel 2003 del **"The European Society of Human Genetics/NaturePublishing Group Award"** per l'articolo più citato nel 2001

- **Vincitore** nel 2010 del **"QNRF award for biomedicine"** al convegno annuale della Qatar National Research Foundation per migliore presentazione nel settorebiomedico

- **Vincitore** nel 2011 del **"Premio Nazionale Ippocrate"** quale migliore ricercatore/divulgatore del 2011

- **Vincitore** nel 2021 del **"Premio Medicina Italia"** per la ricerca scientifica.

● Esperienze professionali (incarichi ricoperti)

2005-oggi

Università degli Studi di Trieste, Trieste.

Dipartimento Universitario Clinico di Scienze Mediche Chirurgiche e della Salute.

- Professore ordinario di Genetica Medica presso il Dipartimento Universitario Clinico di Scienze Mediche Chirurgiche e della Salute e del Dipartimento di Scienze della Vita.
- Direttore della Scuola di Specializzazione in Genetica Medica dell'Università degli Studi di Trieste (fino a ottobre 2024).
- Coordinatore del Dottorato in Medicina Personalizzata e Terapie Innovative dell'università degli Studi di Trieste (fino a ottobre 2024).

- Docente di Genetica Medica presso varie scuole di specializzazione afferenti all'ateneo triestino.

2005-oggi

I.R.C.C.S. Materno Infantile Burlo Garofolo, Trieste

- Direttore della Struttura Complessa Genetica Medica.
- Direttore del Dipartimento dei Servizi e di Diagnostica avanzata (dal 2011).

Durante tale periodo ha svolto un'intesa attività di consulenze genetiche sia interne alla struttura che esterne (ambulatorio per esterni). Nel gennaio 2006 è stato nominato membro della commissione Tecnico-Scientifica del CBM (Consorzio per il Centro di Biomedicina Molecolare) di Trieste, contribuendo all'avvio delle attività del CBM stesso e collaborando con lo stesso per un quinquennio nel quale è stato dato anche avvio al progetto "Parco Genetico del FVG" finalizzato allo studio di sei diverse popolazioni isolate. Dal 1 giugno 2009 al 30 giugno 2010 Direttore Scientifico *ad Interim* dell'IRCCS- Burlo Garofolo, incarico che ha poi di nuovo ricoperto dal 1 agosto 2020 al 31 dicembre 2021. Nel 2009 è stato Visiting Scientist al Sanger Institute, Cambridge UK (2009). Nell'estate del 2010 è stato il promotore e coordinatore scientifico della spedizione scientifica "Marco Polo 2010 che ha attraversato ben 10 nazioni localizzate sulla Via della Seta. Da gennaio 2015 ad aprile 2017 ha trascorso un periodo sabbatico quale responsabile di un team di ricercatori presso il Sidra Medical and Research Center (Doha, Qatar). Le tematiche di ricerca più recenti includono le basi genetiche delle malattie ereditarie, dei caratteri quantitativi e delle malattie complesse, con un focus specifico sugli organi di senso e malattie correlate (es. udito, gusto, preferenze alimentari, olfatto).

Durante questi anni ha contribuito alla realizzazione di aziende:

- G&Life che opera nel settore della nutrigenomica e diete personalizzate (http://www.genex.me/)
- Nanosynthex srl che opera nel settore delle nanotecnologie
- Vergal Beer che opera nel settore alimentare (nutrigenetica) (https://www.vergalbeer.com/)

Incarichi di prestigio

Giugno 2008 – ad oggi

- Dall'estate 2016 è membro del Comitato Nazionale delle Biotecnologie, Biosicurezza e Scienze della Vita della presidenza del Consiglio (CNBBSV).
- Presidente dell'Scientific Advisory Board dell'IDIBELL del l'Hospitalet de Llobregat (Barcellona, Spagna), di cui è stato in precedenza membro.
- Membro del Gruppo di lavoro – Ricerca & Sviluppo dell'AIFA (Agenzia Italiana del Farmaco).
- Delegato italiano presso il CAT (Commitee for Advanced Therapies) dell'EMA (European Medicines Agency) (per nove anni sino al febbraio 2021).
- Dal 2020 è Collaboratore Scientifico presso la Fondazione Cereneo (Center for Neurology and Rehabilitation).

- Nel luglio 2022 è stato nominato Membro (Rappresentante dei Medici Europei) del Comitato per le Terapie Avanzate (CAT) EMA.
- Nel novembre 2022 è stato nominato Presidente della SIGU (Società Italiana di Genetica Umana), in precedenza membro del Consiglio Direttivo (per un triennio fino al 2025)
- Nel 2023 è stato nominato delegato italiano del CHMP (Committee for Medicinal Products for Human Use) dell'EMA
- Nel 2023 è stato nominato Membro del Comitato Nazionale Malattie Rare, membro del Gruppo di Coordinamento Nazionale per la Bioeconomia (Presidenza del Consiglio dei Ministri), membro del Tavolo Tecnico sulle Terapie Avanzate istituito dal Ministero della Salute.
- Nel 2023 è stato nominato come rappresentante italiano in seno al Comitato dei prodotti medicinali per uso umano (CHMP) dell'Agenzia europea per i medicinali (EMA).
- Nel 2024 è stato nominato componente della Commissione Scientifica per la Fondazionie Lorenzini.
- Nel 2024 è stato nominato membro del Comitato Scientifico del Laboratorio sul Management delle Sperimentazioni Cliniche (Lab MSC), con il ruolo di consultente del Lab MSC presso ALTEMS (Alta Scuola di Economia e Management dei Sistemi Sanitari, Università Cattolica del Sacro Cuore).
- Nel 2025 è stato nominato membro del Comitato etico nazionale per le sperimentazioni cliniche relative a terapie avanzate (Advanced Medicinal Therapeutical Products "ATMP"), presso l'Agenzia italiana del farmaco (AIFA).

2001-2005

Seconda Università degli Studi di Napoli, Napoli.

Facoltà di Medicina e Chirurgia.

- Professore associato di Genetica Medica
- Titolare dell'insegnamento di Genetica Medica (sede staccata di Caserta).
- Docente del corso integrato di Biologia, Genetica e Genetica Medica.
- Docente di Genetica Medica presso varie scuole di specializzazione e vari corsi di DU
- Coordinatore del Dottorato di Ricerca in Genetica Medica.

Ha avviato il laboratorio di ricerca di Genetica Medica all'interno del Dipartimento di Patologia Generale. Ha inoltre aderito al Centro di Eccellenza per le Malattie Cardiovascolari della Seconda Università degli Studi di Napoli dove ha realizzato un laboratorio di nanotecnologia. È stato inoltre responsabile dell'Unità di Linkage e Mapping dell'Istituto TIGEM (Telethon Institute of Genetics and Medicine) di Napoli e coordinatore di un gruppo di ricerca sempre all'interno dello stesso TIGEM. Ha proseguito le attività di ricerca sulle perdite uditive, sulla definizione delle basi molecolari delle malattie multifattoriali in popolazioni geneticamente isolate. Nel corso di questi anni ha contribuito all'identificazione dei geni per le seguenti patologie: perdita uditiva da miosina 1A, perdita uditiva da miosina MYH14, perdita uditiva da mutazioni nel gene Espin,

encefalopatia metilmalonica.

È stato coordinatore del Dottorato di Ricerca in Genetica Medica dall'ottobre 2003 sino al giugno 2005**.**

A partire dal novembre 2003 è stato Primario del Servizio di Genetica Medica attivato presso il DAS-Medicina di Laboratorio della Seconda Università degli Studi di Napoli, Azienda Universitaria Policlinico. In questo ruolo ha attivato ed organizzato l'Ambulatorio di Consulenza Genetica, Il laboratorio di Diagnostica Molecolare, le attività di citogenetica, dotando l'AUP di una struttura di cui era carente.

È stato referente aziendale per l'Azienda Universitaria Policlinico della Seconda Università degli Studi di Napoli nell'ambito della Rete Regionale di assistenza a favore dei soggetti affetti dalle Malattie Rare. Ha realizzato SUN-GENS il primo servizio nazionale di genotipizzazione di SNPs, che ha operato offrendo i propri servizi a diverse Università italiane. Membro da novembre 1999 a dicembre 2006 della Commissione di Bioetica dell'IRCCS-Ospedale "Casa Sollievo della Sofferenza

1993-2001

I.R.C.C.S. Ospedale Casa Sollievo della Sofferenza, San Giovanni Rotondo (FG)

- Aiuto Medico e ricercatore presso il Servizio di Genetica Medica (Primario Dott. Leopoldo Zelante).
- Professore a contratto di Genetica Medica e Genetica Molecolare.

Ha proseguito l'attività di ricerca e di diagnostica sull'emocromatosi ereditaria, sulla neurofibromatosi di Tipo 1,sulla fibrosi cistica, sulla distrofia di Duchenne, e sul rene policistico dell'adulto. Ha avviato un progetto di ricerca sulla calcolosi cistinica contribuendo in maniera determinante all'identificazione del gene (aprile 1994) che, una volta alterato, causa tale patologia, e successivamente alla dimostrazione della presenza di eterogeneità genetica. Ha inoltre contribuito all' identificazione del gene che causa una rara sindrome costituita da cataratta ed iperferritinemia (1995).

Ha avviato un importante progetto di studio sulle sordità genetiche, identificando nella connessina-26 il gene che causa la forma più comune di sordità genetica (1997). Nel corso del 1998 ha collaborato ad un progetto di ricerca che ha portato alla identificazione del gene per la Malattia di Leigh. Sempre nel corso del 1998 è stato, dal 15 luglio al 15 ottobre, "Visiting Professor" presso il Department of Haematology dell'Università di Pennsylvania, dove ha acquisito le basi per lo sviluppo delle nuove tecnologie microchip. Nel 1999 ha coordinato i ricercatori del Servizio di Genetica Medica che hanno clonato ed identificato la connessina-30, un nuovo gene della sordità. Sempre nel 1999 ha contribuito in maniera determinante all'identificazione del gene per la calcolosi renale cistinica di tipo non-I. Nel corso dell'anno 2000 ha contribuito all'identificazione di un nuovo gene che causa emocromatosi ereditaria (TFR2). Nel 2001 ha contribuito all'identificazione dei seguenti geni malattia: sindrome BPES, sordità dovuta al gene miosina 6, sindrome di Usher di tipo III, emocromatosi di tipo dominante (HFE4).

In questi anni il suo apporto é stato poi determinante nel gestire e coordinare l'Unità di Mapping del Servizio di Genetica Medica che ha portato all'identificazione di diversi loci malattia. I risultati di questa intensa attività di ricerca sono stati oggetto di un elevato numero di relazioni tenute a diversi congressi nazionali ed internazionali.

Ha inoltre avviato alcuni progetti di ricerca sulle basi molecolari delle malattie multifattoriali. in particolare ha collaborato a progetti di ricerca su Celiachia, Morbo di Chron e Rettocolite Ulcerosa, Osteoporosi ed ha avviato un progetto per lo studio di un isolato genetico-geografico (Progetto Carlantino).

Ha svolto attività di Consulenza Genetica (Medica e Clinica) presso l'Ambulatorio del Servizio di Genetica Medica, e gestito direttamente la sezione di diagnostica molecolare.

Ha inoltre mantenuto un'intensa attività didattica, come segue:

aa 1993-1994

Professore a Contratto di Genetica Molecolare (C.I. Genetica Medica) presso la Scuola di Specializzazione di Ematologia dell'Universita' degli Studi "G.D'Annunzio" di Chieti (Direttore Prof. Torlontano).

aa 1994 –1997

Professore a Contratto di Genetica Molecolare (C.I. Genetica Medica) presso la Scuola di Specializzazione in Ematologia dell'Università degli Studi di Verona (Direttore Prof. Perona).

aa 1996 – 2000

Professore a Contratto di Genetica Medica presso la Scuola per Diploma per Tecnici di Laboratorio dell'Università degli Studi di Foggia.

aa 1997-1998

Professore a Contratto di Genetica Molecolare (C.I. BIOTECNOLOGIE) pressol'Università degli Studi di Ferrara.

Maggio 1992 - Marzo 1993

I.R.C.C.S. Ospedale Casa Sollievo della Sofferenza, San Giovanni Rotondo (FG)

- Assistente medico presso il Servizio di Genetica Medica (Primario Dott. Leopoldo Zelante) con compiti di gestione del Laboratorio di Ricerca in Genetica Medica.

Ha organizzato il laboratorio avviando progetti di ricerca sull'emocromatosi ereditaria, sulla fibrosi cistica, e sulla neurofibromatosi. Ha inoltre concorso allo sviluppo ed applicazione delle tecniche di biologia molecolare alla diagnosi di alcune patologie ereditarie quali la distrofia di Duchenne e il Rene Policistico dell'adulto. I risultati di questi lavori sono stati oggetto di relazioni tenute a diversi congressi Nazionali ed Internazionali

Ha svolto attività di Consulenza Genetica (Medica e Clinica) presso l'Ambulatorio del Servizio di Genetica Medica, e gestito direttamente la sezione di diagnostica molecolare implementando la diagnostica molecolare di diverse patologie ereditarie.

Febbraio 1990 - Maggio 1992

Istituto di Scienze Biologiche dell'Universita' degli Studi di Verona (diretto dal Prof. Pier Franco Pignatti)

- Funzionario Tecnico di VIII Livello

Ha partecipato attivamente alla scoperta di diverse mutazioni presenti nel gene della fibrosi cistica ed ad una approfondita analisi delle frequenze di molte altre mutazioni FC nel bacini del Mediterraneo. Stage di ricerca presso Il Departament de Genetica Molecular dell'"Hospital de St.Pau y S.ta Creu" Barcelona (Spagna) (diretto dal Prof. X.Estivill) nell'ambito di un progetto collaborativo per lo studio delle mutazioni del gene della fibrosi cistica (1990). I risultati delle attività di ricerca sono stati oggetto di una Lecture dal titolo "Screening della fibrosi cistica in Italia: prospettive ad un anno dalla scoperta del gene" tenuta a Perugia nell'ottobre 1990 nell'ambito della sessione sullo screening delle malattie genetiche in Italia del Congresso Nazionale Congiunto A.G.I.- F.I.S.M.E. Ha iniziato ad interessarsi attivamente alla ricerca del difetto genetico coinvolto nello sviluppo di una grave e frequente patologia genetica quale l'emocromatosi primaria. Nell'ambito di tale progetto dopo un ampio lavoro di analisi genetica si è potuto localizzare in maniera più precisa il gene coinvolto in tale patologia, in una piccola porzione di cromosoma 6. I risultati di questi lavori sono stati oggetto di relazioni tenute a diversi congressi nazionali ed internazionali

Febbraio 1987 - Febbraio 1990

Istituto di Scienze Biologiche dell'Università degli Studi di Verona (diretto dal Prof. Pier Franco Pignatti)

- Borsista presso l'Istituto di Scienze Biologiche dell'Università di Verona

Principali campi di ricerca attivamente sviluppati:

a) analisi molecolari del gene della fibrosi cistica (FC), del rene policistico dell'adulto (APKD), e della broncopneumopatia cronica ostruttiva (BPCO).
b) applicazione di sonde molecolari alla diagnostica prenatale e alla individuazione dei portatori del gene FC) sviluppo di metodiche rapide e allo stesso tempo sensibili per indagini molecolari in diverse patologie genetiche (amplificazione del DNA etc.).
c) monitoraggio e follow-up dei trapianti di midollo osseo eterologo.
d) analisi del riarrangiamento bcr-abl nei pazienti affetti da Leucemia Mieloide Cronica (LMC).
e) valutazione della malattia minima residua nei pazienti affetti da LMC dopo terapiacon interferon e/o chemioterapia e/o trapianto di midollo osseo.

f) caratterizzazione e sviluppo di sonde molecolari ipervariabili da utilizzarsi a scopodi identificazione individuale (biologia forense etc.).

Stage di ricerca presso il Department of Biochemistry del "St. Mary's Hospital", Londra (GB) (diretto dal Prof. R.Williamson) nell'ambito di un programma collaborativo per lo studio della fibrosi cistica (1987).
I risultati delle attività di ricerca sono stati oggetto di relazioni tenute a diversicongressi Nazionali ed Internazionali.

Gennaio 1984 - Gennaio 1987

Clinica Medica A dell'Universita' di Torino, Dipartimento di Scienze Biomediche ed Oncologia Umana (diretto dal Prof. Felice Gavosto)

- Interno

Attività svolta:

Frequenza nel reparto ospedaliero e presso il laboratorio di Ematologia, dove si e' interessato attivamente dello studio delle alterazioni molecolari nella patologia ematologica con particolare riguardo alle alterazioni molecolari nelle emoglobinopatie e allo studio degli oncogeni nelle diverse situazioni patologiche. Dall'Ottobre '86 ha collaborato al programma di ricerca "Polimorfismo dei proto-oncogeni umani" presso il Centro CNR di Immunogenetica ed Istocompatibilita' di Torino diretto dal Professor Ruggero Ceppellini.

Capacità linguistiche

Lingua	Livello parlato	Livello Scritto
Inglese	C2	C2
Spagnolo	C1	B2

Altro (partecipazione a convegni e seminari, pubblicazioni, collaborazione a riviste, ecc., ed ogni altra informazione che il dirigente ritiene di dover pubblicare)

Pubblicazioni Scientifiche:

PUBBLICAZIONI SU RIVISTE SCIENTIFICHE INTERNAZIONALI

1. Camaschella C, Bertero MT, Serra A, Dall'Acqua M, Gasparini P, Trento M, Vettore L, Perona G, Saglio G, Mazza U. *A benign form of thalassemia intermedia may be determined by the interaction of triplicated alfa locus and heterozygous beta thalassemia*. **Br J Haematol.**1987 May; 66(1): 103-107.
2. Saglio G, Camaschella C, Giai M, Serra A, Guerrasio A, Peirone B, Gasparini P, Mazza U, Ceppellini R, Biglia N, Cortese P, Sismondi P. *Distribution of Ha-RAS-1 proto-oncogene alleles in breast cancer patients and in a control population.* **Breast Cancer Res Treat.**1988 May; 11(2): 147-153. 2-s2.0-0023914499.
3. Estivill X, Farrall M, Williamson R, Ferrari M, Seia M, Giunta AM, Novelli G, Potenza L, Dallapiccola B, Borgo G, Gasparini P, Pignatta PF, De Benedetti L, Vitale E, Devoto M, Romeo G. *Linkage disequilibrium between Cystic Fibrosis and linked DNA polymorphisms in Italian Families: a collaborative study*. **Am J Hum Genet.**1998; 43: 23-28. 2-s2.0-0023721932.

4. Kitzis A, Chomel JC, Haliassos A, Tesson L, Kaplan J, Feingold J, Giraud G, Labbe A, Dastugue B, Dumur V, Farriaux J, Roussel P, Ferec C, Vidaud M, Goossens M, Bozon D, Auvinet M, Chambon V, Andre J, Lissens W, Liebaers I, Cochaux P, Vassart G, Willems P, Duckworth-Raysiecki G, Kerem B, Tsui LC, Ray P, Krawzak M, Schmidtke J, Novelli G, Dallapiccola B, Gasparini P, Pignatta PF, Seia M, Ferrari M, Devoto M, Romeo G, Schwarz M, Super M, Ivinson A, Read A, Meredith L, Curtis A, Williamson R, Beaudet A, Feldman G, O'Brien W, Bowcock A, Cavalli-Sforza I, Gilbert F, Braman J, King M. *Cystic Fibrosis allele segregation*. **Nature.**1988; 336: 316. 2-s2.0-36849150104.
5. Gasparini P, Savoia S, Pignatti PF, Novelli G, Dallapiccola B. *Amplification of DNA from epithelial cells in urine*. **N Engl J Med.**1989 Mar 23; 320(12): 809. 2-s2.0-0024563721.
6. Gasparini P, Novelli G, Savoia A, Dallapiccola B, Pignatti PF. *First trimester prenatal diagnosis of cystic fibrosis using the polymerase chain reaction: report of eight cases*. **Prenat Diagn.**1989 May; 9(5): 349-355. 2-s2.0-0024602904.
7. Estivill X, Gasparini P, Novelli G, Casals T, Nunes V, Gallano P, Savoia A, Ruzzo A, Dallapiccola B, Pignatti PF. *Linkage disequilibrium for DNA haplotypes near the cystic fibrosis locus in two south European populations*. **Hum Genet.**1989 Sep; 83(2): 175-178. 2-s2.0-0024401333.
8. Gasparini P, Pignatti PF, Mastella G. *Sex difference in D7S8 marker allele distribution in adult cystic fibrosis patients*. **Lancet.**1989 Sep 16, 2(8664): 686-687. 2-s2.0-0024462965.
9. Estivill X, Chillon M, Casals T, Bosch A, Morral N, Nunes V, Gasparini P, Savoia A, Pignatti PF, Novelli G, Dallapiccola B, Fernandez E, Benitez J, Williamson R. *Delta F508 gene deletion in cystic fibrosis in Southern Europe*. **Lancet.**1989 Dec 9; 2(8676): 1404.
10. Gasparini P, Martinelli G, Trabetti E, Ambrosetti A, Benedetti F, Pignatti PF. *Bone marrow transplantation monitoring by DNA analysis*. **Bone Marrow Transplant.**1989 Dec; 4 Suppl: 157-159. 2-s2.0-0024952091.
11. Gasparini P, Novelli G, Estivill X, Olivieri D, Savoia A, Ruzzo A, Nunes V, Borgo G, Antonelli M, Williamson R, Pignatti PF, Dallapiccola B. *The genotype of a new linked DNA marker, MP6-d9, is related to the clinical course of cystic fibrosis.* **J Med Genet.** 1990 Jan; 27(1): 17-20.
12. Ferrari M, Antonelli M, Bellini F, Borgo G, Castiglione O, Curcio L, Dallapiccola B, Devoto M, Estivill X, Gasparini P, Giunta A, Marianelli L, Mastella G, Novelli G, Pignatti PF, Romano L, Romeo G, Seia M, Williamson R. *Genetic differences in cystic fibrosis patients with and without pancreatic insufficiency. An Italian collaborative study*. **Hum Genet.**1990 Apr; 84(5): 435-438. 2-s2.0-0025275639.
13. Gasparini P, Trabetti E, Savoia A, Marigo M, Pignatti PF. *Frequency distribution of the alleles of several variable number of tandem repeat DNA polymorphisms in the Italian*

population. **Hum Hered.**1990; 40(2): 61-68. 2-s2.0-0025239283.

14. Gasparini P, Savoia A, Luisetti M, Peona V, Pignatti PF. *The cystic fibrosis gene is not likely to be involved in chronic obstructive pulmonary disease*. **Am J Respir Cell Mol Biol.**1990 Mar; 2(3): 297-299. 2-s2.0-0025391789.
15. Novelli G, Gasparini P, Savoia A, Pignatti PF, Sangiuolo F, Dallapiccola B. *Polymorphic DNA haplotypes and delta F508 deletion in 212 Italian CF families*. **Hum Genet.**1990 Sep; 85(4): 420-421.
16. Gasparini P, Cappello N, Dallapiccola B, Devoto M, Estivill X, Ferrari M, Leoni G, Novelli G, Piazza A, Pignatti PF, Rosatelli C, Romeo G, Savoia A, Seia M, Williamson R. *Regional distribution of cystic fibrosis linked DNA haplotypes in Italy, a collaborative study*. **Gene Geogr.**1990 Apr; 4(1): 53-64. 2-s2.0-0025417158.
17. Gasparini P, Pignatti PF, Novelli G, Dallapiccola B, Nunes V, Casals T, Estivill X, Fernandez E, Balassopoulos A, Loukopoulos D, Lavinha J, Simova L, Komel R. *Mutation analysis in cystic fibrosis*. **New Engl J Med.**1990; 323: 62-63.
18. Novelli G, Sangiuolo F, Dallapiccola B, Gasparini P, Savoia A, Pignatti PF, Fernandez E, Benitez J, Casals T, Nunes V, Manas P, Estivill X. *Delta F508 gene deletion and prenatal diagnosis of cystic fibrosis in Italian and Spanish families*. **Prenat Diagn.**1990 Jun; 10(6): 413-414. 2-s2.0-0025063540.
19. Borgo G, Mastella G, Gasparini P, Pignatti PF. *Genetic analysis and pancreatic function in cystic fibrosis*. **Lancet.**1990; 1: 1601. 2-s2.0-0025337027.
20. The Cystic Fibrosis Genetic Analysis Consortium. *Worldwide survey of the delta F508*. **Am J Hum Genet.**1990; 47: 354-359.
21. Borgo G, Mastella G, Gasparini P, Zorzanello A, Doro R, Pignatti PF. *Pancreatic function and gene deletion F508 in cystic fibrosis*. **J Med Genet.**1990 Nov; 27(11): 665-669. 2-s2.0-0025155352.
22. Martinelli G, Gasparini P, Ambrosetti A, Perona G, Pignatti PF. *Molecular genetic analysis of bcr breakpoints in Philadelphia chromosome rearrangements and correlation with prognosis and age of CML patients*. **Progress in Clinical and Biological Research**. Hemisphere Publishing Corporation. Washington, D.C.: 1990; 185 – 191.
23. Gasparini P, Pignatti PF, Borgo G, Mastella G. *Sweat cloride concentration in cystic fibrosis patients varies with KM. 19 genotype but not with the presence of the common F508 deletion*. **Am J Med Genet.**1991 May 1; 39(2): 230-231.
24. Gasparini P. *Genetics of cystic fibrosis*. **Dig Dis.**1991; 9(3): 179-188.
25. Gasparini P, Mandich P, Novelli G, Bellone E, Sangiuolo F, De Stefano F, Potenza L, Trabetti E, Marigo M, Pignatti PF, Dallapiccola B, Ajmar F. *Forensic applications of molecular genetic analysis: an Italian collaborative study on paternity testing by the determination of variable number of tandem repeat DNA polymorphisms*. **Hum Hered.**1991; 41(3): 174-181.

2-s2.0-0025854795.

26. Gasparini P, Nunes V, Savoia A, Dognini M, Morral N, Gaona A, Bonizzato A, Chillon M, Sangiuolo F, Novelli G, Dallapiccola B, Pignatti PF, Estivill X. *The search for south European cystic fibrosis mutations: identification of two new mutations, four variants, and intronic sequences*. **Genomics.**1991 May; 10(1): 193-200. 2-s2.0-0025762972.
27. Gasparini P, Dognini M, Bonizzato A, Morral N, Estivill X. *A tetranucleotide repeat polymorphism in the cystic fibrosis gene*. **Hum Genet.**1991 Apr; 86(6): 625. 2-s2.0-0025729981.
28. Trabetti E, De Leo D,Gasparini P, Pignatti PF, Marigo M. *DNA amplification (PCR) of several hypervariable loci in disputed paternity cases.* **DNA Technology and its Forensic Application**. Springer-Verlag Editor. Berlin: 1991; 92-95
29. Hughes T, Ambrosetti A, Barbu V, Bartram C, Battista R, Biondi A, Chiamenti A, Cimino G, Ernst P, Frassoni F, Gasparini P, Gentilini I, Glukman E, Grosveld G, Guerrasio A, Hegewich S, Janssen J, Keating A, Lo Coco F, Martiat P, Martinelli G, Mills K, Morgan G, Nadali G, Pelicci PG, Perona G, Pignatti PF, Richard P, Saglio G, Trabetti E, Turco A, Veneri D, Zaccaria A, Zander A, Goldman J. *Clinical value of PCR in diagnosis and follow-up of leukemia and lymphoma: report of the Third Workshop of the Molecular Biology/BMT Study Group*. **Leukemia**5: 101-104, 1991. 2-s2.0-0025829828.
30. Guerrasio A, Martinelli G, Ambrosetti A, Falda M, Paolino F, Rege-Cambrin G, Rosso C, Pignatti PF, Gasparini P, Perona G, Resegotti L, Saglio G. *Different suppression of the Ph1 positive haemopoiesis induced by intensive chemotherapy in lymphoid and myeloid blast crisis of CML*. **Haematologica.**1991 Mar-Apr; 76(2): 126-130. 2-s2.0-0025921623.
31. Gasparini P, Dognini M, Nunes V, Estivill X. *High conservation of sequences involved in cystic fibrosis mutations in five mammalian species*. **Genomics.**1991 Aug; 10(4): 1070-1072. 2-s2.0-0025799904.
32. Nunes V, Gasparini P, Novelli G, Gaona A, Bonizzato A, Sangiuolo F, Balassopoulos A, Gimenez F, Dognini M, Ravnik-Glavac M, Di Marco E, Komel R, Dallapiccola B, Pignatti PF, Loukopoulos D, Casals T, Estivill X. *Analysis of 14 cystic fibrosis mutations in five south European populations*. **Hum Genet.**1991 Oct; 87(6): 737-738.
33. Gasparini P, Bonizzato A, Dognini M, Pignatti PF. *Restriction site generating-polymerase chain reaction (RG-PCR) for the probeless detection of hidden genetic variation: application to the study of some common cystic fibrosis mutations*. **Mol Cell Probes.**1992 Feb; 6(1): 1-7. 2-s2.0-0026004723.
34. Martinelli G, Chiamenti A, Gasparini P, Pignatti PF, Ambrosetti A, Zaccaria A, Buzzi M, Testoni N, Tura S, Guerrasio A., Rosso C, Saglio G. *BCR breakpoint subregions and blast crisis lineage in CML patients*. **Blood.** 1992 Feb 1; 79(3): 838-839. 2-s2.0-0026513141.
35. Nunes V, Bonizzato A, Gaona A, Dognini M, Chillon M, Casals T,

Pignatti PF, Novelli G, Estivill X, Gasparini P. *A frameshift mutation (2869insG) in the second transmembrane domain of the CFTR gene: identification, regional distribution, and clinical presentation*. **Am J Hum Genet.**1992 May; 50(5): 1140-1142.

36. Gasparini P, Borgo G, Mastella G, Bonizzato A, Dognini M, Pignatti PF. *Nine cystic fibrosis patients homozygous for the CFTR nonsense mutation R1162X have mild or moderate lung disease*. **J Med Genet.**1992 Aug; 29(8): 558-562. 2-s2.0-0026693947.
37. Guerrasio A, Martinelli G, Saglio G, Rosso C, Zaccaria A, Rosti G, Testoni N, Ambrosetti A, Izzi T, Sessarego M, Frassoni F, Gasparini P, Chiamenti A, Di Bartolomeo P, Pignatti PF. *Minimal residual disease status in transplanted CML patients: low incidence of PCR positive cases among 48 disease-free subjects who received unmanipulated allogeneic bone marrow transplants*. **Leukemia.**1992; 6: 507-512.
38. Ravnik-Glavac M, Gasparini P, Peterlin B, Strukelj M, Glavac D, Canki-Klain N, Pignatti PF, Komel R. *Cystic Fibrosis gene mutations and linked RFLPs in the Slovenian population*. **Ann Genet.**1992; 35(2): 85-88.
39. Osborne L, Santis G, Schwarz M, Klinger K, Dork T, McIntosh I, Schwartz M, Nunes V, Macek Jr M, Reiss J, Highsmith W, McMahon R, Novelli G, Malik N, Burger J, Anvret M, Wallace M, Williams C, Mathew C, Rozen R, Graham C, Gasparini P, Bal J, Cassiman J, Balassopoulou A, Davidow L, Raskin S, Kalaydjieva L, Kerem B, Richard S, Simon-Bouy B, Super M, Walbrand U, Keston M, Estivill X, Vavrova V, Friedman K, Barton D, Dallapiccola B, Stuhrmann M, Beards F, Hill A, Pignatti PF, Cuppens H, Angelicheva D, Tummler B, Brock D, Casals T, Macek M, Schmidtke J, Magee A, Bonizzato A, De Boeck C, Kuffardjieva A, Hodson M, Knight R. *Incidence and expression of the N1303K mutation of the cystic fibrosis (CFTR) gene*. **Hum Genet.**1992; 89: 653-658.
40. Bonizzato A, Nicolis E, Gasparini P. *Identification of a new splicing mutation (406-1 GC) in the CFTR gene*. **Hum Mutat.**1992; 1(5): 432-433.
41. Gasparini P, Borgato L, Girelli D, Piperno A, Olivieri O, Gottardi E, Roetto A, Dianzani I, Fargion S, Schinaia G, Cappellini MD, Gandini G, Pignatti PF, Fiorelli G, De Sandre G, Camaschella C. *Linkage analysis of 6p21 polymorphic markers and the hereditary hemochromatosis: localization of the gene centromeric to HLA-F*. **Hum Mol Genet.**1993 May; 2(5): 571-576. 2-s2.0-0027159742.
42. Borgo G, Gasparini P, Bonizzato A, Cabrini G, Mastella G, Pignatti PF. *Cystic fibrosis: the delta F508 mutation does not lead to an exceptionally severe phenotype. A cohort study*. **Eur J Pediatr.**1993 Dec; 152(12): 1006-1011.
43. Gasparini P, Bonizzato A, Totaro A, Zelante L. *Identification of a new DNA polymorphism tightly linked to DQ-alpha locus*. **Exp Clin Immunogenet.**1993; 10(3): 176-178. 2-s2.0-0027134524.
44. Gasparini P, Marigo C, Bisceglia L, Nicolis E, Zelante L, Bombieri

C, Borgo G, Pignatti PF, Cabrini G. *Screening of 62 mutations in a cohort of cystic fibrosis patients from north eastern Italy: their incidence and clinical features of defined genotypes*. **Hum Mutat.**1993; 2(5): 389-394. 2-s2.0-0027384235.

45. Camaschella C, Roetto A, De Sandre G, Piperno A, Totaro A, Dianzani I, Gasparini P. *Construction of a genetic map telomeric to HLA-A by microsatellite analysis*. **Mol Cell Probes.**1993 Oct; 7(5): 411-414. 2-s2.0-0027361646.
46. Trabetti E, Galavotti R, Zanini L, Zardini E, Zatti N, Bernardi F, Notarangelo A, Croce A, Pignatti PF, Gasparini P. *The parental origin of hydatidiform moles and blighted ova: molecular probing with hypervariable DNA polymorphisms*. **Mol Cell Probes.**1993 Aug; 7(4): 325-329. 2-s2.0-0027293078.
47. Gasparini P, Grifa A, Origone P, Coviello D, Antonacci R, Rocchi M. *Detection of a Neurofibromatosis type 1 (NF1) homologous sequence by PCR: implications for the diagnosis and screening of genetic diseases*. **Mol Cell Probes.**1993 Oct; 7(5): 415-418.
48. Ramsay M, Williamson R, Estivill X, Wainwright B, Ho MF, Halford S, Kere J, Savilahti E, De la Chapelle A, Schwarz M, Schwartz M, Super M, Farndon P, Harding C, Meredith L, Al-Jador L, Ferec C, Claustres M, Casals T, Nunes V, Gasparini P, Savoia A, Pignatti PF, Novelli G, Gennarelli M, Dallapiccola B, Kalaydjieva L, Scambler P. *Haplotype analysis to determine the position of a mutation among closely linked DNA markers*. **Hum Mol Genet.**1993; 7: 1007-1014. 2-s2.0-0027292096.
49. Piperno A, Sampietro M, Arosio C, Camaschella C, Gasparini P, Fiorelli G. *Genetic Hemochromatosis: molecular study in the HLA-A class I region*. **Molecular Bases of Human Diseases**, Excerpta Medica, Elsevier Science Publishers. 1993, 273-277.
50. The Cystic Fibrosis Genotype-Phenotype Consortium. *Correlation between genotype and phenotype in patients with cystic fibrosis*. **New Engl J Med.**1993; 329: 1308-1312. 2-s2.0-0346678536.
51. Martinelli G, Trabetti E, Zaccaria A, Farabegoli P, Buzzi M, Testoni N, Calori E, Bandini G, Rosti G, Belardinelli A, Gasparini P, Galavotti R, Ambrosetti A, Tura S, Pignatti PF. *In vitro amplification of hypervariable DNA regions for the evaluation of chimerism after allogeneic BMT*. **Bone Marrow Transpl.**1993; 12: 115-120. 2-s2.0-0027283464.
52. Gasparini P, Grifa A, Oggiano N, Fabbrizzi E, Giorgi PL. *Immotile cilia syndrome: a recombinant family at HLA-linked gene locus*. **Am J Med Genet.**1994 feb 15; 49(4): 450-451. 2-s2.0-0028008349.
53. Zelante L, Notarangelo A, Croce AI, Piemontese MR, Gasparini P. *Cytogenetic and molecular analysis of Trisomy 9. Case report and review*. **Ann Genet.** 1994; 37(1): 21-25.
54. Calonge M, Gasparini P, Chillaron J, Chillon M, Gallucci M, Rousaud F, Zelante L, Testar X, Dallapiccola B, Di Silverio F, Barcelò P, Estivill X, Zorzano A, Nunes V, Palacin M. *Cystinuria caused by mutations in the rBAT, a gene involved in the transport of cystine*. **Nat Genet.**1994 Apr; 6(4): 420-425. 2-

s2.0-0028237714.

55. Eoli M, Pandolfo M, Gasparini P, Milanese C, Salmaggi A, Zeviani M. *The myelin basic protein gene is not a major susceptibility locus for multiple sclerosis in Italian patients*. **J Neurol.**1994 Oct; 241(10): 615-619. 2-s2.0-0028147010.
56. Bisceglia L, Grifa A, Zelante L, Gasparini P. *Development of RNA-SSCP protocols for the identification and screening of CFTR mutations: identification of two new mutations*. **Hum Mutat.**1994; 4(2): 136-140. 2-s2.0-0028141430.
57. Totaro A, Grifa A, Roetto A, Zelante L, Camaschella C, Gasparini P. *A new complex polymorphic repeat close to the HLA-A and HLA-E loci*. **Hum Genet.**1994 Nov; 94(5): 578. 2-s2.0-0028046961.
58. Camaschella C, Gasparini P. *Hunting the hemochromatosis gene: progress and problems*. **Eur J Hum Genet.**1994; 2(3): 141-147. 2-s2.0-0028074334.
59. Gasparini P, Grifa A, Savasta S, Merlo I, Bisceglia L, Totaro A, Zelante L. *The motilin gene: subregional localisation, tissue expression, DNA polymorphisms, and exclusion as a candidate gene for the HLA-associated immotile cilia syndrome*. **Hum Genet.**1994 Dec; 94(6): 671-674. 2-s2.0-0027971307.
60. Morral N, Llevadot R, Casals T, Gasparini P, Macek M, Dork T, Estivill X. *Independent origins of cystic fibrosis mutations R334W, R347P, R1162X, and 3849+10KbC-T provide evidence of mutation recurrence in the CFTR gene*. **Am J Hum Genet.**1994 Nov; 55(5): 890-898.
61. The Cystic Fibrosis Genetic Analysis Consortium. *Population variation of Common Cystic Fibrosis Mutations*. **Hum Mutat.**1994; 4: 167-177. 2-s2.0-0028033069.
62. Totaro A, Grifa A, Roetto A, Lunardi C, D'Agruma L, Sbaiz L, Zelante L, De Sandre G, Camaschella C, Gasparini P. *New polymorphisms and markers in the HLA class I region: relevance to the Hereditary Hemochromatosis (HFE)*. **Hum Genet.**1995 Apr; 95(4): 429-434. 2-s2.0-0028904125.
63. Worwood M, Gasparini P, Camaschella C. *International workshop on molecular genetics of haemochromatosis*, held at Villa Feltrinelli, Gargnano (BS), Italy, 25 September 1994. **J Med Genet.** 1995 Apr; 32(4): 320-323. 2-s2.0-0028987225.
64. Bonizzato A, Bisceglia L, Marigo C, Nicolis E, Bombieri C, Castellani C, Borgo G, Zelante L, Mastella G, Cabrini G, Gasparini P, Pignatti PF. *Analysis of the complete coding region of the CFTR gene in a cohort of CF patients from North-Eastern Italy: identification of 90 % of the mutations*. **Hum Genet.**1995; 95: 397-402. 2-s2.0-0028902968.
65. Marigo C, Bombieri C, Bisceglia L, Zelante L, Gasparini P, Pignatti PF. *Homozygosity for a novel splice site mutation (2790-2 A--->G) preceding exon 15 of the CFTR gene in a cystic fibrosis patient of North-East Italian descent*. **Mol Cell Probes.**1995 Apr; 9(2): 139-145.
66. Calonge M, Nadal M, Calvano S, Testar X, Zelante L, Zorzano A, Estivill X, Gasparini P, Palacin M, Nunes V. *Assignment of the*

gene responsible for cystinuria (rBAT) and of markers D2S1119 and D2S177 to 2p16.3 by fluoresecence in situ hybridization. **Hum Genet.** 1995 Jun; 95(6): 633-636.

67. Grifa A, Piemontese MR, Melchionda S, Origone P, Zelante L, Coviello D, Fratta G, Dallapiccola B, Balestrazzi P, Ajmar F, Gasparini P. *Screening of Neurofibromatosis Type I gene: identification of a large deletion and of an intronic variant*. **Clin Genet.**1995; 47: 281-284.
68. Calonge M, Volpini V, Bisceglia L, Rousaud F, de Sanctis L, Beccia E, Zelante L, Testar X, Zorzano A, Estivill X, Gasparini P, Nunes V, Palacin M. *Genetic heterogeneity in cystinuria: the rBAT gene is linked to type I but not to type III cystinuria*. **Proceedings of the National Academy of Sciences**USA. 1995; 92: 9667-9671.
69. Kobayashi M, Kaplan B, Bellah R, Sartore M, Rappaport E, Steele M, Mansfield E, Gasparini P, Surrey S, Fortina P. *Infundibolopelvic stenosis, multicystic kidney and calyectasis in a kindred: clinical observations and genetic analysis*. **Am J Med Genet.**1995 Nov 6; 59(2): 218-224.
70. Giannattasio S, Lattanzio P, Bisceglia L, Gasparini P, Marra E. *Molecular screening of genetic defects with RNA-SSCP analysis: the PKU and cystinuria Model*. **Mol Cell Probes.**1995 Jun; 9(3): 201-205. 2-s2.0-0029068969.
71. Eoli M, Pandolfo M, Amoroso A, Salmaggi A, Zaffaroni M, Gasparini P, DiDonato S, Milanese C, Zeviani M. *Evidence of linkage between susceptibility to multiple sclerosis and HLA-class II loci in italian multiplex families*. **Eur J Hum Genet.**1995; 3(5): 303-311. 2-s2.0-0028810117.
72. Gasparini P, Calonge M, Bisceglia L, Purroy J, Dianzani I, Notarangelo A, Rousaud F, Gallucci M, Testar X, Ponzone A, Estivill X, Zorzano A, Palacin M, Nunes V, Zelante L. *Molecular genetics of cystinuria: identification of four new mutations and seven polymorphisms, and evidence for genetic heterogeneity*. **Am J Hum Genet.**1995 Oct; 57(4): 781-788. 2-s2.0-0029100842.
73. Ciafre' S, Rinaldi M,Gasparini P, Seripa D, Bisceglia L, Zelante L, Farace M, Fazio V. *Stability and functional effectiveness of phosphorothioate modified duplex DNA and synthetic "mini-genes"*. **Nucleic Acids Res.**1995 Oct 25; 23(20): 4134-4142. 2-s2.0-0028784475.
74. Girelli D, Bisceglia L, Olivieri O, De Franceschi L, Zelante L, Corrocher R, Gasparini P. *Molecular basis for the recently described hereditary hyperferritinemia-cataract syndrome: a mutation in the iron-responsive element of ferritin L-subunit gene (the "Verona mutation")*. **Blood.** 1995 Dec 1; 86(11): 4050-4053.
75. Camaschella C, Roetto A, Gasparini P, Piperno A, Fortina P, Surrey S, Rappaport E. *Allelic association of microsatellites of 6p in Italian hemochromatosis patients*. **Hum Genet.** 1996 Apr; 97(4): 476-481. 2-s2.0-0029987959.
76. Gasparini P, D'Agruma L, de Cillis G, Balestrazzi P, Mingarelli R, Zelante L. *Scanning the first part of neurofibromatosis type 1*

gene by RNA-SSCP: identification of three novel mutations and of two new polymorphisms. **Hum Genet.** 1996 Apr; 97(4): 492-495. 2-s2.0-0029880743.

77. Palacin M, Mora C, Chillaron J, Calonge MJ, Estevez R, Torrents D, Testar X, Zorzano A, Nunes V, Purroy J, Gasparini P, Testar X, Zorzano A. *The molecular basis of cystinuria: the role of the rBAT gene*. **Amino Acid Transport.**1996; 11: 225-246. 2-s2.0-10144243969.
78. Totaro A, Rommens J, Grifa A, Lunardi C, Carella M, Huizenga J, Roetto A, Camaschella C, De Sandre G, Gasparini P. *Hereditary hemochromatosis: generation of a transcription map within a refined and extended map of the HLA class I region*. **Genomics.**1996 Feb 1; 31(3): 319-326. 2-s2.0-19244376894.
79. Amati P, Gasparini P, Zlotogora J, Zelante L, Chomel JC, Kitzis A, Kaplan J, Bonneau D. *A gene for premature ovarian failure associated with eyelid malformation maps to chromosome 3q22-q23*. **Am J Hum Genet.**1996 May; 58(5): 1089-1092. 2-s2.0-0029864808.
80. Bisceglia L, Calonge MJ, Dello Strologo L, Rizzoni G, de Sanctis L, Gallucci M, Beccia E, Testar X, Zorzano A, Estivill X, Zelante L, Palacin M, Gasparini P, Nunes V. *Molecular analysis of the cystinuria disease gene: identification of four new mutations, one large deletion, and one polymorphism*. **Hum Genet.**1996 Oct; 98(4): 447-451. 2-s2.0-10144242646.
81. Girelli D, Olivieri O, Gasparini P, Corrocher R. *Molecular basis for the hereditary hyperferritinemia-cataract syndrome*. **Blood.**1996 Jun 1; 87(11): 4912-4913. 2-s2.0-0030011316.
82. Purroy J, Bisceglia L, Calonge MJ, Zelante L, Testar X, Zorzano A, Estivill X, Palacin M, Nunes V, Gasparini P. *Genomic structure and organization of the human rBAT gene (SLC3A1)*. **Genomics.**1996 Oct 15; 37(2): 249-252.
83. Piperno A, Arosio C, Fargion S, Roetto A, Nicoli C, Girelli D, Sbaiz L, Gasparini P, Boari G, Sampietro M, Camaschella C. *The ancestral hemochromatosis haplotypeis is associated with a severe phenotype expression in Italian patients*. **Hepatology.**1996 Jul; 24(1): 43-46.
84. Grifa A, Totaro A, Carella M, D'Ambrosio L, Sbaiz L, Roetto A, Camaschella C, Gasparini P. *Two polymorphic repeats in the candidate region for the haemochromatosis gene.* **Mol Cellular Probes.**1996 Dec; 10(6): 469-470. 2-s2.0-0030477329.
85. Carella M, D'Ambrosio L, Totaro A, Grifa A, Valentino M, Piperno A, Girelli D, Roetto A, Franco B, Gasparini P, Camaschella C. *Mutation analysis of HLA-H gene in Italian hemochromatosis patients*. **Am J Hum Genet.**1997 Apr; 60(4): 828-832. 2-s2.0-16944363480.
86. Bisceglia L, Calonge MJ, Totaro A, Feliubadalò L, Melchionda S, Garcia J, Testar X, Gallucci M, Ponzone A, Zelante L, Zorzano A, Estivill X, Gasparini P, Nunes V, Palacin M. *Localization, by linkage analysis, of the cystinuria Type III gene to chromosome 19q13.1*. **Am J Hum Genet.**1997 Mar; 60(3): 611-616. 2-s2.0-

16944366211.

87. Genuardi M, Gasparini P,Neri G, Zelante L. *Limb-pelvis hypoplasia/aplasia: a discrete entity in the fibuloulnar developmental field complex*. **Am J Med Genet.** 1997 Jan 20; 68(2): 190-194. 2-s2.0-0031044288.
88. Zelante L, Dallapiccola B, Calvano S, Memeo E, Gasparini P,Simi P, Rossi S, Finelli P, Rocchi M, Archidiacono N. *Two mosaic-YY males carrying asymmetric Y chromosomes*. **Clin Genet.**1997 Jan; 51(1): 65-68. 2-s2.0-0031059701.
89. Zelante L, Gasparini P,Castriota Scandeberg A, Dimitri L, Criconia M, Gorlin JR. *Goldenhar complex: a further case with uncommon associated anomalies*. **Am J Med Genet.** 1997 Apr 14; 69(4): 418-421. 2-s2.0-0030951351.
90. Gasparini P, Estivill X, Volpini V, Totaro A, Stanziale P, Della Monica M, Ventruto V, De Benedetto M, Castellvi-Bel S, Govea N, Mila M, Zelante L, Mansfield E, Surrey S, Fortina P. *Linkage of DFNB1 to non-syndromic neurosensory autosomal recessive deafness in Mediterranean families*. **Eur J Hum Genet.**1997 Mar-Apr; 5(2): 83-88. 2-s2.0-12644276408.
91. Pellicano R, Lomuto M, Ciavarella G, Di Giorgio G, Gasparini P. *Fixed drug eruptions with feprazone are linked to HLA-B22*. **J Am Acad Dermatol.**1997 May; 36(5 Pt 1): 782-784. 2-s2.0-0030919878.
92. Totaro A, Grifa A, Carella M, D'Ambrosio L, Valentino M, Roth M, Borot N, Coppin H, Roetto A, Camaschella C, Gasparini P. *Hereditary Hemochromatosis: a HpaI polymorphism within the HLA-H gene*. **Mol Cell Probes.**1997 Jun; 11(3): 229-230. 2-s2.0-0031170905.
93. Piemontese MR, Memeo E, Carella M, Amati P, Chomel JC, Bonneau D, Pilia G, Cao A, Drabkin H, Gemmill R, Rommens J, Zelante L, Gasparini P, Bisceglia L. *A YAC contig spanning the blepharophimosis-ptosis-epicanthus inversus syndrome and propionic acidemia loci*. **Eur J Hum Genet.**1997 May-Jun; 5(3):171-174. 2-s2.0-12644293812.
94. Girelli D, Bisceglia L, Olivieri O, De Franceschi L, Zelante L, Corrocher R, Gasparini P. *Hereditary hyperferritinemia-cataract syndrome caused by a 29-base pair deletion in the iron responsive element of ferritin L-subunit gene*. **Blood.** 1997 Sep 1; 90(5): 2084-2088. 2-s2.0-0030921671.
95. Mansfield E, Vainer M, Harris D,Gasparini P, Estivill X, Surrey S, Fortina P. *Rapid sizing of polymorphic microsatellite markers by capillary array electrophoresis*. **J Chromatogr A**. 1997 Sep 26; 781(1-2): 295-305. 2-s2.0-0030664528.
96. Zelante L, Gasparini P, Estivill X, Melchionda S, D'Agruma L, Govea N, Milà M, Della Monica M, Lutfi J, Shohat M, Mansfield E, Delgrosso K, Rappaport E, Surrey S, Fortina P. *Connexin26 mutations associated with the most common form of non-syndromic neurosensory autosomal recessive deafness (DFNB1) in Mediterraneans*. **Hum Mol Genet.**1997 Sep; 6(9): 1605-1609.
97. Roetto A, Cicilano M, Gottardi E, Totaro A, Carella M, Gasparini P, Camaschella C. *A frequent polymorphism in the 5' region of the*

BCMA gene. **Mol Cell Probes**. 1997 Aug; 11(4): 311-312. 2-s2.0-0030881175.

98. Calvano S, Memeo E, Piemontese MR, Melchionda S, Bisceglia L, Gasparini P, Zelante L. *Detection of dystrophin deletion carriers using FISH analysis*. **Clin Genet.**1997 Jul; 52(1): 17-22. 2-s2.0-0030741377.
99. Iolascon A, Miraglia del Giudice E, Perrotta S, Granatiero M, Zelante L, Gasparini P. *Exclusion of three candidate genes as determinants of congenital dyserythropoietic anemia type II (CDA-II)*. **Blood**. 1997 Nov 15; 90(10): 4197-4200.
100. Gasparini P, Miraglia del Giudice E, Delaunay J, Totaro A, Granatiero M, Melchionda S, Zelante L, Iolascon A. *Localization of the congenital dyserythropoietic anemia II locus to chromosome 20q11.2 by genomewide search*. **Am J Hum Genet.**1997 Nov; 61(5): 1112-1116. 2-s2.0-16944367512.
101. Roetto A, Sbaiz L, Bosio S, Piperno A, Fargion S, Carella M, Totaro A, Grifa A, Gasparini P, Camaschella C. *A recombination event close to HFE gene in hereditary hemochromatosis*. **Ann Genet.**1997; 40(3): 150-153. 2-s2.0-0030685088.
102. Camaschella C, Roetto A, Cicilano M, Pasquero P, Bosio S, Gubetta L, Di Vito F, Girelli D, Totaro A, Carella M, Grifa A, Gasparini P. *Juvenile and adult hemochromatosis are distinct genetic disorders*. **Eur J Hum Genet.**1997 Nov-Dec; 5(6): 371-375. 2-s2.0-13144259692.
103. Gasparini P,Calvano S, Memeo E, Bisceglia L, Zelante L. *Assignment of ferritin L gene (FTL) to human chromosome band 19q13.3 by in situ hybridization*. **Ann Genet.**1997; 40(4): 227-228.
104. Rizzoni G, Dello Strologo L, Gasparini P, Zelante L, Bisceglia L, Nunes V, Palacin M. *La cystinurie-lysinurie, progres recents*. **Annales de Pediatrie**. 1997; 44: 688-691.
105. Italian Collaborative Group on CF. *Genetic History of cystic fibrosis mutations in Italy. 1. regional distribution*. **Ann Hum Genet.**1997; 61: 411-424.
106. Estivill X, Fortina P, Surrey S, Rabionet R, Melchionda S, D'Agruma L, Mansfield E, Rappaport E, Govea N, Milà M, Zelante L, Gasparini P. *Connexin-26 mutations in sporadic and inherited sensorineural deafness*. **Lancet.** 1998 Feb 7; 351(9100): 394-398. 2-s2.0-0032492217.
107. Annese V, Piepoli A, Andriulli A, Napoletano G, Bisceglia L, Zelante L, Gasparini P. *Polymorphism of motilin gene in patients with Chron's disease*. **Dig Dis Sci**. 1998 Apr; 43(4): 715-719.
108. Totano A, Roetto A, Rommens J, Grifa A, Carella M, d'Agruma L, Valentino M, D'Ambrosio L, Cicalano M, Camaschella C, Franco B, Gasparini P. *Generation of a transcription map of a 1 Mbase region containing the HFE gene (6p22)*. **Eur J Hum Genet.**1998 Mar-Apr; 6(2): 105-113.
109. Stephens C, Reich D, Goldstein D, Shin H, Smith M, Carryngton M, Winkler C, Huttley G, Allikmets R, Schriml L, Gerrard B, Malasky M, Ramos M, Merlot S, Tzetis M, Oddoux C, di Giovine F, Nasioulas G, Chandler D, Aseev M, Hanson M,

Kalaydjieva L, Glavac D, Gasparini P,Kanavakis E, Claustres M, Kambouris M, Ostrer H, Duff G, Baranov V, Sibul H, Metspalu A, Goldman D, Schmidtke J, Estivill X, O'Brien S, Dean M. *Dating the origin of the CCR5-delta32 AIDS resistance allele by the coalescence of haplotypes*. **Am J Hum Genet.**1998 Jun; 62(6): 1507-15.

110. Tiranti V, D'Agruma L, Pareyson D, Mora M, Carrara F, Zelante L, Gasparini P, Zeviani M. *A novel mutation in the mitochondrial tRNA(Val)gene associated with a complex neurological presentation*. **Ann Neurol.** 1998 Jan; 43(1): 98-101.
111. Melchionda S, Seri M, Carella M, Piemontese MR, Zhang XX, Zelante L, Romeo G, Gasparini P**.** *Linkage analysis in two large Italian pedigrees affected with nail patella syndrome*. **Eur J Hum Genet.**1998 Jul-Aug; 6(4):345-349. 2-s2.0-0031877022.
112. Grifa A, Totaro A, Rommens JM, Carella M, Roetto A, Borgato L, Zelante L, Gasparini P. *GABA (gamma-amino-butyric acid) neurotransmission: identification and fine mapping of the human GABAB receptor gene*. **Biochem Biophys Res Commun.**1998 Sep 18; 250(2): 240-245.
113. Totaro A, Grifa A, Carella M, Rommens JM, Valentino MA, Roetto A, Zelante L, Gasparini P. *Cloning of a new gene (FB19) within HLA class I region*. **Biochem Biophys Res Commun.**1998 Sep 29; 250(3): 555-557.
114. Iolascon A, De Mattia D, Perrotta S, Carella M, Gasparini P, Lambertenghi Deliliers G. *Genetic heterogeneity of congenital dyserythropoietic anemia type II.* **Blood.**1998 Oct 1; 92(7): 2593-2594. 2-s2.0-0032189079.
115. Bisceglia L, d'Ambrosio L, Piemontese MR, Carella M, Amati P, Bonneau D, Pilia G, Gasparini P, Zelante L. *Cellular Retinol Binding Protein 1 (RBP1): a frequent polymorphism, refined map position and exclusion as the Blepharophimosis Ptosis Epicanthus inversus Syndrome gene*. **Mol Cell Probes.**1998 Aug; 12(4): 255-258. 2-s2.0-0032142771.
116. Tiranti V, Hoertnagel K, Carrozzo R, Galimberti C, Munaro M, Granatiero M, Zelante L, Gasparini P, Marzella R, Rocchi M, Bayona-Bafaluy MP, Enriquez JA, Uziel G, Bertini E, Dionisi-Vici C, Franco B, Meitinger T, Zeviani M. *Mutations of SURF-1 in Leigh disease associated with cytochrome c oxidase deficiency*. **Am J Hum Genet.**1998 Dec; 63(6): 1609-1621. 2-s2.0-0032470811.
117. Gasparini P, De Fazio A, Croce AI, Stanziale P, Zelante L. *Usher syndrome type III (USH3) linked to chromosome 3q in an Italian family*. **J Med Genet.**1998 Aug; 35(8): 666-667.
118. Carella M, Stewart G, Ajetunmobi JF, Perrotta S, Grootenboer S, Tchernia G, Delaunay J, Totaro A, Zelante L, Gasparini P, Iolascon A. *Genomewide search for dehydrated hereditary stomatocytosis (hereditary xerocytosis): mapping of locus to chromosome 16 (16q23-qter).* **Am J Hum Genet.**1998 sep; 63(3): 810-816.
119. Piperno A, Sampietro M, Pietrangelo A, Arosio C, Lupica L, Montosi G, Vergani A, Fraquelli M, Girelli D, Pasquero P, Roetto

A, Gasparini P, Fargion S, Conte D, Camaschella C. *Heterogeneity of hemochromatosis in Italy*. **Gastroenterology**. 1998 May; 114(5): 996-1002. 2-s2.0-0031957721.

120. Santorelli FM, Bertini E, Petruzzella V, Di Capua M, Calvieri S, Gasparini P, Zeviani M. *A novel insertion mutation (A169i) in the CLN1 gene is associated with infantile neuronal ceroid lipofuscinosis in an Italian patient*. **Biochem Biophys Res Commun.**1998 Apr 17; 245(2): 519-522.
121. Lauteala T, Mykkanen J, Sperandeo MP, Gasparini P, Savontaus ML, Simell O, Andria G, Sebastio G, Aula P. *Genetic homogeneity of lysinuric protein intolerance*. **Eur J Hum Genet.**1998 Nov-Dec; 6(6): 612-615. 2-s2.0-0032420658.
122. Delaunay J, Grootenboer S, Schischmanoff PO, Cynober T, Tchernia G, Dommergues JP, Bost M, Stewart G, Perrota S, Carella M, Gasparini P, Iolascon A. *Dehydrated hereditary stomatocytosis revisited*. **Cellular and Molecular Biology Letters.**1998; 3: 435-441. 2-s2.0-0032446666.
123. Roetto A, Totaro A, Cazzola M, Cicilano M, Bosio S, D'Ascola G, Carella M, Zelante L, Kelly AT, Cox TM, Gasparini P, Camaschella C. *Juvenile hemochromatosis locus maps to chromosome 1q*. **Am J Hum Genet.**1999 May; 64(5): 1388-1393. 2-s2.0-0033358675.
124. Castriota-Scanderberg A, Zelante L, Masala S, Gasparini P, Lachman R. *Acrodysplasia, severe ossification abnormalities with short stature, and fibular hypoplasia*. **Am J Med Genet.**1999 May 7; 84(1): 68-73. 2-s2.0-0033531975.
125. Iolascon A, Stewart GW, Ajetunmobi JF, Perrotta S, Delaunay J, Carella M, Zelante L, Gasparini P. *Familial pseudohyperkalemia maps to the same locus as dehydrated hereditary stomatocytosis (hereditary xerocytosis)*. **Blood.**1999 May 1; 93(9): 3120-3123.
126. Gasparini P, Estivill X, Fortina P. *Vestibular and hearing loss in genetic and metabolic disorders*. **Curr Opin Neurol.**1999 Feb; 12(1): 35-39. 2-s2.0-0033015992.
127. Zelante L, Candela MA, Savoia A, Gasparini P. *Confirmation of Kapur-Toriello syndrome in an Italian patient*. **Clin Dysmorphol.**1999 Apr, 8(2): 151-153. 2-s2.0-0032929184.
128. Seri M, Melchionda S, Dreyer S, Marini M, Carella M, Cusano R, Piemontese MR, Caroli F, Silengo M, Zelante L, Romeo G, Ravazzolo R, Gasparini P, Lee B. *Identification of LMX1B gene point mutations in Italian patients affected with Nail-Patella syndrome*. **Int J Mol Med.**1999 Sep; 4(3): 285-290. 2-s2.0-0033192624.
129. Gasparini P, Arbustini E, Restagno G, Zelante L, Stanziale P, Gatta L, Sbaiz L, Sedita AM, Banchieri N, Sapone L, Fiorucci G, Brinson E, Shilse E, Rappaport E, Fortina P. *Analysis of 31 CFTR mutations by polymerase chain reaction/oligonucleotide ligation assay in a pilot screening of 4476 newborns for cystic fibrosis*. **J Med Screen.**1999; 6(2): 67-69. 2-s2.0-0032993421.
130. Annese V, Latiano A, Bovio P, Forabosco P, Piepoli A, Lombardi G, Andreoli A, Astegiano M, Gionchetti P, Riegler G, Sturniolo G, Clementi M, Rappaport E, Fortina P, Devoto

M, Gasparini P, Andriulli A. *Genetic analysis in Italian families with inflammatory bowel disease supports linkage to the IBD1 locus -- a GISC study.* **Eur J Hum Genet.**1999 Jul; 7(5): 567-573. 2-s2.0-0032784580.

131. Grifa A, Wagner C, D'Ambrosio L, Melchionda S, Bernardi F, Lopez-Bigas N, Rabionet R, Arbones M, Della Monica M, Estivill X, Zelante L, Lang F, Gasparini P. *Mutations in GJB6 cause nonsyndromic autosomal dominant deafness at DFNA3 locus*. **Nat Genet.**1999 Sep; 23(1): 16-18. 2-s2.0-0032846415.
132. International Cystinuria Consortium (Group C: Gasparini P, Bisceglia L, Beccia E, Gallucci M, de Sanctis L, Ponzone A, Rizzoni GF, Zelante L). *Non-type I cystinuria caused by mutations in SLC7A9, encoding a subunit (bo,+AT) of rBAT*. **Nat Genet.**1999 Sep; 23(1): 52-57.
133. Feliubadalo L, Bisceglia L, Font M, Dello Strologo L, Beccia E, Arslan-Kirchner M, Steinmann B, Zelante L, Estivill X, Zorzano A, Palacin M, Gasparini P, Nunes V. *Recombinant families locate the gene for non-type I cystinuria between markers C13 and D19S587 on chromosome 19q13.1*. **Genomics.**1999 Sep 15; 60(3): 362-365. 2-s2.0-0033567939.
134. Pietrangelo A, Montosi G, Totaro A, Garuti C, Conte D, Cassanelli S, Fraquelli M, Sardini C, Vasta F, Gasparini P. *Hereditary hemochromatosis in adults without pathogenic mutations in the hemochromatosis gene*. **N Engl J Med.**1999 Sep 2; 341(10): 725-732. 2-s2.0-0033517341.
135. Lopez-Bigas N, Rabionet R, de Cid R, Govea N, Gasparini P, Zelante L, Arbones ML, Estivill X. *Splice-site mutation in the PDS gene may result in intrafamilial variability for deafness in Pendred syndrome*. **Hum Mutat.**1999; 14(6): 520-526. 2-s2.0-0344577853.
136. Merryweather-Clarke AT, Pointon JJ, Shearman JD, Robson KJ, Jouanolle AM, Mosser A, David V, Le Gall JY, Halsall DJ, Elsey TS, Kelly A, Cox TM, Clare M, Bomford A, Vandwalle JL, Rochette J, Borot N, Coppin H, Roth MP, Ryan E, Crowe J, Totaro A, Gasparini P, Roetto A, Camaschella C, Darke C, Wallace DF, Saeb-Parsy K, Dooley JS, Worwood M, Walker AP; The European Haemochromatosis Consortium. *Polymorphism in intron 4 of HFE does not compromise haemochromatosis mutation results*. **Nat Genet.**1999 Nov; 23(3): 271. 2-s2.0-0008266874.
137. Beauchamp-Nicoud A, Schischmanoff PO, Alloisio N, Boivin P, Parsons SF, Mielot F, Tchernia G, Legrand A, Miraglia Del Giudice E, Gasparini P, Iolascon A, Wickramasinghe SN, Delaunay J. *Suppression of CDA II expression in a homozygote*. **Br J Haematol.**1999 Sep; 106(4): 948-953. 2-s2.0-0032881056.
138. Tiranti V, Lamantea F, Uziel G, Zeviani M, Gasparini P, Marzella R, Rocchi M, Fried M. *Leigh syndrome transmitted by uniparental disomy of chromosome 9*. **J Med Genet.**1999 Dec; 36(12): 927-928. 2-s2.0-0033462139.
139. Robson K, Merryweather-Clarke A, Pointon J, Shearman J, Halsall D, Kelly A, Cox T, Rosenberg W, Howell M, Eccles D, Patch C, Fowler A, Wallace D, Camschella C, Roetto A, Zecchina G, De

Gobbi M, Gasparini P, Cadet E, Vandwalle J, Capron D, Rochette J, Borot N, Demangel C, Dery R, Vinel J. *Diagnosis and management of hemochromatosis since the discovery of the HFE gene: a European experience*. **Brit J Haematol.**2000; 108: 31-39. 2-s2.0-0033963016.

140. Bertoldo F, D'Agruma L, Furlan F, Colapietro F, Lorenzi MT, Maiorano N, Iolascon A, Zelante L, Locascio V, Gasparini P. *Transforming growth factor-beta1 gene polymorphism, bone turnover, and bose mass in Italian postmenopausal women*. **J Bone Miner Res.**2000 Apr; 15(4): 634-639.
141. Purroy J, Bisceglia L, Jaeken J, Gasparini P, Palacin M, Nunes V. *Detection of two novel large deletions in SLC3A1 by semi-quantitative fluorescent multiplex PCR*. **Hum Mutat.**2000; 15(4): 373-379. 2-s2.0-0034127946.
142. Gasparini P, Rabionet R, Barbujani G, Melchionda S, Petersen M, Brondum-Nielsen K, Metspalu A, Oitmaa E, Pisano M, Fortina P, Zelante L, Estivill X. *High carrier frequency of the 35delG deafness mutation in European populations. Genetic Analysis Consortium of GJB2 35delG*. **Eur J Hum Genet.** 2000 Jan; 8(1):19-23.
143. Camaschella C, Roetto A, Calì A, De Gobbi M, Garozzo G, Carella M, Majorano N, Totaro A, Gasparini P. *The gene TFR2 is mutated in a new type of haemochromatosis mapping to 7q22*. **Nat Genet.**2000 May; 25(1): 14-15. 2-s2.0-0034022636.
144. Angius A, Spinelli P, Ghilotti G, Casu G, Sole G, Loi A, Totaro A, Zelante L, Gasparini P, Orzalesi N, Pirastu M, Bonomi L. *Myocilin Gln368stop mutation and advanced age as risk factors for late-onset primary open-angle glaucoma*. **Arch Ophthalmol.**2000 May; 118(5): 674-9. 2-s2.0-19244382595.
145. Iolascon A, Servedio V, Carbone R, Totaro A, Carella M, Perrotta S, Wickramasinghe SN, Delaunay J, Heimpel H, Gasparini P. *Geographic distribution of CDA-II: did a founder effect operate in Southern Italy?* **Haematologica.** 2000 May; 85(5): 470-474.
146. Rabionet R, Zelante L, Lopez-Bigas N, D'Agruma L, Melchionda S, Restagno G, Arbones ML, Gasparini P, Estivill X. *Molecular basis of childhood deafness resulting from mutations in the GJB2 (connexin 26) gene.* **Hum Genet.** 2000 Jan; 106(1): 40-44.
147. Perrotta S, Cappellini MD, Bertoldo F, Servedio V, Iolascon G, D'Agruma L, Gasparini P,Siciliani MC, Iolascon A. *Osteoporosis in beta-thalassaemia major patients: analysis of the genetic background*. **Br J Haematol.**2000 Nov; 111(2): 461-466. 2-s2.0-0034531861.
148. Grootenboer S, Schischmanoff PO, Laurendeau I, Cynober T, Tchernia G, Dommergues JP, Dhermy D, Bost M, Varet B, Snyder M, Ballas SK, Ducot B, Babron MC, Stewart GW, Gasparini P, Iolascon A, Delaunay. *Pleiotropic syndrome of dehydrated hereditary stomatocytosis, pseudohyperkalemia, and perinatal edema maps to 16q23-q24.* **Blood.**2000 Oct 1; 96(7): 2599-2605. 2-s2.0-0034307365.

149. Rabionet R, Gasparini P, Estivill X. *Molecular genetics of hearing impairment due to mutations in gap junction genes encoding beta connexins.* **Hum Mutat.**2000 Sep; 16(3):190-202. Review. 2-s2.0-0033850250.

150. Roetto A, Alberti F, Daraio F, Cali A, Cazzola M, Totaro A, Gasparini P, Camaschella C. *Exclusion of ZIRTL as candidate gene of juvenile hemochromatosis and refinement of the critical interval on 1q21*. **Blood Cells Mol Dis.**2000 Jun; 26(3): 205-210. 2-s2.0-0033812756.

151. Zelante L, Gasparini P, Savoia A, Lomuto M, Pellicano R. *A new case of acromegaloid facial appearance (AFA) syndrome with an expanded phenotype*. **Clin Dysmorphol**. 2000 Jul; 9(3): 221-222.

152. Crisponi L, Deiana M, Loi A, Chiappe F, Uda M, Amati P, Bisceglia L, Zelante L, Nagaraja R, Porcu S, Serafina Ristaldi M, Marzella R, Rocchi M, Nicolino M, Lienhardt-Roussie A, Nivelon A, Verloes A, Schlessinger D, Gasparini P, Bonneau D, Cao A, Pilia G. *The putative forkhead transcription factor FOXL2 is mutated in blepharophimosis/ptosis/epicanthus inversus syndrome*. **Nat Genet.** 2001 Feb; 27(2): 159-166. 2-s2.0-0035131812.

153. Font M, Feliubadalo L, Estivill X, Nunes V, Golomb E, Kreiss Y, Pras E, Bisceglia L, d'Adamo AP, Zelante L, Gasparini P, Bassi MT, George Jr AL, Manzoni M, Riboni M, Ballabio A, Borsani G, Reig N, Fernandez E, Zorzano A, Bertran J, Palacin. *Functional analysis of mutations in SLC7A9, and genotype-phenotype correlation in non-Type I cystinuria*. **Hum Mol Genet.** 2001 Feb 15; 10(4): 305-316. 2-s2.0-0035865031.

154. Bisceglia L, Purroy J, Jimenez-Vidal M, D'Adamo AP, Rousaud F, Beccia E, Penza R, Rizzoni G, Gallucci M, Palacin M, Gasparini P, Nunes V, Zelante L. *Cystinuria type I: Identification of eight new mutations in SLC3A1*. **Kidney Int.**2001 Apr; 59(4): 1250-1256. 2-s2.0-0035072947.

155. Persico AM, D'Agruma L, Maiorano N, Totaro A, Militerni R, Bravaccio C, Wassink TH, Schneider C, Melmed R, Trillo S, Montecchi F, Palermo M, Pascucci T, Puglisi-Allegra S, Reichelt KL, Conciatori M, Marino R, Quattrocchi CC, Baldi A, Zelante L, Gasparini P, Keller F. *Reelin gene alleles and haplotypes as a factor predisposing to autistic disorder*. **Mol Psychiatry.**2001 Mar; 6(2): 150-159. 2-s2.0-17744393442.

156. Roetto A, Totaro A, Piperno A, Piga A, Longo F, Garozzo G, Cali A, De Gobbi M, Gasparini P, Camaschella C. *New mutations inactivating transferrin receptor 2 in hemochromatosis type 3*. **Blood.**2001 May 1; 97(9): 2555-2560. 2-s2.0-0035353167.

157. Lopez-Bigas N, Olive M, Rabionet R, Ben-David O, Martinez-Matos JA, Bravo O, Banchs I, Volpini V, Gasparini P, Avraham KB, Ferrer I, Arbones ML, Estivill X. *Connexin 31 (GJB3) is expressed in the peripheral and auditory nerves and causes neuropathy and hearing impairment*. **Hum Mol Genet.**2001 Apr 15; 10(9): 947-952. 2-s2.0-0035871208.

158. Greco L, Babron MC, Corazza GR, Percopo S, Sica R, Clot F, Fulchignoni-Lataud MC, Zavattari P, Momigliano-Richiardi P,

Casari G, Gasparini P, Tosi R, Mantovani V, De Virgiliis S, Iacono G, D'Alfonso A, Selinger-Leneman H, Lemainque A, Serre JL, Clerget-Darpoux F. *Existence of a genetic risk factor on chromosome 5q in Italian coeliac disease families*. **Ann Hum Genet.**2001 Jan; 65(Pt 1): 35-41. 2-s2.0-0034744360.

159. Nicolao P, Carella M, Giometto B, Tavolato B, Cattin R, Giovannucci-Uzielli ML, Vacca M, Regione FD, Piva S, Bortoluzzi S, Gasparini P. *DHPLC analysis of the MECP2 gene in Italian Rett patients*. **Hum Mutat.**2001 Aug; 18(2): 132-40. 2-s2.0-0034910716.

160. Melchionda S, Ahituv N, Bisceglia L, Sobe T, Glaser F, Rabionet R, Arbones ML, Notarangelo A, Di Iorio E, Carella M, Zelante L, Estivill X, Avraham KB, Gasparini P. *MYO6, the human homologue of the gene responsible for deafness in Snell's waltzer mice, is mutated in autosomal dominant nonsyndromic hearing loss*. **Am J Hum Genet.**2001 Sep; 69(3): 635-640. 2-s2.0-0034887805.

161. Montosi G, Donovan A, Totaro A, Garuti C, Pignatti E, Cassanelli S, Trenor CC, Gasparini P, Andrews NC, Pietrangelo A. *Autosomal-dominant hemochromatosis is associated with a mutation in the ferroportin (SLC11A3) gene*. **J Clin Invest.**2001 Aug; 108(4): 619-623.

162. Giunti L, Pelagatti S, Lazzerini V, Guarducci S, Lapi E, Coviello S, Cecconi A, Ombroni L, Andreucci E, Sani I, Brusaferri A, Lasagni A, Ricotti G, Giometto B, Nicolao P, Gasparini P, Granatiero M, Uzielli ML. *Spectrum and distribution of MECP2 mutations in 64 Italian Rett syndrome girls: tentative genotype/phenotype correlation*. **Brain Dev.**2001 Dec; 23 Suppl 1: S242-5. 2-s2.0-0035192501.

163. Joensuu T, Hamalainen R, Yuan B, Johnson C, Tegelberg S, Gasparini P, Zelante L, Pirvola U, Pakarinen L, Lehesjoki AE, de la Chapelle A, Sankila EM. *Mutations in a novel gene with transmembrane domains underlie Usher syndrome type 3*. **Am J Hum Genet.** 2001 Oct; 69(4): 673-684. Erratum in: Am J Hum Genet 2001 Nov; 69(5): 1160. 2-s2.0-0034835042.

164. Lopez-Bigas N, Melchionda S, de Cid R, Grifa A, Zelante L, Govea N, Arbones ML, Gasparini P, Estivill X. *Identification of five new mutations of PDS/SLC26A4 in Mediterranean families with hearing impairment*. **Hum Mutat.**2001 Dec; ;18(6): 548. Corrected and republished in: Hum Mutat. 2002 Jul; 20(1): 77-78. 2-s2.0-0035650797.

165. Wattenhofer M, Di Iorio MV, Rabionet R, Dougherty L, Pampanos A, Schwede T, Montserrat-Sentis B, Arbones ML, Iliades T, Pasquadibisceglie A, D'Amelio M, Alwan S, Rossier C, Dahl HH, Petersen MB, Estivill X, Gasparini P, Scott HS, Antonarakis SE. *Mutations in the TMPRSS3 gene are a rare cause of childhood nonsyndromic deafness in Caucasian patients*. **J Mol Med.**2002 feb; 80(2): 124-131. 2-s2.0-0036176041.

166. Lopez-Bigas N, Melchionda S, Gasparini P, Borragan A, Arbones ML, Estivill X. *A common frameshift mutation and other variants in GJB4 (connexin 30.3): Analysis of hearing impairment*

families. **Hum Mutat.**2002 Apr; 19(4): 458.

167. De Gobbi M, Roetto A, Piperno A, Mariani R, Alberti F, Papanikolaou G, Politou M, Lockitch G, Girelli D, Fargion S, Cox TM, Gasparini P, Cazzola M, Camaschella C. *Natural history of juvenile haemochromatosis*. **Br J Haematol.**2002 Jun; 117(4): 973-979. 2-s2.0-0036075097.

168. D'Andrea P, Veronesi V, Bicego M, Melchionda S, Zelante L, Di Iorio E, Bruzzone R, Gasparini P. *Hearing loss: frequency and functional studies of the most common connexin26 alleles*. **Biochem Biophys Res Commun.**2002 Aug 23; 296(3): 685-691. 2-s2.0-0036384216.

169. Dello Strologo L, Pras E, Pontesilli C, Beccia E, Ricci-Barbini V, de Sanctis L, Ponzone A, Gallucci M, Bisceglia L, Zelante L, Jimenez-Vidal M, Font M, Zorzano A, Rousaud F, Nunes V, Gasparini P, Palacin M, Rizzoni G. *Comparison between SLC3A1 and SLC7A9 cystinuria patients and carriers: a need for a new classification*. **J Am Soc Nephrol.**2002 Oct; 13(10): 2547-2553. 2-s2.0-18544381349.

170. Carella M, Stanziale P, Gasparini P. *Development of a rapid protocol for mutation detection by microarray analysis*. **Minerva Biotecnol.**2002; 14: 237-240. 2-s2.0-0036975785.

171. Piluso G, Carella M, D'Avanzo M, Santinelli R, Carrano EM, D'Avanzo A, D'Adamo P, Gasparini P, Nigro V. *Genetic heterogeneity of FG syndrome: a fourth locus (FGS4) maps to Xp11.4-p11.3 in an Italian family*. **Hum Genet.**2003 Feb; 112(2): 124-130.

172. D'Adamo P, Pinna M, Capobianco S, Cesarani A, D'Eustacchio A, Fogu P, Carella M, Seri M, Gasparini P. *A novel autosomal dominant non-syndromic deafness locus (DFNA48) maps to 12q13-q14 in a large Italian family*. **Hum Genet.**2003 Mar; 112(3): 319-320.

173. D'Adamo P, Donaudy F, D'Eustacchio A, Di Iorio E, Melchionda S, Gasparini P. *A new locus (DFNA47) for autosomal dominant non-syndromic inherited hearing loss maps to 9p21-22 in a large Italian family*. **Eur J Hum Genet.** 2003 Feb; 11(2): 121-124.

174. Lanzara C, Ficarella R, Totano A, Chen X, Roberto R, Perrotta S, Lasalandra C, Gasparini P, Iolascon A, Carella M. *Congenital dyserythropoietic anemia type II: exclusion of seven candidate genes*. **Blood Cells Mol Dis.** 2003 Jan-Feb; 30(1): 22-29. 2-s2.0-12244284608.

175. Donaudy F, Ferrara A, Esposito L, Hertzano R, Ben-David O, Bell RE, Melchionda S, Zelante L, Avraham KB, Gasparini P. *Multiple mutations of MYO1A, a cochlear-expressed gene, in sensorineural hearing loss*. **Am J Hum Genet.** 2003 Jun; 72(6): 1571-1577. 2-s2.0-0037677643.

176. Rivard SR, Lanzara C, Grimard D, Carella M, Simard H, Ficarella R, Simard R, D'Adamo AP, De Braekeleer M, Gasparini P. *Autosomal dominant reticuloendotelial iron overload (HFE type 4) due to a new missense mutation in the FERROPORTIN 1 gene (SLC11A3) in a large French-Canadian family*. **Haematologica.**2003 Jul; 88(7): 824-826.

177. Rivard SR, Lanzara C, Grimard D, Carella M, Simard H, Ficarella R, Simard R, D'Adamo AP, Férec C, Camaschella C, Mura C, Roetto A, De Braekeleer M, Bechner L, Gasparini P. *Juvenile hemochromatosis locus maps to chromosome 1q in a French Canadian population*. **Eur J Hum Genet**. 2003 Aug; 11(8): 585-589. 2-s2.0-10744230412.

178. Di Leva F, D'Adamo AP, Strollo L, Auletta G, Caravelli A, Carella M, Mari F, Livi W, Renieri A, Gasparini P, D'Urso M, Marciano E, Franze A. *Otosclerosis: exclusion of linkage to the OTSC1 and OTSC2 loci in four Italian families*. **Int J Audiol**. 2003 Dec; 42(8): 475-80. 2-s2.0-10744230545.

179. Del Castillo I, Moreno-Pelayo MA, Del Castillo FJ, Brownstein Z, Marlin S, Adina Q, Cockburn DJ, Pandya A, Siemering KR, Chamberlin GP, Ballana E, Wuyts W, Maciel-Guerra AT, Alvarez A, Villamar M, Shohat M, Abeliovich D, Dahl HH, Estivill X, Gasparini P, Hutchin T, Nance WE, Sartorato EL, Smith RJ, Van Camp G, Avraham KB, Petit C, Moreno F. *Prevalence and evolutionary origins of the del(GJB6-D13S1830) mutation in the DFNB1 locus in hearing-impaired subjects: a multicenter study*. **Am J Hum Genet**. 2003 Dec; 73(6): 1452-1458. 2-s2.0-9144251659.

180. Carella M, Volinia S, Gasparini P. *Nanotechnologies and microchips in genetic diseases*. **J Nephrol**. 2003 Jul-Aug; 16(4): 597-602. Review. 2-s2.0-1642435643.

181. Criscuolo C, Banfi S, Orio M, Gasparini P, Monticelli A, Scarano V, Santorelli FM, Perretti A, Santoro L, De Michele G, Filla A. *A novel mutation in SACS gene in a family from southern Italy*. **Neurology**. 2004 Jan 3; 62(1): 100-102. 2-s2.0-9144241657.

182. Tiranti V, D'Adamo P, Briem E, Ferrari G, Mineri R, Lamantea E, Mandel H,Balestri P, Garcia-Silva MT, Vollmer B, Rinaldo P, Hahn SH, Leonard J, Rahman S,Dionisi-Vici C, Garavaglia B, Gasparini P, Zeviani M. *Ethylmalonic encephalopathy is caused by mutations in ETHE1, a gene encoding a mitochondrial matrix protein*. **Am J Hum Genet**. 2004 Feb; 74(2): 239-252. 2-s2.0-10744232283.

183. Muglia M, Criscuolo C, Magariello A, De Michele G, Scarano V, D'Adamo P,Ambrosio G, Gabriele AL, Patitucci A, Mazzei R, Conforti FL, Sprovieri T,Morgante L, Epifanio A, La Spina P, Valentino P, Gasparini P, Filla A, QuattroneA. *Narrowing of the critical region in autosomal recessive spastic paraplegia linked to the SPG5 locus*. **Neurogenetics**. 2004 Feb; 5(1): 49-54. 2-s2.0-10744225170.

184. Roetto A, Daraio F, Porporato P, Caruso R, Cox TM, Cazzola M, Gasparini P, Piperno A, Camaschella C. *Screening hepcidin for mutations in juvenile hemochromatosis: identification of a new mutation (C70R)*. **Blood**. 2004 Mar 15; 103(6): 2407-2409.

185. Donaudy F, Snoeckx R, Pfister M, Zenner HP, Blin N, Di Stazio M, Ferrara A, Lanzara C, Ficarella R, Declau F, Pusch CM, Nurnberg P, Melchionda S, Zelante L, Ballana E, Estivill X, Van Camp G, Gasparini P, Savoia A. *Nonmuscle myosin heavy-chain gene MYH14 is expressed in cochlea and mutated in patients*

affected by autosomal dominant hearing impairment (DFNA4). **Am J Hum Genet**. 2004 Apr; 74(4): 770-776.

186. Lanzara C, Roetto A, Daraio F, Rivard S, Ficarella R, Simard H, Cox T, Cazzola M, Piperno A, Gimenez-Roqueplo AP, Grammatico P, Volinia S, Gasparini P, Camaschella C. *Spectrum of hemojuvelin gene mutations in 1q-linked juvenile hemochromatosis*. **Blood**. 2004 Jun 1; 103(11): 4317-4321. 2-s2.0-2542468736.
187. Gasparini P, Camaschella C. *Hereditary hemochromatosis: is the gene race over?* **Eur J Hum Genet**. 2004 May; 12(5): 341-342.
188. Buniello A, Montanaro D, Volinia S, Gasparini P, Marigo V. *An expression atlas of connexin genes in the mouse*. **Genomics**. 2004 May; 83(5): 812-820. 2-s2.0-1842665165.
189. Volinia S, Evangelisti R, Francioso F, Arcelli D, Carella M, Gasparini P. *GOAL: automated Gene Ontology analysis of expression profiles*. **Nucleic Acids Res**. 2004 Jul 1; 32(Web Server Issue): W492-9. 2-s2.0-3242886987.
190. Iolascon A, Volinia S, Borriello A, Giordani L, Moretti A, Servedio V, Maiorano N, Cucciolla V, Criniti V, Gasparini P, Indaco S, Della Ragione F. *Genes transcriptionally modulated by interferon alpha2a correlate with the cytokine activity*. **Haematologica**. 2004 Sep; 89(9): 1046-53.
191. Carella M, d'Adamo AP, Grootenboer-Mignot S, Vantyghem MC, Esposito L, D'Eustacchio A, Ficarella R, Stewart GW, Gasparini P, Delaunay J, Iolascon A. *A second locus mapping to 2q35-36 for familial pseudohyperkalaemia*. **Eur J Hum Genet**. 2004 Dec; 12(12): 1073-1076. 2-s2.0-10044259553.
192. Mazzoli M, Newton V, Murgia A, Bitner-Glindzicz M, Gasparini P, Read A, Parving A. *Guidelines and recommendations for testing of Cx26 mutations and interpretation of results*. **Int J Pediatr Otorhinolaryngol**. 2004 Nov; 68(11): 1397-1398. 2-s2.0-5644241852.
193. Jacobs HT, Hutchin TP, Kappi T, Gillies G, Minkkinen K, Walker J, Thompson K, Rovio AT, Carella M, Melchionda S, Zelante L, Gasparini P, Pyykko I I, Shah ZH, Zeviani M, Mueller RF. *Mitochondrial DNA mutations in patients with postlingual, nonsyndromic hearing impairment*. **Eur J Hum Genet**. 2005 Jan; 13(1): 26-33. 2-s2.0-19944432928.
194. Vitiello C, D'Adamo P, Gentile F, Vingolo EM, Gasparini P, Banfi S. *A novel GJA1 mutation causes oculodentodigital dysplasia without syndactyly*. **Am J Med Genet.** 2005 Feb 15; 133(1): 58-60. 2-s2.0-12944252968.
195. Coppola G, Criscuolo C, De Michele G, Striano S, Barbieri F, Striano P,Perretti A, Santoro L, Brescia Morra V, Sacca F, Scarano V, D'Adamo AP, Banfi S, Gasparini P, Santorelli FM, Lehesjoki AE, Filla A. *Autosomal recessive progressive myoclonus epilepsy with ataxia and mental retardation.* **J Neurol**. 2005 Aug, 252(8): 897-900. 2-s2.0-23944475187.
196. Franze A, Caravelli A, Di Leva F, Marciano E, Auletta G, D'Aulos F, Saulino C, Esposito L, Carella M, Gasparini P.

Audiometric evaluation of carriers of the connexin 26 mutation 35delG. **Eur Arch Otorhinolaryngol**. 2005 Nov; 262 (11): 921-924. 2-s2.0-28844498743.

197. Donaudy F, Zheng L, Ficarella R, Ballana E, Carella M, Melchionda S, Estivill X, Bartles J, Gasparini P. *Espin gene (ESPN) mutations associated with autosomal dominant hearing loss cause defects in microvillar elongation or organization*. **J Med Genet**. 2006 Feb; 43(2): 157-161.

198. Gangarossa S, Seri M, Pecci A, Di Bari F, Cusano R, Balduini C, Gasparini P, Savoia A. *Dissecting clinical findings: platelet defects segregate independently of deafness and cataract in a family affected by an apparent syndromic form of macrothrombocytopenia*. **Int J Mol Med**. 2005 Sep; 16(3): 437-441. 2-s2.0-25644445330.

199. Rees DC, Iolascon A, Carella M, O'marcaigh AS, Kendra JR, Jowitt SN, Wales JK, Vora A, Makris M, Manning N, Nicolaou A, Fisher J, Mann A, Machin SJ, Clayton PT, Gasparini P, Stewart GW. *Stomatocytic haemolysis and macrothrombocytopenia (Mediterranean stomatocytosis/macrothrombocytopenia) is the haematological presentation of phytosterolaemia*. **Br J Haematol**. 2005 Jul; 130(2): 297-309.

200. Di Leva F, D'Adamo P, Cubellis MV, D'Eustacchio A, Erricchiello M, Saulino C, Auletta G, Giannini P, Donaudy F, Ciccodicola A, Gasparini P, Franze A, Marciano E. *Identification of a novel mutation in the myosin VIIA motor domain in a family with autosomal dominant hearing loss (DFNA11)*. **Audiol Neurootol**. 2006; 11(3): 157–164.

201. Ballana E, Morales E, Rabionet R, Monteserrat B, Ventayol M, Bravo O, Gasparini P, Estivill X. *Mitochondrial 12S rRNA gene mutations affect RNA secondary structure and lead to variable penetrance in hearing impairment*. **Biochem Biophys Res Commun**. 2006 Mar 24; 341(4): 950-957. 2-s2.0-32344452298

202. Spinazzola A, Viscomi C, Fernandez-Vizarra E, Carrara F, D'Adamo P, Calov S, Marsano RM, Donnina C, Weiher H, Strisciuglio P, Parini R, Sarzi E, Chan A, Di Mauro S, Rötig A, Gasparini P, Ferrero I, Mootha VK, Tiranti V, Zeviani M. *MPV17 encodes an inner mitochondrial membrane protein and is mutated in infantile hepatic mitochondrial DNA depletion*. **Nat Genet**. 2006 May; 38(5): 570-575. 2-s2.0-33646376465

203. Fagoonee S, Di Cunto F, Vozzi D, Volinia S, Pellegrino M, Gasparini P, Silengo L, Atruda F, Tolosano E. *Microarray and large-scale in silico-based identification of genes functionally related to Haptoglobin and/or Hemopexin*. **DNA Cell Biol**. 2006 Jun; 25(6): 323-330.

204. Bertelli M, Cecchin S, Fabbri A, Lapucci C, Gasparini P. *Exclusion of chromosome region 22q12.1-q12.3 as a second locus for Costello syndrome*. **Panminerva Med**. 2006 Jun; 48(2): 145. 2-s2.0-33750547598

205. Ficarella R, Di Leva F, Bortolozzi M, Ortolano S, Donaudy F, Petrillo M, Melchionda S, Lelli A, Domi T, Fedrizzi L, Lim D, Shull GE, Gasparini P, Brini M, Mammano F, Carafoli E. *A functional*

study of plasma-membrane calcium-pump isoform 2 mutants causing digenic deafness. **Proc Natl Acad Sci U S A**. 2007 30;104(5):1516-21 2-s2.0-33846810090

206. Massa V, Fernandez-Vizarra E, Alshahwan S, Bakhsh E, Goffrini P, Ferrero I, Mereghetti P, D'Adamo P, Gasparini P, Zeviani M. *Severe Infantile Encephalomyopathy Caused by a Mutation in COX6B1, a Nucleus-Encoded Subunit of Cytochrome C Oxidase*. **Am J Hum Genet**. 2008 82:1281-9. 2-s2.0-45449121006

207. TepperB, KoellikerY, ZhaoL, UllrichN, LanzaraC, d'AdamoP, FerraraA, UliviS, EspositoL,Gasparini P. *Variation in the Bitter-Taste Receptor Gene TAS2R38, and Adiposity in a Genetically-Isolated Population in Southern Italy*. **Obesity**, 2008 Oct;16(10):2289-95. 2-s2.0-53849098349

208. Sala C, Ciullo M, Lanzara C, Nutile T, Bione S, Massacane R, d'Adamo P, Gasparini P, Toniolo D, Camaschella C. *Variation of Hb levels in normal Italian populations from genetic isolates*. **Haematologica,**2008 Sep;93(9):1372-5.

209. Bugiani M, Gyftodimou Y, Tsimpouka P, Lamantea E, Katzaki E, d'Adamo P, Nakou S, Georgoudi N, Grigoriadou M, Tsina E, Kabolis N, Milani D, Pandelia E, Kokotas H, Gasparini P, Giannoulia-Karantana A, Renieri A, Zeviani M, Petersen MB. *Cohen syndrome resulting from a novel large intragenic COH1 deletion segregating in an isolated Greek island population*. **Am J Med Genet** A. 2008 Sep 1;146A(17):2221-6. 2-s2.0-51449102096

210. Ghezzi D, Saada A, D'Adamo P, Fernandez-Vizarra E, Gasparini P,Tiranti V, Elpeleg O, Zeviani M. *FASTKD2 nonsense mutation in an infantile mitochondrial encephalomyopathy associated with cytochrome c oxidase deficiency.* **Am J Hum Genet**. 2008 Sep;83(3):415-23 2-s2.0-50949096038

211. D'Adamo P, Ulivi S, Beneduci A, Pontoni G, Capasso G, Lanzara C, Andrighetto G, Hladnik U, Nunes V, Palacin M, Gasparini P. *Metabonomics and population studies: age-related amino acids excretion and inferring networks through the study of urine samples in two Italian isolated populations*. **Amino Acids**. 2008 Dec 9. 2-s2.0-76449113994

212. D'Adamo P, Guerci VI, Fabretto A, Faletra F, Grasso DL, Ronfani L, Montico M, Morgutti M, Guastalla P, Gasparini P. *Does epidermal thickening explain GJB2 high carrier frequency and heterozygote advantage?* **Eur J Hum Genet**. 2008 Dec 3.

213. Zadro C, Alemanno MS, Bellacchio E, Ficarella R, Donaudy F, Melchionda S, Zelante L, Rabionet R, Hilgert N, Estivill X, Van Camp G, Gasparini P, Carella M. *Are MYO1C and MYO1F associated with hearing loss?* **Biochim Biophys Acta**. 2009 Jan;1792(1):27-32.

214. Guastalla P, Guerci VI, Fabretto A, Faletra F, Grasso DL, Zocconi E, Stefanidou D, D'Adamo P, Ronfani L, Montico M, Morgutti M, Gasparini P. *Detection of Epidermal Thickening in GJB2 Carriers with Epidermal US*. **Radiology**. 2009 Apr;251(1):280-6.

215. Ghezzi D, Goffrini P, Uziel G, Horvath R, Klopstock T, Lochmüller H, D'Adamo P, Gasparini P,Strom TM, Prokisch H, Invernizzi F, Ferrero I, Zeviani M. *SDHAF1, encoding a LYR complex-II specific assembly factor, is mutated in SDH-defective infantile leukoencephalopathy*. **Nature Genet**. 2009 May 24. 2-s2.0-67349189168

216. Nelis M, Esko T, Mägi R, Zimprich F, Zimprich A, Toncheva D, Karachanak S, Piskácková T, Balascák I, Peltonen L, Jakkula E, Rehnström K, Lathrop M, Heath S, Galan P, Schreiber S, Meitinger T, Pfeufer A, Wichmann HE, Melegh B, Polgár N, Toniolo D, Gasparini P,D'Adamo P, Klovins J, Nikitina-Zake L, Kucinskas V, Kasnauskiene J, Lubinski J, Debniak T, Limborska S, Khrunin A, Estivill X, Rabionet R, Marsal S, Julià A, Antonarakis SE, Deutsch S, Borel C, Attar H, Gagnebin M, Macek M, Krawczak M, Remm M, Metspalu A. *Genetic structure of Europeans: a view from the North-East*. **PLoS One.** 2009;4(5):e5472. 2-s2.0-65549126774

217. Tepper BJ, White EA, Koelliker Y, Lanzara C, d'Adamo P, Gasparini P. *Genetic variation in taste sensitivity to 6-n-propylthiouracil and its relationship to taste perception and food selection*. **Ann N Y Acad Sci.** 2009 Jul;1170:126-39. Review. 2-s2.0-68649098537

218. van Bon BW, Koolen DA, Brueton L, McMullan D, Lichtenbelt KD, Adès LC, Peters G, Gibson K, Novara F, Pramparo T, Bernardina BD, Zoccante L, Balottin U, Piazza F, Pecile V, Gasparini P, Guerci V, Kets M, Pfundt R, de Brouwer AP, Veltman JA, de Leeuw N, Wilson M, Antony J, Reitano S, Luciano D, Fichera M, Romano C, Brunner HG, Zuffardi O, de Vries BB. *The 2q23.1 microdeletion syndrome: clinical and behavioural phenotype*. **Eur J Hum Genet**. 2009 Oct 7. 2-s2.0-74449084036

219. Bedin E, Franzè A, Zadro C, Persico MG, Ciullo M, Hladnik U, Dolcetta D, Grasso DL, Riccardi P, Nutile T, Andrighetto G, D'Adamo P, Gasparini P, Marciano E. *Age-related hearing loss in four Italian genetic isolates: an epidemiological study*. **Int J Audiol**. 2009;48(7):465-72. 2-s2.0-70350475316

220. Traglia M, Sala C, Masciullo C, Cverhova V, Lori F, Pistis G, Bione S, Gasparini P,Ulivi S, Ciullo M, Nutile T, Bosi E, Sirtori M, Mignogna G, Rubinacci A, Buetti I, Camaschella C, Petretto E, Toniolo D. *Heritability and demographic analyses in the large isolated population of val borbera suggest advantages in mapping complex traits genes*. **PLoS One.** 2009 Oct 22;4(10):e7554. 2-s2.0-70449597270

221. Bahloul A, Simmler MC, Michel V, Leibovici M, Perfettini I, Roux I, Weil D, Nouaille S, Zuo J, Zadro C, Licastro D, Gasparini P, Avan P, Hardelin JP, Petit C. *Vezatin, an integral membrane protein of adherens junctions, is required for the sound resilience of cochlear hair cells*. **EMBO Mol Med**. 2009 May;1(2):125-38. 2-s2.0-77953658421

222. Chinetti V, Iossa S, Auletta G, Laria C, De Luca M, Leva FD, Riccardi P, Giannini P, Gasparini P, Ciccodicola A, Marciano E, Franzè A. *Screening for GJB2 and GJB6 gene mutations in patients from Campania region with sensorineural hearing*

loss. **Int J Audiol**. 2010 Apr;49(4):326-31. 2-s2.0-77949666282

223. Zadro C, Ciorba A, Fabris A, Morgutti M, Trevisi P, Gasparini P, Martini A. *Five new OTOF gene mutations and auditory neuropathy*. **Int J Pediatr Otorhinolaryngol**. 2010 May;74(5):494-8. 2-s2.0-77951645790

224. Fabretto A, Shardlow A, Faletra F, Lepore L, Hladnik U, Gasparini P. *A case of lymphedema-distichiasis syndrome carrying a new de novo frameshift FOXC2 mutation*. **Ophthalmic Genet.**2010 Jun;31(2):98-100. 2-s2.0-77952167573

225. Biarnés X, Marchiori A, Giorgetti A, Lanzara C, Gasparini P, Carloni P, Born S, Brockhoff A, Behrens M, Meyerhof W. *Insights into the binding of Phenyltiocarbamide (PTC) agonist to its target human TAS2R38 bitter receptor*. **PLoS One**. 2010 Aug 25;5(8). pii: e12394.

226. Bembich S, Lanzara C, Clarici A, Demarini S, Tepper BJ, Gasparini P, Grasso DL. *Individual Differences in Prefrontal Cortex Activity during Perception of Bitter Taste Using fNIRS Methodology*. **Chem Senses.**2010 Aug 26. 2-s2.0-78649364355

227. Fabretto A, Kutsche K, Harmsen MB, Demarini S, Gasparini P, Fertz MC, Zenker M. *Two cases of Noonan syndrome with severe respiratory and gastroenteral involvement and the SOS1 mutation F623I*. **Eur J Med Genet.**2010 Sep-Oct;53(5):322-4. 2-s2.0-77956927727

228. Faletra F, Devescovi R, Pecile V, Fabretto A, Carrozzi M, Gasparini P. *A new case of duplication of the MDS region identified by high-density SNP arrays and a review of the literature*. **J Appl Genet.**2010 Nov 3. 2-s2.0-79954990400

229. Elks CE, Perry JR, Sulem P, Chasman DI, Franceschini N, He C, Lunetta KL, Visser JA, Byrne EM, Cousminer DL, Gudbjartsson DF, Esko T, Feenstra B, Hottenga JJ, Koller DL, Kutalik Z, Lin P, Mangino M, Marongiu M, McArdle PF, Smith AV, Stolk L, van Wingerden SH, Zhao JH, Albrecht E, Corre T, Ingelsson E, Hayward C, Magnusson PK, Smith EN, Ulivi S, Warrington NM, Zgaga L, Alavere H, Amin N, Aspelund T, Bandinelli S, Barroso I, Berenson GS, Bergmann S, Blackburn H, Boerwinkle E, Buring JE, Busonero F, Campbell H, Chanock SJ, Chen W, Cornelis MC, Couper D, Coviello AD, d'Adamo P, de Faire U, de Geus EJ, Deloukas P, Döring A, Smith GD, Easton DF, Eiriksdottir G, Emilsson V, Eriksson J, Ferrucci L, Folsom AR, Foroud T, Garcia M, Gasparini P,Geller F, Gieger C; GIANT Consortium, Gudnason V, Hall P, Hankinson SE, Ferreli L, Heath AC, Hernandez DG, Hofman A, Hu FB, Illig T, Järvelin MR, Johnson AD, Karasik D, Khaw KT, Kiel DP, Kilpeläinen TO, Kolcic I, Kraft P, Launer LJ, Laven JS, Li S, Liu J, Levy D, Martin NG, McArdle WL, Melbye M, Mooser V, Murray JC, Murray SS, Nalls MA, Navarro P, Nelis M, Ness AR, Northstone K, Oostra BA, Peacock M, Palmer LJ, Palotie A, Paré G, Parker AN, Pedersen NL, Peltonen L, Pennell CE, Pharoah P, Polasek O, Plump AS, Pouta A, Porcu E, Rafnar T, Rice JP, Ring SM, Rivadeneira F, Rudan I, Sala C, Salomaa V, Sanna S, Schlessinger D, Schork NJ, Scuteri A, Segrè AV, Shuldiner AR, Soranzo N, Sovio U, Srinivasan SR, Strachan DP, Tammesoo ML, Tikkanen E, Toniolo D, Tsui K, Tryggvadottir L, Tyrer J, Uda M,

van Dam RM, van Meurs JB, Vollenweider P, Waeber G, Wareham NJ, Waterworth DM, Weedon MN, Wichmann HE, Willemsen G, Wilson JF, Wright AF, Young L, Zhai G, Zhuang WV, Bierut LJ, Boomsma DI, Boyd HA, Crisponi L, Demerath EW, van Duijn CM, Econs MJ, Harris TB, Hunter DJ, Loos RJ, Metspalu A, Montgomery GW, Ridker PM, Spector TD, Streeten EA, Stefansson K, Thorsteinsdottir U, Uitterlinden AG, Widen E, Murabito JM, Ong KK, Murray A. *Thirty new loci for age at menarche identified by a meta-analysis of genome-wide association studies*. **Nat Genet.**2010 Dec;42(12):1077-85. 2-s2.0-84947899470

230. Crovella S, Segat L, Amato A, Athanasakis E, Bezzerri V, Braggion C, Casciaro R, Castaldo G, Colombo C, Covone AE, De Rose V, Gagliardini R, Lanzara C, Minicucci L, Morgutti M, Nicolis E, Pardo F, Quattrucci S, Raia V, Ravazzolo R, Seia M, Stanzial V, Termini L, Zazzeron L, Cabrini G, Gasparini P. *A polymorphism in the 5' UTR of the DEFB1 gene is associated with the lung phenotype in F508del homozygous Italian cystic fibrosis patients*. **Clin Chem Lab Med.** 2010 Nov 16. 2-s2.0-79953145166

231. Sorice R, Bione S, Sansanelli S, Ulivi S, Athanasakis E, Lanzara C, Nutile T, Sala C, Camaschella C, D'Adamo P, Gasparini P, Ciullo M, Toniolo D. *Association of a variant in the CHRNA5-A3-B4 gene cluster region to heavy smoking in the Italian population*. **Eur J Hum Genet**. 2011 Jan 19. doi:10.1038/ejhg.2010.240 2-s2.0-79955767494

232. Bezzerri V, d'Adamo P, Rimessi A, Lanzara C, Crovella S, Nicolis E, Tamanini A, Athanasakis E, Tebon M, Bisoffi G, Drumm ML, Knowles MR, Pinton P, Gasparini P, Berton G, Cabrini G. *Phospholipase C-{beta}3 Is a Key Modulator of IL-8 Expression in Cystic Fibrosis Bronchial Epithelial Cells*. **J Immunol.** 2011 Apr 15;186(8):4946-58.

233. Girotto G, Pirastu N, Sorice R, Biino G, Campbell H, d'Adamo AP, Hastie ND, Nutile T, Polasek O, Portas L, Rudan I, Ulivi S, Zemunik T, Wright AF, Ciullo M, Hayward C, Pirastu M, Gasparini P. *Hearing function and thresholds: a genome-wide association study in European isolated populations identifies new loci and pathways*. **J Med Genet**. 2011 Apr 14. 2-s2.0-79958067518

234. Abu-Amero KK, Faletra F, Gasparini P, Parentin F, Pensiero S, Alorainy IA, Hellani AM, Catalano D, Bosley TM. *Horizontal gaze palsy and progressive scoliosis without ROBO3 mutations*. **Ophthalmic Genet**. 2011 Apr 21. 2-s2.0-80054866145

235. Faletra F, Athanasakis E, Minen F, Fornasier F, Marchetti F, Gasparini P. *Vertebral Defects in Patients with Peters Plus Syndrome and Mutations in B3GALTL*. **Ophthalmic Genet.** 2011 Jun 14. 2-s2.0-80054860616

236. Vozzi D, Aaspõllu A, Athanasakis E, Berto A, Fabretto A, Licastro D, Külm M, Testa F, Trevisi P, Vahter M, Ziviello C, Martini A, Simonelli F, Banfi S, Gasparini P. *Molecular epidemiology of Usher syndrome in Italy*. **Mol Vis.** 2011;17:1662-8. 2-s2.0-79960011547

237. Nalls MA, Couper DJ, Tanaka T, van Rooij FJ, Chen MH, Smith

AV, Toniolo D, Zakai NA, Yang Q, Greinacher A, Wood AR, Garcia M, Gasparini P,Liu Y, Lumley T, Folsom AR, Reiner AP, Gieger C, Lagou V, Felix JF, Völzke H, Gouskova NA, Biffi A, Döring A, Völker U, Chong S, Wiggins KL, Rendon A, Dehghan A, Moore M, Taylor K, Wilson JG, Lettre G, Hofman A, Bis JC, Pirastu N, Fox CS, Meisinger C, Sambrook J, Arepalli S, Nauck M, Prokisch H, Stephens J, Glazer NL, Cupples LA, Okada Y, Takahashi A, Kamatani Y, Matsuda K, Tsunoda T, Tanaka T, Kubo M, Nakamura Y, Yamamoto K, Kamatani N, Stumvoll M, Tönjes A, Prokopenko I, Illig T, Patel KV, Garner SF, Kuhnel B, Mangino M, Oostra BA, Thein SL, Coresh J, Wichmann HE, Menzel S, Lin J, Pistis G, Uitterlinden AG, Spector TD, Teumer A, Eiriksdottir G, Gudnason V, Bandinelli S, Frayling TM, Chakravarti A, van Duijn CM, Melzer D, Ouwehand WH, Levy D, Boerwinkle E, Singleton AB, Hernandez DG, Longo DL, Soranzo N, Witteman JC, Psaty BM, Ferrucci L, Harris TB, O'Donnell CJ, Ganesh SK. *Multiple loci are associated with white blood cell phenotypes*. **PLoS Genet.** 2011 Jun;7(6):e1002113. 2-s2.0-79959824142

238. Wain LV, Verwoert GC, O'Reilly PF, Shi G, Johnson T, Johnson AD, Bochud M, Rice KM, Henneman P, Smith AV, Ehret GB, Amin N, Larson MG, Mooser V, Hadley D, Dörr M, Bis JC, Aspelund T, Esko T, Janssens AC, Zhao JH, Heath S, Laan M, Fu J, Pistis G, Luan J, Arora P, Lucas G, Pirastu N, Pichler I, Jackson AU, Webster RJ, Zhang F, Peden JF, Schmidt H, Tanaka T, Campbell H, Igl W, Milaneschi Y, Hottenga JJ, Vitart V, Chasman DI, Trompet S, Bragg-Gresham JL, Alizadeh BZ, Chambers JC, Guo X, Lehtimäki T, Kühnel B, Lopez LM, Polašek O, Boban M, Nelson CP, Morrison AC, Pihur V, Ganesh SK, Hofman A, Kundu S, Mattace-Raso FU, Rivadeneira F, Sijbrands EJ, Uitterlinden AG, Hwang SJ, Vasan RS, Wang TJ, Bergmann S, Vollenweider P, Waeber G, Laitinen J, Pouta A, Zitting P, McArdle WL, Kroemer HK, Völker U, Völzke H, Glazer NL, Taylor KD, Harris TB, Alavere H, Haller T, Keis A, Tammesoo ML, Aulchenko Y, Barroso I, Khaw KT, Galan P, Hercberg S, Lathrop M, Eyheramendy S, Org E, Sõber S, Lu X, Nolte IM, Penninx BW, Corre T, Masciullo C, Sala C, Groop L, Voight BF, Melander O, O'Donnell CJ, Salomaa V, d'Adamo AP, Fabretto A, Faletra F, Ulivi S, Del Greco M F, Facheris M, Collins FS, Bergman RN, Beilby JP, Hung J, Musk AW, Mangino M, Shin SY, Soranzo N, Watkins H, Goel A, Hamsten A, Gider P, Loitfelder M, Zeginigg M, Hernandez D, Najjar SS, Navarro P, Wild SH, Corsi AM, Singleton A, de Geus EJ, Willemsen G, Parker AN, Rose LM, Buckley B, Stott D, Orru M, Uda M; LifeLines Cohort Study, van der Klauw MM, Zhang W, Li X, Scott J, Chen YD, Burke GL, Kähönen M, Viikari J, Döring A, Meitinger T, Davies G, Starr JM, Emilsson V, Plump A, Lindeman JH, Hoen PA, König IR; EchoGen consortium, Felix JF, Clarke R, Hopewell JC, Ongen H, Breteler M, Debette S, Destefano AL, Fornage M; AortaGen Consortium, Mitchell GF; CHARGE Consortium Heart Failure Working Group, Smith NL; KidneyGen consortium, Holm H, Stefansson K, Thorleifsson G, Thorsteinsdottir U; CKDGen consortium; Cardiogenics consortium; CardioGram, Samani NJ, Preuss M, Rudan I,

Hayward C, Deary IJ, Wichmann HE, Raitakari OT, Palmas W, Kooner JS, Stolk RP, Jukema JW, Wright AF, Boomsma DI, Bandinelli S, Gyllensten UB, Wilson JF, Ferrucci L, Schmidt R, Farrall M, Spector TD, Palmer LJ, Tuomilehto J, Pfeufer A, Gasparini P,Siscovick D, Altshuler D, Loos RJ, Toniolo D, Snieder H, Gieger C, Meneton P, Wareham NJ, Oostra BA, Metspalu A, Launer L, Rettig R, Strachan DP, Beckmann JS, Witteman JC, Erdmann J, van Dijk KW, Boerwinkle E, Boehnke M, Ridker PM, Jarvelin MR, Chakravarti A, Abecasis GR, Gudnason V, Newton-Cheh C, Levy D, Munroe PB, Psaty BM, Caulfield MJ, Rao DC, Tobin MD, Elliott P, van Duijn CM. *Genome-wide association study identifies six new loci influencing pulse pressure and mean arterial pressure*. **Nat Genet.** 2011 Sep 11. doi: 10.1038/ng.922. 2-s2.0-80053383403

239. Rocca MS, Fabretto A, Faletra F, Carlet O, Skabar A, Gasparini P, Pecile V. *Contribution of SNP arrays in diagnosis of deletion 2p11.2-p12*. **Gene.**2012 Jan 15;492(1):315-8. 2-s2.0-84155186334

240. Gieger C, Radhakrishnan A, Cvejic A, Tang W, Porcu E, Pistis G, Serbanovic-Canic J, Elling U, Goodall AH, Labrune Y, Lopez LM, Mägi R, Meacham S, Okada Y, Pirastu N, Sorice R, Teumer A, Voss K, Zhang W, Ramirez-Solis R, Bis JC, Ellinghaus D, Gögele M, Hottenga JJ, Langenberg C, Kovacs P, O'Reilly PF, Shin SY, Esko T, Hartiala J, Kanoni S, Murgia F, Parsa A, Stephens J, van der Harst P, Ellen van der Schoot C, Allayee H, Attwood A, Balkau B, Bastardot F, Basu S, Baumeister SE, Biino G, Bomba L, Bonnefond A, Cambien F, Chambers JC, Cucca F, D'Adamo P, Davies G, de Boer RA, de Geus EJ, Döring A, Elliott P, Erdmann J, Evans DM, Falchi M, Feng W, Folsom AR, Frazer IH, Gibson QD, Glazer NL, Hammond C, Hartikainen AL, Heckbert SR, Hengstenberg C, Hersch M, Illig T, Loos RJ, Jolley J, Tee Khaw K, Kühnel B, Kyrtsonis MC, Lagou V, Lloyd-Jones H, Lumley T, Mangino M, Maschio A, Mateo Leach I, McKnight B, Memari Y, Mitchell BD, Montgomery GW, Nakamura Y, Nauck M, Navis G, Nöthlings U, Nolte IM, Porteous DJ, Pouta A, Pramstaller PP, Pullat J, Ring SM, Rotter JI, Ruggiero D, Ruokonen A, Sala C, Samani NJ, Sambrook J, Schlessinger D, Schreiber S, Schunkert H, Scott J, Smith NL, Snieder H, Starr JM, Stumvoll M, Takahashi A, Tang WH, Taylor K, Tenesa A, Lay Thein S, Tönjes A, Uda M, Ulivi S, van Veldhuisen DJ, Visscher PM, Völker U, Wichmann HE, Wiggins KL, Willemsen G, Yang TP, Hua Zhao J, Zitting P, Bradley JR, Dedoussis GV, Gasparini P, Hazen SL, Metspalu A, Pirastu M, Shuldiner AR, Joost van Pelt L, Zwaginga JJ, Boomsma DI, Deary IJ, Franke A, Froguel P, Ganesh SK, Jarvelin MR, Martin NG, Meisinger C, Psaty BM, Spector TD, Wareham NJ, Akkerman JW, Ciullo M, Deloukas P, Greinacher A, Jupe S, Kamatani N, Khadake J, Kooner JS, Penninger J, Prokopenko I, Stemple D, Toniolo D, Wernisch L, Sanna S, Hicks AA, Rendon A, Ferreira MA, Ouwehand WH, Soranzo N. *New gene functions in megakaryopoiesis and platelet formation*. **Nature.**2011 Nov 30;480(7376):201-8. 2-s2.0-83055161438

241. Khalifa Alkowari M, Girotto G, Abdulhadi K, Dipresa S, Siam

R, Najjar N, Badii R, Gasparini P. *GJB2 and GJB6 genes and the A1555G mitochondrial mutation are only minor causes of nonsyndromic hearing loss in the Qatari population*. **Int J Audiol.**2012 Mar;51(3):181-5. 2-s2.0-84857200820

242. Nicchia GP, Ficarella R, Rossi A, Giangreco I, Nicolotti O, Carotti A, Pisani F, Estivill X, Gasparini P, Svelto M, Frigeri A. *D184E mutation in aquaporin-4 gene impairs water permeability and links to deafness*. **Neuroscience.** 2011 Dec 1;197:80-8. 2-s2.0-82855165043

243. Girotto G, Pirastu P, Gasparini A, d'Adamo P, Gasparini P. *Frequency of hearing loss in a series of rural communities of five developing countries located along the Silk Road*. **Audiological Medicine**, 9, 4: 135–140, 2011 2-s2.0-81255167101

244. Biino G, Gasparini P, D'Adamo P, Ciullo M, Nutile T, Toniolo D, Sala C, Minelli C, Gögele M, Balduini CL. *Influence of age, sex and ethnicity on platelet count in five Italian geographic isolates: mild thrombocytopenia may be physiological*. **Br J Haematol.**2011 Dec 15. doi: 10.1111/j.1365-2141.2011.08981.x. 2-s2.0-84859502866

245. Stolk L, Perry JR, Chasman DI, He C, Mangino M, Sulem P, Barbalic M, Broer L, Byrne EM, Ernst F, Esko T, Franceschini N, Gudbjartsson DF, Hottenga JJ, Kraft P, McArdle PF, Porcu E, Shin SY, Smith AV, van Wingerden S, Zhai G, Zhuang WV, Albrecht E, Alizadeh BZ, Aspelund T, Bandinelli S, Lauc LB, Beckmann JS, Boban M, Boerwinkle E, Broekmans FJ, Burri A, Campbell H, Chanock SJ, Chen C, Cornelis MC, Corre T, Coviello AD, d'Adamo P, Davies G, de Faire U, de Geus EJ, Deary IJ, Dedoussis GV, Deloukas P, Ebrahim S, Eiriksdottir G, Emilsson V, Eriksson JG, Fauser BC, Ferreli L, Ferrucci L, Fischer K, Folsom AR, Garcia ME, Gasparini P,Gieger C, Glazer N, Grobbee DE, Hall P, Haller T, Hankinson SE, Hass M, Hayward C, Heath AC, Hofman A, Ingelsson E, Janssens AC, Johnson AD, Karasik D, Kardia SL, Keyzer J, Kiel DP, Kolcic I, Kutalik Z, Lahti J, Lai S, Laisk T, Laven JS, Lawlor DA, Liu J, Lopez LM, Louwers YV, Magnusson PK, Marongiu M, Martin NG, Klaric IM, Masciullo C, McKnight B, Medland SE, Melzer D, Mooser V, Navarro P, Newman AB, Nyholt DR, Onland-Moret NC, Palotie A, Paré G, Parker AN, Pedersen NL, Peeters PH, Pistis G, Plump AS, Polasek O, Pop VJ, Psaty BM, Räikkönen K, Rehnberg E, Rotter JI, Rudan I, Sala C, Salumets A, Scuteri A, Singleton A, Smith JA, Snieder H, Soranzo N, Stacey SN, Starr JM, Stathopoulou MG, Stirrups K, Stolk RP, Styrkarsdottir U, Sun YV, Tenesa A, Thorand B, Toniolo D, Tryggvadottir L, Tsui K, Ulivi S, van Dam RM, van der Schouw YT, van Gils CH, van Nierop P, Vink JM, Visscher PM, Voorhuis M, Waeber G, Wallaschofski H, Wichmann HE, Widen E, Wijnands-van Gent CJ, Willemsen G, Wilson JF, Wolffenbuttel BH, Wright AF, Yerges-Armstrong LM, Zemunik T, Zgaga L, Zillikens MC, Zygmunt M, Study TL, Arnold AM, Boomsma DI, Buring JE, Crisponi L, Demerath EW, Gudnason V, Harris TB, Hu FB, Hunter DJ, Launer LJ, Metspalu A, Montgomery GW, Oostra BA, Ridker PM, Sanna S, Schlessinger D, Spector TD, Stefansson K, Streeten EA, Thorsteinsdottir U, Uda M, Uitterlinden AG, van Duijn CM,

Völzke H, Murray A, Murabito JM, Visser JA, Lunetta KL. *Meta-analyses identify 13 loci associated with age at menopause and highlight DNA repair and immune pathways*. **Nat Genet.**2012 Jan 22;44(3):260-8. doi: 10.1038/ng.1051. 2-s2.0-84862777492

246. Perrone MD, Rocca MS, Bruno I, Faletra F, Pecile V, Gasparini P. *De novo 911 Kb interstitial deletion on chromosome 1q43 in a boy with mental retardation and short stature*. **Eur J Med Genet.**2012 Feb;55(2):117-9. 2-s2.0-84857440662

247. Faletra F, D'Adamo AP, Santa Rocca M, Carrozzi M, Perrone MD, Pecile V, Gasparini P. *Does the 1.5 Mb microduplication in chromosome band Xp22.31 have a pathogenetic role? New contribution and a review of the literature*. **Am J Med Genet A.**2012 Feb;158A(2):461-4. doi: 10.1002/ajmg.a.34398.

248. Minen F, Cont G, De Cunto A, Martelossi S, Ventura A, Maggiore G, Faletra F, Gasparini P, Cassandrini D. *Delayed diagnosis of glycogen storage disease type III*. **J Pediatr Gastroenterol Nutr.** 2012 Jan;54(1):122-4. 2-s2.0-84855352459

249. van der Harst P, Zhang W, Mateo Leach I, Rendon A, Verweij N, Sehmi J, Paul DS, Elling U, Allayee H, Li X, Radhakrishnan A, Tan ST, Voss K, Weichenberger CX, Albers CA, Al-Hussani A, Asselbergs FW, Ciullo M, Danjou F, Dina C, Esko T, Evans DM, Franke L, Gögele M, Hartiala J, Hersch M, Holm H, Hottenga JJ, Kanoni S, Kleber ME, Lagou V, Langenberg C, Lopez LM, Lyytikäinen LP, Melander O, Murgia F, Nolte IM, O'Reilly PF, Padmanabhan S, Parsa A, Pirastu N, Porcu E, Portas L, Prokopenko I, Ried JS, Shin SY, Tang CS, Teumer A, Traglia M, Ulivi S, Westra HJ, Yang J, Zhao JH, Anni F, Abdellaoui A, Attwood A, Balkau B, Bandinelli S, Bastardot F, Benyamin B, Boehm BO, Cookson WO, Das D, de Bakker PI, de Boer RA, de Geus EJ, de Moor MH, Dimitriou M, Domingues FS, Döring A, Engström G, Eyjolfsson GI, Ferrucci L, Fischer K, Galanello R, Garner SF, Genser B, Gibson QD, Girotto G, Gudbjartsson DF, Harris SE, Hartikainen AL, Hastie CE, Hedblad B, Illig T, Jolley J, Kähönen M, Kema IP, Kemp JP, Liang L, Lloyd-Jones H, Loos RJ, Meacham S, Medland SE, Meisinger C, Memari Y, Mihailov E, Miller K, Moffatt MF, Nauck M, Novatchkova M, Nutile T, Olafsson I, Onundarson PT, Parracciani D, Penninx BW, Perseu L, Piga A, Pistis G, Pouta A, Puc U, Raitakari O, Ring SM, Robino A, Ruggiero D, Ruokonen A, Saint-Pierre A, Sala C, Salumets A, Sambrook J, Schepers H, Schmidt CO, Silljé HH, Sladek R, Smit JH, Starr JM, Stephens J, Sulem P, Tanaka T, Thorsteinsdottir U, Tragante V, van Gilst WH, van Pelt LJ, van Veldhuisen DJ, Völker U, Whitfield JB, Willemsen G, Winkelmann BR, Wirnsberger G, Algra A, Cucca F, d'Adamo AP, Danesh J, Deary IJ, Dominiczak AF, Elliott P, Fortina P, Froguel P, Gasparini P, Greinacher A, Hazen SL, Jarvelin MR, Khaw KT, Lehtimäki T, Maerz W, Martin NG, Metspalu A, Mitchell BD, Montgomery GW, Moore C, Navis G, Pirastu M, Pramstaller PP, Ramirez-Solis R, Schadt E, Scott J, Shuldiner AR, Smith GD, Smith JG, Snieder H, Sorice R, Spector TD, Stefansson K, Stumvoll M, Tang WH, Toniolo D, Tönjes A, Visscher PM, Vollenweider P, Wareham NJ, Wolffenbuttel BH,

Boomsma DI, Beckmann JS, Dedoussis GV, Deloukas P, Ferreira MA, Sanna S, Uda M, Hicks AA, Penninger JM, Gieger C, Kooner JS, Ouwehand WH, Soranzo N, Chambers JC. *Seventy-five genetic loci influencing the human red blood cell*. **Nature.** 2012 Dec 20;492(7429):369-75. doi: 10.1038/nature11677. 2-s2.0-84871464519

250. Faletra F, Berti I, Tommasini A, Pecile V, Cleva L, Alberini E, Bruno I, Gasparini P. *Phylloid pattern of hypomelanosis closely related to chromosomal abnormalities in the 13q detected by SNP array analysis.* **Dermatology.**2012;225(4):294-7. doi: 10.1159/000342884. 2-s2.0-84875217877

251. Nürnberg ST, Rendon A, Smethurst PA, Paul DS, Voss K, Thon JN, Lloyd-Jones H, Sambrook JG, Tijssen MR; HaemGen Consortium, Italiano JE Jr, Deloukas P, Gottgens B, Soranzo N, Ouwehand WH (incl. Gasparini P as collaborator). *A GWAS sequence variant for platelet volume marks an alternative DNM3 promoter in megakaryocytes near a MEIS1 binding site.* **Blood.**2012 Dec 6;120(24):4859-68. doi: 10.1182/blood-2012-01-401893.

252. Chasman DI, Fuchsberger C, Pattaro C, Teumer A, Böger CA, Endlich K, Olden M, Chen MH, Tin A, Taliun D, Li M, Gao X, Gorski M, Yang Q, Hundertmark C, Foster MC, O'Seaghdha CM, Glazer N, Isaacs A, Liu CT, Smith AV, O'Connell JR, Struchalin M, Tanaka T, Li G, Johnson AD, Gierman HJ, Feitosa MF, Hwang SJ, Atkinson EJ, Lohman K, Cornelis MC, Johansson A, Tönjes A, Dehghan A, Lambert JC, Holliday EG, Sorice R, Kutalik Z, Lehtimäki T, Esko T, Deshmukh H, Ulivi S, Chu AY, Murgia F, Trompet S, Imboden M, Coassin S, Pistis G; CARDIoGRAM Consortium; ICBP Consortium; CARe Consortium; WTCCC2, Harris TB, Launer LJ, Aspelund T, Eiriksdottir G, Mitchell BD, Boerwinkle E, Schmidt H, Cavalieri M, Rao M, Hu F, Demirkan A, Oostra BA, de Andrade M, Turner ST, Ding J, Andrews JS, Freedman BI, Giulianini F, Koenig W, Illig T, Meisinger C, Gieger C, Zgaga L, Zemunik T, Boban M, Minelli C, Wheeler HE, Igl W, Zaboli G, Wild SH, Wright AF, Campbell H, Ellinghaus D, Nöthlings U, Jacobs G, Biffar R, Ernst F, Homuth G, Kroemer HK, Nauck M, Stracke S, Völker U, Völzke H, Kovacs P, Stumvoll M, Mägi R, Hofman A, Uitterlinden AG, Rivadeneira F, Aulchenko YS, Polasek O, Hastie N, Vitart V, Helmer C, Wang JJ, Stengel B, Ruggiero D, Bergmann S, Kähönen M, Viikari J, Nikopensius T, Province M, Ketkar S, Colhoun H, Doney A, Robino A, Krämer BK, Portas L, Ford I, Buckley BM, Adam M, Thun GA, Paulweber B, Haun M, Sala C, Mitchell P, Ciullo M, Kim SK, Vollenweider P, Raitakari O, Metspalu A, Palmer C, Gasparini P, Pirastu M, Jukema JW, Probst-Hensch NM, Kronenberg F, Toniolo D, Gudnason V, Shuldiner AR, Coresh J, Schmidt R, Ferrucci L, Siscovick DS, van Duijn CM, Borecki IB, Kardia SL, Liu Y, Curhan GC, Rudan I, Gyllensten U, Wilson JF, Franke A, Pramstaller PP, Rettig R, Prokopenko I, Witteman J, Hayward C, Ridker PM, Parsa A, Bochud M, Heid IM, Kao WH, Fox CS, Köttgen A. *Integration of genome-wide association studies with biological knowledge identifies six novel genes related to kidney function*. **Hum Mol Genet**. 2012 Dec 15;21(24):5329-43. doi:

10.1093/hmg/dds369. 2-s2.0-84870691564

253. Licastro D, Mutarelli M, Peluso I, Neveling K, Wieskamp N, Rispoli R, Vozzi D, Athanasakis E, D'Eustacchio A, Pizzo M, D'Amico F, Ziviello C, Simonelli F, Fabretto A, Scheffer H, Gasparini P,Banfi S, Nigro V. *Molecular diagnosis of Usher syndrome: application of two different next generation sequencing-based procedures*. **PLoS One**. 2012;7(8):e43799. doi: 10.1371/journal.pone.0043799. 2-s2.0-84865585969

254. Pirastu N, Robino A, Lanzara C, Athanasakis E, Esposito L, Tepper BJ, Gasparini P. *Genetics of food preferences: a first view from silk road populations*. **J Food Sci**. 2012 Dec;77(12):S413-8. doi: 10.1111/j.1750-3841.2012.02852.x. 2-s2.0-84871207849

255. McQuillan R, Eklund N, Pirastu N, Kuningas M, McEvoy BP, Esko T, Corre T, Davies G, Kaakinen M, Lyytikäinen LP, Kristiansson K, Havulinna AS, Gögele M, Vitart V, Tenesa A, Aulchenko Y, Hayward C, Johansson A, Boban M, Ulivi S, Robino A, Boraska V, Igl W, Wild SH, Zgaga L, Amin N, Theodoratou E, Polašek O, Girotto G, Lopez LM, Sala C, Lahti J, Laatikainen T, Prokopenko I, Kals M, Viikari J, Yang J, Pouta A, Estrada K, Hofman A, Freimer N, Martin NG, Kähönen M, Milani L, Heliövaara M, Vartiainen E, Räikkönen K, Masciullo C, Starr JM, Hicks AA, Esposito L, Kolčić I, Farrington SM, Oostra B, Zemunik T, Campbell H, Kirin M, Pehlic M, Faletra F, Porteous D, Pistis G, Widén E, Salomaa V, Koskinen S, Fischer K, Lehtimäki T, Heath A, McCarthy MI, Rivadeneira F, Montgomery GW, Tiemeier H, Hartikainen AL, Madden PA, d'Adamo P, Hastie ND, Gyllensten U, Wright AF, van Duijn CM, Dunlop M, Rudan I, Gasparini P,Pramstaller PP, Deary IJ, Toniolo D, Eriksson JG, Jula A, Raitakari OT, Metspalu A, Perola M, Järvelin MR, Uitterlinden A, Visscher PM, Wilson JF; ROHgen Consortium. *Evidence of inbreeding depression on human height*. **PLoS Genet.**2012;8(7):e1002655. 2-s2.0-84864581394

256. Okada Y, Sim X, Go MJ, Wu JY, Gu D, Takeuchi F, Takahashi A, Maeda S, Tsunoda T, Chen P, Lim SC, Wong TY, Liu J, Young TL, Aung T, Seielstad M, Teo YY, Kim YJ, Lee JY, Han BG, Kang D, Chen CH, Tsai FJ, Chang LC, Fann SJ, Mei H, Rao DC, Hixson JE, Chen S, Katsuya T, Isono M, Ogihara T, Chambers JC, Zhang W, Kooner JS; KidneyGen Consortium; CKDGen Consortium, Albrecht E; GUGC consortium, Yamamoto K, Kubo M, Nakamura Y, Kamatani N, Kato N, He J, Chen YT, Cho YS, Tai ES, Tanaka T (incl. Gasparini P as collaborator). *Meta-analysis identifies multiple loci associated with kidney function-related traits in east Asian populations*. **Nat Genet.**2012 Jul 15;44(8):904-9. doi: 10.1038/ng.2352. 2-s2.0-84864413706

257. Sinner MF, Porthan K, Noseworthy PA, Havulinna AS, Tikkanen JT, Müller-Nurasyid M, Peloso G, Ulivi S, Beckmann BM, Brockhaus AC, Cooper RR, Gasparini P,Hengstenberg C, Hwang SJ, Iorio A, Junttila MJ, Klopp N, Kähönen M, Laaksonen MA, Lehtimäki T, Lichtner P, Lyytikäinen LP, Martens E, Meisinger C, Meitinger T, Merchant FM, Nieminen MS, Peters A, Pietilä A, Perz S, Oikarinen L, Raitakari O, Reinhard W, Silander K, Thorand B,

Wichmann HE, Sinagra G, Viikari J, O'Donnell CJ, Ellinor PT, Huikuri HV, Kääb S, Newton-Cheh C, Salomaa V. *A meta-analysis of genome-wide association studies of the electrocardiographic early repolarization pattern*. **Heart Rhythm.**2012 Oct;9(10):1627-34. doi: 10.1016/j.hrthm.2012.06.008. 2-s2.0-84866740007

258. Pattaro C, Köttgen A, Teumer A, Garnaas M, Böger CA, Fuchsberger C, Olden M, Chen MH, Tin A, Taliun D, Li M, Gao X, Gorski M, Yang Q, Hundertmark C, Foster MC, O'Seaghdha CM, Glazer N, Isaacs A, Liu CT, Smith AV, O'Connell JR, Struchalin M, Tanaka T, Li G, Johnson AD, Gierman HJ, Feitosa M, Hwang SJ, Atkinson EJ, Lohman K, Cornelis MC, Johansson Å, Tönjes A, Dehghan A, Chouraki V, Holliday EG, Sorice R, Kutalik Z, Lehtimäki T, Esko T, Deshmukh H, Ulivi S, Chu AY, Murgia F, Trompet S, Imboden M, Kollerits B, Pistis G; CARDIoGRAM Consortium; ICBP Consortium; CARe Consortium; Wellcome Trust Case Control Consortium 2 (WTCCC2), Harris TB, Launer LJ, Aspelund T, Eiriksdottir G, Mitchell BD, Boerwinkle E, Schmidt H, Cavalieri M, Rao M, Hu FB, Demirkan A, Oostra BA, de Andrade M, Turner ST, Ding J, Andrews JS, Freedman BI, Koenig W, Illig T, Döring A, Wichmann HE, Kolcic I, Zemunik T, Boban M, Minelli C, Wheeler HE, Igl W, Zaboli G, Wild SH, Wright AF, Campbell H, Ellinghaus D, Nöthlings U, Jacobs G, Biffar R, Endlich K, Ernst F, Homuth G, Kroemer HK, Nauck M, Stracke S, Völker U, Völzke H, Kovacs P, Stumvoll M, Mägi R, Hofman A, Uitterlinden AG, Rivadeneira F, Aulchenko YS, Polasek O, Hastie N, Vitart V, Helmer C, Wang JJ, Ruggiero D, Bergmann S, Kähönen M, Viikari J, Nikopensius T, Province M, Ketkar S, Colhoun H, Doney A, Robino A, Giulianini F, Krämer BK, Portas L, Ford I, Buckley BM, Adam M, Thun GA, Paulweber B, Haun M, Sala C, Metzger M, Mitchell P, Ciullo M, Kim SK, Vollenweider P, Raitakari O, Metspalu A, Palmer C, Gasparini P,Pirastu M, Jukema JW, Probst-Hensch NM, Kronenberg F, Toniolo D, Gudnason V, Shuldiner AR, Coresh J, Schmidt R, Ferrucci L, Siscovick DS, van Duijn CM, Borecki I, Kardia SL, Liu Y, Curhan GC, Rudan I, Gyllensten U, Wilson JF, Franke A, Pramstaller PP, Rettig R, Prokopenko I, Witteman JC, Hayward C, Ridker P, Parsa A, Bochud M, Heid IM, Goessling W, Chasman DI, Kao WH, Fox CS. *Genome-wide association and functional follow-up reveals new loci for kidney function*. **PLoS Genet.**2012;8(3):e1002584. doi: 10.1371/journal.pgen.1002584. 2-s2.0-84859224378

259. Fabretto A, Santa Rocca M, Perrone MD, Skabar A, Pecile V, Gasparini P. *De novo 6.9 Mb interstitial deletion on chromosome 4q31.1-q32.1 in a girl with severe speech delay and dysmorphic features*. **Am J Med Genet A.**2012 Apr;158A(4):882-7. doi: 10.1002/ajmg.a.35239.

260. Athanasakis E, Biarnés X, Bonati MT, Gasparini P, Faletra F. *Identification of a New Mutation (L46P) in the Human NOG Gene in an Italian Patient with Symphalangism Syndrome*. **Mol Syndromol**. 2012 Jun;3(1):21-24.

261. Athanasakis E, Fabretto A, Faletra F, Mocenigo M, Morgan A, Gasparini P. *Two Novel COH1 Mutations in an Italian Patient*

with Cohen Syndrome. **Mol Syndromol**. 2012 Jun;3(1):30-33. 2-s2.0-84862499186

262. Vuckovic D, Biino G, Panu F, Pirastu M, Gasparini P, Girotto G. *Lifestyle and normal hearing function in Italy and Central Asia: The potential role of coffee*. **Hearing, Balance and communication**.11:4, 218-223. 2-s2.0-84894126671

263. Faletra F, d'Adamo AP, Pensiero S, Athanasakis E, Catalano D, Bruno I, Gasparini P.*A Novel CRYBB2 Missense Mutation Causing Congenital Autosomal Dominant Cataract in an Italian Family*. **Ophthalmic Genet.**2013 Mar-Jun;34(1-2):115-7. doi: 10.3109/13816810.2012.70727. 2-s2.0-84876068280

264. Rocca MS, Faletra F, Devescovi R, Gasparini P, Pecile V. *Molecular cytogenetic characterization of 2p23.2p23.3 deletion in a child with developmental delay, hypotonia and cryptorchism*. **Eur J Med Genet.**2013 Jan;56(1):62-5. doi: 10.1016/j.ejmg.2012.10.008. 2-s2.0-84871701218

265. Keating B, Bansal AT, Walsh S, Millman J, Newman J, Kidd K, Budowle B, Eisenberg A, Donfack J, Gasparini P, Budimlija Z, Henders AK, Chandrupatla H, Duffy DL, Gordon SD, Hysi P, Liu F, Medland SE, Rubin L, Martin NG, Spector TD, Kayser M; on behalf of the International Visible Trait Genetics (VisiGen) Consortium. *First all-in-one diagnostic tool for DNA intelligence: genome-wide inference of biogeographic ancestry, appearance, relatedness, and sex with the Identitas v1 Forensic Chip*. **Int J Legal Med.** 2012 Nov 13. 2-s2.0-84877873104

266. Esko T, Mezzavilla M, Nelis M, Borel C, Debniak T, Jakkula E, Julia A, Karachanak S, Khrunin A, Kisfali P, Krulisova V, Aušrelé Kuinskiené Z, Rehnström K, Traglia M, Nikitina-Zake L, Zimprich F, Antonarakis SE, Estivill X, Glava D, Gut I, Klovins J, Krawczak M, Kuinskas V, Lathrop M, Macek M, Marsal S, Meitinger T, Melegh B, Limborska S, Lubinski J, Paolotie A, Schreiber S, Toncheva D, Toniolo D, Wichmann HE, Zimprich A, Metspalu M, Gasparini P, Metspalu A, D'Adamo P. *Genetic characterization of northeastern Italian population isolates in the context of broader European genetic diversity*. **Eur J Hum Genet.**2013 Jun;21(6):659-65. doi: 10.1038/ejhg.2012.229. 2-s2.0-84885927377

267. Köttgen A, Albrecht E, Teumer A, Vitart V, Krumsiek J, Hundertmark C, Pistis G, Ruggiero D, O'Seaghdha CM, Haller T, Yang Q, Tanaka T, Johnson AD, Kutalik Z, Smith AV, Shi J, Struchalin M, Middelberg RP, Brown MJ, Gaffo AL, Pirastu N, Li G, Hayward C, Zemunik T, Huffman J, Yengo L, Zhao JH, Demirkan A, Feitosa MF, Liu X, Malerba G, Lopez LM, van der Harst P, Li X, Kleber ME, Hicks AA, Nolte IM, Johansson A, Murgia F, Wild SH, Bakker SJ, Peden JF, Dehghan A, Steri M, Tenesa A, Lagou V, Salo P, Mangino M, Rose LM, Lehtimäki T, Woodward OM, Okada Y, Tin A, Müller C, Oldmeadow C, Putku M, Czamara D, Kraft P, Frogheri L, Thun GA, Grotevendt A, Gislason GK, Harris TB, Launer LJ, McArdle P, Shuldiner AR, Boerwinkle E, Coresh J, Schmidt H, Schallert M, Martin NG, Montgomery GW, Kubo M, Nakamura Y, Tanaka T, Munroe PB, Samani NJ, Jacobs DR Jr, Liu K, D'Adamo P, Ulivi S, Rotter JI, Psaty BM, Vollenweider P,

Waeber G, Campbell S, Devuyst O, Navarro P, Kolcic I, Hastie N, Balkau B, Froguel P, Esko T, Salumets A, Khaw KT, Langenberg C, Wareham NJ, Isaacs A, Kraja A, Zhang Q, Wild PS, Scott RJ, Holliday EG, Org E, Viigimaa M, Bandinelli S, Metter JE, Lupo A, Trabetti E, Sorice R, Döring A, Lattka E, Strauch K, Theis F, Waldenberger M, Wichmann HE, Davies G, Gow AJ, Bruinenberg M; LifeLines Cohort Study, Stolk RP, Kooner JS, Zhang W, Winkelmann BR, Boehm BO, Lucae S, Penninx BW, Smit JH, Curhan G, Mudgal P, Plenge RM, Portas L, Persico I, Kirin M, Wilson JF, Mateo Leach I, van Gilst WH, Goel A, Ongen H, Hofman A, Rivadeneira F, Uitterlinden AG, Imboden M, von Eckardstein A, Cucca F, Nagaraja R, Piras MG, Nauck M, Schurmann C, Budde K, Ernst F, Farrington SM, Theodoratou E, Prokopenko I, Stumvoll M, Jula A, Perola M, Salomaa V, Shin SY, Spector TD, Sala C, Ridker PM, Kähönen M, Viikari J, Hengstenberg C, Nelson CP; CARDIoGRAM Consortium; DIAGRAM Consortium; ICBP Consortium; MAGIC Consortium, Meschia JF, Nalls MA, Sharma P, Singleton AB, Kamatani N, Zeller T, Burnier M, Attia J, Laan M, Klopp N, Hillege HL, Kloiber S, Choi H, Pirastu M, Tore S, Probst-Hensch NM, Völzke H, Gudnason V, Parsa A, Schmidt R, Whitfield JB, Fornage M, Gasparini P, Siscovick DS, Polašek O, Campbell H, Rudan I, Bouatia-Naji N, Metspalu A, Loos RJ, van Duijn CM, Borecki IB, Ferrucci L, Gambaro G, Deary IJ, Wolffenbuttel BH, Chambers JC, März W, Pramstaller PP, Snieder H, Gyllensten U, Wright AF, Navis G, Watkins H, Witteman JC, Sanna S, Schipf S, Dunlop MG, Tönjes A, Ripatti S, Soranzo N, Toniolo D, Chasman DI, Raitakari O, Kao WH, Ciullo M, Fox CS, Caulfield M, Bochud M, Gieger C. *Genome-wide association analyses identify 18 new loci associated with serum urate concentrations*. **Nat Genet.**2013 Feb;45(2):145-54. doi: 10.1038/ng.2500. 2-s2.0-84947899575

268. Faletra F, Snider K, Shyng SL, Bruno I, Athanasakis E, Gasparini P, Dionisi-Vici C, Ventura A, Zhou Q, Stanley CA, Burlina A. *Co-inheritance of two ABCC8 mutations causing an unresponsive congenital hyperinsulinism: clinical and functional characterization of two novel ABCC8 mutations*. **Gene**. 2013 Mar 1;516(1):122-5. doi: 10.1016/j.gene.2012.12.055. 2-s2.0-84873099173

269. Biino G, Santimone I, Minelli C, Sorice R, Frongia B, Traglia M, Ulivi S, Di Castelnuovo A, Gögele M, Nutile T, Francavilla M, Sala C, Pirastu N, Cerletti C, Iacoviello L, Gasparini P, Toniolo D, Ciullo M, Pramstaller P, Pirastu M, de Gaetano G, Balduini CL. *Age- and sex-related variations in platelet count in Italy: a proposal of reference ranges based on 40987 subjects' data*. **PLoS One**. 2013;8(1):e54289. doi: 10.1371/journal.pone.0054289. 2-s2.0-84873174474

270. Cousminer DL, Berry DJ, Timpson NJ, Ang W, Thiering E, Byrne EM, Taal HR, Huikari V, Bradfield JP, Kerkhof M, Groen-Blokhuis MM, Kreiner-Møller E, Marinelli M, Holst C, Leinonen JT, Perry JR, Surakka I, Pietiläinen O, Kettunen J, Anttila V, Kaakinen M, Sovio U, Pouta A, Das S, Lagou V, Power C, Prokopenko I, Evans DM, Kemp JP, St Pourcain B, Ring S, Palotie A, Kajantie E,

Osmond C, Lehtimäki T, Viikari JS, Kähönen M, Warrington NM, Lye SJ, Palmer LJ, Tiesler CM, Flexeder C, Montgomery GW, Medland SE, Hofman A, Hakonarson H, Guxens M, Bartels M, Salomaa V; ReproGen Consortium, Murabito JM, Kaprio J, Sørensen TI, Ballester F, Bisgaard H, Boomsma DI, Koppelman GH, Grant SF, Jaddoe VW, Martin NG, Heinrich J, Pennell CE, Raitakari OT, Eriksson JG, Smith GD, Hyppönen E, Järvelin MR, McCarthy MI, Ripatti S, Widén E; Early Growth Genetics (EGG) Consortium (incl. Gasparini P as collaborator). *Genome-wide association and longitudinal analyses reveal genetic loci linking pubertal height growth, pubertal timing and childhood adiposity*. **Hum Mol Genet**. 2013 Jul 1;22(13):2735-47. doi: 10.1093/hmg/ddt104. 2-s2.0-84878901283

271. Ulivi S, Mezzavilla M, Gasparini P. *Genetics of eye colours in different rural populations on the Silk Road.* **Eur J Hum Genet**. 2013 Nov;21(11):1320-3. doi: 10.1038/ejhg.2013.41. 2-s2.0-84885924674

272. Faletra F, Athanasakis E, Morgan A, Biarnés X, Fornasier F, Parini R, Furlan F, Boiani A, Maiorana A, Dionisi-Vici C, Giordano L, Burlina A, Ventura A, Gasparini P. *Congenital hyperinsulinism: Clinical and molecular analysis of a large Italian cohort*. **Gene**. 2013 Mar 15. doi:pii: S0378-1119(13)00255-2. 10.1016/j.gene.2013.03.021. 2-s2.0-84876719167

273. Nolan LS, Maier H, Hermans-Borgmeyer I, Girotto G, Ecob R, Pirastu N, Cadge BA, Hübner C, Gasparini P, Strachan DP, Davis A, Dawson SJ. *Estrogen-related receptor gamma and hearing function: evidence of a role in humans and mice*. **Neurobiol Aging**. 2013 Aug;34(8):2077.e1-9. doi:10.1016/j.neurobiolaging.2013.02.009. 2-s2.0-84877620653

274. Paul DS, Albers CA, Rendon A, Voss K, Stephens J; HaemGen Consortium, van der Harst P, Chambers JC, Soranzo N, Ouwehand WH, Deloukas P (incl. Gasparini P as collaborator). *Maps of open chromatin highlight cell type-restricted patterns of regulatory sequence variation at hematological trait loci*. **Genome Res**. 2013 Jul;23(7):1130-41. doi: 10.1101/gr.155127.113. 2-s2.0-84880080558

275. Marchiori A, Capece L, Giorgetti A, Gasparini P, Behrens M, Carloni P, Meyerhof W. *Coarse-grained/molecular mechanics of the TAS2R38 bitter taste receptor: experimentally-validated detailed structural prediction of agonist binding*. **PLoS One**. 2013 May 31;8(5):e64675. doi: 10.1371/journal.pone.0064675. Print 2013. 2-s2.0-84878530705

276. Vozzi D, Licastro D, Martelossi S, Athanasakis E, Gasparini P, Fabretto A. *Alagille Syndrome: A New Missense Mutation Detected by Whole-Exome Sequencing in a Case Previously Found to Be Negative by DHPLC and MLPA*. **Mol Syndromol.** 2013 Apr;4(4):207-10. doi: 10.1159/000347231. 2-s2.0-84877297238

277. Pistis G, Okonkwo SU, Traglia M, Sala C, Shin SY, Masciullo C, Buetti I, Massacane R, Mangino M, Thein SL, Spector TD, Ganesh S; CHARGE Consortium Hematology Working, Pirastu N, Gasparini P, Soranzo N, Camaschella C, Hart D, Green MR,

Toniolo D. *Genome wide association analysis of a founder population identified TAF3 as a gene for MCHC in humans*. **PLoS One**. 2013 Jul 31;8(7):e69206. doi: 10.1371/journal.pone.0069206. Print 2013. 2-s2.0-84881164024

278. Melchiorri D, Pani L, Gasparini P, Cossu G, Ancans J, Borg JJ, Drai C, Fiedor P, Flory E, Hudson I, Leufkens HG, Müller-Berghaus J, Narayanan G, Neugebauer B, Pokrotnieks J, Robert JL, Salmonson T, Schneider CK. *Regulatory evaluation of Glybera in Europe - two committees, one mission.* **Nat Rev Drug Discov.**2013 Sep;12(9):719. doi: 10.1038/nrd3835-c1. 2-s2.0-84883450737

279. Parsa A, Fuchsberger C, Köttgen A, O'Seaghdha CM, Pattaro C, de Andrade M, Chasman DI, Teumer A, Endlich K, Olden M, Chen MH, Tin A, Kim YJ, Taliun D, Li M, Feitosa M, Gorski M, Yang Q, Hundertmark C, Foster MC, Glazer N, Isaacs A, Rao M, Smith AV, O'Connell JR, Struchalin M, Tanaka T, Li G, Hwang SJ, Atkinson EJ, Lohman K, Cornelis MC, Johansson A, Tönjes A, Dehghan A, Couraki V, Holliday EG, Sorice R, Kutalik Z, Lehtimäki T, Esko T, Deshmukh H, Ulivi S, Chu AY, Murgia F, Trompet S, Imboden M, Kollerits B, Pistis G, Harris TB, Launer LJ, Aspelund T, Eiriksdottir G, Mitchell BD, Boerwinkle E, Schmidt H, Hofer E, Hu F, Demirkan A, Oostra BA, Turner ST, Ding J, Andrews JS, Freedman BI, Giulianini F, Koenig W, Illig T, Döring A, Wichmann HE, Zgaga L, Zemunik T, Boban M, Minelli C, Wheeler HE, Igl W, Zaboli G, Wild SH, Wright AF, Campbell H, Ellinghaus D, Nöthlings U, Jacobs G, Biffar R, Ernst F, Homuth G, Kroemer HK, Nauck M, Stracke S, Völker U, Völzke H, Kovacs P, Stumvoll M, Mägi R, Hofman A, Uitterlinden AG, Rivadeneira F, Aulchenko YS, Polasek O, Hastie N, Vitart V, Helmer C, Wang JJ, Stengel B, Ruggiero D, Bergmann S, Kähönen M, Viikari J, Nikopensius T, Province M, Colhoun H, Doney A, Robino A, Krämer BK, Portas L, Ford I, Buckley BM, Adam M, Thun GA, Paulweber B, Haun M, Sala C, Mitchell P, Ciullo M, Vollenweider P, Raitakari O, Metspalu A, Palmer C, Gasparini P, Pirastu M, Jukema JW, Probst-Hensch NM, Kronenberg F, Toniolo D, Gudnason V, Shuldiner AR, Coresh J, Schmidt R, Ferrucci L, van Duijn CM, Borecki I, Kardia SL, Liu Y, Curhan GC, Rudan I, Gyllensten U, Wilson JF, Franke A, Pramstaller PP, Rettig R, Prokopenko I, Witteman J, Hayward C, Ridker PM, Bochud M, Heid IM, Siscovick DS, Fox CS, Kao WL, Böger CA. *Common variants in Mendelian kidney disease genes and their association with renal function.* **J Am Soc Nephrol**. 2013 Dec;24(12):2105-17. doi: 10.1681/ASN.2012100983. 2-s2.0-84889074946

280. O'Seaghdha CM, Wu H, Yang Q, Kapur K, Guessous I, Zuber AM, Köttgen A, Stoudmann C, Teumer A, Kutalik Z, Mangino M, Dehghan A, Zhang W, Eiriksdottir G, Li G, Tanaka T, Portas L, Lopez LM, Hayward C, Lohman K, Matsuda K, Padmanabhan S, Firsov D, Sorice R, Ulivi S, Brockhaus AC, Kleber ME, Mahajan A, Ernst FD, Gudnason V, Launer LJ, Mace A, Boerwinckle E, Arking DE, Tanikawa C, Nakamura Y, Brown MJ, Gaspoz JM, Theler JM, Siscovick DS, Psaty BM, Bergmann S, Vollenweider P, Vitart V, Wright AF, Zemunik T, Boban M, Kolcic I, Navarro P, Brown EM,

Estrada K, Ding J, Harris TB, Bandinelli S, Hernandez D, Singleton AB, Girotto G, Ruggiero D, d'Adamo AP, Robino A, Meitinger T, Meisinger C, Davies G, Starr JM, Chambers JC, Boehm BO, Winkelmann BR, Huang J, Murgia F, Wild SH, Campbell H, Morris AP, Franco OH, Hofman A, Uitterlinden AG, Rivadeneira F, Völker U, Hannemann A, Biffar R, Hoffmann W, Shin SY, Lescuyer P, Henry H, Schurmann C; SUNLIGHT Consortium; GEFOS Consortium, Munroe PB, Gasparini P, Pirastu N, Ciullo M, Gieger C, März W, Lind L, Spector TD, Smith AV, Rudan I, Wilson JF, Polasek O, Deary IJ, Pirastu M, Ferrucci L, Liu Y, Kestenbaum B, Kooner JS, Witteman JC, Nauck M, Kao WH, Wallaschofski H, Bonny O, Fox CS, Bochud M. *Meta-analysis of genome-wide association studies identifies six new Loci for serum calcium concentrations.* **PLoS Genet**. 2013;9(9):e1003796. doi: 10.1371/journal.pgen.1003796. 2-s2.0-84884683733

281. Girotto G, Abdulhadi K, Buniello A, Vozzi D, Licastro D, d'Eustacchio A, Vuckovic D, Alkowari MK, Steel KP, Badii R, Gasparini P. *Linkage Study and Exome Sequencing Identify a BDP1 Mutation Associated with Hereditary Hearing Loss*. **PLoS One**. 2013 Dec 2;8(12):e80323. doi: 10.1371/journal.pone.0080323. eCollection 2013. 2-s2.0-84891781477

282. Athanasakis E, Licastro D, Faletra F, Fabretto A, Dipresa S, Vozzi D, Morgan A, d'Adamo AP, Pecile V, Biarnés X, Gasparini P. *Next generation sequencing in nonsyndromic intellectual disability: From a negative molecular karyotype to a possible causative mutation detection*. **Am J Med Genet A**. 2014 Jan;164(1):170-6. doi: 10.1002/ajmg.a.36274. 2-s2.0-84890792446

283. Faletra F, Girotto G, D'Adamo AP, Vozzi D, Morgan A, Gasparini P. *A novel P2RX2 mutation in an Italian family affected by autosomal dominant nonsyndromic hearing loss*. **Gene**. 2014 Jan 25;534(2):236-9. doi: 10.1016/j.gene.2013.10.052. 2-s2.0-84890428608

284. Faletra F, D'Adamo AP, Bruno I, Athanasakis E, Biskup S, Esposito L, Gasparini P. *Autosomal recessive stickler syndrome due to a loss of function mutation in the COL9A3 gene*. **Am J Med Genet A**. 2014 Jan;164(1):42-7. doi: 10.1002/ajmg.a.36165. 2-s2.0-84890792872

285. Perry JR, Hsu YH, Chasman DI, Johnson AD, Elks C, Albrecht E, Andrulis IL, Beesley J, Berenson GS, Bergmann S, Bojesen SE, Bolla MK, Brown J, Buring JE, Campbell H, Chang-Claude J, Chenevix-Trench G, Corre T, Couch FJ, Cox A, Czene K, D'adamo AP, Davies G, Deary IJ, Dennis J, Easton DF, Engelhardt EG, Eriksson JG, Esko T, Fasching PA, Figueroa JD, Flyger H, Fraser A, Garcia-Closas M, Gasparini P, Gieger C, Giles G, Guenel P, Hägg S, Hall P, Hayward C, Hopper J, Ingelsson E; kConFab investigators, Kardia SL, Kasiman K, Knight JA, Lahti J, Lawlor DA, Magnusson PK, Margolin S, Marsh JA, Metspalu A, Olson JE, Pennell CE, Polasek O, Rahman I, Ridker PM, Robino A, Rudan I, Rudolph A, Salumets A, Schmidt MK, Schoemaker MJ, Smith EN, Smith JA, Southey M, Stöckl D, Swerdlow AJ, Thompson DJ,

Truong T, Ulivi S, Waldenberger M, Wang Q, Wild S, Wilson JF, Wright AF, Zgaga L, Consortium R, Ong KK, Murabito JM, Karasik D, Murray A. *DNA mismatch repair gene MSH6 implicated in determining age at natural menopause*. **Hum Mol Genet**. 2014 May 1;23(9):2490-7. doi: 10.1093/hmg/ddt620. 2-s2.0-84897501936

286. Girotto G, Vuckovic D, Buniello A, Lorente-Cánovas B, Lewis M, Gasparini P, Steel KP. *Expression and replication studies to identify new candidate genes involved in normal hearing function*. **PLoS One**. 2014 Jan 14;9(1):e85352. doi: 10.1371/journal.pone.0085352. eCollection 2014 Jan 14. 2-s2.0-84897433140

287. Mezzavilla M, Vozzi D, Pirastu N, Girotto G, D Adamo P, Gasparini P, Colonna V. *Genetic landscape of populations along the Silk Road: admixture and migration patterns.* **BMC Genet**. 2014 Dec 5;15(1):131. 2-s2.0-84964314396

288. Perry JR, Day F, Elks CE, Sulem P, Thompson DJ, Ferreira T, He C, Chasman DI, Esko T, Thorleifsson G, Albrecht E, Ang WQ, Corre T, Cousminer DL, Feenstra B, Franceschini N, Ganna A, Johnson AD, Kjellqvist S, Lunetta KL, McMahon G, Nolte IM, Paternoster L, Porcu E, Smith AV, Stolk L, Teumer A, Tšernikova N, Tikkanen E, Ulivi S, Wagner EK, Amin N, Bierut LJ, Byrne EM, Hottenga JJ, Koller DL, Mangino M, Pers TH, Yerges-Armstrong LM, Hua Zhao J, Andrulis IL, Anton-Culver H, Atsma F, Bandinelli S, Beckmann MW, Benitez J, Blomqvist C, Bojesen SE, Bolla MK, Bonanni B, Brauch H, Brenner H, Buring JE, Chang-Claude J, Chanock S, Chen J, Chenevix-Trench G, Collée JM, Couch FJ, Couper D, Coviello AD, Cox A, Czene K, D'adamo AP, Davey Smith G, De Vivo I, Demerath EW, Dennis J, Devilee P, Dieffenbach AK, Dunning AM, Eiriksdottir G, Eriksson JG, Fasching PA, Ferrucci L, Flesch-Janys D, Flyger H, Foroud T, Franke L, Garcia ME, García-Closas M, Geller F, de Geus EE, Giles GG, Gudbjartsson DF, Gudnason V, Guénel P, Guo S, Hall P, Hamann U, Haring R, Hartman CA, Heath AC, Hofman A, Hooning MJ, Hopper JL, Hu FB, Hunter DJ, Karasik D, Kiel DP, Knight JA, Kosma VM, Kutalik Z, Lai S, Lambrechts D, Lindblom A, Mägi R, Magnusson PK, Mannermaa A, Martin NG, Masson G, McArdle PF, McArdle WL, Melbye M, Michailidou K, Mihailov E, Milani L, Milne RL, Nevanlinna H, Neven P, Nohr EA, Oldehinkel AJ, Oostra BA, Palotie A, Peacock M, Pedersen NL, Peterlongo P, Peto J, Pharoah PD, Postma DS, Pouta A, Pylkäs K, Radice P, Ring S, Rivadeneira F, Robino A, Rose LM, Rudolph A, Salomaa V, Sanna S, Schlessinger D, Schmidt MK, Southey MC, Sovio U, Stampfer MJ, Stöckl D, Storniolo AM, Timpson NJ, Tyrer J, Visser JA, Vollenweider P, Völzke H, Waeber G, Waldenberger M, Wallaschofski H, Wang Q, Willemsen G, Winqvist R, Wolffenbuttel BH, Wright MJ; Australian Ovarian Cancer Study; GENICA Network; kConFab; LifeLines Cohort Study; InterAct Consortium; Early Growth Genetics (EGG) Consortium, Boomsma DI, Econs MJ, Khaw KT, Loos RJ, McCarthy MI, Montgomery GW, Rice JP, Streeten EA, Thorsteinsdottir U, van Duijn CM, Alizadeh BZ, Bergmann S, Boerwinkle E, Boyd HA, Crisponi L, Gasparini P,

Gieger C, Harris TB, Ingelsson E, Järvelin MR, Kraft P, Lawlor D, Metspalu A, Pennell CE, Ridker PM, Snieder H, Sørensen TI, Spector TD, Strachan DP, Uitterlinden AG, Wareham NJ, Widen E, Zygmunt M, Murray A, Easton DF, Stefansson K, Murabito JM, Ong KK. *Parent-of-origin-specific allelic associations among 106 genomic loci for age at menarche.* **Nature**. 2014 Oct 2;514(7520):92-7. doi: 10.1038/nature13545. 2-s2.0-84908024873

289. Vuckovic D, Dallapiccola B, Franzè A, Mauri L, Perrone MD, Gasparini P. *Connexin 26 variant carriers have a better gastrointestinal health: is this the heterozygote advantage?* **Eur J Hum Genet.**2014 Aug 6. doi: 10.1038/ejhg.2014.151.

290. Wolber LE, Girotto G, Buniello A, Vuckovic D, Pirastu N, Lorente-Cánovas B, Rudan I, Hayward C, Polasek O, Ciullo M, Mangino M, Steves C, Concas MP, Cocca M, Spector TD, Gasparini P, Steel KP, Williams FM. *Salt-inducible kinase 3, SIK3, is a new gene associated with hearing.* **Hum Mol Genet**. 2014 Dec 1;23(23):6407-18. doi: 10.1093/hmg/ddu346. 2-s2.0-84938758651

291. Girotto G, Mezzavilla M, Abdulhadi K, Vuckovic D, Vozzi D, Khalifa Alkowari M, Gasparini P, Badii R. *Consanguinity and hereditary hearing loss in Qatar.* **Hum Hered.**2014;77(1-4):175-82. doi: 10.1159/000360475. 2-s2.0-84904885909

292. Arking DE, Pulit SL, Crotti L, van der Harst P, Munroe PB, Koopmann TT, Sotoodehnia N, Rossin EJ, Morley M, Wang X, Johnson AD, Lundby A, Gudbjartsson DF, Noseworthy PA, Eijgelsheim M, Bradford Y, Tarasov KV, Dörr M, Müller-Nurasyid M, Lahtinen AM, Nolte IM, Smith AV, Bis JC, Isaacs A, Newhouse SJ, Evans DS, Post WS, Waggott D, Lyytikäinen LP, Hicks AA, Eisele L, Ellinghaus D, Hayward C, Navarro P, Ulivi S, Tanaka T, Tester DJ, Chatel S, Gustafsson S, Kumari M, Morris RW, Naluai ÅT, Padmanabhan S, Kluttig A, Strohmer B, Panayiotou AG, Torres M, Knoflach M, Hubacek JA, Slowikowski K, Raychaudhuri S, Kumar RD, Harris TB, Launer LJ, Shuldiner AR, Alonso A, Bader JS, Ehret G, Huang H, Kao WH, Strait JB, Macfarlane PW, Brown M, Caulfield MJ, Samani NJ, Kronenberg F, Willeit J; CARe Consortium; COGENT Consortium, Smith JG, Greiser KH, Meyer Zu Schwabedissen H, Werdan K, Carella M, Zelante L, Heckbert SR, Psaty BM, Rotter JI, Kolcic I, Polašek O, Wright AF, Griffin M, Daly MJ; DCCT/EDIC, Arnar DO, Hólm H, Thorsteinsdottir U; eMERGE Consortium, Denny JC, Roden DM, Zuvich RL, Emilsson V, Plump AS, Larson MG, O'Donnell CJ, Yin X, Bobbo M, D'Adamo AP, Iorio A, Sinagra G, Carracedo A, Cummings SR, Nalls MA, Jula A, Kontula KK, Marjamaa A, Oikarinen L, Perola M, Porthan K, Erbel R, Hoffmann P, Jöckel KH, Kälsch H, Nöthen MM; HRGEN Consortium, den Hoed M, Loos RJ, Thelle DS, Gieger C, Meitinger T, Perz S, Peters A, Prucha H, Sinner MF, Waldenberger M, de Boer RA, Franke L, van der Vleuten PA, Beckmann BM, Martens E, Bardai A, Hofman N, Wilde AA, Behr ER, Dalageorgou C, Giudicessi JR, Medeiros-Domingo A, Barc J, Kyndt F, Probst V, Ghidoni A, Insolia R, Hamilton RM, Scherer SW, Brandimarto J, Margulies K, Moravec CE, del Greco M F, Fuchsberger C,

O'Connell JR, Lee WK, Watt GC, Campbell H, Wild SH, El Mokhtari NE, Frey N, Asselbergs FW, Mateo Leach I, Navis G, van den Berg MP, van Veldhuisen DJ, Kellis M, Krijthe BP, Franco OH, Hofman A, Kors JA, Uitterlinden AG, Witteman JC, Kedenko L, Lamina C, Oostra BA, Abecasis GR, Lakatta EG, Mulas A, Orrú M, Schlessinger D, Uda M, Markus MR, Völker U, Snieder H, Spector TD, Ärnlöv J, Lind L, Sundström J, Syvänen AC, Kivimaki M, Kähönen M, Mononen N, Raitakari OT, Viikari JS, Adamkova V, Kiechl S, Brion M, Nicolaides AN, Paulweber B, Haerting J, Dominiczak AF, Nyberg F, Whincup PH, Hingorani AD, Schott JJ, Bezzina CR, Ingelsson E, Ferrucci L, Gasparini P, Wilson JF, Rudan I, Franke A, Mühleisen TW, Pramstaller PP, Lehtimäki TJ, Paterson AD, Parsa A, Liu Y, van Duijn CM, Siscovick DS, Gudnason V, Jamshidi Y, Salomaa V, Felix SB, Sanna S, Ritchie MD, Stricker BH, Stefansson K, Boyer LA, Cappola TP, Olsen JV, Lage K, Schwartz PJ, Kääb S, Chakravarti A, Ackerman MJ, Pfeufer A, de Bakker PI, Newton-Cheh C. *Genetic association study of QT interval highlights role for calcium signaling pathways in myocardial repolarization.* **Nat Genet.**2014 Aug;46(8):826-36. doi: 10.1038/ng.3014. 2-s2.0-84905594041

293. Rubinato E, Morgan A, D'Eustacchio A, Pecile V, Gortani G, Gasparini P, Faletra F. *A novel deletion mutation involving TMEM38B in a patient with autosomal recessive osteogenesis imperfecta.* **Gene**. 2014 Jul 25;545(2):290-2. doi: 10.1016/j.gene.2014.05.028. 2-s2.0-84901633705

294. O'Connell J, Gurdasani D, Delaneau O, Pirastu N, Ulivi S, Cocca M, Traglia M, Huang J, Huffman JE, Rudan I, McQuillan R, Fraser RM, Campbell H, Polasek O, Asiki G, Ekoru K, Hayward C, Wright AF, Vitart V, Navarro P, Zagury JF, Wilson JF, Toniolo D, Gasparini P, Soranzo N, Sandhu MS, Marchini J. *A general approach for haplotype phasing across the full spectrum of relatedness.* **PLoS Genet.**2014 Apr 17;10(4):e1004234. doi: 10.1371/journal.pgen.1004234. eCollection 2014 Apr. 2-s2.0-84901370399

295. Mezzavilla M, Iorio A, Bobbo M, D'Eustacchio A, Merlo M, Gasparini P, Ulivi S, Sinagra G. *Insight into genetic determinants of resting heart rate.* **Gene**. 2014 Jul 15;545(1):170-4. doi: 10.1016/j.gene.2014.03.045. 2-s2.0-84901200114

296. Vozzi D, Morgan A, Vuckovic D, D'Eustacchio A, Abdulhadi K, Rubinato E, Badii R, Gasparini P, Girotto G. *Hereditary hearing loss: a 96 gene targeted sequencing protocol reveals novel alleles in a series of Italian and Qatari patients.* **Gene**. 2014 Jun 1;542(2):209-16. doi: 10.1016/j.gene.2014.03.033. 2-s2.0-84898781283

297. Pirastu N, Kooyman M, Traglia M, Robino A, Willems SM, Pistis G, d'Adamo P, Amin N, d'Eustacchio A, Navarini L, Sala C, Karssen LC, van Duijn C, Toniolo D, Gasparini P. *Association analysis of bitter receptor genes in five isolated populations identifies a significant correlation between TAS2R43 variants and coffee liking.* **PLoS One**. 2014 Mar 19;9(3):e92065. doi: 10.1371/journal.pone.0092065. eCollection 2014. 2-s2.0-

84898656773

298. Robino A, Mezzavilla M, Pirastu N, Dognini M, Tepper BJ, Gasparini P. *A population-based approach to study the impact of PROP perception on food liking in populations along the Silk Road.* **PLoS One**. 2014 Mar 13;9(3):e91716. doi: 10.1371/journal.pone.0091716. eCollection 2014. 2-s2.0-84897999085

299. Olden M, Corre T, Hayward C, Toniolo D, Ulivi S, Gasparini P, Pistis G, Hwang SJ, Bergmann S, Campbell H, Cocca M, Gandin I, Girotto G, Glaudemans B, Hastie ND, Loffing J, Polasek O, Rampoldi L, Rudan I, Sala C, Traglia M, Vollenweider P, Vuckovic D, Youhanna S, Weber J, Wright AF, Kutalik Z, Bochud M, Fox CS, Devuyst O. *Common variants in UMOD associate with urinary uromodulin levels: a meta-analysis.* **J Am Soc Nephrol.**2014 Aug;25(8):1869-82. doi: 10.1681/ASN.2013070781. 2-s2.0-84910148385

300. Wolf NI, Vanderver A, van Spaendonk RM, Schiffmann R, Brais B, Bugiani M, Sistermans E, Catsman-Berrevoets C, Kros JM, Pinto PS, Pohl D, Tirupathi S, Strømme P, de Grauw T, Fribourg S, Demos M, Pizzino A, Naidu S, Guerrero K, van der Knaap MS, Bernard G; 4H Research Group(incl. Gasparini P as collaborator). *Clinical spectrum of 4H leukodystrophy caused by POLR3A and POLR3B mutations.* **Neurology**.2014 Nov 18;83(21):1898-905. doi: 10.1212/WNL.0000000000001002. 2-s2.0-84961290480

301. Vuckovic D, Dawson S, Scheffer DI, Rantanen T, Morgan A, Di Stazio M, Vozzi D, Nutile T, Concas MP, Biino G, Nolan L, Bahl A, Loukola A, Viljanen A, Davis A, Ciullo M, Corey DP, Pirastu M, Gasparini P, Girotto G. *Genome-wide association analysis on normal hearing function identifies PCDH20 and SLC28A3 as candidates for hearing function and loss*. **Hum Mol Genet**. 2015 Jul 17. pii: ddv279. 2-s2.0-84943762980

302. Joshi PK, Esko T, Mattsson H, Eklund N, Gandin I, Nutile T, Jackson AU, Schurmann C, Smith AV, Zhang W, Okada Y, Stančáková A, Faul JD, Zhao W, Bartz TM, Concas MP, Franceschini N, Enroth S, Vitart V, Trompet S, Guo X, Chasman DI, O'Connel JR, Corre T, Nongmaithem SS, Chen Y, Mangino M, Ruggiero D, Traglia M, Farmaki AE, Kacprowski T, Bjonnes A, van der Spek A, Wu Y, Giri AK, Yanek LR, Wang L, Hofer E, Rietveld CA, McLeod O, Cornelis MC, Pattaro C, Verweij N, Baumbach C, Abdellaoui A, Warren HR, Vuckovic D, Mei H, Bouchard C, Perry JR, Cappellani S, Mirza SS, Benton MC, Broeckel U, Medland SE, Lind PA, Malerba G, Drong A, Yengo L, Bielak LF, Zhi D, van der Most PJ, Shriner D, Mägi R, Hemani G, Karaderi T, Wang Z, Liu T, Demuth I, Zhao JH, Meng W, Lataniotis L, van der Laan SW, Bradfield JP, Wood AR, Bonnefond A, Ahluwalia TS, Hall LM, Salvi E, Yazar S, Carstensen L, de Haan HG, Abney M, Afzal U, Allison MA, Amin N, Asselbergs FW, Bakker SJ, Barr RG, Baumeister SE, Benjamin DJ, Bergmann S, Boerwinkle E, Bottinger EP, Campbell A, Chakravarti A, Chan Y, Chanock SJ, Chen C, Chen YD, Collins FS, Connell J, Correa A, Cupples LA, Smith GD, Davies G, Dörr M, Ehret G, Ellis SB, Feenstra B, Feitosa MF, Ford I, Fox CS, Frayling

TM, Friedrich N, Geller F, Scotland G, Gillham-Nasenya I, Gottesman O, Graff M, Grodstein F, Gu C, Haley C, Hammond CJ, Harris SE, Harris TB, Hastie ND, Heard-Costa NL, Heikkilä K, Hocking LJ, Homuth G, Hottenga JJ, Huang J, Huffman JE, Hysi PG, Ikram MA, Ingelsson E, Joensuu A, Johansson Å, Jousilahti P, Jukema JW, Kähönen M, Kamatani Y, Kanoni S, Kerr SM, Khan NM, Koellinger P, Koistinen HA, Kooner MK, Kubo M, Kuusisto J, Lahti J, Launer LJ, Lea RA, Lehne B, Lehtimäki T, Liewald DC, Lind L, Loh M, Lokki ML, London SJ, Loomis SJ, Loukola A, Lu Y, Lumley T, Lundqvist A, Männistö S, Marques-Vidal P, Masciullo C, Matchan A, Mathias RA, Matsuda K, Meigs JB, Meisinger C, Meitinger T, Menni C, Mentch FD, Mihailov E, Milani L, Montasser ME, Montgomery GW, Morrison A, Myers RH, Nadukuru R, Navarro P, Nelis M, Nieminen MS, Nolte IM, O'Connor GT, Ogunniyi A, Padmanabhan S, Palmas WR, Pankow JS, Patarcic I, Pavani F, Peyser PA, Pietilainen K, Poulter N, Prokopenko I, Ralhan S, Redmond P, Rich SS, Rissanen H, Robino A, Rose LM, Rose R, Sala C, Salako B, Salomaa V, Sarin AP, Saxena R, Schmidt H, Scott LJ, Scott WR, Sennblad B, Seshadri S, Sever P, Shrestha S, Smith BH, Smith JA, Soranzo N, Sotoodehnia N, Southam L, Stanton AV, Stathopoulou MG, Strauch K, Strawbridge RJ, Suderman MJ, Tandon N, Tang ST, Taylor KD, Tayo BO, Töglhofer AM, Tomaszewski M, Tšernikova N, Tuomilehto J, Uitterlinden AG, Vaidya D, van Hylckama Vlieg A, van Setten J, Vasankari T, Vedantam S, Vlachopoulou E, Vozzi D, Vuoksimaa E, Waldenberger M, Ware EB, Wentworth-Shields W, Whitfield JB, Wild S, Willemsen G, Yajnik CS, Yao J, Zaza G, Zhu X; BioBank Japan Project, Salem RM, Melbye M, Bisgaard H, Samani NJ, Cusi D, Mackey DA, Cooper RS, Froguel P, Pasterkamp G, Grant SF, Hakonarson H, Ferrucci L, Scott RA, Morris AD, Palmer CN, Dedoussis G, Deloukas P, Bertram L, Lindenberger U, Berndt SI, Lindgren CM, Timpson NJ, Tönjes A, Munroe PB, Sørensen TI, Rotimi CN, Arnett DK, Oldehinkel AJ, Kardia SL, Balkau B, Gambaro G, Morris AP, Eriksson JG, Wright MJ, Martin NG, Hunt SC, Starr JM, Deary IJ, Griffiths LR, Tiemeier H, Pirastu N, Kaprio J, Wareham NJ, Pérusse L, Wilson JG, Girotto G, Caulfield MJ, Raitakari O, Boomsma DI, Gieger C, van der Harst P, Hicks AA, Kraft P, Sinisalo J, Knekt P, Johannesson M, Magnusson PK, Hamsten A, Schmidt R, Borecki IB, Vartiainen E, Becker DM, Bharadwaj D, Mohlke KL, Boehnke M, van Duijn CM, Sanghera DK, Teumer A, Zeggini E, Metspalu A, Gasparini P, Ulivi S, Ober C, Toniolo D, Rudan I, Porteous DJ, Ciullo M, Spector TD, Hayward C, Dupuis J, Loos RJ, Wright AF, Chandak GR, Vollenweider P, Shuldiner AR, Ridker PM, Rotter JI, Sattar N, Gyllensten U, North KE, Pirastu M, Psaty BM, Weir DR, Laakso M, Gudnason V, Takahashi A, Chambers JC, Kooner JS, Strachan DP, Campbell H, Hirschhorn JN, Perola M, Polašek O, Wilson JF. *Directional dominance on stature and cognition in diverse human populations.* **Nature**. 2015 Jul 23;523(7561):459-62. doi: 10.1038/nature14618. 2-s2.0-84937889764

303. Vuckovic D, Gasparini P, Soranzo N, Iotchkova V. *MultiMeta: an R package for meta-analyzing multi-phenotype genome-wide*

association studies. **Bioinformatics**. 2015 Apr 22. pii: btv222. 2-s2.0-84939534434

304. Bertok S, Žerjav Tanšek M, Kotnik P, Battelino T, Volk M, Pecile V, Cleva L, Gasparini P, Kovač J, Hovnik T. *Clinical and Molecular Cytogenetic Characterisation of Children with Developmental Delay and Dysmorphic Features*. **Slovenian Journal of Public Health.** Published online 2015 Mar 13. doi:10.1515/sjph-2015-0010. 2-s2.0-84991393466

305. Mezzavilla M, Ulivi S, Bianca ML, Carlino D, Gasparini P, Robino A. *Analysis of functional variants reveals new candidate genes associated with alexithymia*. **Psychiatry Res**. 2015 Jun 30;227(2-3):363-5. doi: 10.1016/j.psychres.2015.03.018. 2-s2.0-84929131728

306. Iossa S, Costa V, Corvino V, Auletta G, Barruffo L, Cappellani S, Ceglia C, Cennamo G, D'Adamo AP, D'Amico A, Di Paolo N, Forte R, Gasparini P, Laria C, Lombardo B, Malesci R, Vitale A, Marciano E, Franzè A. *Phenotypic and genetic characterization of a family carrying two Xq21.1-21.3 interstitial deletions associated with syndromic hearing loss*. **Mol Cytogenet.**2015 Mar 20;8:18. doi: 10.1186/s13039-015-0120-0. eCollection 2015. 2-s2.0-84961300589

307. Huffman JE, Albrecht E, Teumer A, Mangino M, Kapur K, Johnson T, Kutalik Z, Pirastu N, Pistis G, Lopez LM, Haller T, Salo P, Goel A, Li M, Tanaka T, Dehghan A, Ruggiero D, Malerba G, Smith AV, Nolte IM, Portas L, Phipps-Green A, Boteva L, Navarro P, Johansson A, Hicks AA, Polasek O, Esko T, Peden JF, Harris SE, Murgia F, Wild SH, Tenesa A, Tin A, Mihailov E, Grotevendt A, Gislason GK, Coresh J, D'Adamo P, Ulivi S, Vollenweider P, Waeber G, Campbell S, Kolcic I, Fisher K, Viigimaa M, Metter JE, Masciullo C, Trabetti E, Bombieri C, Sorice R, Döring A, Reischl E, Strauch K, Hofman A, Uitterlinden AG, Waldenberger M, Wichmann HE, Davies G, Gow AJ, Dalbeth N, Stamp L, Smit JH, Kirin M, Nagaraja R, Nauck M, Schurmann C, Budde K, Farrington SM, Theodoratou E, Jula A, Salomaa V, Sala C, Hengstenberg C, Burnier M, Mägi R, Klopp N, Kloiber S, Schipf S, Ripatti S, Cabras S, Soranzo N, Homuth G, Nutile T, Munroe PB, Hastie N, Campbell H, Rudan I, Cabrera C, Haley C, Franco OH, Merriman TR, Gudnason V, Pirastu M, Penninx BW, Snieder H, Metspalu A, Ciullo M, Pramstaller PP, van Duijn CM, Ferrucci L, Gambaro G, Deary IJ, Dunlop MG, Wilson JF, Gasparini P, Gyllensten U, Spector TD, Wright AF, Hayward C, Watkins H, Perola M, Bochud M, Kao WH, Caulfield M, Toniolo D, Völzke H, Gieger C, Köttgen A, Vitart V. *Modulation of genetic associations with serum urate levels by body-mass-index in humans*. **PLoS One**. 2015 Mar 26;10(3):e0119752. doi: 10.1371/journal.pone.0119752. eCollection 2015. 2-s2.0-84926688627

308. Rusconi D, Negri G, Colapietro P, Picinelli C, Milani D, Spena S, Magnani C, Silengo MC, Sorasio L, Curtisova V, Cavaliere ML, Prontera P, Stangoni G, Ferrero GB, Biamino E, Fischetto R, Piccione M, Gasparini P, Salviati L, Selicorni A, Finelli P, Larizza L, Gervasini C. *Characterization of 14 novel deletions underlying Rubinstein-Taybi syndrome: an update of the CREBBP deletion*

repertoire. **Hum Genet.** 2015 Jun;134(6):613-26. doi: 10.1007/s00439-015-1542-9. 2-s2.0-84936758346

309. Pirastu N, Kooyman M, Traglia M, Robino A, Willems SM, Pistis G, Amin N, Sala C, Karssen LC, van Duijn CM, Toniolo D, Gasparini P. *Genome-wide association analysis on five isolated populations identifies variants of the HLA-DOA gene associated with white wine liking*. **Eur J Hum Genet.** 2015 Mar 11. doi: 10.1038/ejhg.2015.34. 2-s2.0-84948715641
310. Mezzavilla M, Vozzi D, Badii R, Alkowari MK, Abdulhadi K, Girotto G, Gasparini P. *Increased rate of deleterious variants in long runs of homozygosity of an inbred population from Qatar*. **Hum Hered**. 2015;79(1):14-9. 2-s2.0-84924092013
311. Locke AE, Kahali B, Berndt SI, Justice AE, Pers TH, Day FR, Powell C, Vedantam S, Buchkovich ML, Yang J, Croteau-Chonka DC, Esko T, Fall T, Ferreira T, Gustafsson S, Kutalik Z, Luan J, Mägi R, Randall JC, Winkler TW, Wood AR, Workalemahu T, Faul JD, Smith JA, Hua Zhao J, Zhao W, Chen J, Fehrmann R, Hedman ÅK, Karjalainen J, Schmidt EM, Absher D, Amin N, Anderson D, Beekman M, Bolton JL, Bragg-Gresham JL, Buyske S, Demirkan A, Deng G, Ehret GB, Feenstra B, Feitosa MF, Fischer K, Goel A, Gong J, Jackson AU, Kanoni S, Kleber ME, Kristiansson K, Lim U, Lotay V, Mangino M, Mateo Leach I, Medina-Gomez C, Medland SE, Nalls MA, Palmer CD, Pasko D, Pechlivanis S, Peters MJ, Prokopenko I, Shungin D, Stančáková A, Strawbridge RJ, Ju Sung Y, Tanaka T, Teumer A, Trompet S, van der Laan SW, van Setten J, Van Vliet-Ostaptchouk JV, Wang Z, Yengo L, Zhang W, Isaacs A, Albrecht E, Ärnlöv J, Arscott GM, Attwood AP, Bandinelli S, Barrett A, Bas IN, Bellis C, Bennett AJ, Berne C, Blagieva R, Blüher M, Böhringer S, Bonnycastle LL, Böttcher Y, Boyd HA, Bruinenberg M, Caspersen IH, Ida Chen YD, Clarke R, Daw EW, de Craen AJ, Delgado G, Dimitriou M, Doney AS, Eklund N, Estrada K, Eury E, Folkersen L, Fraser RM, Garcia ME, Geller F, Giedraitis V, Gigante B, Go AS, Golay A, Goodall AH, Gordon SD, Gorski M, Grabe HJ, Grallert H, Grammer TB, Gräßler J, Grönberg H, Groves CJ, Gusto G, Haessler J, Hall P, Haller T, Hallmans G, Hartman CA, Hassinen M, Hayward C, Heard-Costa NL, Helmer Q, Hengstenberg C, Holmen O, Hottenga JJ, James AL, Jeff JM, Johansson Å, Jolley J, Juliusdottir T, Kinnunen L, Koenig W, Koskenvuo M, Kratzer W, Laitinen J, Lamina C, Leander K, Lee NR, Lichtner P, Lind L, Lindström J, Sin Lo K, Lobbens S, Lorbeer R, Lu Y, Mach F, Magnusson PK, Mahajan A, McArdle WL, McLachlan S, Menni C, Merger S, Mihailov E, Milani L, Moayyeri A, Monda KL, Morken MA, Mulas A, Müller G, Müller-Nurasyid M, Musk AW, Nagaraja R, Nöthen MM, Nolte IM, Pilz S, Rayner NW, Renstrom F, Rettig R, Ried JS, Ripke S, Robertson NR, Rose LM, Sanna S, Scharnagl H, Scholtens S, Schumacher FR, Scott WR, Seufferlein T, Shi J, Vernon Smith A, Smolonska J, Stanton AV, Steinthorsdottir V, Stirrups K, Stringham HM, Sundström J, Swertz MA, Swift AJ, Syvänen AC, Tan ST, Tayo BO, Thorand B, Thorleifsson G, Tyrer JP, Uh HW, Vandenput L, Verhulst FC, Vermeulen SH, Verweij N, Vonk JM, Waite LL, Warren HR, Waterworth D, Weedon MN, Wilkens LR, Willenborg C, Wilsgaard

T, Wojczynski MK, Wong A, Wright AF, Zhang Q; LifeLines Cohort Study, Brennan EP, Choi M, Dastani Z, Drong AW, Eriksson P, Franco-Cereceda A, Gådin JR, Gharavi AG, Goddard ME, Handsaker RE, Huang J, Karpe F, Kathiresan S, Keildson S, Kiryluk K, Kubo M, Lee JY, Liang L, Lifton RP, Ma B, McCarroll SA, McKnight AJ, Min JL, Moffatt MF, Montgomery GW, Murabito JM, Nicholson G, Nyholt DR, Okada Y, Perry JR, Dorajoo R, Reinmaa E, Salem RM, Sandholm N, Scott RA, Stolk L, Takahashi A, Tanaka T, Van't Hooft FM, Vinkhuyzen AA, Westra HJ, Zheng W, Zondervan KT; ADIPOGen Consortium; AGEN-BMI Working Group; CARDIOGRAMplusC4D Consortium; CKDGen Consortium; GLGC; ICBP; MAGIC Investigators; MuTHER Consortium; MIGen Consortium; PAGE Consortium; ReproGen Consortium; GENIE Consortium; International Endogene Consortium, Heath AC, Arveiler D, Bakker SJ, Beilby J, Bergman RN, Blangero J, Bovet P, Campbell H, Caulfield MJ, Cesana G, Chakravarti A, Chasman DI, Chines PS, Collins FS, Crawford DC, Cupples LA, Cusi D, Danesh J, de Faire U, den Ruijter HM, Dominiczak AF, Erbel R, Erdmann J, Eriksson JG, Farrall M, Felix SB, Ferrannini E, Ferrières J, Ford I, Forouhi NG, Forrester T, Franco OH, Gansevoort RT, Gejman PV, Gieger C, Gottesman O, Gudnason V, Gyllensten U, Hall AS, Harris TB, Hattersley AT, Hicks AA, Hindorff LA, Hingorani AD, Hofman A, Homuth G, Hovingh GK, Humphries SE, Hunt SC, Hyppönen E, Illig T, Jacobs KB, Jarvelin MR, Jöckel KH, Johansen B, Jousilahti P, Jukema JW, Jula AM, Kaprio J, Kastelein JJ, Keinanen-Kiukaanniemi SM, Kiemeney LA, Knekt P, Kooner JS, Kooperberg C, Kovacs P, Kraja AT, Kumari M, Kuusisto J, Lakka TA, Langenberg C, Le Marchand L, Lehtimäki T, Lyssenko V, Männistö S, Marette A, Matise TC, McKenzie CA, McKnight B, Moll FL, Morris AD, Morris AP, Murray JC, Nelis M, Ohlsson C, Oldehinkel AJ, Ong KK, Madden PA, Pasterkamp G, Peden JF, Peters A, Postma DS, Pramstaller PP, Price JF, Qi L, Raitakari OT, Rankinen T, Rao DC, Rice TK, Ridker PM, Rioux JD, Ritchie MD, Rudan I, Salomaa V, Samani NJ, Saramies J, Sarzynski MA, Schunkert H, Schwarz PE, Sever P, Shuldiner AR, Sinisalo J, Stolk RP, Strauch K, Tönjes A, Trégouët DA, Tremblay A, Tremoli E, Virtamo J, Vohl MC, Völker U, Waeber G, Willemsen G, Witteman JC, Zillikens MC, Adair LS, Amouyel P, Asselbergs FW, Assimes TL, Bochud M, Boehm BO, Boerwinkle E, Bornstein SR, Bottinger EP, Bouchard C, Cauchi S, Chambers JC, Chanock SJ, Cooper RS, de Bakker PI, Dedoussis G, Ferrucci L, Franks PW, Froguel P, Groop LC, Haiman CA, Hamsten A, Hui J, Hunter DJ, Hveem K, Kaplan RC, Kivimaki M, Kuh D, Laakso M, Liu Y, Martin NG, März W, Melbye M, Metspalu A, Moebus S, Munroe PB, Njølstad I, Oostra BA, Palmer CN, Pedersen NL, Perola M, Pérusse L, Peters U, Power C, Quertermous T, Rauramaa R, Rivadeneira F, Saaristo TE, Saleheen D, Sattar N, Schadt EE, Schlessinger D, Slagboom PE, Snieder H, Spector TD, Thorsteinsdottir U, Stumvoll M, Tuomilehto J, Uitterlinden AG, Uusitupa M, van der Harst P, Walker M, Wallaschofski H, Wareham NJ, Watkins H, Weir DR, Wichmann HE, Wilson JF, Zanen P, Borecki IB, Deloukas P, Fox CS, Heid IM, O'Connell JR, Strachan DP, Stefansson K, van Duijn

CM, Abecasis GR, Franke L, Frayling TM, McCarthy MI, Visscher PM, Scherag A, Willer CJ, Boehnke M, Mohlke KL, Lindgren CM, Beckmann JS, Barroso I, North KE, Ingelsson E, Hirschhorn JN, Loos RJ, Speliotes EK (incl. Gasparini P as collaborator). *Genetic studies of body mass index yield new insights for obesity biology.* **Nature**. 2015 Feb 12;518(7538):197-206. doi: 10.1038/nature14177. 2-s2.0-84923171580

312. Shungin D, Winkler TW, Croteau-Chonka DC, Ferreira T, Locke AE, Mägi R, Strawbridge RJ, Pers TH, Fischer K, Justice AE, Workalemahu T, Wu JM, Buchkovich ML, Heard-Costa NL, Roman TS, Drong AW, Song C, Gustafsson S, Day FR, Esko T, Fall T, Kutalik Z, Luan J, Randall JC, Scherag A, Vedantam S, Wood AR, Chen J, Fehrmann R, Karjalainen J, Kahali B, Liu CT, Schmidt EM, Absher D, Amin N, Anderson D, Beekman M, Bragg-Gresham JL, Buyske S, Demirkan A, Ehret GB, Feitosa MF, Goel A, Jackson AU, Johnson T, Kleber ME, Kristiansson K, Mangino M, Mateo Leach I, Medina-Gomez C, Palmer CD, Pasko D, Pechlivanis S, Peters MJ, Prokopenko I, Stančáková A, Ju Sung Y, Tanaka T, Teumer A, Van Vliet-Ostaptchouk JV, Yengo L, Zhang W, Albrecht E, Ärnlöv J, Arscott GM, Bandinelli S, Barrett A, Bellis C, Bennett AJ, Berne C, Blüher M, Böhringer S, Bonnet F, Böttcher Y, Bruinenberg M, Carba DB, Caspersen IH, Clarke R, Daw EW, Deelen J, Deelman E, Delgado G, Doney AS, Eklund N, Erdos MR, Estrada K, Eury E, Friedrich N, Garcia ME, Giedraitis V, Gigante B, Go AS, Golay A, Grallert H, Grammer TB, Gräßler J, Grewal J, Groves CJ, Haller T, Hallmans G, Hartman CA, Hassinen M, Hayward C, Heikkilä K, Herzig KH, Helmer Q, Hillege HL, Holmen O, Hunt SC, Isaacs A, Ittermann T, James AL, Johansson I, Juliusdottir T, Kalafati IP, Kinnunen L, Koenig W, Kooner IK, Kratzer W, Lamina C, Leander K, Lee NR, Lichtner P, Lind L, Lindström J, Lobbens S, Lorentzon M, Mach F, Magnusson PK, Mahajan A, McArdle WL, Menni C, Merger S, Mihailov E, Milani L, Mills R, Moayyeri A, Monda KL, Mooijaart SP, Mühleisen TW, Mulas A, Müller G, Müller-Nurasyid M, Nagaraja R, Nalls MA, Narisu N, Glorioso N, Nolte IM, Olden M, Rayner NW, Renstrom F, Ried JS, Robertson NR, Rose LM, Sanna S, Scharnagl H, Scholtens S, Sennblad B, Seufferlein T, Sitlani CM, Vernon Smith A, Stirrups K, Stringham HM, Sundström J, Swertz MA, Swift AJ, Syvänen AC, Tayo BO, Thorand B, Thorleifsson G, Tomaschitz A, Troffa C, van Oort FV, Verweij N, Vonk JM, Waite LL, Wennauer R, Wilsgaard T, Wojczynski MK, Wong A, Zhang Q, Hua Zhao J, Brennan EP, Choi M, Eriksson P, Folkersen L, Franco-Cereceda A, Gharavi AG, Hedman ÅK, Hivert MF, Huang J, Kanoni S, Karpe F, Keildson S, Kiryluk K, Liang L, Lifton RP, Ma B, McKnight AJ, McPherson R, Metspalu A, Min JL, Moffatt MF, Montgomery GW, Murabito JM, Nicholson G, Nyholt DR, Olsson C, Perry JR, Reinmaa E, Salem RM, Sandholm N, Schadt EE, Scott RA, Stolk L, Vallejo EE, Westra HJ, Zondervan KT; ADIPOGen Consortium; CARDIOGRAMplusC4D Consortium; CKDGen Consortium; GEFOS Consortium; GENIE Consortium; GLGC; ICBP; International Endogene Consortium; LifeLines Cohort Study; MAGIC Investigators; MuTHER Consortium; PAGE Consortium; ReproGen

Consortium, Amouyel P, Arveiler D, Bakker SJ, Beilby J, Bergman RN, Blangero J, Brown MJ, Burnier M, Campbell H, Chakravarti A, Chines PS, Claudi-Boehm S, Collins FS, Crawford DC, Danesh J, de Faire U, de Geus EJ, Dörr M, Erbel R, Eriksson JG, Farrall M, Ferrannini E, Ferrières J, Forouhi NG, Forrester T, Franco OH, Gansevoort RT, Gieger C, Gudnason V, Haiman CA, Harris TB, Hattersley AT, Heliövaara M, Hicks AA, Hingorani AD, Hoffmann W, Hofman A, Homuth G, Humphries SE, Hyppönen E, Illig T, Jarvelin MR, Johansen B, Jousilahti P, Jula AM, Kaprio J, Kee F, Keinanen-Kiukaanniemi SM, Kooner JS, Kooperberg C, Kovacs P, Kraja AT, Kumari M, Kuulasmaa K, Kuusisto J, Lakka TA, Langenberg C, Le Marchand L, Lehtimäki T, Lyssenko V, Männistö S, Marette A, Matise TC, McKenzie CA, McKnight B, Musk AW, Möhlenkamp S, Morris AD, Nelis M, Ohlsson C, Oldehinkel AJ, Ong KK, Palmer LJ, Penninx BW, Peters A, Pramstaller PP, Raitakari OT, Rankinen T, Rao DC, Rice TK, Ridker PM, Ritchie MD, Rudan I, Salomaa V, Samani NJ, Saramies J, Sarzynski MA, Schwarz PE, Shuldiner AR, Staessen JA, Steinthorsdottir V, Stolk RP, Strauch K, Tönjes A, Tremblay A, Tremoli E, Vohl MC, Völker U, Vollenweider P, Wilson JF, Witteman JC, Adair LS, Bochud M, Boehm BO, Bornstein SR, Bouchard C, Cauchi S, Caulfield MJ, Chambers JC, Chasman DI, Cooper RS, Dedoussis G, Ferrucci L, Froguel P, Grabe HJ, Hamsten A, Hui J, Hveem K, Jöckel KH, Kivimaki M, Kuh D, Laakso M, Liu Y, März W, Munroe PB, Njølstad I, Oostra BA, Palmer CN, Pedersen NL, Perola M, Pérusse L, Peters U, Power C, Quertermous T, Rauramaa R, Rivadeneira F, Saaristo TE, Saleheen D, Sinisalo J, Slagboom PE, Snieder H, Spector TD, Thorsteinsdottir U, Stumvoll M, Tuomilehto J, Uitterlinden AG, Uusitupa M, van der Harst P, Veronesi G, Walker M, Wareham NJ, Watkins H, Wichmann HE, Abecasis GR, Assimes TL, Berndt SI, Boehnke M, Borecki IB, Deloukas P, Franke L, Frayling TM, Groop LC, Hunter DJ, Kaplan RC, O'Connell JR, Qi L, Schlessinger D, Strachan DP, Stefansson K, van Duijn CM, Willer CJ, Visscher PM, Yang J, Hirschhorn JN, Zillikens MC, McCarthy MI, Speliotes EK, North KE, Fox CS, Barroso I, Franks PW, Ingelsson E, Heid IM, Loos RJ, Cupples LA, Morris AP, Lindgren CM, Mohlke KL (incl. Gasparini P as collaborator). *New genetic loci link adipose and insulin biology to body fat distribution.* **Nature**. 2015 Feb 12;518(7538):187-96. doi: 10.1038/nature14132. 2-s2.0-84923169934

313. Cenedese V, Mezzavilla M, Morgan A, Marino R, Ettorre CP, Margaglione M, Gasparini P, Menini A. *Assessment of the olfactory function in Italian patients with type 3 von Willebrand disease caused by a homozygous 253 Kb deletion involving VWF and TMEM16B/ANO2*. **PLoS One**. 2015 Jan 30;10(1):e0116483. doi: 10.1371/journal.pone.0116483. eCollection 2015. 2-s2.0-84922149803

314. Zonta F, Girotto G, Buratto D, Crispino G, Morgan A, Abdulhadi K, Alkowari M, Badii R, Gasparini P, Mammano F. *The p.Cys169Tyr variant of connexin 26 is not a polymorphism*. **Hum Mol Genet**. 2015 May 1;24(9):2641-8. doi: 10.1093/hmg/ddv026. 2-s2.0-84938845904

315. Day FR, Ruth KS, Thompson DJ, Lunetta KL, Pervjakova N, Chasman DI, Stolk L, Finucane HK, Sulem P, Bulik-Sullivan B, Esko T, Johnson AD, Elks CE, Franceschini N, He C, Altmaier E, Brody JA, Franke LL, Huffman JE, Keller MF, McArdle PF, Nutile T, Porcu E, Robino A, Rose LM, Schick UM, Smith JA, Teumer A, Traglia M, Vuckovic D, Yao J, Zhao W, Albrecht E, Amin N, Corre T, Hottenga JJ, Mangino M, Smith AV, Tanaka T, Abecasis GR, Andrulis IL, Anton-Culver H, Antoniou AC, Arndt V, Arnold AM, Barbieri C, Beckmann MW, Beeghly-Fadiel A, Benitez J, Bernstein L, Bielinski SJ, Blomqvist C, Boerwinkle E, Bogdanova NV, Bojesen SE, Bolla MK, Borresen-Dale AL, Boutin TS, Brauch H, Brenner H, Brüning T, Burwinkel B, Campbell A, Campbell H, Chanock SJ, Chapman JR, Chen YI, Chenevix-Trench G, Couch FJ, Coviello AD, Cox A, Czene K, Darabi H, De Vivo I, Demerath EW, Dennis J, Devilee P, Dörk T, Dos-Santos-Silva I, Dunning AM, Eicher JD, Fasching PA, Faul JD, Figueroa J, Flesch-Janys D, Gandin I, Garcia ME, García-Closas M, Giles GG, Girotto GG, Goldberg MS, González-Neira A, Goodarzi MO, Grove ML, Gudbjartsson DF, Guénel P, Guo X, Haiman CA, Hall P, Hamann U, Henderson BE, Hocking LJ, Hofman A, Homuth G, Hooning MJ, Hopper JL, Hu FB, Huang J, Humphreys K, Hunter DJ, Jakubowska A, Jones SE, Kabisch M, Karasik D, Knight JA, Kolcic I, Kooperberg C, Kosma VM, Kriebel J, Kristensen V, Lambrechts D, Langenberg C, Li J, Li X, Lindström S, Liu Y, Luan J, Lubinski J, Mägi R, Mannermaa A, Manz J, Margolin S, Marten J, Martin NG, Masciullo C, Meindl A, Michailidou K, Mihailov E, Milani L, Milne RL, Müller-Nurasyid M, Nalls M, Neale BM, Nevanlinna H, Neven P, Newman AB, Nordestgaard BG, Olson JE, Padmanabhan S, Peterlongo P, Peters U, Petersmann A, Peto J, Pharoah PD, Pirastu NN, Pirie A, Pistis G, Polasek O, Porteous D, Psaty BM, Pylkäs K, Radice P, Raffel LJ, Rivadeneira F, Rudan I, Rudolph A, Ruggiero D, Sala CF, Sanna S, Sawyer EJ, Schlessinger D, Schmidt MK, Schmidt F, Schmutzler RK, Schoemaker MJ, Scott RA, Seynaeve CM, Simard J, Sorice R, Southey MC, Stöckl D, Strauch K, Swerdlow A, Taylor KD, Thorsteinsdottir U, Toland AE, Tomlinson I, Truong T, Tryggvadottir L, Turner ST, Vozzi D, Wang Q, Wellons M, Willemsen G, Wilson JF, Winqvist R, Wolffenbuttel BB, Wright AF, Yannoukakos D, Zemunik T, Zheng W, Zygmunt M, Bergmann S, Boomsma DI, Buring JE, Ferrucci L, Montgomery GW, Gudnason V, Spector TD, van Duijn CM, Alizadeh BZ, Ciullo M, Crisponi L, Easton DF, Gasparini PP, Gieger C, Harris TB, Hayward C, Kardia SL, Kraft P, McKnight B, Metspalu A, Morrison AC, Reiner AP, Ridker PM, Rotter JI, Toniolo D, Uitterlinden AG, Ulivi S, Völzke H, Wareham NJ, Weir DR, Yerges-Armstrong LM; PRACTICAL Consortium; kConFab Investigators; AOCS Investigators; Generation Scotland; EPIC-InterAct Consortium; LifeLines Cohort Study, Price AL, Stefansson K, Visser JA, Ong KK, Chang-Claude J, Murabito JM, Perry JR, Murray A. *Large-scale genomic analyses link reproductive aging to hypothalamic signaling, breast cancer susceptibility and BRCA1-mediated DNA repair*. **Nat Genet**. 2015 Sep 28. doi: 10.1038/ng.3412. 2-s2.0-85000348717

316. Huang J, Howie B, McCarthy S, Memari Y, Walter K, Min JL, Danecek P, Malerba G, Trabetti E, Zheng HF; UK10K Consortium, Gambaro G, Richards JB, Durbin R, Timpson NJ, Marchini J, Soranzo N. *Improved imputation of low-frequency and rare variants using the UK10K haplotype reference panel*. **Nat Commun.**2015 Sep 14;6:8111. doi: 10.1038/ncomms9111. 2-s2.0-84941702459

317. Sidore C, Busonero F, Maschio A, Porcu E, Naitza S, Zoledziewska M, Mulas A, Pistis G, Steri M, Danjou F, Kwong A, Ortega Del Vecchyo VD, Chiang CW, Bragg-Gresham J, Pitzalis M, Nagaraja R, Tarrier B, Brennan C, Uzzau S, Fuchsberger C, Atzeni R, Reinier F, Berutti R, Huang J, Timpson NJ, Toniolo D, Gasparini P, Malerba G, Dedoussis G, Zeggini E, Soranzo N, Jones C, Lyons R, Angius A, Kang HM, Novembre J, Sanna S, Schlessinger D, Cucca F, Abecasis GR. *Genome sequencing elucidates Sardinian genetic architecture and augments association analyses for lipid and blood inflammatory markers*. **Nat Genet.**2015 Sep 14. doi: 10.1038/ng.3368. 2-s2.0-84945370533

318. Robinson MR, Hemani G, Medina-Gomez C, Mezzavilla M, Esko T, Shakhbazov K, Powell JE, Vinkhuyzen A, Berndt SI, Gustafsson S, Justice AE, Kahali B, Locke AE, Pers TH, Vedantam S, Wood AR, van Rheenen W, Andreassen OA, Gasparini P, Metspalu A, Berg LH, Veldink JH, Rivadeneira F, Werge TM, Abecasis GR, Boomsma DI, Chasman DI, de Geus EJ, Frayling TM, Hirschhorn JN, Hottenga JJ, Ingelsson E, Loos RJ, Magnusson PK, Martin NG, Montgomery GW, North KE, Pedersen NL, Spector TD, Speliotes EK, Goddard ME, Yang J, Visscher PM. *Population genetic differentiation of height and body mass index across Europe*. **Nat Genet**. 2015 Sep 14. doi: 10.1038/ng.3401. 2-s2.0-84945484144

319. Robino A, Bevilacqua L, Pirastu N, Situlin R, Di Lenarda R, Gasparini P, Navarra CO. *Polymorphisms in sweet taste genes (TAS1R2 and GLUT2), sweet liking, and dental caries prevalence in an adult Italian population.* **Genes Nutr.**2015 Sep;10(5):485. doi: 10.1007/s12263-015-0485-z.

320. Martin HC, Christ R, Hussin JG, O'Connell J, Gordon S, Mbarek H, Hottenga JJ, McAloney K, Willemsen G, Gasparini P, Pirastu N, Montgomery GW, Navarro P, Soranzo N, Toniolo D, Vitart V, Wilson JF, Marchini J, Boomsma DI, Martin NG, Donnelly P. *Multicohort analysis of the maternal age effect on recombination*. **Nat Commun**. 2015 Aug 5;6:7846. doi: 10.1038/ncomms8846. 2-s2.0-84938904285

321. UK10K Consortium, Walter K, Min JL, Huang J, Crooks L, Memari Y, McCarthy S, Perry JR, Xu C, Futema M, Lawson D, Iotchkova V, Schiffels S, Hendricks AE, Danecek P, Li R, Floyd J, Wain LV, Barroso I, Humphries SE, Hurles ME, Zeggini E, Barrett JC, Plagnol V, Richards JB, Greenwood CM, Timpson NJ, Durbin R, Soranzo N. *The UK10K project identifies rare variants in health and disease*. **Nature**. 2015 Oct 1;526(7571):82-90. doi: 10.1038/nature14962. 2-s2.0-84943182742

322. Carlino D, Francavilla R, Baj G, Kulak K, d'Adamo P, Ulivi S, Cappellani S, Gasparini P, Tongiorgi E. *Brain-derived*

neurotrophic factor serum levels in genetically isolated populations: gender-specific association with anxiety disorder subtypes but not with anxiety levels or Val66Met polymorphism. **PeerJ.** 2015 Oct 29;3:e1252. doi: 10.7717/peerj.1252. eCollection 2015. 2-s2.0-84946196370

323. Girotto G, Scheffer DI, Morgan A, Vozzi D, Rubinato E, Di Stazio M, Muzzi E, Pensiero S, Giersch AB, Corey DP, Gasparini P. *PSIP1/LEDGF: a new gene likely involved in sensorineural progressive hearing loss.* **Sci Rep.** 2015 Dec 22;5:18568. Doi: 10.1038/srep18568. 2-s2.0-84951756451
324. Haber M, Mezzavilla M, Xue Y, Comas D, Gasparini P, Zalloua P, Tyler-Smith C. *Genetic evidence for an origin of the Armenians from Bronze Age mixing of multiple populations*. **Eur J Hum Genet**. 2016 Jun;24(6):931-6. doi: 10.1038/ejhg.2015.206. 2-s2.0-84944929433
325. d'Apolito M, Pisanelli D, Faletra F, Giardino I, Gigante M, Pettoello-Mantovani M, Goulet O. Gasparini P, Campanozzi A. *Genetic analysis of Italian patients with congenital tufting enteropathy*.**World J Pediatr**. 2016 May;12(2):219-24. doi: 10.1007/s12519-015-0070-y. 2-s2.0-84961063561
326. Pattaro C, Teumer A, Gorski M, Chu AY, Li M, Mijatovic V, Garnaas M, Tin A, Sorice R, Li Y, Taliun D, Olden M, Foster M, Yang Q, Chen MH, Pers TH, Johnson AD, Ko YA, Fuchsberger C, Tayo B, Nalls M, Feitosa MF, Isaacs A, Dehghan A, d'Adamo P, Adeyemo A, Dieffenbach AK, Zonderman AB, Nolte IM, van der Most PJ, Wright AF, Shuldiner AR, Morrison AC, Hofman A, Smith AV, Dreisbach AW, Franke A, Uitterlinden AG, Metspalu A, Tonjes A, Lupo A, Robino A, Johansson Å, Demirkan A, Kollerits B, Freedman BI, Ponte B, Oostra BA, Paulweber B, Krämer BK, Mitchell BD, Buckley BM, Peralta CA, Hayward C, Helmer C, Rotimi CN, Shaffer CM, Müller C, Sala C, van Duijn CM, Saint-Pierre A, Ackermann D, Shriner D, Ruggiero D, Toniolo D, Lu Y, Cusi D, Czamara D, Ellinghaus D, Siscovick DS, Ruderfer D, Gieger C, Grallert H, Rochtchina E, Atkinson EJ, Holliday EG, Boerwinkle E, Salvi E, Bottinger EP, Murgia F, Rivadeneira F, Ernst F, Kronenberg F, Hu FB, Navis GJ, Curhan GC, Ehret GB, Homuth G, Coassin S, Thun GA, Pistis G, Gambaro G, Malerba G, Montgomery GW, Eiriksdottir G, Jacobs G, Li G, Wichmann HE, Campbell H, Schmidt H, Wallaschofski H, Völzke H, Brenner H, Kroemer HK, Kramer H, Lin H, Leach IM, Ford I, Guessous I, Rudan I, Prokopenko I, Borecki I, Heid IM, Kolcic I, Persico I, Jukema JW, Wilson JF, Felix JF, Divers J, Lambert JC, Stafford JM, Gaspoz JM, Smith JA, Faul JD, Wang JJ, Ding J, Hirschhorn JN, Attia J, Whitfield JB, Chalmers J, Viikari J, Coresh J, Denny JC, Karjalainen J, Fernandes JK, Endlich K, Butterbach K, Keene KL, Lohman K, Portas L, Launer LJ, Lyytikäinen LP, Yengo L, Franke L, Ferrucci L, Rose LM, Kedenko L, Rao M, Struchalin M, Kleber ME, Cavalieri M, Haun M, Cornelis MC, Ciullo M, Pirastu M, de Andrade M, McEvoy MA, Woodward M, Adam M, Cocca M, Nauck M, Imboden M, Waldenberger M, Pruijm M, Metzger M, Stumvoll M, Evans MK, Sale MM, Kähönen M, Boban M, Bochud M, Rheinberger M, Verweij N, Bouatia-Naji N, Martin NG, Hastie

N, Probst-Hensch N, Soranzo N, Devuyst O, Raitakari O, Gottesman O, Franco OH, Polasek O, Gasparini P, Munroe PB, Ridker PM, Mitchell P, Muntner P, Meisinger C, Smit JH; ICBP Consortium; AGEN Consortium; CARDIOGRAM; CHARGe-Heart Failure Group; ECHOGen Consortium, Kovacs P, Wild PS, Froguel P, Rettig R, Mägi R, Biffar R, Schmidt R, Middelberg RP, Carroll RJ, Penninx BW, Scott RJ, Katz R, Sedaghat S, Wild SH, Kardia SL, Ulivi S, Hwang SJ, Enroth S, Kloiber S, Trompet S, Stengel B, Hancock SJ, Turner ST, Rosas SE, Stracke S, Harris TB, Zeller T, Zemunik T, Lehtimäki T, Illig T, Aspelund T, Nikopensius T, Esko T, Tanaka T, Gyllensten U, Völker U, Emilsson V, Vitart V, Aalto V, Gudnason V, Chouraki V, Chen WM, Igl W, März W, Koenig W, Lieb W, Loos RJ, Liu Y, Snieder H, Pramstaller PP, Parsa A, O'Connell JR, Susztak K, Hamet P, Tremblay J, de Boer IH, Böger CA, Goessling W, Chasman DI, Köttgen A, Kao WH, Fox CS. *Genetic associations at 53 loci highlight cell types and biological pathways relevant for kidney function*. **Nat Commun**. 2016 Jan 21;7:10023. doi: 10.1038/ncomms10023. 2-s2.0-84957605211

327. Cocca M, Bedognetti D, La Bianca M, Gasparini P, Girotto G. *Pharmacogenetics driving personalized medicine: analysis of genetic polymorphisms related to breast cancer medications in Italian isolated populations.* **J Transl Med**. 2016 Jan 22;14:22. doi: 10.1186/s12967-016-0778-z. 2-s2.0-84955749660

328. Robino A, Mezzavilla M, Pirastu N, La Bianca M, Gasparini P, Carlino D, Tepper BJ. *Understanding the role of personality and alexithymia in food preferences and PROP taste perception.* **Physiol Behav**. 2016 Apr 1;157:72-8. doi: 10.1016/j.physbeh.2016.01.022. 2-s2.0-84956626500

329. Okbay A, Beauchamp JP, Fontana MA, Lee JJ, Pers TH, Rietveld CA, Turley P, Chen GB, Emilsson V, Meddens SF, Oskarsson S, Pickrell JK, Thom K, Timshel P, de Vlaming R, Abdellaoui A, Ahluwalia TS, Bacelis J, Baumbach C, Bjornsdottir G, Brandsma JH, Pina Concas M, Derringer J, Furlotte NA, Galesloot TE, Girotto G, Gupta R, Hall LM, Harris SE, Hofer E, Horikoshi M, Huffman JE, Kaasik K, Kalafati IP, Karlsson R, Kong A, Lahti J, van der Lee SJ, deLeeuw C, Lind PA, Lindgren KO, Liu T, Mangino M, Marten J, Mihailov E, Miller MB, van der Most PJ, Oldmeadow C, Payton A, Pervjakova N, Peyrot WJ, Qian Y, Raitakari O, Rueedi R, Salvi E, Schmidt B, Schraut KE, Shi J, Smith AV, Poot RA, St Pourcain B, Teumer A, Thorleifsson G, Verweij N, Vuckovic D, Wellmann J, Westra HJ, Yang J, Zhao W, Zhu Z, Alizadeh BZ, Amin N, Bakshi A, Baumeister SE, Biino G, Bønnelykke K, Boyle PA, Campbell H, Cappuccio FP, Davies G, De Neve JE, Deloukas P, Demuth I, Ding J, Eibich P, Eisele L, Eklund N, Evans DM, Faul JD, Feitosa MF, Forstner AJ, Gandin I, Gunnarsson B, Halldórsson BV, Harris TB, Heath AC, Hocking LJ, Holliday EG, Homuth G, Horan MA, Hottenga JJ, de Jager PL, Joshi PK, Jugessur A, Kaakinen MA, Kähönen M, Kanoni S, Keltigangas-Järvinen L, Kiemeney LA, Kolcic I, Koskinen S, Kraja AT, Kroh M, Kutalik Z, Latvala A, Launer LJ, Lebreton MP, Levinson DF, Lichtenstein P, Lichtner P, Liewald DC; LifeLines Cohort Study, Loukola A, Madden PA, Mägi R, Mäki-Opas T,

Marioni RE, Marques-Vidal P, Meddens GA, McMahon G, Meisinger C, Meitinger T, Milaneschi Y, Milani L, Montgomery GW, Myhre R, Nelson CP, Nyholt DR, Ollier WE, Palotie A, Paternoster L, Pedersen NL, Petrovic KE, Porteous DJ, Räikkönen K, Ring SM, Robino A, Rostapshova O, Rudan I, Rustichini A, Salomaa V, Sanders AR, Sarin AP, Schmidt H, Scott RJ, Smith BH, Smith JA, Staessen JA, Steinhagen-Thiessen E, Strauch K, Terracciano A, Tobin MD, Ulivi S, Vaccargiu S, Quaye L, van Rooij FJ, Venturini C, Vinkhuyzen AA, Völker U, Völzke H, Vonk JM, Vozzi D, Waage J, Ware EB, Willemsen G, Attia JR, Bennett DA, Berger K, Bertram L, Bisgaard H, Boomsma DI, Borecki IB, Bültmann U, Chabris CF, Cucca F, Cusi D, Deary IJ, Dedoussis GV, van Duijn CM, Eriksson JG, Franke B, Franke L, Gasparini P, Gejman PV, Gieger C, Grabe HJ, Gratten J, Groenen PJ, Gudnason V, van der Harst P, Hayward C, Hinds DA, Hoffmann W, Hyppönen E, Iacono WG, Jacobsson B, Järvelin MR, Jöckel KH, Kaprio J, Kardia SL, Lehtimäki T, Lehrer SF, Magnusson PK, Martin NG, McGue M, Metspalu A, Pendleton N, Penninx BW, Perola M, Pirastu N, Pirastu M, Polasek O, Posthuma D, Power C, Province MA, Samani NJ, Schlessinger D, Schmidt R, Sørensen TI, Spector TD, Stefansson K, Thorsteinsdottir U, Thurik AR, Timpson NJ, Tiemeier H, Tung JY, Uitterlinden AG, Vitart V, Vollenweider P, Weir DR, Wilson JF, Wright AF, Conley DC, Krueger RF, Davey Smith G, Hofman A, Laibson DI, Medland SE, Meyer MN, Yang J, Johannesson M, Visscher PM, Esko T, Koellinger PD, Cesarini D, Benjamin DJ. *Genome-wide association study identifies 74 loci associated with educational attainment*. **Nature**. 2016 May 11;533(7604):539-42. doi: 10.1038/nature17671. 2-s2.0-84969760786

330. Hinney A, Kesselmeier M, Jall S, Volckmar AL, Föcker M, Antel J; GCAN; WTCCC3, Heid IM, Winkler TW; GIANT, Grant SF; EGG, Guo Y, Bergen AW, Kaye W, Berrettini W, Hakonarson H; Price Foundation Collaborative Group; Children's Hospital of Philadelphia/Price Foundation, Herpertz-Dahlmann B, de Zwaan M, Herzog W, Ehrlich S, Zipfel S, Egberts KM, Adan R, Brandys M, van Elburg A, Boraska, Perica V, Franklin CS, Tschöp MH, Zeggini E, Bulik CM, Collier D,Scherag A, Müller TD, Hebebrand J. Perica VB, Franklin CS, Floyd JA, Thornton LM, Huckins LM, Southam L, Rayner NW, Tachmazidou I, Klump KL, Treasure J, Lewis CM, Schmidt U, Tozzi F, Iezebrink K, Hebebrand J, Gorwood P, Adan RA, Kas MJ, Favaro A, Santonastaso P, Fernández-Aranda F, Gratacos M, Rybakowski F, Dmitrzak-Weglarz M, Kaprio J, Keski-Rahkonen A, Raevuori-Helkamaa A, Van Furth EF, Slof-Op't Landt MC, Hudson JI, Reichborn-Kjennerud T, Knudsen GP, Monteleone P, Kaplan AS, Karwautz A, Hakonarson H, Berrettini WH, Guo Y, Li D, Schork NJ, Komaki G, Ando T, Inoko H, Esko T, Fischer K, Männik K, Metspalu A, Baker JH, Cone RD, Dackor J, DeSocio JE, Hilliard CE, O'Toole JK, Pantel J, Szatkiewicz JP, Taico C, Zerwas S, Trace SE, Davis OS, Helder S, Bühren K, Burghardt R, de Zwaan M, Egberts K, Ehrlich S, Herpertz-Dahlmann B, Herzog W, Imgart H,Scherag A, Scherag S, Zipfel S, Boni

C, Ramoz N, Versini A, Brandys MK, Danner UN, de Kove C, Hendriks J, Koeleman BP, Ophoff RA,Strengman E, van Elburg AA, Bruson A, Clementi M, Degortes D, Forzan M, Tenconi E, Docampo E, Escaramí G, Jiménez-Murcia S, Lissowska J,Rajewski A, Szeszenia-Dabrowska N, Slopien A, Hauser J, Karhunen L, Meulenbelt I, Slagboom PE, Tortorella A, Maj M, Dedoussis G, Dikeos D,Gonidakis F, Tziouvas K, Tsitsika A, Papezova H, Slachtova L, Martaskova D, Kennedy JL, Levitan RD, Yilmaz Z, Huemer J, Koubek D, Merl E,Wagner G, Lichtenstein P, Breen G, Cohen-Woods S, Farmer A, McGuffin P, Cichon S, Giegling I, Herms S, Rujescu D, Schreiber S, Wichmann HE, Dina C, Sladek R, Gambaro G, Soranzo N, Julia A, Marsal S, Rabionet R, Gaborieau V, Dick DM, Palotie A, Ripatti S, Widén E, Andreassen OA, Espeseth T, Lundervold A, Reinvang I, Steen VM, Hellard SL, Mattingsdal M, Ntalla I, Bencko V, Foretova L, Janout V, Navratilova M,Gallinger S, Pinto D, Scherer SW, Aschauer H, Carlberg L, Schosser A, Alfredsson L, Ding B, Klareskog L, Padyukov L, Finan C, Kalsi G, Roberts M, Logan DW, Peltonen L, Ritchie GR, Barrett JC, Estivill X, Hinney A, Sullivan PF, Collier DA, Zeggini E, Bulik CM, Anderson CA, Barrett JC,Floyd JA, Franklin CS, McGinnis R, Soranzo N, Zeggini E, Sambrook J, Stephens J, Ouwehand WH, McArdle WL, Ring SM, Strachan DP,Alexander G, Bulik CM, Collier DA, Conlon PJ, Dominiczak A, Duncanson A, Hill A, Langford C, Lord G, Maxwell AP, Morgan L, Peltonen L,Sandford RN, Sheerin N, Vannberg FO, Blackburn H, Chen WM, Edkins S, Gillman M, Gray E, Hunt SE, Nengut-Gumuscu S, Potter S, Rich SS,Simpkin D, Whittaker P, de Bakker P, Bültmann U, Geleijnse M, Harst PV, Koppelman G, Rosmalen JG, van Rossum L, Smidt H, Swertz MA, Stolk RP, Alizadeh B, de Boer R, Boezen HM, Bruinenberg M, Franke L, van der Harst P, Hillege H, van der Klauw M, Navis G, Ormel J, Postma D,Rosmalen J, Slaets J, Snieder H, Stolk R, Wolffenbuttel B, Wijmenga C, Berg J, Blackwood D, Campbell H, Cavanagh J, Connell J, Connor M,Cunningham-Burley S, Deary I, Dominiczak A, Ellis P, FitzPatrick B, Ford I, Gertz R, Grau A, Haddow G, Jackson C, Kerr S, Lindsay R, McGilchrist M, McIntyre D, Morris A, Morton R, Muir W, Murray G, Palmer C, Pell J, Philp A, Porteous D, Porteous M, Procter R, Ralston S, Reid D, Sinnott R,Smith B, Clair DS, Sullivan F, Sweetland M, Ure J, Watt G, Wolf R, Wright A, Berndt SI, Gustafsson S, Mägi R, Ganna A, Wheeler E, Feitosa MF,Justice AE, Monda KL, Croteau-Chonka DC, Day FR, Esko T, Fall T, Ferreira T, Gentilini D, Jackson AU, Luan J, Randall JC, Vedantam S, Willer CJ, Winkler TW, Wood AR, Workalemahu T, Hu YJ, Lee SH, Liang L, Lin DY, Min JL, Neale BM, Thorleifsson G, Yang J, Albrecht E, Amin N,Bragg-Gresham JL, Cadby G, den Heijer M, Eklund N, Fischer K, Goel A, Hottenga JJ, Huffman JE, Jarick I, Johansson A, Johnson T, Kanoni S,Kleber ME, König IR, Kristiansson K, Kutalik Z, Lamina C, Lecoeur C, Li G, Mangino M, McArdle WL, Medina-Gomez C, Müller-Nurasyid M, Ngwa JS, Nolte IM, Paternoster L, Pechlivanis S, Perola M, Peters

MJ, Preuss M, Rose LM, Shi J, Shungin D, Smith AV, Strawbridge RJ, Surakka I,Teumer A, Trip MD, Tyrer J, Van Vliet-Ostaptchouk JV, Vandenput L, Waite LL, Zhao JH, Absher D, Asselbergs FW, Atalay M, Attwood AP,Balmforth AJ, Basart H, Beilby J, Bonnycastle LL, Brambilla P, Bruinenberg M, Campbell H, Chasman DI, Chines PS, Collins FS, Connell JM,Cookson W, de Faire U, de Vegt F, Dei M, Dimitriou M, Edkins S, Estrada K, Evans DM, Farrall M, Ferrario MM, Ferrières J, Franke L, Frau F,Gejman PV, Grallert H, Grönberg H, Gudnason V, Hall AS, Hall P, Hartikainen AL, Hayward C, Heard-Costa NL, Heath AC, Hebebrand J, Homuth G, Hu FB, Hunt SE, Hyppönen E, Iribarren C, Jacobs KB, Jansson JO, Jula A, Kähönen M, Kathiresan S, Kee F, Khaw KT, Kivimaki M, Koenig W,Kraja AT, Kumari M, Kuulasmaa K, Kuusisto J, Laitinen JH, Lakka TA, Langenberg C, Launer LJ, Lind L, Lindström J, Liu J, Liuzzi A, Lokki ML,Lorentzon M, Madden PA, Magnusson PK, Manunta P, Marek D, März W, Mateo Leach I, McKnight B, Medland SE, Mihailov E, Milani L,Montgomery GW, Mooser V, Mühleisen TW, Munroe PB, Musk AW, Narisu N, Navis G, Nicholson G, Nohr EA, Ong KK, Oostra BA, Palmer CN,Palotie A, Peden JF, Pedersen N, Peters A, Polasek O, Pouta A, Pramstaller PP, Prokopenko I, Pütter C, Radhakrishnan A, Raitakari O, Rendon A, Rivadeneira F, Rudan I, Saaristo TE, Sambrook JG, Sanders AR, Sanna S, Saramies J, Schipf S, Schreiber S, Schunkert H, Shin SY, Signorini S, Sinisalo J, Skrobek B, Soranzo N, Stancakova A, Stark K, Stephens JC, Stirrups K, Stolk RP, Stumvoll M, Swift AJ, Theodoraki EV, Thorand B,Tregouet DA, Tremoli E, Van der Klauw MM, van Meurs JB, Vermeulen SH, Viikari J, Virtamo J, Vitart V, Waeber G, Wang Z, Widen E, Wild SH,Willemsen G, Winkelmann BR, Witteman JC, Wolffenbuttel BH, Wong A, Wright AF, Zillikens M, Amouyel P, Boehm BO, Boerwinkle E, Boomsma DI, Caulfield MJ, Chanock SJ, Cupples L, Cusi D, Dedoussis GV, Erdmann J, Eriksson JG, Franks PW, Froguel P, Gieger C, Gyllensten U,Hamsten A, Harris TB, Hengstenberg C, Hicks AA, Hingorani A, Hinney A, Hofman A, Hovingh KG, Hveem K, Illig T, Jarvelin MR, Jöckel KH,Keinanen-Kiukaanniemi SM, Kiemeney LA, Kuh D, Laakso M, Lehtimäki T, Levinson DF, Martin NG, Metspalu A, Morris AD, Nieminen MS,Njølstad I, Ohlsson C, Oldehinkel AJ, Ouwehand WH, Palmer LJ, Penninx B, Power C, Province MA, Psaty BM, Qi L, Rauramaa R, Ridker PM,Ripatti S, Salomaa V, Samani NJ, Snieder H, Sørensen TI, Spector TD, Stefansson K, Tönjes A, Tuomilehto J, Uitterlinden AG, Uusitupa M, van der Harst P, Vollenweider P, Wallaschofski H, Wareham NJ, Watkins H, Wichmann H, Wilson JF, Abecasis GR, Assimes TL, Barroso I, Boehnke M, Borecki IB, Deloukas P, Fox CS, Frayling T, Groop LC, Haritunian T, Heid IM, Hunter D, Kaplan RC, Karpe F, Moffatt M, Mohlke KL, O'Connell JR, Pawitan Y, Schadt EE, Schlessinger D, Steinthorsdottir V, Strachan DP, Thorsteinsdottir U, van Duijn CM, Visscher PM, Di Blasio AM,Hirschhorn JN, Lindgren CM, Morris AP, Meyre D, Scherag A, McCarthy MI, Speliotes EK, North KE, Loos RJ, Adair LS, Ang W, Atalay M, van

Beijsterveldt T, Bergen N, Benke K, Berry DJ, Boomsma DI, Bradfield JP, Charoen P, Coin L, Cooper C, Cousminer DL, Das S, Davis OS,Dedoussis GV, Elliott P, Estivill X, Evans DM, Feenstra B, Flexeder C, Frayling T, Freathy RM, Gaillard R, Geller F, Gillman M, Grant SF, Groen-Blokhuis M, Goh LK, Guxens M, Hakonarson H, Hattersley AT, Haworth CM, Hadley D, Hebebrand J, Heinrich J, Hinney A, Hirschhorn JN, Hocher B, Holloway JW, Holst C, Hottenga JJ, Horikoshi M, Huikari V, Hypponen E, Iñiguez C, Jaddoe VW, Jarvelin MR, Kaakinen M, Kilpeläinen TO,Kirin M, Kowgier M, Lakka HM, Lakka TA, Lange LA, Lawlor DA, Lehtimäki T, Lewin A, Lindgren C, Lindi V, Maggi R, Marsh J, McCarthy MI,Melbye M, Middeldorp C, Millwood I, Mohlke KL, Mook-Kanamori DO, Murray JC, Nivard M, Nohr EA, Ntalla I, Oken E, Ong KK, O'Reilly PF,Palmer LJ, Panoutsopoulou K, Pararajasingham J, Pearson ER, Pennell CE, Power C, Price TS, Prokopenko I, Raitakari OT, Rodriguez A, Salem RM, Saw SM, Scherag A, Sebert S, Siitonen N, Simell O, Sørensen TI, Sovio U, Pourcain BS, Strachan DP, Sunyer J, Taal HR, Teo YY, Thiering E, Tiesler C, Timpson NJ, Uitterlinden AG, Valcárcel B, Warrington NM, White S, Widén E, Willemsen G, Wilson JF, Yaghootkar H, Zeggini E, Elks CE, Perry JR, Sulem P, Chasman DI, Franceschini N, He C, Lunetta KL, Visser JA, Byrne EM, Cousminer DL, Gudbjartsson DF, Esko T, Feenstra B, Hottenga JJ, Koller DL, Kutalik Z, Lin P, Mangino M, Marongiu M, McArdle PF, Smith AV, Stolk L, van Wingerden SH, Zhao JH, Albrecht E,Corre T, Ingelsson E, Hayward C, Magnusson PK, Smith EN, Ulivi S, Warrington M, Zgaga L, Alavere H, Amin N, Aspelund T, Bandinelli S,Barroso I, Berenson GS, Bergmann S, Blackburn H, Boerwinkle E, Buring JE, Busonero F, Campbell H, Chanock SJ, Chen W, Cornelis MC,Couper D, Coviello AD, d'Adamo P, de Faire U, de Geus EJ, Deloukas P, Döring A, Davey Smith G, Easton DF, Eiriksdottir G, Emilsson V,Eriksson J, Ferrucci L, Folsom AR, Foroud T, Garcia M, Gasparini P, Geller F, Gieger C, Gudnason V, Hall P, Hankinson SE, Ferreli L, Heath AC,Hernandez DG, Hofman A, Hu FB, Illig T, Järvelin MR, Johnson AD, Karasik D, Khaw KT, Kiel DP, Kilpeläinen TO, Kolcic I, Kraft P, Launer LJ,Laven JS, Li S, Liu J, Levy D, Martin NG, McArdle WL, Melbye M, Mooser V, Murray JC, Murray SS, Nalls MA, Navarro P, Nelis M, Ness AR,Northstone K, Oostra BA, Peacock M, Palmer LJ, Palotie A, Paré G, Parker AN, Pedersen NL, Peltonen L, Pennell CE, Pharoah P, Polasek O,Plump AS, Pouta A, Porcu E, Rafnar T, Rice JP, Ring SM, Rivadeneira F, Rudan I, Sala C, Salomaa V, Sanna S, Schlessinger D, Schork NJ,Scuteri A, Segrè AV, Shuldiner AR, Soranzo N, Sovio U, Srinivasan SR, Strachan DP, Tammesoo ML, Tikkanen E, Toniolo D, Tsui K, Tryggvadottir L, Tyrer J, Uda M, van Dam RM, van Meurs JB, Vollenweider P, Waeber G, Wareham NJ, Waterworth DM, Weedon MN, Wichmann HE, Willemsen G, Wilson JF, Wright AF, Young L, Zhai G, Zhuang WV, Bierut LJ, Boyd HA, Crisponi L, Demerath EW, van Duijn CM, Econs MJ, Harris TB, Hunter DJ, Loos RJ, Metspalu A, Montgomery GW, Ridker PM, Spector TD, Streeten EA, Stefansson

K, Thorsteinsdottir U, Uitterlinden AG, Widen E,Murabito JM, Ong KK, Murray A, Brandt H, Crawford S, Crow S, Fichter MM, Halmi KA, Johnson C, Kaplan AS, Via M, Mitchell J, Strober M,Rotondo A, Treasure J, Woodside DB, Bulik CM, Keel P, Klump KL, Lilenfeld L, Thornton LM, Plotnicov K, Bergen AW, Berrettini W, Kaye W, Magistretti P. *Evidence for three genetic loci involved in both anorexia nervosa risk and variation of body mass index*. **Mol Psychiatry**. 2016 May 17; doi: 10.1038/mp.2016.71. 2-s2.0-84968552358

331. Pirastu N, Kooyman M, Traglia M, Robino A, Willems SM, Pistis G, Amin N, Sala C, Karssen LC, Van Duijn C, Toniolo D, Gasparini P. *A Genome-Wide Association Study in isolated populations reveals new genes associated to common food likings*. **Rev Endocr Metab Disord**. 2016 Jun;17(2):209-19. doi: 10.1007/s11154-016-9354-3. 2-s2.0-84964684039

332. Risso DS, Mezzavilla M, Pagani L, Robino A, Morini G, Tofanelli S, Carrai M, Campa D, Barale R, Caradonna F, Gasparini P, Luiselli D, Wooding S, Drayna D. *Global diversity in the TAS2R38 bitter taste receptor: revisiting a classic evolutionary PROPosal*. **Sci Rep**. 2016 Jun 27;6:28406. doi: 10.1038/srep28406. 2-s2.0-84966356381

333. McCarthy S, Das S, Kretzschmar W, Delaneau O, Wood AR, Teumer A, Kang HM, Fuchsberger C, Danecek P, Sharp K, Luo Y, Sidore C, Kwong A, Timpson N, Koskinen S, Vrieze S, Scott LJ, Zhang H, Mahajan A, Veldink J, Peters U, Pato C, van Duijn CM, Gillies CE, Gandin I, Mezzavilla M, Gilly A, Cocca M, Traglia M, Angius A, Barrett JC, Boomsma D, Branham K, Breen G, Brummett CM, Busonero F, Campbell H, Chan A, Chen S, Chew E, Collins FS, Corbin LJ, Smith GD, Dedoussis G, Dorr M, Farmaki AE, Ferrucci L, Forer L, Fraser RM, Gabriel S, Levy S, Groop L, Harrison T, Hattersley A, Holmen OL, Hveem K, Kretzler M, Lee JC, McGue M, Meitinger T, Melzer D, Min JL, Mohlke KL, Vincent JB, Nauck M, Nickerson D, Palotie A, Pato M, Pirastu N, McInnis M, Richards JB, Sala C, Salomaa V, Schlessinger D, Schoenherr S, Slagboom PE, Small K, Spector T, Stambolian D, Tuke M, Tuomilehto J, Van den Berg LH, Van Rheenen W, Volker U, Wijmenga C, Toniolo D, Zeggini E, Gasparini P, Sampson MG, Wilson JF, Frayling T, de Bakker PI, Swertz MA, McCarroll S, Kooperberg C, Dekker A, Altshuler D, Willer C, Iacono W, Ripatti S, Soranzo N, Walter K, Swaroop A, Cucca F, Anderson CA, Myers RM, Boehnke M, McCarthy MI, Durbin R; Haplotype Reference Consortium. *A reference panel of 64,976 haplotypes for genotype imputation*. **Nat Genet**. 2016 Oct;48(10):1279-83. doi: 10.1038/ng.3643. 2-s2.0-84983479616

334. Pirastu N, Kooyman M, Robino A, van der Spek A, Navarini L, Amin N, Karssen LC, Van Duijn CM, Gasparini P. *Non-additive genome-wide association scan reveals a new gene associated with habitual coffee consumption*. **Sci Rep**. 2016 Aug 25;6:31590. doi: 10.1038/srep31590. 2-s2.0-84984618888

335. van der Harst, P; van Setten, J; Verweij, N; Vogler, G; Franke, L; Maurano, MT; Wang, X; Leach, IM; Eijgelsheim, M; Sotoodehnia, N; Hayward, C; Sorice, R; Meirelles, O; Lytikäinen,

L-P; Polašek, O; Tanaka, T; Arking, DE; Ulivi, S; Trompet, S;Müller-Nurasyid, M; Smith, AV; Dörr, M; Kerr, KF; Magnani, JW; Del Greco, MF; Zhang, W; Nolte, IA; Silva, CT;Padmanabhan, S; Tragante, V; Esko, T; Abecasis, GR; Adriaens, ME; Andersen, K; Barnett, P; Bis, JC; Bodmer, R; Buckley, BM; Campbell, H; Cannon, MV; Chakravarti, A; Chen, LY; Delitala, A; Devereux, RB; Doevendands, PA; Dominiczak, AF;Ferrucci, L; Ford, I; Gieger, C; Harris, TB; Haugen, E; Heinig, M; Hernandez, DG; Hillege, HL; Hirschhorn, JN; Hofman, A;Hubner, N; Hwang, S-J; Iorio, A; Kähönen, M; Kellis, M; Kolcic, I; Kooner, IK; Kooner, JS; Kors, JA; Lakatta, EG; Lage, K;Launder, LJ; Levy, D; Lundby, A; Macfarlane, PW; May, D; Meitinger, T; Metspalu, A; Nappo, S; Naitza, S; Neph, S; Nord, AS;Nutile, T; Okin, PM; Olsen, JV; Oostra, BA; Penninger, JM; Pennacchio, LA; Raychaudhuri, S; Rice, KM; Rossin, EJ; Rotter, JI; Schafer, S; Schlessinger, D; Schmidt, CO; Sehmi, J; Silljé, HHW; Singara, G; Sinner, MF; Slowikowski, K; Soliman, EZ;Spector, TD; Spiering, W; Stamatoyannopoulos, JA; Stolk, RP; Strauch, K; Tan, S-T; Tarasov, KV; Trinh, B; Uitterlinden, AG;van den Boogaard, M; van Duijn, CM; van Gilst, WH; Siikari, JS; Visscher, PM; Vitart, V; Völker, U; Waldenberger, M; Weichenberger, CX; Westra, H-J; Wijmenga, C; Wolffenbutel, BH; Yang, J; Bezzina, CR; Munroe, PB; Snieder, H; Wright, AF;Rudan, I; Boyer, LA; Asselbergs, FW; van Veldhuisen, DJ; Stricker, BH; Psaty, BM; Ciullo, M; Sanna, S; Lehtimäki, T; Wilson, JF; Bandinelli, S; Alonso, A; Gasparini, P; Wouter Jukema, J; Kääb, S; Gudnason, V; Felix, SB; Heckbert, SR; de Boer, RA;Newton-Cheh, C; Hicks, AA; Chambers, JC; Jamshidi, Y; Visel, A; Christoffels, VM; Isaacs, A; Samani, NJ; de Bakker, PIW. *Fifty-two genetic loci influencing myocardial mass*. **Journal of the American College of Cardiology**. Sep 2016 27;68(13):1435-1448. doi: 10.1016/j.jacc.2016.07.729

336. Palombo F, Al-Wardy N, Ruscone GA, Oppo M, Kindi MN, Angius A, Al Lamki K, Girotto G, Giangregorio T, Benelli M, Magi A, Seri M, Gasparini P, Cucca F, Sazzini M, Al Khabori M, Pippucci T, Romeo G. *A novel founder MYO15A frameshift duplication is the major cause of genetic hearing loss in Oman*. **Journal of Human Genetics**Oct 2016 13; doi: 10.1038/jhg.2016.120. 2-s2.0-85010905715

337. Iotchkova V, Huang J, Morris JA, Jain D, Barbieri C, Walter K, Min JL, Chen L, Astle W, Cocca M, Deelen P, Elding H, Farmaki AE, Franklin CS, Franberg M, Gaunt TR, Hofman A, Jiang T, Kleber ME, Lachance G, Luan J, Malerba G, Matchan A, Mead D, Memari Y, Ntalla I, Panoutsopoulou K, Pazoki R, Perry JRB, Rivadeneira F, Sabater-Lleal M, Sennblad B, Shin SY, Southam L, Traglia M, van Dijk F, van Leeuwen EM, Zaza G, Zhang W; UK10K Consortium, Amin N, Butterworth A, Chambers JC, Dedoussis G, Dehghan A, Franco OH, Franke L, Frontini M, Gambaro G, Gasparini P, Hamsten A, Issacs A, Kooner JS, Kooperberg C, Langenberg C, Marz W, Scott RA, Swertz MA, Toniolo D, Uitterlinden AG, van Duijn CM, Watkins H, Zeggini E, Maurano

MT, Timpson NJ, Reiner AP, Auer PL, Soranzo N. *Discovery and refinement of genetic loci associated with cardiometabolic risk using dense imputation maps*. **Nature Genetics** 2016 Nov; 48(11):1303-1312. doi: 10.1038/ng.3668. 2-s2.0-84988713790

338. Navarra CO, Robino A, Pirastu N, Bevilacqua L, Gasparini P, Di Lenarda R, Crovella S. *Caries and Innate Immunity: DEFB1 Gene Polymorphisms and Caries Susceptibility in Genetic Isolates from North-Eastern Italy*. **Caries Research** 2016;50(6):589-594. doi: 10.1159/000450965. 2-s2.0-84995810912

339. Marioni RE, Ritchie SJ, Joshi PK, Hagenaars SP, Okbay A, Fischer K, Adams MJ, Hill WD, Davies G; Social Science Genetic Association Consortium, Nagy R, Amador C, Läll K, Metspalu A, Liewald DC, Campbell A, Wilson JF, Hayward C, Esko T, Porteous DJ, Gale CR, Deary IJ, Okbay A,Beauchamp JP,Fontana MA,Lee JJ,Pers TH,Rietveld CA,Turley P,Chen GB,Emilsson V,Meddens SF,Oskarsson S,Pickrell JK,Thom K,Timshel P,de Vlaming R,Abdellaoui A,Ahluwalia TS,Bacelis J,Baumbach C,Bjornsdottir G,Brandsma JH,Concas MP,Derringer J,Furlotte NA,Galesloot TE,Girotto G,Gupta R,Hall LM,Harris SE,Hofer E,Horikoshi M,Huffman JE,Kaasik K,Kalafati IP,Karlsson R,Kong A,Lahti J,van der Lee SJ,de Leeuw C,Lind PA,Lindgren KO,Liu T,Mangino M,Marten J,Mihailov E,Miller MB,van der Most PJ,Oldmeadow C,Payton A,Pervjakova N,Peyrot WJ,Qian Y,Raitakari O,Rueedi R,Salvi E,Schmidt B,Schraut KE,Shi J,Smith AV,Poot RA,St Pourcain B,Teumer A,Thorleifsson G,Verweij N,Vuckovic D,Wellmann J,Westra HJ,Yang J,Zhao W,Zhu Z,Alizadeh BZ,Amin N,Bakshi A,Baumeister SE,Biino G,Bønnelykke K,Boyle PA,Campbell H,Cappuccio FP,Davies G,De Neve JE,Deloukas P,Demuth I,Ding J,Eibich P,Eisele L,Eklund N,Evans DM,Faul JD,Feitosa MF,Forstner AJ,Gandin I,Gunnarsson B,Halldórsson BV,Harris TB,Heath AC,Hocking LJ,Holliday EG,Homuth G,Horan MA,Hottenga JJ,de Jager PL,Joshi PK,Jugessur A,Kaakinen MA,Kähönen M,Kanoni S,Keltigangas-Järvinen L, Kiemeney LA,Kolcic I,Koskinen S,Kraja AT,Kroh M,Kutalik Z,Latvala A,Launer LJ,Lebreton MP,Levinson DF,Lichtenstein P,Lichtner P,Liewald DC,Loukola A,Madden PA,Mägi R,Mäki-Opas T,Marioni RE,Marques-Vidal P,Meddens GA,McMahon G,Meisinger C,Meitinger T,Milaneschi Y,Milani L,Montgomery GW,Myhre R,Nelson CP,Nyholt DR,Ollier WE,Palotie A,Paternoster L,Pedersen NL,Petrovic KE,Porteous DJ,Räikkönen K,Ring SM,Robino A,Rostapshova O,Rudan I,Rustichini A,Salomaa V,Sanders AR,Sarin AP,Schmidt H,Scott RJ,Smith BH,Smith JA,Staessen JA,Steinhagen-Thiessen E, Strauch K,Terracciano A,Tobin MD,Ulivi S,Vaccargiu S,Quaye L,van Rooij FJ,Venturini C,Vinkhuyzen AA,Völker U,Völzke H,Vonk JM,Vozzi D,Waage J,Ware EB,Willemsen G,Attia JR,Bennett DA,Berger K,Bertram L,Bisgaard H,Boomsma DI,Borecki IB,Bultmann U,Chabris CF,Cucca F,Cusi D,Deary IJ,Dedoussis GV,van Duijn CM,Eriksson JG,Franke B,Franke L,Gasparini P, Gejman PV,Gieger C,Grabe HJ,Gratten J,Groenen PJ,Gudnason V,van der Harst P,Hayward C,Hinds DA,Hoffmann W,Hypponen E,Iacono WG,Jacobsson B,Järvelin MR,Jöckel KH,Kaprio J,Kardia SL,Lehtimäki T,Lehrer

SF,Magnusson PK,Martin NG,McGue M,Metspalu A,Pendleton N,Penninx BW,Perola M,Pirastu N,Pirastu M,Polasek O,Posthuma D,Power C,Province MA,Samani NJ,Schlessinger D,Schmidt R,Sørensen TI,Spector TD,Stefansson K,Thorsteinsdottir U, Thurik AR,Timpson NJ,Tiemeier H,Tung JY,Uitterlinden AG,Vitart V,Vollenweider P,Weir DR,Wilson JF,Wright AF,Conley DC,Krueger RF,Smith GD,Hofman A,Laibson DI,Medland SE,Meyer MN,Yang J,Johannesson M,Visscher PM,Esko T,Koellinger PD,Cesarini D,Benjamin DJ. *Genetic variants linked to education predict longevity*. **Proceedings of the national Academy of Sciences of United States od America** 2016 Nov 22;113(47):13366-13371. doi: 10.1073/pnas.1605334113. 2-s2.0-84996555363

340. Barban N, Jansen R, de Vlaming R, Vaez A, Mandemakers JJ, Tropf FC, Shen X, Wilson JF, Chasman DI, Nolte IM, Tragante V, van der Laan SW, Perry JR, Kong A; BIOS Consortium, Ahluwalia TS, Albrecht E, Yerges-Armstrong L, Atzmon G, Auro K, Ayers K, Bakshi A, Ben-Avraham D, Berger K, Bergman A, Bertram L, Bielak LF, Bjornsdottir G, Bonder MJ, Broer L, Bui M, Barbieri C, Cavadino A, Chavarro JE, Turman C, Concas MP, Cordell HJ, Davies G, Eibich P, Eriksson N, Esko T, Eriksson J, Falahi F, Felix JF, Fontana MA, Franke L, Gandin I, Gaskins AJ, Gieger C, Gunderson EP, Guo X, Hayward C, He C, Hofer E, Huang H, Joshi PK, Kanoni S, Karlsson R, Kiechl S, Kifley A, Kluttig A, Kraft P, Lagou V, Lecoeur C, Lahti J, Li-Gao R, Lind PA, Liu T, Makalic E, Mamasoula C, Matteson L, Mbarek H, McArdle PF, McMahon G, Meddens SF, Mihailov E, Miller M, Missmer SA, Monnereau C, van der Most PJ, Myhre R, Nalls MA, Nutile T, Kalafati IP, Porcu E, Prokopenko I, Rajan KB, Rich-Edwards J, Rietveld CA, Robino A, Rose LM, Rueedi R, Ryan KA, Saba Y, Schmidt D, Smith JA, Stolk L, Streeten E, Tönjes A, Thorleifsson G, Ulivi S, Wedenoja J, Wellmann J, Willeit P, Yao J, Yengo L, Zhao JH, Zhao W, Zhernakova DV, Amin N, Andrews H, Balkau B, Barzilai N, Bergmann S, Biino G, Bisgaard H, Bønnelykke K, Boomsma DI, Buring JE, Campbell H, Cappellani S, Ciullo M, Cox SR, Cucca F, Toniolo D, Davey-Smith G, Deary IJ, Dedoussis G, Deloukas P, van Duijn CM, de Geus EJ, Eriksson JG, Evans DA, Faul JD, Sala CF, Froguel P, Gasparini P, Girotto G, Grabe HJ, Greiser KH, Groenen PJ, de Haan HG, Haerting J, Harris TB, Heath AC, Heikkilä K, Hofman A, Homuth G, Holliday EG, Hopper J, Hyppönen E, Jacobsson B, Jaddoe VW, Johannesson M, Jugessur A, Kähönen M, Kajantie E, Kardia SL, Keavney B, Kolcic I, Koponen P, Kovacs P, Kronenberg F, Kutalik Z, La Bianca M, Lachance G, Iacono WG, Lai S, Lehtimäki T, Liewald DC; LifeLines Cohort Study, Lindgren CM, Liu Y, Luben R, Lucht M, Luoto R, Magnus P, Magnusson PK, Martin NG, McGue M, McQuillan R, Medland SE, Meisinger C, Mellström D, Metspalu A, Traglia M, Milani L, Mitchell P, Montgomery GW, Mook-Kanamori D, de Mutsert R, Nohr EA, Ohlsson C, Olsen J, Ong KK, Paternoster L, Pattie A, Penninx BW, Perola M, Peyser PA, Pirastu M, Polasek O, Power C, Kaprio J, Raffel LJ, Räikkönen K, Raitakari O, Ridker PM, Ring SM, Roll K, Rudan I, Ruggiero D, Rujescu D, Salomaa V,

Schlessinger D, Schmidt H, Schmidt R, Schupf N, Smit J, Sorice R, Spector TD, Starr JM, Stöckl D, Strauch K, Stumvoll M, Swertz MA, Thorsteinsdottir U, Thurik AR, Timpson NJ, Tung JY, Uitterlinden AG, Vaccargiu S, Viikari J, Vitart V, Völzke H, Vollenweider P, Vuckovic D, Waage J, Wagner GG, Wang JJ, Wareham NJ, Weir DR, Willemsen G, Willeit J, Wright AF, Zondervan KT, Stefansson K, Krueger RF, Lee JJ, Benjamin DJ, Cesarini D, Koellinger PD, den Hoed M, Snieder H, Mills MC. *Genome-wide analysis identifies 12 loci influencing human reproductive behavior*. **Nature Genetics** 2016 Dec;48(12):1462-1472. doi: 10.1038/ng.3698. 2-s2.0-84992744163

341. Corre T, Olinger E, Harris SE, Traglia M, Ulivi S, Lenarduzzi S, Belge H, Youhanna S, Tokonami N, Bonny O, Houillier P, Polasek O, Deary IJ, Starr JM, Toniolo D, Gasparini P, Vollenweider P, Hayward C, Bochud M, Devuyst O. *Common variants in CLDN14 are associated with differential excretion of magnesium over calcium in urine.* **Pflugers Archiv: European journal of physiology** 2017 Jan;469(1):91-103. doi: 10.1007/s00424-016-1913-7. 2-s2.0-85001013366

342. Li M, Li Y, Weeks O, Mijatovic V, Teumer A, Huffman JE, Tromp G, Fuchsberger C, Gorski M, Lyytikäinen LP, Nutile T, Sedaghat S, Sorice R, Tin A, Yang Q, Ahluwalia TS, Arking DE, Bihlmeyer NA, Böger CA, Carroll RJ, Chasman DI, Cornelis MC, Dehghan A, Faul JD, Feitosa MF, Gambaro G, Gasparini P, Giulianini F, Heid I, Huang J, Imboden M, Jackson AU, Jeff J, Jhun MA, Katz R, Kifley A, Kilpeläinen TO, Kumar A, Laakso M, Li-Gao R, Lohman K, Lu Y, Mägi R, Malerba G, Mihailov E, Mohlke KL, Mook-Kanamori DO, Robino A, Ruderfer D, Salvi E, Schick UM, Schulz CA, Smith AV, Smith JA, Traglia M, Yerges-Armstrong LM, Zhao W, Goodarzi MO, Kraja AT, Liu C, Wessel J; CHARGE Glycemic-T2D Working Group,; CHARGE Blood Pressure Working Group,, Boerwinkle E, Borecki IB, Bork-Jensen J, Bottinger EP, Braga D, Brandslund I, Brody JA, Campbell A, Carey DJ, Christensen C, Coresh J, Crook E, Curhan GC, Cusi D, de Boer IH, de Vries AP, Denny JC, Devuyst O, Dreisbach AW, Endlich K, Esko T, Franco OH, Fulop T, Gerhard GS, Glümer C, Gottesman O, Grarup N, Gudnason V, Hansen T, Harris TB, Hayward C, Hocking L, Hofman A, Hu FB, Husemoen LL, Jackson RD, Jørgensen T, Jørgensen ME, Kähönen M, Kardia SL, König W, Kooperberg C, Kriebel J, Launer LJ, Lauritzen T, Lehtimäki T, Levy D, Linksted P, Linneberg A, Liu Y, Loos RJ, Lupo A, Meisinger C, Melander O, Metspalu A, Mitchell P, Nauck M, Nürnberg P, Orho-Melander M, Parsa A, Pedersen O, Peters A, Peters U, Polasek O, Porteous D, Probst-Hensch NM, Psaty BM, Qi L, Raitakari OT, Reiner AP, Rettig R, Ridker PM, Rivadeneira F, Rossouw JE, Schmidt F, Siscovick D, Soranzo N, Strauch K, Toniolo D, Turner ST, Uitterlinden AG, Ulivi S, Velayutham D, Völker U, Völzke H, Waldenberger M, Wang JJ, Weir DR, Witte D, Kuivaniemi H, Fox CS, Franceschini N, Goessling W, Köttgen A, Chu AY, Wessel J,Chu AY,Willems SM,Wang S,Yaghootkar H,Brody JA,Dauriz M,Hivert MF,Raghavan S,Lipovich L,Hidalgo B,Fox K,Huffman JE,An P,Lu Y,Rasmussen-Torvik LJ, Grarup N,Ehm MG,Baldridge

AS,Stančáková A, Abrol R,Besse CL,Boland A,Bork-Jensen J,Fornage M,Freitag DF,Garcia ME,Guo X,Hara K,Isaacs A,Jakobsdottir J,Lange LA,Layton JC,Li M,Zhao JH,Meidtner K,Morrison AC,Nalls MA,Peters MJ,Sabater-Lleal M,Schurmann C,Silveira A,Smith AV,Southam L,Stoiber MH,Strawbridge RJ,Taylor KD,Varga TV,Allin KH,Amin N,Aponte JL,Aung T,Barbieri C,Bihlmeyer NA,Boehnke M,Bombieri C,Bowden DW,Burns SM,Chen Y,Chen YD,Cheng CY,Correa A,Czajkowski J,Dehghan A,Ehret GB,Eiriksdottir G,Escher SA,Farmaki AE,Frånberg M, Gambaro G,Giulianini F,Goddard WA 3rd,Goel A,Gottesman O,Grove ML,Gustafsson S,Hai Y,Hallmans GR,Heo J,Hoffmann P,Ikram MK,Jensen RA,Jørgensen ME, Jørgensen T, Karaleftheri M,Khor CC,Kirkpatrick A,Kraja AT,Kuusisto J,Lange EM 1st,Lee T,Lee WJ,Leong A,Liao J,Liu C,Liu Y,Lindgren CM,Linneberg A,Malerba G,Mamakou V,Marouli E,Maruthur NM,Matchan A,McKean-Cowdin R,McLeod O,Metcalf GA,Mohlke KL,Muzny DM,Ntalla I,Palmer ND,Pasko D,Peter A,Rayner NW,Renstro M F,Rice K,Sala CF,Sennblad B,Serafetinidis I,Smith JA,Soranzo N,Speliotes EK,Stahl EA,Stirrups K,Tentolouris N,Thanopoulou A,Torres M,Traglia M,Tsafantakis E,Javad S,Yanek LR,Zengini E,Becker DM,Bis JC,Brown JB,Cupples LA,Hansen T,Ingelsson E,Karter AJ,Lorenzo C,Mathias RA,Norris JM,Peloso GM,Sheu WH,Toniolo D,Vaidya D,Varma R,Wagenknecht LE,Boeing H,Bottinger EP,Dedoussis G,Deloukas P,Ferrannini E,Franco OH,Franks PW,Gibbs RA,Gudnason V,Hamsten A,Harris TB,Hattersley AT,Hayward C,Hofman A,Jansson JH,Langenberg C,Launer LJ,Levy D,Oostra BA,O'Donnell CJ,O'Rahilly S,Padmanabhan S,Pankow JS,Polasek O,Province MA,Rich SS,Ridker PM,Rudan I,Schulze MB,Smith BH,Uitterlinden AG,Walker M,Watkins H,Wong TY,Zeggini E,Consortiumy TE,Laakso M,Borecki IB,Chasman DI,Pedersen O,Psaty BM,Tai ES,van Duijn CM,Wareham NJ,Waterworth DM,Boerwinkle E,Kao WH,Florez JC,Loos RJ,Wilson JG,Frayling TM,Siscovick DS,Dupuis JE,Rotter JI,Meigs JB,Scott RA,Goodarzi MO,Liu C,Kraja AT,Smith JA,Brody JA,Franceschini N,Bis JC,Rice K,Morrison AC,Lu Y,Weiss S,Guo X,Palmas W,Martin LW,Chen YI,Surendran P,Drenos F,Cook JP,Auer PL,Chu AY,Giri A,Zhao W,Jakobsdottir J,Lin LA,Stafford JM,Amin N,Mei H,Yao J,Voorman A,Larson MG,Grove ML,Smith AV,Hwang SJ,Chen H,Huan T,Kosova G,Stitziel NO,Kathiresan S,Samani N,Schunkert H,Deloukas P,Genetics MI,Li M,Fuchsberger C,Pattaro C,Gorski M,Kooperberg C,Papanicolaou GJ,Rossouw JE,Faul JD,Kardia SL,Bouchard C,Raffel LJ,Uitterlinden AG,Franco OH,Vasan RS,O'Donnell CJ,Taylor KD,Liu K,Bottinger EP,Gottesman O,Daw EW,Giulianini F,Ganesh S,Salfati E,Harris TB,Launer LJ,Dörr M,Felix SB,Rettig R,Völzke H, Kim E,Lee WJ 1st,Lee T,Sheu WH,Tsosie KS,Edwards DR,Liu Y,Correa A,Weir DR,Völker U, Ridker PM,Boerwinkle E,Gudnason V,Reiner AP,van Duijn CM,Borecki IB,Edwards TL,Chakravarti A,Rotter JI,Psaty BM,Loos RJ,Fornage M,Ehret G,Newton-Cheh C,Levy D,Chasman DI. *SOS2 and ACP1 Loci Identified through Large-Scale Exome Chip Analysis Regulate Kidney Development and Function*. **Journal of the American**

Society of Nephrology: JASN 2017 Mar;28(3):981-994. doi: 10.1681/ASN.2016020131. 2-s2.0-85021851118

343. Cole AM, Cox S, Jeong C, Petousi N, Aryal DR, Droma Y, Hanaoka M, Ota M, Kobayashi N, Gasparini P, Montgomery H, Robbins P, Di Rienzo A, Cavalleri GL. *Genetic structure in the Sherpa and neighboring Nepalese populations*. **BMC Genomics** 2017 Jan 19; 18(1):102. doi: 10.1186/s12864-016-3469-5.

344. Zupin L, Navarra CO, Robino A, Bevilacqua L, Di Lenarda R, Gasparini P, Crovella S. *NLRC5 polymorphism is associated with susceptibility to chronic periodontitis*. **Immunobiology** 2017 May;222(5):704-708. doi: 10.1016/j.imbio.2017.01.001.

345. Warren HR, Evangelou E, Cabrera CP, Gao H, Ren M, Mifsud B, Ntalla I, Surendran P, Liu C, Cook JP, Kraja AT, Drenos F, Loh M, Verweij N, Marten J, Karaman I, Lepe MP, O'Reilly PF, Knight J, Snieder H, Kato N, He J, Tai ES, Said MA, Porteous D, Alver M, Poulter N, Farrall M, Gansevoort RT, Padmanabhan S, Mägi R, Stanton A, Connell J, Bakker SJ, Metspalu A, Shields DC, Thom S, Brown M, Sever P, Esko T, Hayward C, van der Harst P, Saleheen D, Chowdhury R, Chambers JC, Chasman DI, Chakravarti A, Newton-Cheh C, Lindgren CM, Levy D, Kooner JS, Keavney B, Tomaszewski M, Samani NJ, Howson JM, Tobin MD, Munroe PB, Ehret GB, Wain LV; International Consortium of Blood Pressure (ICBP) 1000G Analyses; BIOS Consortium; Lifelines Cohort Study; Understanding Society Scientific group; CHD Exome+ Consortium; ExomeBP Consortium; T2D-GENES Consortium; GoT2DGenes Consortium; Cohorts for Heart and Ageing Research in Genome Epidemiology (CHARGE) BP Exome Consortium; International Genomics of Blood Pressure (iGEN-BP) Consortium; UK Biobank CardioMetabolic Consortium BP working group, Völker U,Vollenweider P,Wild S,Willemsen G,Wright AF,Yao J,Thériault S,Conen D,John A,Sever P,Debette S,Mook-Kanamori DO,Zeggini E,Spector TD,van der Harst P,Palmer CN,Vergnaud AC,Loos RJ,Polasek O,Starr JM,Girotto G,Hayward C,Kooner JS,Lindgren CM,Vitart V,Samani NJ,Tuomilehto J,Gyllensten U,Knekt P,Deary IJ,Ciullo M,Elosua R,Keavney BD,Hicks AA,Scott RA,Gasparini P, Laan M,Liu Y,Watkins H,Hartman CA,Salomaa V,Toniolo D,Perola M,Wilson JF,Schmidt H,Zhao JH,Lehtimäki T,van Duijn CM,Gudnason V,Psaty BM,Peters A,Rettig R,James A,Jukema JW,Strachan DP,Palmas W,Metspalu A,Ingelsson E,Boomsma DI,Franco OH,Bochud M,Newton-Cheh C,Munroe PB,Elliott P,Chasman DI,Chakravarti A,Knight J,Morris AP,Levy D,Tobin MD,Snieder H,Caulfield MJ,Ehret GB,Barnes MR,Tzoulaki I,Caulfield MJ, Elliott P. *Genome-wide association analysis identifies novel blood pressure loci and offers biological insights into cardiovascular risk*. **Nature Genetics** 2017 Mar;49(3):403-415. doi: 10.1038/ng.3768.

346. Marouli E, Graff M, Medina-Gomez C, Lo KS, Wood AR, Kjaer TR, Fine RS, Lu Y, Schurmann C, Highland HM, Rüeger S, Thorleifsson G, Justice AE, Lamparter D, Stirrups KE, Turcot V,

Young KL, Winkler TW, Esko T, Karaderi T, Locke AE, Masca NG, Ng MC, Mudgal P, Rivas MA, Vedantam S, Mahajan A, Guo X, Abecasis G, Aben KK, Adair LS, Alam DS, Albrecht E, Allin KH, Allison M, Amouyel P, Appel EV, Arveiler D, Asselbergs FW, Auer PL, Balkau B, Banas B, Bang LE, Benn M, Bergmann S, Bielak LF, Blüher M, Boeing H, Boerwinkle E, Böger CA, Bonnycastle LL, Bork-Jensen J, Bots ML, Bottinger EP, Bowden DW, Brandslund I, Breen G, Brilliant MH, Broer L, Burt AA, Butterworth AS, Carey DJ, Caulfield MJ, Chambers JC, Chasman DI, Chen YI, Chowdhury R, Christensen C, Chu AY, Cocca M, Collins FS, Cook JP, Corley J, Galbany JC, Cox AJ, Cuellar-Partida G, Danesh J, Davies G, de Bakker PI, de Borst GJ, de Denus S, de Groot MC, de Mutsert R, Deary IJ, Dedoussis G, Demerath EW, den Hollander AI, Dennis JG, Di Angelantonio E, Drenos F, Du M, Dunning AM, Easton DF, Ebeling T, Edwards TL, Ellinor PT, Elliott P, Evangelou E, Farmaki AE, Faul JD, Feitosa MF, Feng S, Ferrannini E, Ferrario MM, Ferrieres J, Florez JC, Ford I, Fornage M, Franks PW, Frikke-Schmidt R, Galesloot TE, Gan W, Gandin I, Gasparini P, Giedraitis V, Giri A, Girotto G, Gordon SD, Gordon-Larsen P, Gorski M, Grarup N, Grove ML, Gudnason V, Gustafsson S, Hansen T, Harris KM, Harris TB, Hattersley AT, Hayward C, He L, Heid IM, Heikkilä K, Helgeland Ø, Hernesniemi J, Hewitt AW, Hocking LJ, Hollensted M, Holmen OL, Hovingh GK, Howson JM, Hoyng CB, Huang PL, Hveem K, Ikram MA, Ingelsson E, Jackson AU, Jansson JH, Jarvik GP, Jensen GB, Jhun MA, Jia Y, Jiang X, Johansson S, Jørgensen ME, Jørgensen T, Jousilahti P, Jukema JW, Kahali B, Kahn RS, Kähönen M, Kamstrup PR, Kanoni S, Kaprio J, Karaleftheri M, Kardia SL, Karpe F, Kee F, Keeman R, Kiemeney LA, Kitajima H, Kluivers KB, Kocher T, Komulainen P, Kontto J, Kooner JS, Kooperberg C, Kovacs P, Kriebel J, Kuivaniemi H, Küry S, Kuusisto J, La Bianca M, Laakso M, Lakka TA, Lange EM, Lange LA, Langefeld CD, Langenberg C, Larson EB, Lee IT, Lehtimäki T, Lewis CE, Li H, Li J, Li-Gao R, Lin H, Lin LA, Lin X, Lind L, Lindström J, Linneberg A, Liu Y, Liu Y, Lophatananon A, Luan J, Lubitz SA, Lyytikäinen LP, Mackey DA, Madden PA, Manning AK, Männistö S, Marenne G, Marten J, Martin NG, Mazul AL, Meidtner K, Metspalu A, Mitchell P, Mohlke KL, Mook-Kanamori DO, Morgan A, Morris AD, Morris AP, Müller-Nurasyid M, Munroe PB, Nalls MA, Nauck M, Nelson CP, Neville M, Nielsen SF, Nikus K, Njølstad PR, Nordestgaard BG, Ntalla I, O'Connel JR, Oksa H, Loohuis LM, Ophoff RA, Owen KR, Packard CJ, Padmanabhan S, Palmer CN, Pasterkamp G, Patel AP, Pattie A, Pedersen O, Peissig PL, Peloso GM, Pennell CE, Perola M, Perry JA, Perry JR, Person TN, Pirie A, Polasek O, Posthuma D, Raitakari OT, Rasheed A, Rauramaa R, Reilly DF, Reiner AP, Renström F, Ridker PM, Rioux JD, Robertson N, Robino A, Rolandsson O, Rudan I, Ruth KS, Saleheen D, Salomaa V, Samani NJ, Sandow K, Sapkota Y, Sattar N, Schmidt MK, Schreiner PJ, Schulze MB, Scott RA, Segura-Lepe MP, Shah S, Sim X, Sivapalaratnam S, Small KS, Smith AV, Smith JA, Southam L, Spector TD, Speliotes EK, Starr JM, Steinthorsdottir V, Stringham HM, Stumvoll M, Surendran P, 't Hart LM, Tansey

KE, Tardif JC, Taylor KD, Teumer A, Thompson DJ, Thorsteinsdottir U, Thuesen BH, Tönjes A, Tromp G, Trompet S, Tsafantakis E, Tuomilehto J, Tybjaerg-Hansen A, Tyrer JP, Uher R, Uitterlinden AG, Ulivi S, van der Laan SW, Van Der Leij AR, van Duijn CM, van Schoor NM, van Setten J, Varbo A, Varga TV, Varma R, Edwards DR, Vermeulen SH, Vestergaard H, Vitart V, Vogt TF, Vozzi D, Walker M, Wang F, Wang CA, Wang S, Wang Y, Wareham NJ, Warren HR, Wessel J, Willems SM, Wilson JG, Witte DR, Woods MO, Wu Y, Yaghootkar H, Yao J, Yao P, Yerges-Armstrong LM, Young R, Zeggini E, Zhan X, Zhang W, Zhao JH, Zhao W, Zhao W, Zheng H, Zhou W; EPIC-InterAct Consortium; CHD Exome+ Consortium; ExomeBP Consortium; T2D-Genes Consortium; GoT2D Genes Consortium; Global Lipids Genetics Consortium; ReproGen Consortium; MAGIC Investigators, Rotter JI, Boehnke M, Kathiresan S, McCarthy MI, Willer CJ, Stefansson K, Borecki IB, Liu DJ, North KE, Heard-Costa NL, Pers TH, Lindgren CM, Oxvig C, Kutalik Z, Rivadeneira F, Loos RJ, Frayling TM, Hirschhorn JN, Deloukas P, Lettre G. *Rare and low-frequency coding variants alter human adult height*. **Nature**2017 Feb 9;542(7640):186-190. doi: 10.1038/nature21039.

347. Alkowari MK, Vozzi D, Bhagat S, Krishnamoorthy N, Morgan A, Hayder Y, Logendra B, Najjar N, Gandin I, Gasparini P, Badii R, Girotto G, Abdulhadi K. *Targeted sequencing identifies novel variants involved in autosomal recessive hereditary hearing loss in Qatari families*. **Mutation research** 2017 May 4; 800-802:29-36. doi: 10.1016/j.mrfmmm.2017.05.001.

348. Zupin L, Robino A, Navarra CO, Pirastu N, Di Lenarda R, Gasparini P, Crovella S, Bevilacqua L. *LTF and DEFB1 polymorphisms are associated with susceptibility toward chronic periodontitis development*. **Oral diseases** 2017 May 9. doi: 10.1111/odi.12689.

349. Tachmazidou I, Süveges D, Min JL, Ritchie GRS, Steinberg J, Walter K, Iotchkova V, Schwartzentruber J, Huang J, Memari Y, McCarthy S, Crawford AA, Bombieri C, Cocca M, Farmaki AE, Gaunt TR, Jousilahti P, Kooijman MN, Lehne B, Malerba G, Männistö S, Matchan A, Medina-Gomez C, Metrustry SJ, Nag A, Ntalla I, Paternoster L, Rayner NW, Sala C, Scott WR, Shihab HA, Southam L, St Pourcain B, Traglia M, Trajanoska K, Zaza G, Zhang W, Artigas MS, Bansal N, Benn M, Chen Z, Danecek P, Lin WY, Locke A, Luan J, Manning AK, Mulas A, Sidore C, Tybjaerg-Hansen A, Varbo A, Zoledziewska M, Finan C, Hatzikotoulas K, Hendricks AE, Kemp JP, Moayyeri A, Panoutsopoulou K, Szpak M, Wilson SG, Boehnke M, Cucca F, Di Angelantonio E, Langenberg C, Lindgren C, McCarthy MI, Morris AP, Nordestgaard BG, Scott RA, Tobin MD, Wareham NJ; SpiroMeta Consortium; GoT2D Consortium, Burton P, Chambers JC, Smith GD, Dedoussis G, Felix JF, Franco OH, Gambaro G, Gasparini P, Hammond CJ, Hofman A, Jaddoe VWV, Kleber M, Kooner JS, Perola M, Relton C, Ring SM, Rivadeneira F, Salomaa V, Spector TD, Stegle O, Toniolo D, Uitterlinden AG; arcOGEN Consortium; Understanding Society Scientific Group; UK10K Consortium, Barroso I, Greenwood CMT, Perry JRB, Walker BR, Butterworth AS, Xue Y,

Durbin R, Small KS, Soranzo N, Timpson NJ, Zeggini E. *Whole-Genome Sequencing Coupled to Imputation Discovers Genetic Signals for Anthropometric Traits*. **American journal of human genetics.** 2017 Jun 1;100(6):865-884. doi: 10.1016/j.ajhg.2017.04.014.

350. Gorski M, van der Most PJ, Teumer A, Chu AY, Li M, Mijatovic V, Nolte IM, Cocca M, Taliun D, Gomez F, Li Y, Tayo B, Tin A, Feitosa MF, Aspelund T, Attia J, Biffar R, Bochud M, Boerwinkle E, Borecki I, Bottinger EP, Chen MH, Chouraki V, Ciullo M, Coresh J, Cornelis MC, Curhan GC, d'Adamo AP, Dehghan A, Dengler L, Ding J, Eiriksdottir G, Endlich K, Enroth S, Esko T, Franco OH, Gasparini P, Gieger C, Girotto G, Gottesman O, Gudnason V, Gyllensten U, Hancock SJ, Harris TB, Helmer C, Höllerer S, Hofer E, Hofman A, Holliday EG, Homuth G, Hu FB, Huth C, Hutri-Kähönen N, Hwang SJ, Imboden M, Johansson Å, Kähönen M, König W, Kramer H, Krämer BK, Kumar A, Kutalik Z, Lambert JC, Launer LJ, Lehtimäki T, de Borst MH, Navis G, Swertz M, Liu Y, Lohman K, Loos RJF, Lu Y, Lyytikäinen LP, McEvoy MA, Meisinger C, Meitinger T, Metspalu A, Metzger M, Mihailov E, Mitchell P, Nauck M, Oldehinkel AJ, Olden M, Wjh Penninx B, Pistis G, Pramstaller PP, Probst-Hensch N, Raitakari OT, Rettig R, Ridker PM, Rivadeneira F, Robino A, Rosas SE, Ruderfer D, Ruggiero D, Saba Y, Sala C, Schmidt H, Schmidt R, Scott RJ, Sedaghat S, Smith AV, Sorice R, Stengel B, Stracke S, Strauch K, Toniolo D, Uitterlinden AG, Ulivi S, Viikari JS, Völker U, Vollenweider P, Völzke H, Vuckovic D, Waldenberger M, Jin Wang J, Yang Q, Chasman DI, Tromp G, Snieder H, Heid IM, Fox CS, Köttgen A, Pattaro C, Böger CA, Fuchsberger C. *1000 Genomes-based meta-analysis identifies 10 novel loci for kidney function*. **Scientific reports**2017 May 26;7:46835. doi: 10.1038/srep46835.

351. Xue Y, Mezzavilla M, Haber M, McCarthy S, Chen Y, Narasimhan V, Gilly A, Ayub Q, Colonna V, Southam L, Finan C, Massaia A, Chheda H, Palta P, Ritchie G, Asimit J, Dedoussis G, Gasparini P, Palotie A, Ripatti S, Soranzo N, Toniolo D, Wilson JF, Durbin R, Tyler-Smith C, Zeggini E. *Enrichment of low-frequency functional variants revealed by whole-genome sequencing of multiple isolated European populations*. **Nature communications.**2017 Jun 23;8:15927. doi: 10.1038/ncomms15927.

352. Wain LV, Vaez A, Jansen R, Joehanes R, van der Most PJ, Erzurumluoglu AM, O'Reilly PF, Cabrera CP, Warren HR, Rose LM, Verwoert GC, Hottenga JJ, Strawbridge RJ, Esko T, Arking DE, Hwang SJ, Guo X, Kutalik Z, Trompet S, Shrine N, Teumer A, Ried JS, Bis JC, Smith AV, Amin N, Nolte IM, Lyytikäinen LP, Mahajan A, Wareham NJ, Hofer E, Joshi PK, Kristiansson K, Traglia M, Havulinna AS, Goel A, Nalls MA, Sõber S, Vuckovic D, Luan J, Del Greco M F, Ayers KL, Marrugat J, Ruggiero D, Lopez LM, Niiranen T, Enroth S, Jackson AU, Nelson CP, Huffman JE, Zhang W, Marten J, Gandin I, Harris SE, Zemunik T, Lu Y, Evangelou E, Shah N, de Borst MH, Mangino M, Prins BP, Campbell A, Li-Gao R, Chauhan G, Oldmeadow C, Abecasis G, Abedi M, Barbieri CM, Barnes MR, Batini C, Beilby J, Blake T,

Boehnke M, Bottinger EP, Braund PS, Brown M, Brumat M, Campbell H, Chambers JC, Cocca M, Collins F, Connell J, Cordell HJ, Damman JJ, Davies G, de Geus EJ, de Mutsert R, Deelen J, Demirkale Y, Doney ASF, Dörr M, Farrall M, Ferreira T, Frånberg M, Gao H, Giedraitis V, Gieger C, Giulianini F, Gow AJ, Hamsten A, Harris TB, Hofman A, Holliday EG, Hui J, Jarvelin MR, Johansson Å, Johnson AD, Jousilahti P, Jula A, Kähönen M, Kathiresan S, Khaw KT, Kolcic I, Koskinen S, Langenberg C, Larson M, Launer LJ, Lehne B, Liewald DCM, Lin L, Lind L, Mach F, Mamasoula C, Menni C, Mifsud B, Milaneschi Y, Morgan A, Morris AD, Morrison AC, Munson PJ, Nandakumar P, Nguyen QT, Nutile T, Oldehinkel AJ, Oostra BA, Org E, Padmanabhan S, Palotie A, Paré G, Pattie A, Penninx BWJH, Poulter N, Pramstaller PP, Raitakari OT, Ren M, Rice K, Ridker PM, Riese H, Ripatti S, Robino A, Rotter JI, Rudan I, Saba Y, Saint Pierre A, Sala CF, Sarin AP, Schmidt R, Scott R, Seelen MA, Shields DC, Siscovick D, Sorice R, Stanton A, Stott DJ, Sundström J, Swertz M, Taylor KD, Thom S, Tzoulaki I, Tzourio C, Uitterlinden AG, Völker U, Vollenweider P, Wild S, Willemsen G, Wright AF, Yao J, Thériault S, Conen D, Attia J, Sever P, Debette S, Mook-Kanamori DO, Zeggini E, Spector TD, van der Harst P, Palmer CNA, Vergnaud AC, Loos RJF, Polasek O, Starr JM, Girotto G, Hayward C, Kooner JS, Lindgren CM, Vitart V, Samani NJ, Tuomilehto J, Gyllensten U, Knekt P, Deary IJ, Ciullo M, Elosua R, Keavney BD, Hicks AA, Scott RA, Gasparini P, Laan M, Liu Y, Watkins H, Hartman CA, Salomaa V, Toniolo D, Perola M, Wilson JF, Schmidt H, Zhao JH, Lehtimäki T, van Duijn CM, Gudnason V, Psaty BM, Peters A, Rettig R, James A, Jukema JW, Strachan DP, Palmas W, Metspalu A, Ingelsson E, Boomsma DI, Franco OH, Bochud M, Newton-Cheh C, Munroe PB, Elliott P, Chasman DI, Chakravarti A, Knight J, Morris AP, Levy D, Tobin MD, Snieder H, Caulfield MJ, Ehret GB. *Novel Blood Pressure Locus and Gene Discovery Using Genome-Wide Association Study and Expression Data Sets From Blood and the Kidney*. **Hypertension (Dallas,Tex: 1979)** 2017 Jul 24. pii: HYPERTENSIONAHA.117.09438. doi: 10.1161/HYPERTENSIONAHA.117.09438.

353. Warren HR, Evangelou E, Cabrera CP, Gao H, Ren M, Mifsud B, Ntalla I, Surendran P, Liu C, Cook JP, Kraja AT, Drenos F, Loh M, Verweij N, Marten J, Karaman I, Lepe MP, O'Reilly PF, Knight J, Snieder H, Kato N, He J, Tai ES, Said MA, Porteous D, Alver M, Poulter N, Farrall M, Gansevoort RT, Padmanabhan S, Mägi R, Stanton A, Connell J, Bakker SJ, Metspalu A, Shields DC, Thom S, Brown M, Sever P, Esko T, Hayward C, van der Harst P, Saleheen D, Chowdhury R, Chambers JC, Chasman DI, Chakravarti A, Newton-Cheh C, Lindgren CM, Levy D, Kooner JS, Keavney B, Tomaszewski M, Samani NJ, Howson JM, Tobin MD, Munroe PB, Ehret GB, Wain LV; International Consortium of Blood Pressure (ICBP) 1000G Analyses; BIOS Consortium; Lifelines Cohort Study; Understanding Society Scientific group; CHD Exome+ Consortium; ExomeBP Consortium; T2D-GENES Consortium; GoT2DGenes Consortium; Cohorts for Heart and Ageing Research in Genome Epidemiology (CHARGE) BP Exome Consortium;

International Genomics of Blood Pressure (iGEN-BP) Consortium; UK Biobank CardioMetabolic Consortium BP working group. Völker U, Vollenweider P, Wild S, Willemsen G, Wright AF, Yao J, Thériault S, Conen D, John A, Sever P, Debette S, Mook-Kanamori DO, Zeggini E, Spector TD, van der Harst P, Palmer CN, Vergnaud AC, Loos RJ, Polasek O, Starr JM, Girotto G, Hayward C, Kooner JS, Lindgren CM, Vitart V, Samani NJ, Tuomilehto J, Gyllensten U, Knekt P, Deary IJ, Ciullo M, Elosua R, Keavney BD, Hicks AA, Scott RA, Gasparini P, Laan M, Liu Y, Watkins H, Hartman CA, Salomaa V, Toniolo D, Perola M, Wilson JF, Schmidt H, Zhao JH, Lehtimäki T, van Duijn CM, Gudnason V, Psaty BM, Peters A, Rettig R, James A, Jukema JW, Strachan DP, Palmas W, Metspalu A, Ingelsson E, Boomsma DI, Franco OH, Bochud M, Newton-Cheh C, Munroe PB, Elliott P, Chasman DI, Chakravarti A, Knight J, Morris AP, Levy D, Tobin MD, Snieder H, Caulfield MJ, Ehret GB, Barnes MR, Tzoulaki I, Caulfield MJ, Elliott P. *Genome-wide association analysis identifies novel blood pressure loci and offers biological insights into cardiovascular risk.* **Nat Genet**. 2017 Mar;49(3):403-415. doi: 10.1038/ng.3768.

354. Marouli E, Graff M, Medina-Gomez C, Lo KS, Wood AR, Kjaer TR, Fine RS, Lu Y, Schurmann C, Highland HM, Rüeger S, Thorleifsson G, Justice AE, Lamparter D, Stirrups KE, Turcot V, Young KL, Winkler TW, Esko T, Karaderi T, Locke AE, Masca NG, Ng MC, Mudgal P, Rivas MA, Vedantam S, Mahajan A, Guo X, Abecasis G, Aben KK, Adair LS, Alam DS, Albrecht E, Allin KH, Allison M, Amouyel P, Appel EV, Arveiler D, Asselbergs FW, Auer PL, Balkau B, Banas B, Bang LE, Benn M, Bergmann S, Bielak LF, Blüher M, Boeing H, Boerwinkle E, Böger CA, Bonnycastle LL, Bork-Jensen J, Bots ML, Bottinger EP, Bowden DW, Brandslund I, Breen G, Brilliant MH, Broer L, Burt AA, Butterworth AS, Carey DJ, Caulfield MJ, Chambers JC, Chasman DI, Chen YI, Chowdhury R, Christensen C, Chu AY, Cocca M, Collins FS, Cook JP, Corley J, Galbany JC, Cox AJ, Cuellar-Partida G, Danesh J, Davies G, de Bakker PI, de Borst GJ, de Denus S, de Groot MC, de Mutsert R, Deary IJ, Dedoussis G, Demerath EW, den Hollander AI, Dennis JG, Di Angelantonio E, Drenos F, Du M, Dunning AM, Easton DF, Ebeling T, Edwards TL, Ellinor PT, Elliott P, Evangelou E, Farmaki AE, Faul JD, Feitosa MF, Feng S, Ferrannini E, Ferrario MM, Ferrieres J, Florez JC, Ford I, Fornage M, Franks PW, Frikke-Schmidt R, Galesloot TE, Gan W, Gandin I, Gasparini P, Giedraitis V, Giri A, Girotto G, Gordon SD, Gordon-Larsen P, Gorski M, Grarup N, Grove ML, Gudnason V, Gustafsson S, Hansen T, Harris KM, Harris TB, Hattersley AT, Hayward C, He L, Heid IM, Heikkilä K, Helgeland Ø, Hernesniemi J, Hewitt AW, Hocking LJ, Hollensted M, Holmen OL, Hovingh GK, Howson JM, Hoyng CB, Huang PL, Hveem K, Ikram MA, Ingelsson E, Jackson AU, Jansson JH, Jarvik GP, Jensen GB, Jhun MA, Jia Y, Jiang X, Johansson S, Jørgensen ME, Jørgensen T, Jousilahti P, Jukema JW, Kahali B, Kahn RS, Kähönen M, Kamstrup PR, Kanoni S, Kaprio J, Karaleftheri M, Kardia SL, Karpe F, Kee F, Keeman R, Kiemeney LA, Kitajima H, Kluivers KB, Kocher T, Komulainen P, Kontto J,

Kooner JS, Kooperberg C, Kovacs P, Kriebel J, Kuivaniemi H, Küry S, Kuusisto J, La Bianca M, Laakso M, Lakka TA, Lange EM, Lange LA, Langefeld CD, Langenberg C, Larson EB, Lee IT, Lehtimäki T, Lewis CE, Li H, Li J, Li-Gao R, Lin H, Lin LA, Lin X, Lind L, Lindström J, Linneberg A, Liu Y, Liu Y, Lophatananon A, Luan J, Lubitz SA, Lyytikäinen LP, Mackey DA, Madden PA, Manning AK, Männistö S, Marenne G, Marten J, Martin NG, Mazul AL, Meidtner K, Metspalu A, Mitchell P, Mohlke KL, Mook-Kanamori DO, Morgan A, Morris AD, Morris AP, Müller-Nurasyid M, Munroe PB, Nalls MA, Nauck M, Nelson CP, Neville M, Nielsen SF, Nikus K, Njølstad PR, Nordestgaard BG, Ntalla I, O'Connel JR, Oksa H, Loohuis LM, Ophoff RA, Owen KR, Packard CJ, Padmanabhan S, Palmer CN, Pasterkamp G, Patel AP, Pattie A, Pedersen O, Peissig PL, Peloso GM, Pennell CE, Perola M, Perry JA, Perry JR, Person TN, Pirie A, Polasek O, Posthuma D, Raitakari OT, Rasheed A, Rauramaa R, Reilly DF, Reiner AP, Renström F, Ridker PM, Rioux JD, Robertson N, Robino A, Rolandsson O, Rudan I, Ruth KS, Saleheen D, Salomaa V, Samani NJ, Sandow K, Sapkota Y, Sattar N, Schmidt MK, Schreiner PJ, Schulze MB, Scott RA, Segura-Lepe MP, Shah S, Sim X, Sivapalaratnam S, Small KS, Smith AV, Smith JA, Southam L, Spector TD, Speliotes EK, Starr JM, Steinthorsdottir V, Stringham HM, Stumvoll M, Surendran P, 't Hart LM, Tansey KE, Tardif JC, Taylor KD, Teumer A, Thompson DJ, Thorsteinsdottir U, Thuesen BH, Tönjes A, Tromp G, Trompet S, Tsafantakis E, Tuomilehto J, Tybjaerg-Hansen A, Tyrer JP, Uher R, Uitterlinden AG, Ulivi S, van der Laan SW, Van Der Leij AR, van Duijn CM, van Schoor NM, van Setten J, Varbo A, Varga TV, Varma R, Edwards DR, Vermeulen SH, Vestergaard H, Vitart V, Vogt TF, Vozzi D, Walker M, Wang F, Wang CA, Wang S, Wang Y, Wareham NJ, Warren HR, Wessel J, Willems SM, Wilson JG, Witte DR, Woods MO, Wu Y, Yaghootkar H, Yao J, Yao P, Yerges-Armstrong LM, Young R, Zeggini E, Zhan X, Zhang W, Zhao JH, Zhao W, Zhao W, Zheng H, Zhou W; EPIC-InterAct Consortium; CHD Exome+ Consortium; ExomeBP Consortium; T2D-Genes Consortium; GoT2D Genes Consortium; Global Lipids Genetics Consortium; ReproGen Consortium; MAGIC Investigators, Rotter JI, Boehnke M, Kathiresan S, McCarthy MI, Willer CJ, Stefansson K, Borecki IB, Liu DJ, North KE, Heard-Costa NL, Pers TH, Lindgren CM, Oxvig C, Kutalik Z, Rivadeneira F, Loos RJ, Frayling TM, Hirschhorn JN, Deloukas P, Lettre G. *Rare and low-frequency coding variants alter human adult height*. **Nature.**2017 Feb 9;542(7640):186-190. doi: 10.1038/nature21039.

355. Tepper BJ, Melis M, Koelliker Y, Gasparini P, Ahijevych KL, Tomassini Barbarossa I. *Factors Influencing the Phenotypic Characterization of the Oral Marker, PROP*. **Nutrients**. 2017 Nov 23;9(12). pii: E1275. doi: 10.3390/nu9121275.

356. Corre T, Arjona FJ, Hayward C, Youhanna S, de Baaij JHF, Belge H, Nägele N, Debaix H, Blanchard MG, Traglia M, Harris SE, Ulivi S, Rueedi R, Lamparter D, Macé A, Sala C, Lenarduzzi S, Ponte B, Pruijm M, Ackermann D, Ehret G, Baptista D, Polasek O, Rudan I, Hurd TW, Hastie ND, Vitart V, Waeber G, Kutalik Z,

Bergmann S, Vargas-Poussou R, Konrad M, Gasparini P, Deary IJ, Starr JM, Toniolo D, Vollenweider P, Hoenderop JGJ, Bindels RJM, Bochud M, Devuyst O. *Genome-Wide Meta-Analysis Unravels Interactions between Magnesium Homeostasis and Metabolic Phenotypes*. **J Am Soc Nephrol**. 2018 Jan;29(1):335-348. doi: 10.1681/ASN.2017030267.

357. Trimouille A, Houcinat N, Vuillaume ML, Fergelot P, Boucher C, Toutain J, Caignec CL, Vincent M, Nizon M, Andrieux J, Vanlerberghe C, Delobel B, Duban B, Mansour S, Baple E, McKeown C, Poke G, Robertshaw K, Fifield E, Fabretto A, Pecile V, Gasparini P, Carrozzi M, Lacombe D, Arveiler B, Rooryck C, Moutton S. *19p13 microduplications encompassing NFIX are responsible for intellectual disability, short stature and small head circumference*. **Eur J Hum Genet**. 2018 Jan;26(1):85-93. doi: 10.1038/s41431-017-0037-7.

358. Turcot V, Lu Y, Highland HM, Schurmann C, Justice AE, Fine RS, Bradfield JP, Esko T, Giri A, Graff M, Guo X, Hendricks AE, Karaderi T, Lempradl A, Locke AE, Mahajan A, Marouli E, Sivapalaratnam S, Young KL, Alfred T, Feitosa MF, Masca NGD, Manning AK, Medina-Gomez C, Mudgal P, Ng MCY, Reiner AP, Vedantam S, Willems SM, Winkler TW, Abecasis G, Aben KK, Alam DS, Alharthi SE, Allison M, Amouyel P, Asselbergs FW, Auer PL, Balkau B, Bang LE, Barroso I, Bastarache L, Benn M, Bergmann S, Bielak LF, Blüher M, Boehnke M, Boeing H, Boerwinkle E, Böger CA, Bork-Jensen J, Bots ML, Bottinger EP, Bowden DW, Brandslund I, Breen G, Brilliant MH, Broer L, Brumat M, Burt AA, Butterworth AS, Campbell PT, Cappellani S, Carey DJ, Catamo E, Caulfield MJ, Chambers JC, Chasman DI, Chen YI, Chowdhury R, Christensen C, Chu AY, Cocca M, Collins FS, Cook JP, Corley J, Corominas Galbany J, Cox AJ, Crosslin DS, Cuellar-Partida G, D'Eustacchio A, Danesh J, Davies G, Bakker PIW, Groot MCH, Mutsert R, Deary IJ, Dedoussis G, Demerath EW, Heijer M, Hollander AI, Ruijter HM, Dennis JG, Denny JC, Di Angelantonio E, Drenos F, Du M, Dubé MP, Dunning AM, Easton DF, Edwards TL, Ellinghaus D, Ellinor PT, Elliott P, Evangelou E, Farmaki AE, Farooqi IS, Faul JD, Fauser S, Feng S, Ferrannini E, Ferrieres J, Florez JC, Ford I, Fornage M, Franco OH, Franke A, Franks PW, Friedrich N, Frikke-Schmidt R, Galesloot TE, Gan W, Gandin I, Gasparini P, Gibson J, Giedraitis V, Gjesing AP, Gordon-Larsen P, Gorski M, Grabe HJ, Grant SFA, Grarup N, Griffiths HL, Grove ML, Gudnason V, Gustafsson S, Haessler J, Hakonarson H, Hammerschlag AR, Hansen T, Harris KM, Harris TB, Hattersley AT, Have CT, Hayward C, He L, Heard-Costa NL, Heath AC, Heid IM, Helgeland Ø, Hernesniemi J, Hewitt AW, Holmen OL, Hovingh GK, Howson JMM, Hu Y, Huang PL, Huffman JE, Ikram MA, Ingelsson E, Jackson AU, Jansson JH, Jarvik GP, Jensen GB, Jia Y, Johansson S, Jørgensen ME, Jørgensen T, Jukema JW, Kahali B, Kahn RS, Kähönen M, Kamstrup PR, Kanoni S, Kaprio J, Karaleftheri M, Kardia SLR, Karpe F, Kathiresan S, Kee F, Kiemeney LA, Kim E, Kitajima H, Komulainen P, Kooner JS, Kooperberg C, Korhonen T, Kovacs P, Kuivaniemi H, Kutalik Z, Kuulasmaa K, Kuusisto J, Laakso M, Lakka TA, Lamparter D,

Lange EM, Lange LA, Langenberg C, Larson EB, Lee NR, Lehtimäki T, Lewis CE, Li H, Li J, Li-Gao R, Lin H, Lin KH, Lin LA, Lin X, Lind L, Lindström J, Linneberg A, Liu CT, Liu DJ, Liu Y, Lo KS, Lophatananon A, Lotery AJ, Loukola A, Luan J, Lubitz SA, Lyytikäinen LP, Männistö S, Marenne G, Mazul AL, McCarthy MI, McKean-Cowdin R, Medland SE, Meidtner K, Milani L, Mistry V, Mitchell P, Mohlke KL, Moilanen L, Moitry M, Montgomery GW, Mook-Kanamori DO, Moore C, Mori TA, Morris AD, Morris AP, Müller-Nurasyid M, Munroe PB, Nalls MA, Narisu N, Nelson CP, Neville M, Nielsen SF, Nikus K, Njølstad PR, Nordestgaard BG, Nyholt DR, O'Connel JR, O'Donoghue ML, Olde Loohuis LM, Ophoff RA, Owen KR, Packard CJ, Padmanabhan S, Palmer CNA, Palmer ND, Pasterkamp G, Patel AP, Pattie A, Pedersen O, Peissig PL, Peloso GM, Pennell CE, Perola M, Perry JA, Perry JRB, Pers TH, Person TN, Peters A, Petersen ERB, Peyser PA, Pirie A, Polasek O, Polderman TJ, Puolijoki H, Raitakari OT, Rasheed A, Rauramaa R, Reilly DF, Renström F, Rheinberger M, Ridker PM, Rioux JD, Rivas MA, Roberts DJ, Robertson NR, Robino A, Rolandsson O, Rudan I, Ruth KS, Saleheen D, Salomaa V, Samani NJ, Sapkota Y, Sattar N, Schoen RE, Schreiner PJ, Schulze MB, Scott RA, Segura-Lepe MP, Shah SH, Sheu WH, Sim X, Slater AJ, Small KS, Smith AV, Southam L, Spector TD, Speliotes EK, Starr JM, Stefansson K, Steinthorsdottir V, Stirrups KE, Strauch K, Stringham HM, Stumvoll M, Sun L, Surendran P, Swift AJ, Tada H, Tansey KE, Tardif JC, Taylor KD, Teumer A, Thompson DJ, Thorleifsson G, Thorsteinsdottir U, Thuesen BH, Tönjes A, Tromp G, Trompet S, Tsafantakis E, Tuomilehto J, Tybjaerg-Hansen A, Tyrer JP, Uher R, Uitterlinden AG, Uusitupa M, Laan SW, Duijn CM, Leeuwen N, van Setten J, Vanhala M, Varbo A, Varga TV, Varma R, Velez Edwards DR, Vermeulen SH, Veronesi G, Vestergaard H, Vitart V, Vogt TF, Völker U, Vuckovic D, Wagenknecht LE, Walker M, Wallentin L, Wang F, Wang CA, Wang S, Wang Y, Ware EB, Wareham NJ, Warren HR, Waterworth DM, Wessel J, White HD, Willer CJ, Wilson JG, Witte DR, Wood AR, Wu Y, Yaghootkar H, Yao J, Yao P, Yerges-Armstrong LM, Young R, Zeggini E, Zhan X, Zhang W, Zhao JH, Zhao W, Zhao W, Zhou W, Zondervan KT, Rotter JI, Pospisilik JA, Rivadeneira F, Borecki IB, Deloukas P, Frayling TM, Lettre G, North KE, Lindgren CM, Hirschhorn JN, Loos RJF; CHD Exome+ Consortium; EPIC-CVD Consortium; ExomeBP Consortium; Global Lipids Genetic Consortium; GoT2D Genes Consortium; EPIC InterAct Consortium; INTERVAL Study; ReproGen Consortium; T2D-Genes Consortium; MAGIC Investigators; Understanding Society Scientific Group. *Protein-altering variants associated with body mass index implicate pathways that control energy intake and expenditure in obesity*. **Nat Genet**. 2018 Jan;50(1):26-41. doi: 10.1038/s41588-017-0011-x.

359. Espino Guarch M, Font-Llitjós M, Murillo-Cuesta S, Errasti-Murugarren E, Celaya AM, Girotto G, Vuckovic D, Mezzavilla M, Vilches C, Bodoy S, Sahún I, González L, Prat E, Zorzano A, Dierssen M, Varela-Nieto I, Gasparini P, Palacín M, Nunes V. *Mutations in L-type amino acid transporter-2 support SLC7A8*

as a novel gene involved in age-related hearing loss. **Elife**. 2018 Jan 22;7. pii: e31511. doi: 10.7554/eLife.31511.

360. Sung YJ, Winkler TW, de Las Fuentes L, Bentley AR, Brown MR, Kraja AT, Schwander K, Ntalla I, Guo X, Franceschini N, Lu Y, Cheng CY, Sim X, Vojinovic D, Marten J, Musani SK, Li C, Feitosa MF, Kilpeläinen TO, Richard MA, Noordam R, Aslibekyan S, Aschard H, Bartz TM, Dorajoo R, Liu Y, Manning AK, Rankinen T, Smith AV, Tajuddin SM, Tayo BO, Warren HR, Zhao W, Zhou Y, Matoba N, Sofer T, Alver M, Amini M, Boissel M, Chai JF, Chen X, Divers J, Gandin I, Gao C, Giulianini F, Goel A, Harris SE, Hartwig FP, Horimoto ARVR, Hsu FC, Jackson AU, Kähönen M, Kasturiratne A, Kühnel B, Leander K, Lee WJ, Lin KH, 'an Luan J, McKenzie CA, Meian H, Nelson CP, Rauramaa R, Schupf N, Scott RA, Sheu WHH, Stančáková A, Takeuchi F, van der Most PJ, Varga TV, Wang H, Wang Y, Ware EB, Weiss S, Wen W, Yanek LR, Zhang W, Zhao JH, Afaq S, Alfred T, Amin N, Arking D, Aung T, Barr RG, Bielak LF, Boerwinkle E, Bottinger EP, Braund PS, Brody JA, Broeckel U, Cabrera CP, Cade B, Caizheng Y, Campbell A, Canouil M, Chakravarti A; CHARGE Neurology Working Group, Chauhan G, Christensen K, Cocca M; COGENT-Kidney Consortium, Collins FS, Connell JM, de Mutsert R, de Silva HJ, Debette S, Dörr M, Duan Q, Eaton CB, Ehret G, Evangelou E, Faul JD, Fisher VA, Forouhi NG, Franco OH, Friedlander Y, Gao H; GIANT Consortium, Gigante B, Graff M, Gu CC, Gu D, Gupta P, Hagenaars SP, Harris TB, He J, Heikkinen S, Heng CK, Hirata M, Hofman A, Howard BV, Hunt S, Irvin MR, Jia Y, Joehanes R, Justice AE, Katsuya T, Kaufman J, Kerrison ND, Khor CC, Koh WP, Koistinen HA, Komulainen P, Kooperberg C, Krieger JE, Kubo M, Kuusisto J, Langefeld CD, Langenberg C, Launer LJ, Lehne B, Lewis CE, Li Y; Lifelines Cohort Study, Lim SH, Lin S, Liu CT, Liu J, Liu J, Liu K, Liu Y, Loh M, Lohman KK, Long J, Louie T, Mägi R, Mahajan A, Meitinger T, Metspalu A, Milani L, Momozawa Y, Morris AP, Mosley TH Jr, Munson P, Murray AD, Nalls MA, Nasri U, Norris JM, North K, Ogunniyi A, Padmanabhan S, Palmas WR, Palmer ND, Pankow JS, Pedersen NL, Peters A, Peyser PA, Polasek O, Raitakari OT, Renström F, Rice TK, Ridker PM, Robino A, Robinson JG, Rose LM, Rudan I, Sabanayagam C, Salako BL, Sandow K, Schmidt CO, Schreiner PJ, Scott WR, Seshadri S, Sever P, Sitlani CM, Smith JA, Snieder H, Starr JM, Strauch K, Tang H, Taylor KD, Teo YY, Tham YC, Uitterlinden AG, Waldenberger M, Wang L, Wang YX, Wei WB, Williams C, Wilson G, Wojczynski MK, Yao J, Yuan JM, Zonderman AB, Becker DM, Boehnke M, Bowden DW, Chambers JC, Chen YI, de Faire U, Deary IJ, Esko T, Farrall M, Forrester T, Franks PW, Freedman BI, Froguel P, Gasparini P, Gieger C, Horta BL, Hung YJ, Jonas JB, Kato N, Kooner JS, Laakso M, Lehtimäki T, Liang KW, Magnusson PKE, Newman AB, Oldehinkel AJ, Pereira AC, Redline S, Rettig R, Samani NJ, Scott J, Shu XO, van der Harst P, Wagenknecht LE, Wareham NJ, Watkins H, Weir DR, Wickremasinghe AR, Wu T, Zheng W, Kamatani Y, Laurie CC, Bouchard C, Cooper RS, Evans MK, Gudnason V, Kardia SLR, Kritchevsky SB, Levy D, O'Connell JR, Psaty BM, van Dam RM, Sims M, Arnett DK, Mook-Kanamori

DO, Kelly TN, Fox ER, Hayward C, Fornage M, Rotimi CN, Province MA, van Duijn CM, Tai ES, Wong TY, Loos RJF, Reiner AP, Rotter JI, Zhu X, Bierut LJ, Gauderman WJ, Caulfield MJ, Elliott P, Rice K, Munroe PB, Morrison AC, Cupples LA, Rao DC, Chasman DI. *A Large-Scale Multi-ancestry Genome-wide Study Accounting for Smoking Behavior Identifies Multiple Significant Loci for Blood Pressure*. **Am J Hum Genet**. 2018 Mar 1;102(3):375-400. doi: 10.1016/j.ajhg.2018.01.015.

361. Pinart M, Nimptsch K, Bouwman J, Dragsted LO, Yang C, De Cock N, Lachat C, Perozzi G, Canali R, Lombardo R, D'Archivio M, Guillaume M, Donneau AF, Jeran S, Linseisen J, Kleiser C, Nöthlings U, Barbaresko J, Boeing H, Stelmach-Mardas M, Heuer T, Laird E, Walton J, Gasparini P, Robino A, Castaño L, Rojo-Martínez G, Merino J, Masana L, Standl M, Schulz H, Biagi E, Nurk E, Matthys C, Gobbetti M, de Angelis M, Windler E, Zyriax BC, Tafforeau J, Pischon T. *Joint Data Analysis in Nutritional Epidemiology: Identification of Observational Studies and Minimal Requirements*. **J Nutr**. 2018 Feb 1;148(2):285-297. doi: 10.1093/jn/nxx037.

362. Hysi PG, Valdes AM, Liu F, Furlotte NA, Evans DM, Bataille V, Visconti A, Hemani G, McMahon G, Ring SM, Smith GD, Duffy DL, Zhu G, Gordon SD, Medland SE, Lin BD, Willemsen G, Jan Hottenga J, Vuckovic D, Girotto G, Gandin I, Sala C, Concas MP, Brumat M, Gasparini P, Toniolo D, Cocca M, Robino A, Yazar S, Hewitt AW, Chen Y, Zeng C, Uitterlinden AG, Ikram MA, Hamer MA, van Duijn CM, Nijsten T, Mackey DA, Falchi M, Boomsma DI, Martin NG, Hinds DA, Kayser M, Spector TD; International Visible Trait Genetics Consortium; International Visible Trait Genetics Consortium. *Genome-wide association meta-analysis of individuals of European ancestry identifies new loci explaining a substantial fraction of hair color variation and heritability*. **Nat Genet**. 2018 May;50(5):652-656. doi: 10.1038/s41588-018-0100-5.

363. Vuckovic D, Mezzavilla M, Cocca M, Morgan A, Brumat M, Catamo E, Concas MP, Biino G, Franzè A, Ambrosetti U, Pirastu M, Gasparini P, Girotto G. *Whole-genome sequencing reveals new insights into age-related hearing loss: cumulative effects, pleiotropy and the role of selection*. **Eur J Hum Genet**. 2018 Aug;26(8):1167-1179. doi: 10.1038/s41431-018-0126-2.

364. Feitosa MF, Kraja AT, Chasman DI, Sung YJ, Winkler TW, Ntalla I, Guo X, Franceschini N, Cheng CY, Sim X, Vojinovic D, Marten J, Musani SK, Li C, Bentley AR, Brown MR, Schwander K, Richard MA, Noordam R, Aschard H, Bartz TM, Bielak LF, Dorajoo R, Fisher V, Hartwig FP, Horimoto ARVR, Lohman KK, Manning AK, Rankinen T, Smith AV, Tajuddin SM, Wojczynski MK, Alver M, Boissel M, Cai Q, Campbell A, Chai JF, Chen X, Divers J, Gao C, Goel A, Hagemeijer Y, Harris SE, He M, Hsu FC, Jackson AU, Kähönen M, Kasturiratne A, Komulainen P, Kühnel B, Laguzzi F, Luan J, Matoba N, Nolte IM, Padmanabhan S, Riaz M, Rueedi R, Robino A, Said MA, Scott RA, Sofer T, Stančáková A, Takeuchi F, Tayo BO, van der Most PJ, Varga TV, Vitart V, Wang Y, Ware EB, Warren HR, Weiss S, Wen W, Yanek LR, Zhang W, Zhao JH, Afaq

S, Amin N, Amini M, Arking DE, Aung T, Boerwinkle E, Borecki I, Broeckel U, Brown M, Brumat M, Burke GL, Canouil M, Chakravarti A, Charumathi S, Ida Chen YD, Connell JM, Correa A, de Las Fuentes L, de Mutsert R, de Silva HJ, Deng X, Ding J, Duan Q, Eaton CB, Ehret G, Eppinga RN, Evangelou E, Faul JD, Felix SB, Forouhi NG, Forrester T, Franco OH, Friedlander Y, Gandin I, Gao H, Ghanbari M, Gigante B, Gu CC, Gu D, Hagenaars SP, Hallmans G, Harris TB, He J, Heikkinen S, Heng CK, Hirata M, Howard BV, Ikram MA; InterAct Consortium, John U, Katsuya T, Khor CC, Kilpeläinen TO, Koh WP, Krieger JE, Kritchevsky SB, Kubo M, Kuusisto J, Lakka TA, Langefeld CD, Langenberg C, Launer LJ, Lehne B, Lewis CE, Li Y, Lin S, Liu J, Liu J, Loh M, Louie T, Mägi R, McKenzie CA, Meitinger T, Metspalu A, Milaneschi Y, Milani L, Mohlke KL, Momozawa Y, Nalls MA, Nelson CP, Sotoodehnia N, Norris JM, O'Connell JR, Palmer ND, Perls T, Pedersen NL, Peters A, Peyser PA, Poulter N, Raffel LJ, Raitakari OT, Roll K, Rose LM, Rosendaal FR, Rotter JI, Schmidt CO, Schreiner PJ, Schupf N, Scott WR, Sever PS, Shi Y, Sidney S, Sims M, Sitlani CM, Smith JA, Snieder H, Starr JM, Strauch K, Stringham HM, Tan NYQ, Tang H, Taylor KD, Teo YY, Tham YC, Turner ST, Uitterlinden AG, Vollenweider P, Waldenberger M, Wang L, Wang YX, Wei WB, Williams C, Yao J, Yu C, Yuan JM, Zhao W, Zonderman AB, Becker DM, Boehnke M, Bowden DW, Chambers JC, Deary IJ, Esko T, Farrall M, Franks PW, Freedman BI, Froguel P, Gasparini P, Gieger C, Jonas JB, Kamatani Y, Kato N, Kooner JS, Kutalik Z, Laakso M, Laurie CC, Leander K, Lehtimäki T, Study LC, Magnusson PKE, Oldehinkel AJ, Penninx BWJH, Polasek O, Porteous DJ, Rauramaa R, Samani NJ, Scott J, Shu XO, van der Harst P, Wagenknecht LE, Wareham NJ, Watkins H, Weir DR, Wickremasinghe AR, Wu T, Zheng W, Bouchard C, Christensen K, Evans MK, Gudnason V, Horta BL, Kardia SLR, Liu Y, Pereira AC, Psaty BM, Ridker PM, van Dam RM, Gauderman WJ, Zhu X, Mook-Kanamori DO, Fornage M, Rotimi CN, Cupples LA, Kelly TN, Fox ER, Hayward C, van Duijn CM, Tai ES, Wong TY, Kooperberg C, Palmas W, Rice K, Morrison AC, Elliott P, Caulfield MJ, Munroe PB, Rao DC, Province MA, Levy D. *Novel genetic associations for blood pressure identified via gene-alcohol interaction in up to 570K individuals across multiple ancestries*. **PLoS One**. 2018 Jun 18;13(6):e0198166. doi: 10.1371/journal.pone.0198166. eCollection 2018.

365. Prins BP, Mead TJ, Brody JA, Sveinbjornsson G, Ntalla I, Bihlmeyer NA, van den Berg M, Bork-Jensen J, Cappellani S, Van Duijvenboden S, Klena NT, Gabriel GC, Liu X, Gulec C, Grarup N, Haessler J, Hall LM, Iorio A, Isaacs A, Li-Gao R, Lin H, Liu CT, Lyytikäinen LP, Marten J, Mei H, Müller-Nurasyid M, Orini M, Padmanabhan S, Radmanesh F, Ramirez J, Robino A, Schwartz M, van Setten J, Smith AV, Verweij N, Warren HR, Weiss S, Alonso A, Arnar DO, Bots ML, de Boer RA, Dominiczak AF, Eijgelsheim M, Ellinor PT, Guo X, Felix SB, Harris TB, Hayward C, Heckbert SR, Huang PL, Jukema JW, Kähönen M, Kors JA, Lambiase PD, Launer LJ, Li M, Linneberg A, Nelson CP, Pedersen O, Perez M, Peters A, Polasek O, Psaty BM, Raitakari OT, Rice KM,

Rotter JI, Sinner MF, Soliman EZ, Spector TD, Strauch K, Thorsteinsdottir U, Tinker A, Trompet S, Uitterlinden A, Vaartjes I, van der Meer P, Völker U, Völzke H, Waldenberger M, Wilson JG, Xie Z, Asselbergs FW, Dörr M, van Duijn CM, Gasparini P, Gudbjartsson DF, Gudnason V, Hansen T, Kääb S, Kanters JK, Kooperberg C, Lehtimäki T, Lin HJ, Lubitz SA, Mook-Kanamori DO, Conti FJ, Newton-Cheh CH, Rosand J, Rudan I, Samani NJ, Sinagra G, Smith BH, Holm H, Stricker BH, Ulivi S, Sotoodehnia N, Apte SS, van der Harst P, Stefansson K, Munroe PB, Arking DE, Lo CW, Jamshidi Y. *Exome-chip meta-analysis identifies novel loci associated with cardiac conduction, including ADAMTS6*. **Genome Biol**. 2018 Jul 17;19(1):87. doi: 10.1186/s13059-018-1457-6.

366. Lee JJ, Wedow R, Okbay A, Kong E, Maghzian O, Zacher M, Nguyen-Viet TA, Bowers P, Sidorenko J, Karlsson Linnér R, Fontana MA, Kundu T, Lee C, Li H, Li R, Royer R, Timshel PN, Walters RK, Willoughby EA, Yengo L; 23andMe Research Team; COGENT (Cognitive Genomics Consortium); Social Science Genetic Association Consortium, Alver M, Bao Y, Clark DW, Day FR, Furlotte NA, Joshi PK, Kemper KE, Kleinman A, Langenberg C, Mägi R, Trampush JW, Verma SS, Wu Y, Lam M, Zhao JH, Zheng Z, Boardman JD, Campbell H, Freese J, Harris KM, Hayward C, Herd P, Kumari M, Lencz T, Luan J, Malhotra AK, Metspalu A, Milani L, Ong KK, Perry JRB, Porteous DJ, Ritchie MD, Smart MC, Smith BH, Tung JY, Wareham NJ, Wilson JF, Beauchamp JP, Conley DC, Esko T, Lehrer SF, Magnusson PKE, Oskarsson S, Pers TH, Robinson MR, Thom K, Watson C, Chabris CF, Meyer MN, Laibson DI, Yang J, Johannesson M, Koellinger PD, Turley P, Visscher PM, Benjamin DJ, Cesarini D. Agee M, Alipanahi B, Auton A, Bell RK, Bryc K, Elson SL, Fontanillas P, Hinds DA, McCreight JC, Huber KE, Litterman NK, McIntyre MH, Mountain JL, Noblin ES, Northover CAM, Pitts SJ, Sathirapongsasuti JF, Sazonova OV, Shelton JF, Shringarpure S, Tian C, Vacic V, Wilson CH, Okbay A, Beauchamp JP, Fontana MA, Lee JJ, Pers TH, Rietveld CA, Turley P, Chen GB, Emilsson V, Meddens SFW, Oskarsson S, Pickrell JK, Thom K, Timshel P, Vlaming R, Abdellaoui A, Ahluwalia TS, Bacelis J, Baumbach C, Bjornsdottir G, Brandsma JH, Concas MP, Derringer J, Furlotte NA, Galesloot TE, Girotto G, Gupta R, Hall LM, Harris SE, Hofer E, Horikoshi M, Huffman JE, Kaasik K, Kalafati IP, Karlsson R, Kong A, Lahti J, van der Lee SJ, Leeuw C, Lind PA, Lindgren KO, Liu T, Mangino M, Marten J, Mihailov E, Miller MB, van der Most PJ, Oldmeadow C, Payton A, Pervjakova N, Peyrot WJ, Qian Y, Raitakari O, Rueedi R, Salvi E, Schmidt B, Schraut KE, Shi J, Smith AV, Poot RA, St Pourcain B, Teumer A, Thorleifsson G, Verweij N, Vuckovic D, Wellmann J, Westra HJ, Yang J, Zhao W, Zhu Z, Alizadeh BZ, Amin N, Bakshi A, Baumeister SE, Biino G, Bønnelykke K, Boyle PA, Campbell H, Cappuccio FP, Davies G, De Neve JE, Deloukas P, Demuth I, Ding J, Eibich P, Eisele L, Eklund N, Evans DM, Faul JD, Feitosa MF, Forstner AJ, Gandin I, Gunnarsson B, Halldórsson BV, Harris TB, Heath AC, Hocking LJ, Holliday EG, Homuth G, Horan MA, Hottenga JJ, de Jager PL,

Joshi PK, Jugessur A, Kaakinen MA, Kähönen M, Kanoni S, Keltigangas-Järvinen L, Kiemeney LALM, Kolcic I, Koskinen S, Kraja AT, Kroh M, Kutalik Z, Latvala A, Launer LJ, Lebreton MP, Levinson DF, Lichtenstein P, Lichtner P, Liewald DCM, Loukola LCSA, Madden PA, Mägi R, Mäki-Opas T, Marioni RE, Marques-Vidal P, Meddens GA, McMahon G, Meisinger C, Meitinger T, Milaneschi Y, Milani L, Montgomery GW, Myhre R, Nelson CP, Nyholt DR, Ollier WER, Palotie A, Paternoster L, Pedersen NL, Petrovic KE, Porteous DJ, Räikkönen K, Ring SM, Robino A, Rostapshova O, Rudan I, Rustichini A, Salomaa V, Sanders AR, Sarin AP, Schmidt H, Scott RJ, Smith BH, Smith JA, Staessen JA, Steinhagen-Thiessen E, Strauch K, Terracciano A, Tobin MD, Ulivi S, Vaccargiu S, Quaye L, van Rooij FJA, Venturini C, Vinkhuyzen AAE, Völker U, Völzke H, Vonk JM, Vozzi D, Waage J, Ware EB, Willemsen G, Attia JR, Bennett DA, Berger K, Bertram L, Bisgaard H, Boomsma DI, Borecki IB, Bültmann U, Chabris CF, Cucca F, Cusi D, Deary IJ, Dedoussis GV, van Duijn CM, Eriksson JG, Franke B, Franke L, Gasparini P, Gejman PV, Gieger C, Grabe HJ, Gratten J, Groenen PJF, Gudnason V, van der Harst P, Hayward C, Hinds DA, Hoffmann W, Hyppönen E, Iacono WG, Jacobsson B, Järvelin MR, Jöckel KH, Kaprio J, Kardia SLR, Lehtimäki T, Lehrer SF, Magnusson PKE, Martin NG, McGue M, Metspalu A, Pendleton N, Penninx BWJH, Perola M, Pirastu N, Pirastu M, Polasek O, Posthuma D, Power C, Province MA, Samani NJ, Schlessinger D, Schmidt R, Sørensen TIA, Spector TD, Stefansson K, Thorsteinsdottir U, Thurik AR, Timpson NJ, Tiemeier H, Tung JY, Uitterlinden AG, Vitart V, Vollenweider P, Weir DR, Wilson JF, Wright AF, Conley DC, Krueger RF, Smith GD, Hofman A, Laibson DI, Medland SE, Meyer MN, Yang J, Johannesson M, Visscher PM, Esko T, Koellinger PD, Cesarini D, Benjamin DJ. *Gene discovery and polygenic prediction from a genome-wide association study of educational attainment in 1.1 million individuals*. **Nat Genet**. 2018 Aug;50(8):1112-1121. doi: 10.1038/s41588-018-0147-3.

367. van Setten J, Brody JA, Jamshidi Y, Swenson BR, Butler AM, Campbell H, Del Greco FM, Evans DS, Gibson Q, Gudbjartsson DF, Kerr KF, Krijthe BP, Lyytikäinen LP, Müller C, Müller-Nurasyid M, Nolte IM, Padmanabhan S, Ritchie MD, Robino A, Smith AV, Steri M, Tanaka T, Teumer A, Trompet S, Ulivi S, Verweij N, Yin X, Arnar DO, Asselbergs FW, Bader JS, Barnard J, Bis J, Blankenberg S, Boerwinkle E, Bradford Y, Buckley BM, Chung MK, Crawford D, den Hoed M, Denny JC, Dominiczak AF, Ehret GB, Eijgelsheim M, Ellinor PT, Felix SB, Franco OH, Franke L, Harris TB, Holm H, Ilaria G, Iorio A, Kähönen M, Kolcic I, Kors JA, Lakatta EG, Launer LJ, Lin H, Lin HJ, Loos RJF, Lubitz SA, Macfarlane PW, Magnani JW, Leach IM, Meitinger T, Mitchell BD, Munzel T, Papanicolaou GJ, Peters A, Pfeufer A, Pramstaller PP, Raitakari OT, Rotter JI, Rudan I, Samani NJ, Schlessinger D, Silva Aldana CT, Sinner MF, Smith JD, Snieder H, Soliman EZ, Spector TD, Stott DJ, Strauch K, Tarasov KV, Thorsteinsdottir U, Uitterlinden AG, Van Wagoner DR, Völker U, Völzke H, Waldenberger M, Jan Westra H, Wild PS, Zeller T, Alonso A, Avery CL, Bandinelli S, Benjamin EJ, Cucca F, Dörr M, Ferrucci L,

Gasparini P, Gudnason V, Hayward C, Heckbert SR, Hicks AA, Jukema JW, Kääb S, Lehtimäki T, Liu Y, Munroe PB, Parsa A, Polasek O, Psaty BM, Roden DM, Schnabel RB, Sinagra G, Stefansson K, Stricker BH, van der Harst P, van Duijn CM, Wilson JF, Gharib SA, de Bakker PIW, Isaacs A, Arking DE, Sotoodehnia N. *PR interval genome-wide association meta-analysis identifies 50 loci associated with atrial and atrioventricular electrical activity*. **Nat Commun**. 2018 Jul 25;9(1):2904. doi: 10.1038/s41467-018-04766-9.

368. Fetoni AR, Zorzi V, Paciello F, Ziraldo G, Peres C, Raspa M, Scavizzi F, Salvatore AM, Crispino G, Tognola G, Gentile G, Spampinato AG, Cuccaro D, Guarnaccia M, Morello G, Van Camp G, Fransen E, Brumat M, Girotto G, Paludetti G, Gasparini P, Cavallaro S, Mammano F. *Cx26 partial loss causes accelerated presbycusis by redox imbalance and dysregulation of Nfr2 pathway*. **Redox Biol**. 2018 Oct;19:301-317. doi: 10.1016/j.redox.2018.08.002.

369. Evangelou E, Warren HR, Mosen-Ansorena D, Mifsud B, Pazoki R, Gao H, Ntritsos G, Dimou N, Cabrera CP, Karaman I, Ng FL, Evangelou M, Witkowska K, Tzanis E, Hellwege JN, Giri A, Velez Edwards DR, Sun YV, Cho K, Gaziano JM, Wilson PWF, Tsao PS, Kovesdy CP, Esko T, Mägi R, Milani L, Almgren P, Boutin T, Debette S, Ding J, Giulianini F, Holliday EG, Jackson AU, Li-Gao R, Lin WY, Luan J, Mangino M, Oldmeadow C, Prins BP, Qian Y, Sargurupremraj M, Shah N, Surendran P, Thériault S, Verweij N, Willems SM, Zhao JH, Amouyel P, Connell J, de Mutsert R, Doney ASF, Farrall M, Menni C, Morris AD, Noordam R, Paré G, Poulter NR, Shields DC, Stanton A, Thom S, Abecasis G, Amin N, Arking DE, Ayers KL, Barbieri CM, Batini C, Bis JC, Blake T, Bochud M, Boehnke M, Boerwinkle E, Boomsma DI, Bottinger EP, Braund PS, Brumat M, Campbell A, Campbell H, Chakravarti A, Chambers JC, Chauhan G, Ciullo M, Cocca M, Collins F, Cordell HJ, Davies G, de Borst MH, de Geus EJ, Deary IJ, Deelen J, Del Greco M F, Demirkale CY, Dörr M, Ehret GB, Elosua R, Enroth S, Erzurumluoglu AM, Ferreira T, Frånberg M, Franco OH, Gandin I, Gasparini P, Giedraitis V, Gieger C, Girotto G, Goel A, Gow AJ, Gudnason V, Guo X, Gyllensten U, Hamsten A, Harris TB, Harris SE, Hartman CA, Havulinna AS, Hicks AA, Hofer E, Hofman A, Hottenga JJ, Huffman JE, Hwang SJ, Ingelsson E, James A, Jansen R, Jarvelin MR, Joehanes R, Johansson Å, Johnson AD, Joshi PK, Jousilahti P, Jukema JW, Jula A, Kähönen M, Kathiresan S, Keavney BD, Khaw KT, Knekt P, Knight J, Kolcic I, Kooner JS, Koskinen S, Kristiansson K, Kutalik Z, Laan M, Larson M, Launer LJ, Lehne B, Lehtimäki T, Liewald DCM, Lin L, Lind L, Lindgren CM, Liu Y, Loos RJF, Lopez LM, Lu Y, Lyytikäinen LP, Mahajan A, Mamasoula C, Marrugat J, Marten J, Milaneschi Y, Morgan A, Morris AP, Morrison AC, Munson PJ, Nalls MA, Nandakumar P, Nelson CP, Niiranen T, Nolte IM, Nutile T, Oldehinkel AJ, Oostra BA, O'Reilly PF, Org E, Padmanabhan S, Palmas W, Palotie A, Pattie A, Penninx BWJH, Perola M, Peters A, Polasek O, Pramstaller PP, Nguyen QT, Raitakari OT, Ren M, Rettig R, Rice K, Ridker PM, Ried JS, Riese H, Ripatti S, Robino A, Rose LM, Rotter

JI, Rudan I, Ruggiero D, Saba Y, Sala CF, Salomaa V, Samani NJ, Sarin AP, Schmidt R, Schmidt H, Shrine N, Siscovick D, Smith AV, Snieder H, Sõber S, Sorice R, Starr JM, Stott DJ, Strachan DP, Strawbridge RJ, Sundström J, Swertz MA, Taylor KD, Teumer A, Tobin MD, Tomaszewski M, Toniolo D, Traglia M, Trompet S, Tuomilehto J, Tzourio C, Uitterlinden AG, Vaez A, van der Most PJ, van Duijn CM, Vergnaud AC, Verwoert GC, Vitart V, Völker U, Vollenweider P, Vuckovic D, Watkins H, Wild SH, Willemsen G, Wilson JF, Wright AF, Yao J, Zemunik T, Zhang W, Attia JR, Butterworth AS, Chasman DI, Conen D, Cucca F, Danesh J, Hayward C, Howson JMM, Laakso M, Lakatta EG, Langenberg C, Melander O, Mook-Kanamori DO, Palmer CNA, Risch L, Scott RA, Scott RJ, Sever P, Spector TD, van der Harst P, Wareham NJ, Zeggini E, Levy D, Munroe PB, Newton-Cheh C, Brown MJ, Metspalu A, Hung AM, O'Donnell CJ, Edwards TL, Psaty BM, Tzoulaki I, Barnes MR, Wain LV, Elliott P, Caulfield MJ; Million Veteran Program. *Genetic analysis of over 1 million people identifies 535 new loci associated with blood pressure traits*. **Nat Genet**. 2018 Oct;50(10):1412-1425. doi: 10.1038/s41588-018-0205-x.

370. Bevilacqua L, Navarra CO, Pirastu N, Lenarda RD, Gasparini P, Robino A. *A genome-wide association study identifies an association between variants in EFCAB4B gene and periodontal disease in an Italian isolated population*. **J Periodontal Res**. 2018 Dec;53(6):992-998. doi: 10.1111/jre.12598.

371. Maisano Delser P, Ravnik-Glavač M, Gasparini P, Glavač D, Mezzavilla M. Genetic *Landscape of Slovenians: Past Admixture and Natural Selection Pattern***. Front Genet**. 2018 Nov 19;9:551. doi: 10.3389/fgene.2018.00551.

372. Morgan A, Lenarduzzi S, Cappellani S, Pecile V, Morgutti M, Orzan E, Ghiselli S, Ambrosetti U, Brumat M, Gajendrarao P, La Bianca M, Faletra F, Grosso E, Sirchia F, Sensi A, Graziano C, Seri M, Gasparini P, Girotto G. *Genomic Studies in a Large Cohort of Hearing Impaired Italian Patients Revealed Several New Alleles, a Rare Case of Uniparental Disomy (UPD) and the Importance to Search for Copy Number Variations*. **Front Genet**. 2018 Dec 21;9:681. doi: 10.3389/fgene.2018.00681.

373. Morgan A, Vuckovic D, Krishnamoorthy N, Rubinato E, Ambrosetti U, Castorina P, Franzè A, Vozzi D, La Bianca M, Cappellani S, Di Stazio M, Gasparini P, Girotto G. *Next-generation sequencing identified SPATC1L as a possible candidate gene for both early-onset and age-related hearing loss*. **Eur J Hum Genet**. 2019 Jan;27(1):70-79. doi: 10.1038/s41431-018-0229-9.

374. Girotto G., Morgan A., Krishnamoorthy N., Cocca M., Brumat M., Bassani S., Bianca M.L., Di Stazio M., Gasparini P. *Next generation sequencing and animal models reveal SLC9A3R1 as a new gene involved in human age-related hearing loss*. **Frontiers in Genetics**. 2019;10. doi: 10.3389/fgene.2019.00142

375. Robino A., Concas M.P., Catamo E., Gasparini P. *A brief review of genetic approaches to the study of food preferences: Current knowledge and future directions*. **Nutrients**. 2019;11(8).

doi: 10.3390/nu11081735

376. Karlsson Linnér R, Biroli P, Kong E, Meddens SFW, Wedow R, Fontana MA, Lebreton M, Tino SP, Abdellaoui A, Hammerschlag AR, Nivard MG, Okbay A, Rietveld CA, Timshel PN, Trzaskowski M, Vlaming R, Zünd CL, Bao Y, Buzdugan L, Caplin AH, Chen CY, Eibich P, Fontanillas P, Gonzalez JR, Joshi PK, Karhunen V, Kleinman A, Levin RZ, Lill CM, Meddens GA, Muntané G, Sanchez-Roige S, Rooij FJV, Taskesen E, Wu Y, Zhang F; 23and Me Research Team; eQTLgen Consortium; International Cannabis Consortium; Social Science Genetic Association Consortium, Auton A, Boardman JD, Clark DW, Conlin A, Dolan CC, Fischbacher U, Groenen PJF, Harris KM, Hasler G, Hofman A, Ikram MA, Jain S, Karlsson R, Kessler RC, Kooyman M, MacKillop J, Männikkö M, Morcillo-Suarez C, McQueen MB, Schmidt KM, Smart MC, Sutter M, Thurik AR, Uitterlinden AG, White J, Wit H, Yang J, Bertram L, Boomsma DI, Esko T, Fehr E, Hinds DA, Johannesson M, Kumari M, Laibson D, Magnusson PKE, Meyer MN, Navarro A, Palmer AA, Pers TH, Posthuma D, Schunk D, Stein MB, Svento R, Tiemeier H, Timmers PRHJ, Turley P, Ursano RJ, Wagner GG, Wilson JF, Gratten J, Lee JJ, Cesarini D, Benjamin DJ, Koellinger PD, Beauchamp JP. Agee M, Alipanahi B, Bell RK, Bryc K, Elson SL, Furlotte NA, Huber KE, Litterman NK, McCreight JC, McIntyre MH, Mountain JL, Northover CAM, Pitts SJ, Sathirapongsasuti JF, Sazonova OV, Shelton JF, Shringarpure S, Tian C, Tung JY, Vacic V, Wilson CH, Agbessi M, Ahsan H, Alves I, Andiappan A, Awadalla P, Battle A, Beutner F, Jan Bonder M, Boomsma DI, Christiansen M, Claringbould A, Deelen P, Esko T, Favé MJ, Franke L, Frayling T, Gharib SA, Gibson G, Heijmans B, Hemani G, Jansen R, Kähönen M, Kalnapenkis A, Kasela S, Kettunen J, Kim Y, Kirsten H, Kovacs P, Krohn K, Kronberg-Guzman J, Kukushkina V, Kutalik Z, Lee B, Lehtimäki T, Loeffler M, Marigorta UM, Metspalu A, Milani L, Montgomery GW, Müller-Nurasyid M, Nauck M, Nivard MG, Penninx B, Perola M, Pervjakova N, Pierce B, Powell J, Prokisch H, Psaty BM, Raitakari O, Ring S, Ripatti S, Rotzchke O, Rüeger S, Saha A, Scholz M, Schramm K, Seppälä I, Stumvoll M, Sullivan P, Hoen PT, Teumer A, Thiery J, Tong L, Tönjes A, Dongen JV, Meurs JV, Verlouw J, Visscher PM, Völker U, Võsa U, Westra HJ, Yaghootkar H, Yang J, Zeng B, Zhang F, Okbay A, Beauchamp JP, Fontana MA, Lee JJ, Pers TH, Rietveld CA, Turley P, Chen GB, Emilsson V, Meddens SFW, Oskarsson S, Pickrell JK, Thom K, Timshel P, de Vlaming R, Abdellaoui A, Ahluwalia TS, Bacelis J, Baumbach C, Bjornsdottir G, Brandsma JH, Concas MP, Derringer J, Furlotte NA, Galesloot TE, Girotto G, Gupta R, Hall LM, Harris SE, Hofer E, Horikoshi M, Huffman JE, Kaasik K, Kalafati IP, Karlsson R, Kong A, Lahti J, Lee SJV, de Leeuw C, Lind PA, Lindgren KO, Liu T, Mangino M, Marten J, Mihailov E, Miller MB, Most PJV, Oldmeadow C, Payton A, Pervjakova N, Peyrot WJ, Qian Y, Raitakari O, Rueedi R, Salvi E, Schmidt B, Schraut KE, Shi J, Smith AV, Poot RA, Pourcain BS, Teumer A, Thorleifsson G, Verweij N, Vuckovic D, Wellmann J, Westra HJ, Yang J, Zhao W, Zhu Z, Alizadeh BZ, Amin N, Bakshi A, Baumeister SE, Biino G, Bønnelykke K, Boyle PA, Campbell H,

Cappuccio FP, Davies G, De Neve JE, Deloukas P, Demuth I, Ding J, Eibich P, Eisele L, Eklund N, Evans DM, Faul JD, Feitosa MF, Forstner AJ, Gandin I, Gunnarsson B, Halldórsson BV, Harris TB, Heath AC, Hocking LJ, Holliday EG, Homuth G, Horan MA, Hottenga JJ, de Jager PL, Joshi PK, Jugessur A, Kaakinen MA, Kähönen M, Kanoni S, Keltigangas-Järvinen L, Kiemeney LALM, Kolcic I, Koskinen S, Kraja AT, Kroh M, Kutalik Z, Latvala A, Launer LJ, Lebreton MP, Levinson DF, Lichtenstein P, Lichtner P, Liewald DCM, Loukola A, Madden PA, Mägi R, Mäki-Opas T, Marioni RE, Marques-Vidal P, Meddens GA, McMahon G, Meisinger C, Meitinger T, Milaneschi Y, Milani L, Montgomery GW, Myhre R, Nelson CP, Nyholt DR, Ollier WER, Palotie A, Paternoster L, Pedersen NL, Petrovic KE, Porteous DJ, Räikkönen K, Ring SM, Robino A, Rostapshova O, Rudan I, Rustichini A, Salomaa V, Sanders AR, Sarin AP, Schmidt H, Scott RJ, Smith BH, Smith JA, Staessen JA, Steinhagen-Thiessen E, Strauch K, Terracciano A, Tobin MD, Ulivi S, Vaccargiu S, Quaye L, Rooij FJV, Venturini C, Vinkhuyzen AAE, Völker U, Völzke H, Vonk JM, Vozzi D, Waage J, Ware EB, Willemsen G, Attia JR, Bennett DA, Berger K, Bertram L, Bisgaard H, Boomsma DI, Borecki IB, Bültmann U, Chabris CF, Cucca F, Cusi D, Deary IJ, Dedoussis GV, Duijn CMV, Eriksson JG, Franke B, Franke L, Gasparini P, Gejman PV, Gieger C, Grabe HJ, Gratten J, Groenen PJF, Gudnason V, Harst PV, Hayward C, Hinds DA, Hoffmann W, Hyppönen E, Iacono WG, Jacobsson B, Järvelin MR, Jöckel KH, Kaprio J, Kardia SLR, Lehtimäki T, Lehrer SF, Magnusson PKE, Martin NG, McGue M, Metspalu A, Pendleton N, Penninx B, Perola M, Pirastu N, Pirastu M, Polasek O, Posthuma D, Power C, Province MA, Samani NJ, Schlessinger D, Schmidt R, Sørensen TIA, Spector TD, Stefansson K, Thorsteinsdottir U, Thurik AR, Timpson NJ, Tiemeier H, Tung JY, Uitterlinden AG, Vitart V, Vollenweider P, Weir DR, Wilson JF, Wright AF, Conley DC, Krueger RF, Smith GD, Hofman A, Laibson DI, Medland SE, Meyer MN, Yang J, Johannesson M, Visscher PM, Esko T, Koellinger PD, Cesarini D, Benjamin DJ.*Genome-wide association analyses of risk tolerance and risky behaviors in over 1 million individuals identify hundreds of loci and shared genetic influences*. **Nat Genet**. 2019 Feb;51(2):245-257. doi: 10.1038/s41588-018-0309-3.

377. Justice A.E., Karaderi T., Highland H.M., Young K.L., Graff M., Lu Y., Turcot V., Auer P.L., Fine R.S., Guo X., Schurmann C., Lempradl A., Marouli E., Mahajan A., Winkler T.W., Locke A.E., Medina-Gomez C., Esko T., Vedantam S., Giri A., Lo K.S., Alfred T., Mudgal P., Ng M.C.Y., Heard-Costa N.L., Feitosa M.F., Manning A.K., Willems S.M., Sivapalaratnam S., Abecasis G., Alam D.S., Allison M., Amouyel P., Arzumanyan Z., Balkau B., Bastarache L., Bergmann S., Bielak L.F., Blüher M., Boehnke M., Boeing H., Boerwinkle E., Böger C.A., Bork-Jensen J., Bottinger E.P., Bowden D.W., Brandslund I., Broer L., Burt A.A., Butterworth A.S., Caulfield M.J., Cesana G., Chambers J.C., Chasman D.I., Chen Y.-D.I., Chowdhury R., Christensen C., Chu A.Y., Collins F.S., Cook J.P., Cox A.J., Crosslin D.S., Danesh J., de Bakker P.I.W., Denus S., Mutsert R., Dedoussis G., Demerath

E.W., Dennis J.G., Denny J.C., Angelantonio E.D., Dörr M., Drenos F., Dubé M.-P., Dunning A.M., Easton D.F., Elliott P., Evangelou E., Farmaki A.-E., Feng S., Ferrannini E., Ferrieres J., Florez J.C., Fornage M., Fox C.S., Franks P.W., Friedrich N., Gan W., Gandin I., Gasparini P., Giedraitis V., Girotto G., Gorski M., Grallert H., Grarup N., Grove M.L., Gustafsson S., Haessler J., Hansen T., Hattersley A.T., Hayward C., Heid I.M., Holmen O.L., Hovingh G.K., Howson J.M.M., Hu Y., Hung Y.-J., Hveem K., Ikram M.A., Ingelsson E., Jackson A.U., Jarvik G.P., Jia Y., Jørgensen T., Jousilahti P., Justesen J.M., Kahali B., Karaleftheri M., Kardia S.L.R., Karpe F., Kee F., Kitajima H., Komulainen P., Kooner J.S., Kovacs P., Krämer B.K., Kuulasmaa K., Kuusisto J., Laakso M., Lakka T.A., Lamparter D., Lange L.A., Langenberg C., Larson E.B., Lee N.R., Lee W.-J., Lehtimäki T., Lewis C.E., Li H., Li J., Li-Gao R., Lin L.-A., Lin X., Lind L., Lindström J., Linneberg A., Liu C.-T., Liu D.J., Luan J., Lyytikäinen L.-P., MacGregor S., Mägi R., Männistö S., Marenne G., Marten J., Masca N.G.D., McCarthy M.I., Meidtner K., Mihailov E., Moilanen L., Moitry M., Mook-Kanamori D.O., Morgan A., Morris A.P., Müller-Nurasyid M., Munroe P.B., Narisu N., Nelson C.P., Neville M., Ntalla I., O'Connell J.R., Owen K.R., Pedersen O., Peloso G.M., Pennell C.E., Perola M., Perry J.A., Perry J.R.B., Pers T.H., Ewing A., Polasek O., Raitakari O.T., Rasheed A., Raulerson C.K., Rauramaa R., Reilly D.F., Reiner A.P., Ridker P.M., Rivas M.A., Robertson N.R., Robino A., Rudan I., Ruth K.S., Saleheen D., Salomaa V., Samani N.J., Schreiner P.J., Schulze M.B., Scott R.A., Segura-Lepe M., Sim X., Slater A.J., Small K.S., Smith B.H., Smith J.A., Southam L., Spector T.D., Speliotes E.K., Stefansson K., Steinthorsdottir V., Stirrups K.E., Strauch K., Stringham H.M., Stumvoll M., Sun L., Surendran P., Swart K.M.A., Tardif J.-C., Taylor K.D., Teumer A., Thompson D.J., Thorleifsson G., Thorsteinsdottir U., Thuesen B.H., Tönjes A., Torres M., Tsafantakis E., Tuomilehto J., Uitterlinden A.G., Uusitupa M., van Duijn C.M., Vanhala M., Varma R., Vermeulen S.H., Vestergaard H., Vitart V., Vogt T.F., Vuckovic D., Wagenknecht L.E., Walker M., Wallentin L., Wang F., Wang C.A., Wang S., Wareham N.J., Warren H.R., Waterworth D.M., Wessel J., White H.D., Willer C.J., Wilson J.G., Wood A.R., Wu Y., Yaghootkar H., Yao J., Yerges-Armstrong L.M., Young R., Zeggini E., Zhan X., Zhang W., Zhao J.H., Zhao W., Zheng H., Zhou W., Zillikens M.C., Rivadeneira F., Borecki I.B., Pospisilik J.A., Deloukas P., Frayling T.M., Lettre G., Mohlke K.L., Rotter J.I., Kutalik Z., Hirschhorn J.N., Cupples L.A., Loos R.J.F., North K.E., Lindgren C.M., CHD Exome+ Consortium, Cohorts for Heart and Aging Research in Genomic Epidemiology (CHARGE) Consortium, EPIC-CVD Consortium, ExomeBP Consortium, Global Lipids Genetic Consortium, GoT2D Genes Consortium, InterAct, ReproGen Consortium, T2D-Genes Consortium, The MAGIC Investigators. *Protein-coding variants implicate novel genes related to lipid homeostasis contributing to body-fat distribution*. **Nature Genetics**. 2019;51(3):452-469. doi: 10.1038/s41588-018-0334-2

378. Bentley A.R., Sung Y.J., Brown M.R., Winkler T.W., Kraja

A.T., Ntalla I., Schwander K., Chasman D.I., Lim E., Deng X., Guo X., Liu J., Lu Y., Cheng C.-Y., Sim X., Vojinovic D., Huffman J.E., Musani S.K., Li C., Feitosa M.F., Richard M.A., Noordam R., Baker J., Chen G., Aschard H., Bartz T.M., Ding J., Dorajoo R., Manning A.K., Rankinen T., Smith A.V., Tajuddin S.M., Zhao W., Graff M., Alver M., Boissel M., Chai J.F., Chen X., Divers J., Evangelou E., Gao C., Goel A., Hagemeijer Y., Harris S.E., Hartwig F.P., He M., Horimoto A.R.V.R., Hsu F.-C., Hung Y.-J., Jackson A.U., Kasturiratne A., Komulainen P., Kühnel B., Leander K., Lin K.-H., Luan J., Lyytikäinen L.-P., Matoba N., Nolte I.M., Pietzner M., Prins B., Riaz M., Robino A., Said M.A., Schupf N., Scott R.A., Sofer T., Stancáková A., Takeuchi F., Tayo B.O., van der Most P.J., Varga T.V., Wang T.-D., Wang Y., Ware E.B., Wen W., Xiang Y.-B., Yanek L.R., Zhang W., Zhao J.H., Adeyemo A., Afaq S., Amin N., Amini M., Arking D.E., Arzumanyan Z., Aung T., Ballantyne C., Barr R.G., Bielak L.F., Boerwinkle E., Bottinger E.P., Broeckel U., Brown M., Cade B.E., Campbell A., Canouil M., Charumathi S., Chen Y.-D.I., Christensen K., Concas M.P., Connell J.M., de las Fuentes L., de Silva H.J., de Vries P.S., Doumatey A., Duan Q., Eaton C.B., Eppinga R.N., Faul J.D., Floyd J.S., Forouhi N.G., Forrester T., Friedlander Y., Gandin I., Gao H., Ghanbari M., Gharib S.A., Gigante B., Giulianini F., Grabe H.J., Gu C.C., Harris T.B., Heikkinen S., Heng C.-K., Hirata M., Hixson J.E., Ikram M.A., Jia Y., Joehanes R., Johnson C., Jonas J.B., Justice A.E., Katsuya T., Khor C.C., Kilpeläinen T.O., Koh W.-P., Kolcic I., Kooperberg C., Krieger J.E., Kritchevsky S.B., Kubo M., Kuusisto J., Lakka T.A., Langefeld C.D., Langenberg C., Launer L.J., Lehne B., Lewis C.E., Li Y., Liang J., Lin S., Liu C.-T., Liu J., Liu K., Loh M., Lohman K.K., Louie T., Luzzi A., Mägi R., Mahajan A., Manichaikul A.W., McKenzie C.A., Meitinger T., Metspalu A., Milaneschi Y., Milani L., Mohlke K.L., Momozawa Y., Morris A.P., Murray A.D., Nalls M.A., Nauck M., Nelson C.P., North K.E., O'Connell J.R., Palmer N.D., Papanicolau G.J., Pedersen N.L., Peters A., Peyser P.A., Polasek O., Poulter N., Raitakari O.T., Reiner A.P., Renström F., Rice T.K., Rich S.S., Robinson J.G., Rose L.M., Rosendaal F.R., Rudan I., Schmidt C.O., Schreiner P.J., Scott W.R., Sever P., Shi Y., Sidney S., Sims M., Smith J.A., Snieder H., Starr J.M., Strauch K., Stringham H.M., Tan N.Y.Q., Tang H., Taylor K.D., Teo Y.Y., Tham Y.C., Tiemeier H., Turner S.T., Uitterlinden A.G., van Heemst D., Waldenberger M., Wang H., Wang L., Wang L., Wei W.B., Williams C.A., Wilson G., Sr, Wojczynski M.K., Yao J., Young K., Yu C., Yuan J.-M., Zhou J., Zonderman A.B., Becker D.M., Boehnke M., Bowden D.W., Chambers J.C., Cooper R.S., de Faire U., Deary I.J., Elliott P., Esko T., Farrall M., Franks P.W., Freedman B.I., Froguel P., Gasparini P., Gieger C., Horta B.L., Juang J.-M.J., Kamatani Y., Kammerer C.M., Kato N., Kooner J.S., Laakso M., Laurie C.C., Lee I.-T., Lehtimäki T., Magnusson P.K.E., Oldehinkel A.J., Penninx B.W.J.H., Pereira A.C., Rauramaa R., Redline S., Samani N.J., Scott J., Shu X.-O., van der Harst P., Wagenknecht L.E., Wang J.-S., Wang Y.X., Wareham N.J., Watkins H., Weir D.R., Wickremasinghe A.R., Wu T., Zeggini E., Zheng W., Bouchard C.,

Evans M.K., Gudnason V., Kardia S.L.R., Liu Y., Psaty B.M., Ridker P.M., van Dam R.M., Mook-Kanamori D.O., Fornage M., Province M.A., Kelly T.N., Fox E.R., Hayward C., van Duijn C.M., Tai E.S., Wong T.Y., Loos R.J.F., Franceschini N., Rotter J.I., Zhu X., Bierut L.J., Gauderman W.J., Rice K., Munroe P.B., Morrison A.C., Rao D.C., Rotimi C.N., Cupples L.A., COGENT-Kidney Consortium, EPIC-InterAct Consortium, Understanding Society Scientific Group, Lifelines Cohort. *Multi-ancestry genome-wide gene–smoking interaction study of 387,272 individuals identifies new loci associated with serum lipids*. **Nature Genetics**. 2019;51(4):636-648. doi: 10.1038/s41588-019-0378-y.

379. Di Stazio M, Collesi C, Vozzi D, Liu W, Myers M, Morgan A, D Adamo PA, Girotto G, Rubinato E, Giacca M, Gasparini P. *TBL1Y: a new gene involved in syndromic hearing loss*. **Eur J Hum Genet**. 2019;27(3):466-474. doi: 10.1038/s41431-018-0282-4.

380. Agnoletto C., Corrà F., Minotti L., Baldassari F., Crudele F., Cook W.J.J., Di Leva G., D'Adamo A.P., Gasparini P., Volinia S. *Heterogeneity in circulating tumor cells: The relevance of the stem-cell subset*. **Cancers**. 2019;11(4). doi: 10.3390/cancers11040483.

381. Zupin L, Moura Rodrigues R, Navarra CO, Bevilacqua L, Catamo E, Di Lenarda R, Gasparini P, Crovella S, Robino A. *Association of LTA gene haploblock with periodontal disease in Italian adults*. **J Periodontal Res**. 2019;54(2):128-133. doi: 10.1111/jre.12609.

382. Bottega R., Napolitano L.M.R., Carbone A., Cappelli E., Corsolini F., Onesti S., Savoia A., Gasparini P., Faletra F. *Two further patients with Warsaw breakage syndrome. Is a mild phenotype possible?* **Molecular Genetics and Genomic Medicine**. 2019;7(5). doi: 10.1002/mgg3.639.

383. van Setten J, Verweij N, Mbarek H, Niemeijer MN, Trompet S, Arking DE, Brody JA, Gandin I, Grarup N, Hall LM, Hemerich D, Lyytikäinen LP, Mei H, Müller-Nurasyid M, Prins BP, Robino A, Smith AV, Warren HR, Asselbergs FW, Boomsma DI, Caulfield MJ, Eijgelsheim M, Ford I, Hansen T, Harris TB, Heckbert SR, Hottenga JJ, Iorio A, Kors JA, Linneberg A, MacFarlane PW, Meitinger T, Nelson CP, Raitakari OT, Silva Aldana CT, Sinagra G, Sinner M, Soliman EZ, Stoll M, Uitterlinden A, van Duijn CM, Waldenberger M, Alonso A, Gasparini P, Gudnason V, Jamshidi Y, Kääb S, Kanters JK, Lehtimäki T, Munroe PB, Peters A, Samani NJ, Sotoodehnia N, Ulivi S, Wilson JG, de Geus EJC, Jukema JW, Stricker B, van der Harst P, de Bakker PIW, Isaacs A. *Genome-wide association meta-analysis of 30,000 samples identifies seven novel loci for quantitative ECG traits*. **Eur J Hum Genet**. 2019 Jan 24;27(6):952-962. doi: 10.1038/s41431-018-0295-z.

384. de Vries PS, Brown MR, Bentley AR, Sung YJ, Winkler TW, Ntalla I, Schwander K, Kraja AT, Guo X, Franceschini N, Cheng CY, Sim X, Vojinovic D, Huffman JE, Musani SK, Li C, Feitosa MF, Richard MA, Noordam R, Aschard H, Bartz TM, Bielak LF, Deng X, Dorajoo R, Lohman KK, Manning AK, Rankinen T, Smith AV, Tajuddin SM, Evangelou E, Graff M, Alver M, Boissel M, Chai JF, Chen X, Divers J, Gandin I, Gao C, Goel A, Hagemeijer Y, Harris

SE, Hartwig FP, He M, Horimoto ARVR, Hsu FC, Jackson AU, Kasturiratne A, Komulainen P, Kühnel B, Laguzzi F, Lee JH, Luan J, Lyytikäinen LP, Matoba N, Nolte IM, Pietzner M, Riaz M, Said MA, Scott RA, Sofer T, Stancáková A, Takeuchi F, Tayo BO, van der Most PJ, Varga TV, Wang Y, Ware EB, Wen W, Yanek LR, Zhang W, Zhao JH, Afaq S, Amin N, Amini M, Arking DE, Aung T, Ballantyne C, Boerwinkle E, Broeckel U, Campbell A, Canouil M, Charumathi S, Chen YI, Connell JM, de Faire U, de Las Fuentes L, de Mutsert R, de Silva HJ, Ding J, Dominiczak AF, Duan Q, Eaton CB, Eppinga RN, Faul JD, Fisher V, Forrester T, Franco OH, Friedlander Y, Ghanbari M, Giulianini F, Grabe HJ, Grove ML, Gu CC, Harris TB, Heikkinen S, Heng CK, Hirata M, Hixson JE, Howard BV, Ikram MA; InterAct Consortium, Jacobs DR Jr, Johnson C, Jonas JB, Kammerer CM, Katsuya T, Khor CC, Kilpeläinen TO, Koh WP, Koistinen HA, Kolcic I, Kooperberg C, Krieger JE, Kritchevsky SB, Kubo M, Kuusisto J, Lakka TA, Langefeld CD, Langenberg C, Launer LJ, Lehne B, Lemaitre RN, Li Y, Liang J, Liu J, Liu K, Loh M, Louie T, Mägi R, Manichaikul AW, McKenzie CA, Meitinger T, Metspalu A, Milaneschi Y, Milani L, Mohlke KL, Mosley TH Jr, Mukamal KJ, Nalls MA, Nauck M, Nelson CP, Sotoodehnia N, O'Connell JR, Palmer ND, Pazoki R, Pedersen NL, Peters A, Peyser PA, Polasek O, Poulter N, Raffel LJ, Raitakari OT, Reiner AP, Rice TK, Rich SS, Robino A, Robinson JG, Rose LM, Rudan I, Schmidt CO, Schreiner PJ, Scott WR, Sever P, Shi Y, Sidney S, Sims M, Smith BH, Smith JA, Snieder H, Starr JM, Strauch K, Tan N, Taylor KD, Teo YY, Tham YC, Uitterlinden AG, van Heemst D, Vuckovic D, Waldenberger M, Wang L, Wang Y, Wang Z, Wei WB, Williams C, Wilson G Sr, Wojczynski MK, Yao J, Yu B, Yu C, Yuan JM, Zhao W, Zonderman AB, Becker DM, Boehnke M, Bowden DW, Chambers JC, Deary IJ, Esko T, Farrall M, Franks PW, Freedman BI, Froguel P, Gasparini P, Gieger C, Horta BL, Kamatani Y, Kato N, Kooner JS, Laakso M, Leander K, Lehtimäki T; Lifelines Cohort, Groningen, The Netherlands (Lifelines Cohort Study), Magnusson PKE, Penninx B, Pereira AC, Rauramaa R, Samani NJ, Scott J, Shu XO, van der Harst P, Wagenknecht LE, Wang YX, Wareham NJ, Watkins H, Weir DR, Wickremasinghe AR, Zheng W, Elliott P, North KE, Bouchard C, Evans MK, Gudnason V, Liu CT, Liu Y, Psaty BM, Ridker PM, van Dam RM, Kardia SLR, Zhu X, Rotimi CN, Mook-Kanamori DO, Fornage M, Kelly TN, Fox ER, Hayward C, van Duijn CM, Tai ES, Wong TY, Liu J, Rotter JI, Gauderman WJ, Province MA, Munroe PB, Rice K, Chasman DI, Cupples LA, Rao DC, Morrison AC. *Multi-Ancestry Genome-Wide Association Study of Lipid Levels Incorporating Gene-Alcohol Interactions*. **Am J Epidemiol**. 2019 Jan 29;188(6):1033-1054. doi: 10.1093/aje/kwz005.

385. Cont G, Paviotti G., Montico M., Paganin P., Guerra M., Trappan A., Demarini S., Gasparini P., Robino A. *TAS2R38 bitter taste genotype is associated with complementary feeding behavior in infants*. **Genes and Nutrition**. 2019;14(1). doi: 10.1186/s12263-019-0640-z

386. Mezzavilla M, Notarangelo M, Concas MP, Catamo E, Gasparini P, Grillotti MG, Robino A. *Investigation of the link between PROP*

taste perception and vegetables consumption using FAOSTAT data. **Int J Food Sci Nutr**. 2019;70(4):484-490. doi: 10.1080/09637486.2018.1519527.

387. Wuttke M., Li Y., Li M., Sieber K.B., Feitosa M.F., Gorski M., Tin A., Wang L., Chu A.Y., Hoppmann A., Kirsten H., Giri A., Chai J.-F., Sveinbjornsson G., Tayo B.O., Nutile T., Fuchsberger C., Marten J., Cocca M., Ghasemi S., Xu Y., Horn K., Noce D., van der Most P.J., Sedaghat S., Yu Z., Akiyama M., Afaq S., Ahluwalia T.S., Almgren P., Amin N., Ärnlöv J., Bakker S.J.L., Bansal N., Baptista D., Bergmann S., Biggs M.L., Biino G., Boehnke M., Boerwinkle E., Boissel M., Bottinger E.P., Boutin T.S., Brenner H., Brumat M., Burkhardt R., Butterworth A.S., Campana E., Campbell A., Campbell H., Canouil M., Carroll R.J., Catamo E., Chambers J.C., Chee M.-L., Chee M.-L., Chen X., Cheng C.-Y., Cheng Y., Christensen K., Cifkova R., Ciullo M., Concas M.P., Cook J.P., Coresh J., Corre T., Sala C.F., Cusi D., Danesh J., Daw E.W., de Borst M.H., De Grandi A., de Mutsert R., de Vries A.P.J., Degenhardt F., Delgado G., Demirkan A., Di Angelantonio E., Dittrich K., Divers J., Dorajoo R., Eckardt K.-U., Ehret G., Elliott P., Endlich K., Evans M.K., Felix J.F., Foo V.H.X., Franco O.H., Franke A., Freedman B.I., Freitag-Wolf S., Friedlander Y., Froguel P., Gansevoort R.T., Gao H., Gasparini P., Gaziano J.M., Giedraitis V., Gieger C., Girotto G., Giulianini F., Gögele M., Gordon S.D., Gudbjartsson D.F., Gudnason V., Haller T., Hamet P., Harris T.B., Hartman C.A., Hayward C., Hellwege J.N., Heng C.-K., Hicks A.A., Hofer E., Huang W., Hutri-Kähönen N., Hwang S.-J., Ikram M.A., Indridason O.S., Ingelsson E., Ising M., Jaddoe V.W.V., Jakobsdottir J., Jonas J.B., Joshi P.K., Josyula N.S., Jung B., Kähönen M., Kamatani Y., Kammerer C.M., Kanai M., Kastarinen M., Kerr S.M., Khor C.-C., Kiess W., Kleber M.E., Koenig W., Kooner J.S., Körner A., Kovacs P., Kraja A.T., Krajcoviechova A., Kramer H., Krämer B.K., Kronenberg F., Kubo M., Kühnel B., Kuokkanen M., Kuusisto J., La Bianca M., Laakso M., Lange L.A., Langefeld C.D., Lee J.J.-M., Lehne B., Lehtimäki T., Lieb W., Lim S.-C., Lind L., Lindgren C.M., Liu J., Liu J., Loeffler M., Loos R.J.F., Lucae S., Lukas M.A., Lyytikäinen L.-P., Mägi R., Magnusson P.K.E., Mahajan A., Martin N.G., Martins J., März W., Mascalzoni D., Matsuda K., Meisinger C., Meitinger T., Melander O., Metspalu A., Mikaelsdottir E.K., Milaneschi Y., Miliku K., Mishra P.P., Mohlke K.L., Mononen N., Montgomery G.W., Mook-Kanamori D.O., Mychaleckyj J.C., Nadkarni G.N., Nalls M.A., Nauck M., Nikus K., Ning B., Nolte I.M., Noordam R., O'Connell J., O'Donoghue M.L., Olafsson I., Oldehinkel A.J., Orho-Melander M., Ouwehand W.H., Padmanabhan S., Palmer N.D., Palsson R., Penninx B.W.J.H., Perls T., Perola M., Pirastu M., Pirastu N., Pistis G., Podgornaia A.I., Polasek O., Ponte B., Porteous D.J., Poulain T., Pramstaller P.P., Preuss M.H., Prins B.P., Province M.A., Rabelink T.J., Raffield L.M., Raitakari O.T., Reilly D.F., Rettig R., Rheinberger M., Rice K.M., Ridker P.M., Rivadeneira F., Rizzi F., Roberts D.J., Robino A., Rossing P., Rudan I., Rueedi R., Ruggiero D., Ryan K.A., Saba Y., Sabanayagam C., Salomaa V., Salvi E., Saum K.-U., Schmidt H.,

Schmidt R., Schöttker B., Schulz C.-A., Schupf N., Shaffer C.M., Shi Y., Smith A.V., Smith B.H., Soranzo N., Spracklen C.N., Strauch K., Stringham H.M., Stumvoll M., Svensson P.O., Szymczak S., Tai E.-S., Tajuddin S.M., Tan N.Y.Q., Taylor K.D., Teren A., Tham Y.-C., Thiery J., Thio C.H.L., Thomsen H., Thorleifsson G., Toniolo D., Tönjes A., Tremblay J., Tzoulaki I., Uitterlinden A.G., Vaccargiu S., van Dam R.M., van der Harst P., van Duijn C.M., Velez Edward D.R., Verweij N., Vogelezang S., Völker U., Vollenweider P., Waeber G., Waldenberger M., Wallentin L., Wang Y.X., Wang C., Waterworth D.M., Bin Wei W., White H., Whitfield J.B., Wild S.H., Wilson J.F., Wojczynski M.K., Wong C., Wong T.-Y., Xu L., Yang Q., Yasuda M., Yerges-Armstrong L.M., Zhang W., Zonderman A.B., Rotter J.I., Bochud M., Psaty B.M., Vitart V., Wilson J.G., Dehghan A., Parsa A., Chasman D.I., Ho K., Morris A.P., Devuyst O., Akilesh S., Pendergrass S.A., Sim X., Böger C.A., Okada Y., Edwards T.L., Snieder H., Stefansson K., Hung A.M., Heid I.M., Scholz M., Teumer A., Köttgen A., Pattaro C., Lifelines Cohort Study, V. A. Million Veteran Program. *A catalog of genetic loci associated with kidney function from analyses of a million individuals*. **Nature Genetics**. 2019;51(6):957-972. doi: 10.1038/s41588-019-0407-x.

388. Concas M.P., Catamo E., Biino G., Toniolo D., Gasparini P., Robino A. *Factors associated with food liking and their relationship with metabolic traits in Italian cohorts*. **Food Quality and Preference**. 2019;75:64-70. doi: 10.1016/j.foodqual.2019.02.010.

389. Sung Y.J., De Las Fuentes L., Winkler T.W., Chasman D.I., Bentley A.R., Kraja A.T., Ntalla I., Warren H.R., Guo X., Schwander K., Manning A.K., Brown M.R., Aschard H., Feitosa M.F., Franceschini N., Lu Y., Cheng C.-Y., Sim X., Vojinovic D., Marten J., Musani S.K., Kilpeläinen T.O., Richard M.A., Aslibekyan S., Bartz T.M., Dorajoo R., Li C., Liu Y., Rankinen T., Smith A.V., Tajuddin S.M., Tayo B.O., Zhao W., Zhou Y., Matoba N., Sofer T., Alver M., Amini M., Boissel M., Chai J.F., Chen X., Divers J., Gandin I., Gao C., Giulianini F., Goel A., Harris S.E., Hartwig F.P., He M., Horimoto A.R.V.R., Hsu F.-C., Jackson A.U., Kammerer C.M., Kasturiratne A., Komulainen P., Kühnel B., Leander K., Lee W.-J., Lin K.-H., Luan J., Lyytikäinen L.-P., McKenzie C.A., Nelson C.P., Noordam R., Scott R.A., Sheu W.H.H., Stančáková A., Takeuchi F., Van Der Most P.J., Varga T.V., Waken R.J., Wang H., Wang Y., Ware E.B., Weiss S., Wen W., Yanek L.R., Zhang W., Zhao J.H., Afaq S., Alfred T., Amin N., Arking D.E., Aung T., Barr R.G., Bielak L.F., Boerwinkle E., Bottinger E.P., Braund P.S., Brody J.A., Broeckel U., Cade B., Campbell A., Canouil M., Chakravarti A., Cocca M., Collins F.S., Connell J.M., De Mutsert R., De Silva H.J., Dörr M., Duan Q., Eaton C.B., Ehret G., Evangelou E., Faul J.D., Forouhi N.G., Franco O.H., Friedlander Y., Gao H., Gigante B., Gu C.C., Gupta P., Hagenaars S.P., Harris T.B., He J., Heikkinen S., Heng C.-K., Hofman A., Howard B.V., Hunt S.C., Irvin M.R., Jia Y., Katsuya T., Kaufman J., Kerrison N.D., Khor C.C., Koh W.-P., Koistinen H.A., Kooperberg C.B.,

Krieger J.E., Kubo M., Kutalik Z., Kuusisto J., Lakka T.A., Langefeld C.D., Langenberg C., Launer L.J., Lee J.H., Lehne B., Levy D., Lewis C.E., Li Y., Lim S.H., Liu C.-T., Liu J., Liu J., Liu Y., Loh M., Lohman K.K., Louie T., Mägi R., Matsuda K., Meitinger T., Metspalu A., Milani L., Momozawa Y., Mosley, J.r. T.H., Nalls M.A., Nasri U., O'Connell J.R., Ogunniyi A., Palmas W.R., Palmer N.D., Pankow J.S., Pedersen N.L., Peters A., Peyser P.A., Polasek O., Porteous D., Raitakari O.T., Renström F., Rice T.K., Ridker P.M., Robino A., Robinson J.G., Rose L.M., Rudan I., Sabanayagam C., Salako B.L., Sandow K., Schmidt C.O., Schreiner P.J., Scott W.R., Sever P., Sims M., Sitlani C.M., Smith B.H., Smith J.A., Snieder H., Starr J.M., Strauch K., Tang H., Taylor K.D., Teo Y.Y., Tham Y.C., Uitterlinden A.G., Waldenberger M., Wang L., Wang Y.X., Wei W.B., Wilson G., Wojczynski M.K., Xiang Y.-B., Yao J., Yuan J.-M., Zonderman A.B., Becker D.M., Boehnke M., Bowden D.W., Chambers J.C., Chen Y.-D.I., Weir D.R., De Faire U., Deary I.J., Esko T., Farrall M., Forrester T., Freedman B.I., Froguel P., Gasparini P., Gieger C., Horta B.L., Hung Y.-J., Jonas J.B., Kato N., Kooner J.S., Laakso M., Lehtimäki T., Liang K.-W., Magnusson P.K.E., Oldehinkel A.J., Pereira A.C., Perls T., Rauramaa R., Redline S., Rettig R., Samani N.J., Scott J., Shu X.-O., Van Der Harst P., Wagenknecht L.E., Wareham N.J., Watkins H., Wickremasinghe A.R., Wu T., Kamatani Y., Laurie C.C., Bouchard C., Cooper R.S., Evans M.K., Gudnason V., Hixson J., Kardia S.L.R., Kritchevsky S.B., Psaty B.M., Van Dam R.M., Arnett D.K., Mook-Kanamori D.O., Fornage M., Fox E.R., Hayward C., Van Duijn C.M., Tai E.S., Wong T.Y., Loos R.J.F., Reiner A.P., Rotimi C.N., Bierut L.J., Zhu X., Cupples L.A., Province M.A., Rotter J.I., Franks P.W., Rice K., Elliott P., Caulfield M.J., Gauderman W.J., Munroe P.B., Rao D.C., Morrison A.C. *A multi-ancestry genome-wide study incorporating gene-smoking interactions identifies multiple new loci for pulse pressure and mean arterial pressure*. **Human Molecular Genetics**. 2019;28(15):2615-2633. doi: 10.1093/hmg/ddz070

390. Lenarduzzi S., Morgan A., Faletra F., Cappellani S., Morgutti M., Mezzavilla M., Peruzzi A., Ghiselli S., Ambrosetti U., Graziano C., Seri M., Gasparini P., Girotto G. *Next generation sequencing study in a cohort of Italian patients with syndromic hearing loss*. **Hearing Research**. 2019;381. doi: 10.1016/j.heares.2019.07.006

391. Tin A., Marten J., Halperin Kuhns V.L., Li Y., Wuttke M., Kirsten H., Sieber K.B., Qiu C., Gorski M., Yu Z., Giri A., Sveinbjornsson G., Li M., Chu A.Y., Hoppmann A., O'Connor L.J., Prins B., Nutile T., Noce D., Akiyama M., Cocca M., Ghasemi S., van der Most P.J., Horn K., Xu Y., Fuchsberger C., Sedaghat S., Afaq S., Amin N., Ärnlöv J., Bakker S.J.L., Bansal N., Baptista D., Bergmann S., Biggs M.L., Biino G., Boerwinkle E., Bottinger E.P., Boutin T.S., Brumat M., Burkhardt R., Campana E., Campbell A., Campbell H., Carroll R.J., Catamo E., Chambers J.C., Ciullo M., Concas M.P., Coresh J., Corre T., Cusi D., Felicita S.C., de Borst M.H., De Grandi A., de Mutsert R., de Vries A.P.J., Delgado G., Demirkan A., Devuyst O., Dittrich K., Eckardt K.-U., Ehret G.,

Endlich K., Evans M.K., Gansevoort R.T., Gasparini P., Giedraitis V., Gieger C., Girotto G., Gögele M., Gordon S.D., Gudbjartsson D.F., Gudnason V., Haller T., Hamet P., Harris T.B., Hayward C., Hicks A.A., Hofer E., Holm H., Huang W., Hutri-Kähönen N., Hwang S.-J., Ikram M.A., Lewis R.M., Ingelsson E., Jakobsdottir J., Jonsdottir I., Jonsson H., Joshi P.K., Josyula N.S., Jung B., Kähönen M., Kamatani Y., Kanai M., Kerr S.M., Kiess W., Kleber M.E., Koenig W., Kooner J.S., Körner A., Kovacs P., Krämer B.K., Kronenberg F., Kubo M., Kühnel B., La Bianca M., Lange L.A., Lehne B., Lehtimäki T., Liu J., Loeffler M., Loos R.J.F., Lyytikäinen L.-P., Magi R., Mahajan A., Martin N.G., März W., Mascalzoni D., Matsuda K., Meisinger C., Meitinger T., Metspalu A., Milaneschi Y., O'Donnell C.J., Wilson O.D., Gaziano J.M., Mishra P.P., Mohlke K.L., Mononen N., Montgomery G.W., Mook-Kanamori D.O., Müller-Nurasyid M., Nadkarni G.N., Nalls M.A., Nauck M., Nikus K., Ning B., Nolte I.M., Noordam R., O'Connell J.R., Olafsson I., Padmanabhan S., Penninx B.W.J.H., Perls T., Peters A., Pirastu M., Pirastu N., Pistis G., Polasek O., Ponte B., Porteous D.J., Poulain T., Preuss M.H., Rabelink T.J., Raffield L.M., Raitakari O.T., Rettig R., Rheinberger M., Rice K.M., Rizzi F., Robino A., Rudan I., Krajcoviechova A., Cifkova R., Rueedi R., Ruggiero D., Ryan K.A., Saba Y., Salvi E., Schmidt H., Schmidt R., Shaffer C.M., Smith A.V., Smith B.H., Spracklen C.N., Strauch K., Stumvoll M., Sulem P., Tajuddin S.M., Teren A., Thiery J., Thio C.H.L., Thorsteinsdottir U., Toniolo D., Tönjes A., Tremblay J., Uitterlinden A.G., Vaccargiu S., van der Harst P., van Duijn C.M., Verweij N., Völker U., Vollenweider P., Waeber G., Waldenberger M., Whitfield J.B., Wild S.H., Wilson J.F., Yang Q., Zhang W., Zonderman A.B., Bochud M., Wilson J.G., Pendergrass S.A., Ho K., Parsa A., Pramstaller P.P., Psaty B.M., Böger C.A., Snieder H., Butterworth A.S., Okada Y., Edwards T.L., Stefansson K., Susztak K., Scholz M., Heid I.M., Hung A.M., Teumer A., Pattaro C., Woodward O.M., Vitart V., Köttgen A., German Chronic Kidney Disease Study, Lifelines Cohort Study, V. A. Million Veteran Program. *Target genes, variants, tissues and transcriptional pathways influencing human serum urate levels*. **Nature Genetics**. 2019;51(10):1459-1474. doi: 10.1038/s41588-019-0504-x.

392. Clark D.W., Okada Y., Moore K.H.S., Mason D., Pirastu N., Gandin I., Mattsson H., Barnes C.L.K., Lin K., Zhao J.H., Deelen P., Rohde R., Schurmann C., Guo X., Giulianini F., Zhang W., Medina-Gomez C., Karlsson R., Bao Y., Bartz T.M., Baumbach C., Biino G., Bixley M.J., Brumat M., Chai J.-F., Corre T., Cousminer D.L., Dekker A.M., Eccles D.A., van Eijk K.R., Fuchsberger C., Gao H., Germain M., Gordon S.D., de Haan H.G., Harris S.E., Hofer E., Huerta-Chagoya A., Igartua C., Jansen I.E., Jia Y., Kacprowski T., Karlsson T., Kleber M.E., Li S.A., Li-Gao R., Mahajan A., Matsuda K., Meidtner K., Meng W., Montasser M.E., van der Most P.J., Munz M., Nutile T., Palviainen T., Prasad G., Prasad R.B., Priyanka T.D.S., Rizzi F., Salvi E., Sapkota B.R., Shriner D., Skotte L., Smart M.C., Smith A.V., van der Spek A., Spracklen C.N., Strawbridge R.J., Tajuddin S.M., Trompet S.,

Turman C., Verweij N., Viberti C., Wang L., Warren H.R., Wootton R.E., Yanek L.R., Yao J., Yousri N.A., Zhao W., Adeyemo A.A., Afaq S., Aguilar-Salinas C.A., Akiyama M., Albert M.L., Allison M.A., Alver M., Aung T., Azizi F., Bentley A.R., Boeing H., Boerwinkle E., Borja J.B., de Borst G.J., Bottinger E.P., Broer L., Campbell H., Chanock S., Chee M.-L., Chen G., Chen Y.-D.I., Chen Z., Chiu Y.-F., Cocca M., Collins F.S., Concas M.P., Corley J., Cugliari G., van Dam R.M., Damulina A., Daneshpour M.S., Day F.R., Delgado G.E., Dhana K., Doney A.S.F., Dörr M., Doumatey A.P., Dzimiri N., Ebenesersdóttir S.S., Elliott J., Elliott P., Ewert R., Felix J.F., Fischer K., Freedman B.I., Girotto G., Goel A., Gögele M., Goodarzi M.O., Graff M., Granot-Hershkovitz E., Grodstein F., Guarrera S., Gudbjartsson D.F., Guity K., Gunnarsson B., Guo Y., Hagenaars S.P., Haiman C.A., Halevy A., Harris T.B., Hedayati M., van Heel D.A., Hirata M., Höfer I., Hsiung C.A., Huang J., Hung Y.-J., Ikram M.A., Jagadeesan A., Jousilahti P., Kamatani Y., Kanai M., Kerrison N.D., Kessler T., Khaw K.-T., Khor C.C., de Kleijn D.P.V., Koh W.-P., Kolcic I., Kraft P., Krämer B.K., Kutalik Z., Kuusisto J., Langenberg C., Launer L.J., Lawlor D.A., Lee I.-T., Lee W.-J., Lerch M.M., Li L., Liu J., Loh M., London S.J., Loomis S., Lu Y., Luan J., Mägi R., Manichaikul A.W., Manunta P., Másson G., Matoba N., Mei X.W., Meisinger C., Meitinger T., Mezzavilla M., Milani L., Millwood I.Y., Momozawa Y., Moore A., Morange P.-E., Moreno-Macías H., Mori T.A., Morrison A.C., Muka T., Murakami Y., Murray A.D., de Mutsert R., Mychaleckyj J.C., Nalls M.A., Nauck M., Neville M.J., Nolte I.M., Ong K.K., Orozco L., Padmanabhan S., Pálsson G., Pankow J.S., Pattaro C., Pattie A., Polasek O., Poulter N., Pramstaller P.P., Quintana-Murci L., Räikkönen K., Ralhan S., Rao D.C., van Rheenen W., Rich S.S., Ridker P.M., Rietveld C.A., Robino A., van Rooij F.J.A., Ruggiero D., Saba Y., Sabanayagam C., Sabater-Lleal M., Sala C.F., Salomaa V., Sandow K., Schmidt H., Scott L.J., Scott W.R., Sedaghati-Khayat B., Sennblad B., van Setten J., Sever P.J., Sheu W.H.-H., Shi Y., Shrestha S., Shukla S.R., Sigurdsson J.K., Sikka T.T., Singh J.R., Smith B.H., Stančáková A., Stanton A., Starr J.M., Stefansdottir L., Straker L., Sulem P., Sveinbjornsson G., Swertz M.A., Taylor A.M., Taylor K.D., Terzikhan N., Tham Y.-C., Thorleifsson G., Thorsteinsdottir U., Tillander A., Tracy R.P., Tusié-Luna T., Tzoulaki I., Vaccargiu S., Vangipurapu J., Veldink J.H., Vitart V., Völker U., Vuoksimaa E., Wakil S.M., Waldenberger M., Wander G.S., Wang Y.X., Wareham N.J., Wild S., Yajnik C.S., Yuan J.-M., Zeng L., Zhang L., Zhou J., Amin N., Asselbergs F.W., Bakker S.J.L., Becker D.M., Lehne B., Bennett D.A., van den Berg L.H., Berndt S.I., Bharadwaj D., Bielak L.F., Bochud M., Boehnke M., Bouchard C., Bradfield J.P., Brody J.A., Campbell A., Carmi S., Caulfield M.J., Cesarini D., Chambers J.C., Chandak G.R., Cheng C.-Y., Ciullo M., Cornelis M., Cusi D., Smith G.D., Deary I.J., Dorajoo R., van Duijn C.M., Ellinghaus D., Erdmann J., Eriksson J.G., Evangelou E., Evans M.K., Faul J.D., Feenstra B., Feitosa M., Foisy S., Franke A., Friedlander Y., Gasparini P., Gieger C., Gonzalez C., Goyette P., Grant S.F.A., Griffiths L.R., Groop L., Gudnason V.,

Gyllensten U., Hakonarson H., Hamsten A., van der Harst P., Heng C.-K., Hicks A.A., Hochner H., Huikuri H., Hunt S.C., Jaddoe V.W.V., De Jager P.L., Johannesson M., Johansson Å., Jonas J.B., Jukema J.W., Junttila J., Kaprio J., Kardia S.L.R., Karpe F., Kumari M., Laakso M., van der Laan S.W., Lahti J., Laudes M., Lea R.A., Lieb W., Lumley T., Martin N.G., März W., Matullo G., McCarthy M.I., Medland S.E., Merriman T.R., Metspalu A., Meyer B.F., Mohlke K.L., Montgomery G.W., Mook-Kanamori D., Munroe P.B., North K.E., Nyholt D.R., O'connell J.R., Ober C., Oldehinkel A.J., Palmas W., Palmer C., Pasterkamp G.G., Patin E., Pennell C.E., Perusse L., Peyser P.A., Pirastu M., Polderman T.J.C., Porteous D.J., Posthuma D., Psaty B.M., Rioux J.D., Rivadeneira F., Rotimi C., Rotter J.I., Rudan I., Den Ruijter H.M., Sanghera D.K., Sattar N., Schmidt R., Schulze M.B., Schunkert H., Scott R.A., Shuldiner A.R., Sim X., Small N., Smith J.A., Sotoodehnia N., Tai E.-S., Teumer A., Timpson N.J., Toniolo D., Tregouet D.-A., Tuomi T., Vollenweider P., Wang C.A., Weir D.R., Whitfield J.B., Wijmenga C., Wong T.-Y., Wright J., Yang J., Yu L., Zemel B.S., Zonderman A.B., Perola M., Magnusson P.K.E., Uitterlinden A.G., Kooner J.S., Chasman D.I., Loos R.J.F., Franceschini N., Franke L., Haley C.S., Hayward C., Walters R.G., Perry J.R.B., Esko T., Helgason A., Stefansson K., Joshi P.K., Kubo M., Wilson J.F. *Associations of autozygosity with a broad range of human phenotypes*. **Nature Communications**. 2019;10(1). doi: 10.1038/s41467-019-12283-6.

393. Kilpeläinen T.O., Bentley A.R., Noordam R., Sung Y.J., Schwander K., Winkler T.W., Jakupović H., Chasman D.I., Manning A., Ntalla I., Aschard H., Brown M.R., de las Fuentes L., Franceschini N., Guo X., Vojinovic D., Aslibekyan S., Feitosa M.F., Kho M., Musani S.K., Richard M., Wang H., Wang Z., Bartz T.M., Bielak L.F., Campbell A., Dorajoo R., Fisher V., Hartwig F.P., Horimoto A.R.V.R., Li C., Lohman K.K., Marten J., Sim X., Smith A.V., Tajuddin S.M., Alver M., Amini M., Boissel M., Chai J.F., Chen X., Divers J., Evangelou E., Gao C., Graff M., Harris S.E., He M., Hsu F.-C., Jackson A.U., Zhao J.H., Kraja A.T., Kühnel B., Laguzzi F., Lyytikäinen L.-P., Nolte I.M., Rauramaa R., Riaz M., Robino A., Rueedi R., Stringham H.M., Takeuchi F., van der Most P.J., Varga T.V., Verweij N., Ware E.B., Wen W., Li X., Yanek L.R., Amin N., Arnett D.K., Boerwinkle E., Brumat M., Cade B., Canouil M., Chen Y.-D.I., Concas M.P., Connell J., de Mutsert R., de Silva H.J., de Vries P.S., Demirkan A., Ding J., Eaton C.B., Faul J.D., Friedlander Y., Gabriel K.P., Ghanbari M., Giulianini F., Gu C.C., Gu D., Harris T.B., He J., Heikkinen S., Heng C.-K., Hunt S.C., Ikram M.A., Jonas J.B., Koh W.-P., Komulainen P., Krieger J.E., Kritchevsky S.B., Kutalik Z., Kuusisto J., Langefeld C.D., Langenberg C., Launer L.J., Leander K., Lemaitre R.N., Lewis C.E., Liang J., Alizadeh B.Z., Boezen H.M., Franke L., Navis G., Rots M., Swertz M., Wolffenbuttel B.H.R., Wijmenga C., Liu J., Mägi R., Manichaikul A., Meitinger T., Metspalu A., Milaneschi Y., Mohlke K.L., Mosley T.H., Jr., Murray A.D., Nalls M.A., Nang E.-E.K., Nelson C.P., Nona S., Norris J.M., Nwuba C.V., O'Connell J., Palmer N.D., Papanicolau G.J., Pazoki R., Pedersen N.L., Peters

A., Peyser P.A., Polasek O., Porteous D.J., Poveda A., Raitakari O.T., Rich S.S., Risch N., Robinson J.G., Rose L.M., Rudan I., Schreiner P.J., Scott R.A., Sidney S.S., Sims M., Smith J.A., Snieder H., Sofer T., Starr J.M., Sternfeld B., Strauch K., Tang H., Taylor K.D., Tsai M.Y., Tuomilehto J., Uitterlinden A.G., van der Ende M.Y., van Heemst D., Voortman T., Waldenberger M., Wennberg P., Wilson G., Xiang Y.-B., Yao J., Yu C., Yuan J.-M., Zhao W., Zonderman A.B., Becker D.M., Boehnke M., Bowden D.W., de Faire U., Deary I.J., Elliott P., Esko T., Freedman B.I., Froguel P., Gasparini P., Gieger C., Kato N., Laakso M., Lakka T.A., Lehtimäki T., Magnusson P.K.E., Oldehinkel A.J., Penninx B.W.J.H., Samani N.J., Shu X.-O., van der Harst P., Van Vliet-Ostaptchouk J.V., Vollenweider P., Wagenknecht L.E., Wang Y.X., Wareham N.J., Weir D.R., Wu T., Zheng W., Zhu X., Evans M.K., Franks P.W., Gudnason V., Hayward C., Horta B.L., Kelly T.N., Liu Y., North K.E., Pereira A.C., Ridker P.M., Tai E.S., van Dam R.M., Fox E.R., Kardia S.L.R., Liu C.-T., Mook-Kanamori D.O., Province M.A., Redline S., van Duijn C.M., Rotter J.I., Kooperberg C.B., Gauderman W.J., Psaty B.M., Rice K., Munroe P.B., Fornage M., Cupples L.A., Rotimi C.N., Morrison A.C., Rao D.C., Loos R.J.F., Lifelines Cohort Study. *Multi-ancestry study of blood lipid levels identifies four loci interacting with physical activity*. **Nature Communications**. 2019 Jan 22;10(1). doi: 10.1038/s41467-018-08008-w.

394. Nagtegaal A.P., Broer L., Zilhao N.R., Jakobsdottir J., Bishop C.E., Brumat M., Christiansen M.W., Cocca M., Gao Y., Heard-Costa N.L., Evans D.S., Pankratz N., Pratt S.R., Price T.R., Spankovich C., Stimson M.R., Valle K., Vuckovic D., Wells H., Eiriksdottir G., Fransen E., Ikram M.A., Li C.-M., Longstreth W.T., Jr., Steves C., Van Camp G., Correa A., Cruickshanks K.J., Gasparini P., Girotto G., Kaplan R.C., Nalls M., Schweinfurth J.M., Seshadri S., Sotoodehnia N., Tranah G.J., Uitterlinden A.G., Wilson J.G., Gudnason V., Hoffman H.J., Williams F.M.K., Goedegebure A. *Genome-wide association meta-analysis identifies five novel loci for age-related hearing impairment*. **Scientific Reports**. 2019;9(1). doi. 10.1038/s41598-019-51630-x.

395. Morgan A., Koboldt D.C., Barrie E.S., Crist E.R., García García G., Mezzavilla M., Faletra F., Mihalic Mosher T., Wilson R.K., Blanchet C., Manickam K., Roux A.-F., Gasparini P., Dell'Orco D., Girotto G. *Mutations in PLS1, encoding fimbrin, cause autosomal dominant nonsyndromic hearing loss*. **Human Mutation**. 2019;40(12):2286-2295. doi: 10.1002/humu.23891.

396. Mezzavilla M., Cocca M., Guidolin F., Gasparini P. *A population-based approach for gene prioritization in understanding complex traits*. **Human Genetics**. 2020. doi: 10.1007/s00439-020-02152-4.

397. Cocca M., Barbieri C., Concas M.P., Robino A., Brumat M., Gandin I., Trudu M., Sala C.F., Vuckovic D., Girotto G., Matullo G., Polasek O., Kolčić I., Gasparini P., Soranzo N., Toniolo D., Mezzavilla M. *A bird's-eye view of Italian genomic variation through whole-genome sequencing*. **European Journal of**

Human Genetics. 2020;28(4):435-444. doi: 10.1038/s41431-019-0551-x.

398. Mezzavilla M., Zupin L., Navarra C.O., Di Lenarda R., Gasparini P., Crovella S., Robino A., Bevilacqua L. *Deleterious variants in genes associated with bone mineral density are linked to susceptibility to periodontitis development.* **Meta Gene**. 2020; 24. doi: 10.1016/j.mgene.2020.100670.

399. Di Stazio M., Morgan A., Brumat M., Bassani S., Dell'Orco D., Marino V., Garagnani P., Giuliani C., Gasparini P., Girotto G. *New age-related hearing loss candidate genes in humans: an ongoing challenge*. **Gene**. 2020; 742. doi: 10.1016/j.gene.2020.144561.

400. Nistico' D., Guidolin F., Navarra C.O., Bobbo M., Magnolato A., D'Adamo A.P., Giorgio E., Pivetta B., Barbi E., Gasparini P., Cadenaro M., Sirchia F. *Dental anomalies as a possible clue of 1p36 deletion syndrome due to germline mosaicism: a case report*. **BMC pediatrics**. 2020;20(1):201. doi: 10.1186/s12887-020-02049-1

401. de las Fuentes, L., Sung, Y.J., Noordam, R., Winkler, T., Feitosa, M.F., Schwander, K., Bentley, A.R., Brown, M.R., Guo, X., Manning, A., Chasman, D.I., Aschard, H., Bartz, T.M., Bielak, L.F., Campbell, A., Cheng, C.-Y., Dorajoo, R., Hartwig, F.P., Horimoto, A.R.V.R., Li, C., Li-Gao, R., Liu, Y., Marten, J., Musani, S.K., Ntalla, I., Rankinen, T., Richard, M., Sim, X., Smith, A.V., Tajuddin, S.M., Tayo, B.O., Vojinovic, D., Warren, H.R., Xuan, D., Alver, M., Boissel, M., Chai, J.-F., Chen, X., Christensen, K., Divers, J., Evangelou, E., Gao, C., Girotto, G., Harris, S.E., He, M., Hsu, F.-C., Kühnel, B., Laguzzi, F., Li, X., Lyytikäinen, L.-P., Nolte, I.M., Poveda, A., Rauramaa, R., Riaz, M., Rueedi, R., Shu, X.-O., Snieder, H., Sofer, T., Takeuchi, F., Verweij, N., Ware, E.B., Weiss, S., Yanek, L.R., Amin, N., Arking, D.E., Arnett, D.K., Bergmann, S., Boerwinkle, E., Brody, J.A., Broeckel, U., Brumat, M., Burke, G., Cabrera, C.P., Canouil, M., Chee, M.L., Chen, Y.-D.I., Cocca, M., Connell, J., de Silva, H.J., de Vries, P.S., Eiriksdottir, G., Faul, J.D., Fisher, V., Forrester, T., Fox, E.F., Friedlander, Y., Gao, H., Gigante, B., Giulianini, F., Gu, C.C., Gu, D., Harris, T.B., He, J., Heikkinen, S., Heng, C.-K., Hunt, S., Ikram, M.A., Irvin, M.R., Kähönen, M., Kavousi, M., Khor, C.C., Kilpeläinen, T.O., Koh, W.-P., Komulainen, P., Kraja, A.T., Krieger, J.E., Langefeld, C.D., Li, Y., Liang, J., Liewald, D.C.M., Liu, C.-T., Liu, J., Lohman, K.K., Mägi, R., McKenzie, C.A., Meitinger, T., Metspalu, A., Milaneschi, Y., Milani, L., Mook-Kanamori, D.O., Nalls, M.A., Nelson, C.P., Norris, J.M., O'Connell, J., Ogunniyi, A., Padmanabhan, S., Palmer, N.D., Pedersen, N.L., Perls, T., Peters, A., Petersmann, A., Peyser, P.A., Polasek, O., Porteous, D.J., Raffel, L.J., Rice, T.K., Rotter, J.I., Rudan, I., Rueda-Ochoa, O.-L., Sabanayagam, C., Salako, B.L., Schreiner, P.J., Shikany, J.M., Sidney, S.S., Sims, M., Sitlani, C.M., Smith, J.A., Starr, J.M., Strauch, K., Swertz, M.A., Teumer, A., Tham, Y.C., Uitterlinden, A.G., Vaidya, D., van der Ende, M.Y., Waldenberger, M., Wang, L., Wang, Y.-X., Wei, W.-B., Weir, D.R., Wen, W., Yao, J., Yu, B., Yu, C., Yuan, J.-M., Zhao, W., Zonderman, A.B., Becker, D.M., Bowden, D.W., Deary, I.J., Dörr,

M., Esko, T., Freedman, B.I., Froguel, P., Gasparini, P., Gieger, C., Jonas, J.B., Kammerer, C.M., Kato, N., Lakka, T.A., Leander, K., Lehtimäki, T., Magnusson, P.K.E., Marques-Vidal, P., Penninx, B.W.J.H., Samani, N.J., van der Harst, P., Wagenknecht, L.E., Wu, T., Zheng, W., Zhu, X., Bouchard, C., Cooper, R.S., Correa, A., Evans, M.K., Gudnason, V., Hayward, C., Horta, B.L., Kelly, T.N., Kritchevsky, S.B., Levy, D., Palmas, W.R., Pereira, A.C., Province, M.M., Psaty, B.M., Ridker, P.M., Rotimi, C.N., Tai, E.S., van Dam, R.M., van Duijn, C.M., Wong, T.Y., Rice, K., Gauderman, W.J., Morrison, A.C., North, K.E., Kardia, S.L.R., Caulfield, M.J., Elliott, P., Munroe, P.B., Franks, P.W., Rao, D.C., Fornage, M., Lifelines Cohort Study. *Gene-educational attainment interactions in a multi-ancestry genome-wide meta-analysis identify novel blood pressure loci*. **Molecular Psychiatry**. 2021; 26(6): 2111-2125. doi: 10.1038/s41380-020-0719-3.

402. Morgan, A., Gasparini, P., Girotto, G. *Molecular testing for the study of non-syndromic hearing loss*. **Hearing, Balance and Communication**. 2020; 18 (4), pp. 270-277. doi: 10.1080/21695717.2020.1807260

403. Piatti, G., Ambrosetti, U., Robino, A., Girotto, G., Gasparini, P. *Primary Ciliary Dyskinesia: The Impact of Taste Receptor (TAS2R38) Gene Polymorphisms on Disease Outcome and Severity*. **International Archives of Allergy and Immunology**. 2020; 181 (9), pp. 727-731. doi: 10.1159/000508938.

404. Ntalla, I., Weng, L.-C., Cartwright, J.H., Hall, A.W., Sveinbjornsson, G., Tucker, N.R., Choi, S.H., Chaffin, M.D., Roselli, C., Barnes, M.R., Mifsud, B., Warren, H.R., Hayward, C., Marten, J., Cranley, J.J., Concas, M.P., Gasparini, P., Boutin, T., Kolcic, I., Polasek, O., Rudan, I., Araujo, N.M., Lima-Costa, M.F., Ribeiro, A.L.P., Souza, R.P., Tarazona-Santos, E., Giedraitis, V., Ingelsson, E., Mahajan, A., Morris, A.P., Del Greco M, F., Foco, L., Gögele, M., Hicks, A.A., Cook, J.P., Lind, L., Lindgren, C.M., Sundström, J., Nelson, C.P., Riaz, M.B., Samani, N.J., Sinagra, G., Ulivi, S., Kähönen, M., Mishra, P.P., Mononen, N., Nikus, K., Caulfield, M.J., Dominiczak, A., Padmanabhan, S., Montasser, M.E., O'Connell, J.R., Ryan, K., Shuldiner, A.R., Aeschbacher, S., Conen, D., Risch, L., Thériault, S., Hutri-Kähönen, N., Lehtimäki, T., Lyytikäinen, L.-P., Raitakari, O.T., Barnes, C.L.K., Campbell, H., Joshi, P.K., Wilson, J.F., Isaacs, A., Kors, J.A., van Duijn, C.M., Huang, P.L., Gudnason, V., Harris, T.B., Launer, L.J., Smith, A.V., Bottinger, E.P., Loos, R.J.F., Nadkarni, G.N., Preuss, M.H., Correa, A., Mei, H., Wilson, J., Meitinger, T., Müller-Nurasyid, M., Peters, A., Waldenberger, M., Mangino, M., Spector, T.D., Rienstra, M., van de Vegte, Y.J., van der Harst, P., Verweij, N., Kääb, S., Schramm, K., Sinner, M.F., Strauch, K., Cutler, M.J., Fatkin, D., London, B., Olesen, M., Roden, D.M., Benjamin Shoemaker, M., Gustav Smith, J., Biggs, M.L., Bis, J.C., Brody, J.A., Psaty, B.M., Rice, K., Sotoodehnia, N., De Grandi, A., Fuchsberger, C., Pattaro, C., Pramstaller, P.P., Ford, I., Wouter Jukema, J., Macfarlane, P.W., Trompet, S., Dörr, M., Felix, S.B., Völker, U., Weiss, S., Havulinna, A.S., Jula, A., Sääksjärvi, K.,

Salomaa, V., Guo, X., Heckbert, S.R., Lin, H.J., Rotter, J.I., Taylor, K.D., Yao, J., de Mutsert, R., Maan, A.C., Mook-Kanamori, D.O., Noordam, R., Cucca, F., Ding, J., Lakatta, E.G., Qian, Y., Tarasov, K.V., Levy, D., Lin, H., Newton-Cheh, C.H., Lunetta, K.L., Murray, A.D., Porteous, D.J., Smith, B.H., Stricker, B.H., Uitterlinden, A., van den Berg, M.E., Haessler, J., Jackson, R.D., Kooperberg, C., Peters, U., Reiner, A.P., Whitsel, E.A., Alonso, A., Arking, D.E., Boerwinkle, E., Ehret, G.B., Soliman, E.Z., Avery, C.L., Gogarten, S.M., Kerr, K.F., Laurie, C.C., Seyerle, A.A., Stilp, A., Assa, S., Abdullah Said, M., Yldau van der Ende, M., Lambiase, P.D., Orini, M., Ramirez, J., Van Duijvenboden, S., Arnar, D.O., Gudbjartsson, D.F., Holm, H., Sulem, P., Thorleifsson, G., Thorolfsdottir, R.B., Thorsteinsdottir, U., Benjamin, E.J., Tinker, A., Stefansson, K., Ellinor, P.T., Jamshidi, Y., Lubitz, S.A., Munroe, P.B. *Multi-ancestry GWAS of the electrocardiographic PR interval identifies 202 loci underlying cardiac conduction*. **Nature Communications**. 2020; 11 (1), art. no. 2542. doi: 10.1038/s41467-020-15706-x.

405. Catamo, E., Tornese, G., Concas, M.P., Gasparini, P., Robino, A. *Differences in taste and smell perception between type 2 diabetes mellitus patients and healthy controls*. **Nutrition, Metabolism and Cardiovascular Diseases**. 2021; 31 (1), pp. 193-200. doi: 10.1016/j.numecd.2020.08.025.

406. Spedicati B, Cocca M, Palmisano R, Faletra F, Barbieri C, Francescatto M, Mezzavilla M, Morgan A, Pelliccione G, Gasparini P, Girotto G. *Natural Human Knockouts and Mendelian disorders: deep phenotyping in Italian isolates*. **European Journal of Human Genetics.** 2021. doi: 10.1038/s41431-021-00850-9.

407. Simcoe, M., Valdes, A., Liu, F., Furlotte, N.A., Evans, D.M., Hemani, G., Ring, S.M., Smith, G.D., Duffy, D.L., Zhu, G., Gordon, S.D., Medland, S.E., Vuckovic, D., Girotto, G., Sala, C., Catamo, E., Concas, M.P., Brumat, M., Gasparini, P., Toniolo, D., Cocca, M., Robino, A., Yazar, S., Hewitt, A., Wu, W., Kraft, P., Hammond, C.J., Shi, Y., Chen, Y., Zeng, C., Klaver, C.C.W., Uitterlinden, A.G., Ikram, M.A., Hamer, M.A., van Duijn, C.M., Nijsten, T., Han, J., Mackey, D.A., Martin, N.G., Cheng, C.-Y., Hinds, D.A., Spector, T.D., Kayser, M., Hysi, P.G., the 23andMe Research Team; the International Visible Trait Genetics Consortium. *Genome-wide association study in almost 195,000 individuals identifies 50 previously unidentified genetic loci for eye color*. **Science Advances** 2021, 7(11), art. no. abd1239. doi: 10.1126/sciadv.abd1239

408. Concas MP, Cocca M, Francescatto M, Battistuzzi T, Spedicati B, Feresin A, Morgan A, Gasparini P, Girotto G, *The role of knockout Olfactory Receptors Genes in Smell Perception*. **Genes** 2021, 12(5), 631. doi: https://doi.org/10.3390/genes12050631

409. Concas MP, Morgan A, Pelliccione G, Gasparini P, Girotto G. *Genetics, odor perception and food liking: the intriguing role of cinnamon*. **Food Quality and Preference** 2021, 93. doi: 10.1016/j.foodqual.2021.104277

410. Di Stazio, M., Foschi, N., Athanasakis, E., Gasparini, P., d'Adamo, A.P. *Systematic analysis of factors that improve*

homologous direct repair (HDR) efficiency in CRISPR/Cas9 technique. **PLoS ONE** 2021, 16, art. no. e0247603. doi: 10.1371/journal.pone.0247603

411. Radio, FC, Pang, K, Ciolfi, A, et al., including Gasparini, P.. *SPEN haploinsufficiency causes a neurodevelopmental disorder overlapping proximal 1p36 deletion syndrome with an episignature of X chromosomes in females*. **American Journal of Human Genetics**, 2021, 108(3): 502-516. doi: 10.1016/j.ajhg.2021.01.015

412. Concas, M.P., Morgan, A., Serra, F., Nagtegaal, A.P., Oosterloo, B.C., Seshadri, S., Heard-Costa, N., Van Camp, G., Fransen, E., Francescatto, M., Logroscino, G., Sardone, R., Quaranta, N., Gasparini, P., Girotto, G. *Hearing function: Identification of new candidate genes further explaining the complexity of this sensory ability*. **Genes,** 2021, 12 (8), art. no. 1228, DOI: 10.3390/genes12081228.

413. Robino, A., Rosso, N., Guerra, M., Corleone, P., Casagranda, B., Giraudi, P.J., Tiribelli, C., Simeth, C., Monica, F., La Bianca, M., Gasparini, P., de Manzini, N., Palmisano, S. *Taste perception and expression in stomach of bitter taste receptor tas2r38 in obese and lean subjects*. **Appetite**, 2021, 166, art. no. 105595. DOI: 10.1016/j.appet.2021.105595

414. Gallì, A., Todisco, G., Catamo, E., Sala, C., Elena, C., Pozzi, S., Bono, E., Ferretti, V.V., Rizzo, E., Molteni, E., Zibellini, S., Sarchi, M., Boveri, E., Ferrari, J., Fiorelli, N., Camaschella, C., Gasparini, P., Toniolo, D., Cazzola, M., Malcovati, L. *Relationship between clone metrics and clinical outcome in clonal cytopenia*. **Blood**, 2021, 138 (11), pp. 965-976. DOI: 10.1182/blood.2021011323

415. Pinart, M., Jeran, S., Boeing, H., Stelmach-Mardas, M., Standl, M., Schulz, H., Harris, C., von Berg, A., Herberth, G., Koletzko, S., Linseisen, J., Breuninger, T.A., Nöthlings, U., Barbaresko, J., Benda, S., Lachat, C., Yang, C., Gasparini, P., Robino, A., Rojo-Martínez, G., Castaño, L., Guillaume, M., Donneau, A.-F., Hoge, A., Gillain, N., Avraam, D., Burton, P.R., Bouwman, J., Pischon, T., Nimptsch, K. *Dietary Macronutrient Composition in Relation to Circulating HDL and Non-HDL Cholesterol: A Federated Individual-Level Analysis of Cross-Sectional Data from Adolescents and Adults in 8 European Studies*. **The Journal of nutrition**, 2021, 151 (8), pp. 2317-2329.

416. Mezzavilla, M., Navarra, C.O., Di Lenarda, R., Gasparini, P., Bevilacqua, L., Robino, A. *Runs of homozygosity are associated with staging of periodontitis in isolated populations*. **Human Molecular Genetics**, 2021, 30 (12), pp. 1154-1159. doi: 10.1093/hmg/ddab085

417. Pelletier, F., Perrier, S., Cayami, F.K., Mirchi, A., Saikali, S., Tran, L.T., Ulrick, N., Guerrero, K., Rampakakis, E., Van Spaendonk, R.M.L., Naidu, S., Pohl, D., Gibson, W.T., Demos, M., Goizet, C., Tejera-Martin, I., Potic, A., Fogel, B.L., Brais, B., Sylvain, M., Sébire, G., Lourenço, C.M., Bonkowsky, J.L., Catsman-Berrevoets, C., Pinto, P.S., Tirupathi, S., Strømme, P.,

De Grauw, T., Gieruszczak-Bialek, D., Krägeloh-Mann, I., Mierzewska, H., Philippi, H., Rankin, J., Atik, T., Banwell, B., Benko, W.S., Blaschek, A., Bley, A., Boltshauser, E., Bratkovic, D., Brozova, K., Cimas, I., Clough, C., Corenblum, B., Dinopoulos, A., Dolan, G., Faletra, F., Fernandez, R., Fletcher, J., Garcia Garcia, M.E., Gasparini, P., Gburek-Augustat, J., Gonzalez Moron, D., Hamati, A., Harting, I., Hertzberg, C., Hill, A., Hobson, G.M., Innes, A.M., Kauffman, M., Kirwin, S.M., Kluger, G., Kolditz, P., Kotzaeridou, U., La Piana, R., Liston, E., McClintock, W., McEntagart, M., McKenzie, F., Melançon, S., Misbahuddin, A., Suri, M., Monton, F.I., Moutton, S., Murphy, R.P.J., Nickel, M., Onay, H., Orcesi, S., Özklnay, F., Patzer, S., Pedro, H., Pekic, S., Pineda Marfa, M., Pizzino, A., Plecko, B., Poll-The, B.T., Popovic, V., Rating, D., Rioux, M.-F., Rodriguez Espinosa, N., Ronan, A., Ostergaard, J.R., Rossignol, E., Sanchez-Carpintero, R., Schossig, A., Senbil, N., Sønderberg Roos, L.K., Stevens, C.A., Synofzik, M., Sztriha, L., Tibussek, D., Timmann, D., Tonduti, D., Van De Warrenburg, B.P., Vázquez-López, M., Venkateswaran, S., Wasling, P., Wassmer, E., Webster, R.I., Wiegand, G., Yoon, G., Rotteveel, J., Schiffmann, R., Van Der Knaap, M.S., Vanderver, A., Martos-Moreno, G.Á., Polychronakos, C., Wolf, N.I., Bernard, G. *Endocrine and Growth Abnormalities in 4H Leukodystrophy Caused by Variants in POLR3A, POLR3B, and POLR1C*. **Journal of Clinical Endocrinology and Metabolism**, 2021, 106 (2), pp. E660-E674. doi: 10.1210/clinem/dgaa700

418. Bassani, S., Van Beelen, E., Rossel, M., Voisin, N., Morgan, A., Arribat, Y., Chatron, N., Chrast, J., Cocca, M., Delprat, B., Faletra, F., Giannuzzi, G., Guex, N., Machavoine, R., Pradervand, S., Smits, J.J., Van De Kamp, J.M., Ziegler, A., Amati, F., Marlin, S., Kremer, H., Locher, H., Maurice, T., Gasparini, P., Girotto, G., Reymond, A. *Variants in USP48 encoding ubiquitin hydrolase are associated with autosomal dominant non-syndromic hereditary hearing loss*. **Human Molecular Genetics**, 2021, 30 (19), pp. 1785-1796. doi: 10.1093/hmg/ddab145

419. Robino, A., Concas, M.P., Spinelli, S., Pierguidi, L., Tepper, B.J., Gasparini, P., Prescott, J., Monteleone, E., Gallina Toschi, T., Torri, L., Pagliarini, E., Gasperi, F., Dinnella, C. *Combined influence of TAS2R38 genotype and PROP phenotype on the intensity of basic tastes, astringency and pungency in the Italian taste project*. **Food Quality and Preference**, 2022, 95, art. no. 104361. doi: 10.1016/j.foodqual.2021.104361

420. Concas, M.P., Cocca, M., Catamo, E., Gasparini, P., Robino, A. *Eating disinhibition and food liking are influenced by variants in CAV1 (caveolin 1) gene*. **Food Quality and Preference**, 2022, 96, art. no. 104447. doi: 10.1016/j.foodqual.2021.104447

421. Catamo, E., Robino, A., Tinti, D., Dovc, K., Franceschi, R., Giangreco, M., Gasparini, P., Barbi,E., Cauvin, V., Rabbone, I., Battelino, T., Tornese, G. *Altered Taste Function in Young Individuals With Type 1 Diabetes.* **Front Nutr.** 2022 Jan 12;8:797920. doi: 10.3389/fnut.2021.797920. PMID: 35096943; PMCID: PMC8790289.

422. Catamo, E., Navarini, L., Gasparini, P., Robino, A. *Are taste variations associated with the liking of sweetened and unsweetened coffee?* **Physiol Behav.** 2022 Feb 1;244:113655. doi: 10.1016/j.physbeh.2021.113655. Epub 2021 Nov 20. PMID: 34813822.

423. Concas, M.P., Morgan, A., Tesolin, P., Santin, A., Girotto, G., Gasparini, P. *Sensory Capacities and Eating Behavior: Intriguing Results from a Large Cohort of Italian Individuals.* **Foods.** 2022 Mar 2;11(5):735. doi: 10.3390/foods11050735. PMID: 35267368; PMCID: PMC8909480.

424. Musante, L., Costa, P., Zanus, C., Faletra, F., Murru, F.M., Bianco, A.M., La Bianca, M., Ragusa, G., Athanasakis, E., d'Adamo, A.P., Carrozzi, M., Gasparini, P. *The Genetic Diagnosis of Ultrarare DEEs: An Ongoing Challenge.* **Genes** (Basel). 2022 Mar 12;13(3):500. doi: 10.3390/genes13030500. PMID: 35328054; PMCID: PMC8953579.

425. Okbay, A., Wu, Y., Wang, N.,et al., including Gasparini, P. *Polygenic prediction of educational attainment within and between families from genome-wide association analyses in 3 million individuals.* **Nat Genet.** 2022 Apr;54(4):437-449. doi: 10.1038/s41588-022-01016-z. Epub 2022 Mar 31. PMID: 35361970; PMCID: PMC9005349.

426. May-Wilson. S., Matoba, N., Wade, K.H., Hottenga, J.J., Concas, M.P., Mangino, M., Grzeszkowiak, E.J., Menni, C., Gasparini, P., Timpson, N.J., Veldhuizen, M.G., de Geus, E., Wilson, J.F., Pirastu, N. *Large-scale GWAS of food liking reveals genetic determinants and genetic correlations with distinct neurophysiological traits.* **Nat Commun.** 2022 May 18;13(1):2743. doi: 10.1038/s41467-022-30187-w. PMID: 35585065; PMCID: PMC9117208.

427. Winkler, T.W., Rasheed, H., Teumer, A., et al. including Gasparini, P. *Differential and shared genetic effects on kidney function between diabetic and non-diabetic individuals.* **Commun Biol.** 2022 Jun 13;5(1):580. doi: 10.1038/s42003-022-03448-z. PMID: 35697829; PMCID: PMC9192715.

428. Musante, L., Faletra, F., Meier, K., Tomoum, H., Najarzadeh Torbati, P., Blair, E., North, S., Gärtner, J., Diegmann, S., Beiraghi Toosi, M., Ashrafzadeh, F., Ghayoor Karimiani, E., Murphy, D., Murru, F.M., Zanus, C., Magnolato, A., La Bianca, M., Feresin, A., Girotto, G., Gasparini, P., Costa, P., Carrozzi, M. TTC5 syndrome: *Clinical and molecular spectrum of a severe and recognizable condition*. **Am J Med Genet A.** 2022 Sep;188(9):2652-2665. doi: 10.1002/ajmg.a.62852. Epub 2022 Jun 7. Erratum in: Am J Med Genet A. 2022 Dec 27;: PMID: 35670379; PMCID: PMC9541101.

429. Mezzavilla, M., Cocca, M., Maisano Delser, P., Badii, R., Abbaszadeh, F., Hadi, K.A., Giorgia, G., Gasparini, P. *Ancestry-related distribution of Runs of homozygosity and functional variants in Qatari population.* **BMC Genom Data.** 2022 Sep 21;23(1):73. doi: 10.1186/s12863-022-01087-1. PMID: 36131251; PMCID: PMC9490902.

430. Tesolin, P., Bertinetto, F.E., Sonaglia, A., Cappellani, S., Concas, M.P., Morgan, A., Ferrero, N.M., Zabotti, A., Gasparini, P., Amoroso, A., Quartuccio, L., Girotto, G. *High Throughput Genetic Characterisation of Caucasian Patients Affected by Multi-Drug Resistant Rheumatoid or Psoriatic Arthritis.* **J Pers Med**. 2022 Sep 30;12(10):1618. doi: 10.3390/jpm12101618. PMID: 36294757; PMCID: PMC9605087.

431. Yengo, L., Vedantam, S., Marouli, E., et al. including Gasparini P. *A saturated map of common genetic variants associated with human height*. **Nature.** 2022 Oct;610(7933):704-712. doi: 10.1038/s41586-022-05275-y. Epub 2022 Oct 12. PMID: 36224396; PMCID: PMC9605867.

432. D'Errico, S., Bergamini, P.R., Fattorini, P., Zanconati, F., Bussani, R., Cova, M.A., Pagnan, L., Belgrano, M., Gasparini P, Girotto, G., Lenarduzzi, S., Addobbati, R., Rakar, S., Aleksova, A., Dal Ferro, M., Zecchin, M., Sinagra, G. *Il Registro Regionale delle Morti Cardiache Improvvise in età giovanile del Friuli Venezia Giulia. Protocolli operativi e risultati di un progetto multidisciplinare [The Regional Registry of Sudden Cardiac Death of Friuli Venezia Giulia. Protocols, best practices and results of a multidisciplinary project].* **G Ital Cardiol** (Rome). 2022 Nov;23(11):827-835. Italian. doi: 10.1714/3900.38822. PMID: 36300386.

433. Aneli, S., Mezzavilla, M., Bortolini, E., Pirastu, N., Girotto, G., Spedicati, B., Berchialla, P., Gasparini, P., Pagani, L. *Impact of cultural and genetic structure on food choices along the Silk Road.* **Proc Natl Acad Sci** U S A. 2022 Nov 22;119(47):e2209311119. doi: 10.1073/pnas.2209311119. Epub 2022 Nov 14. PMID: 36375050; PMCID: PMC9704696.

434. Paldino,A., Dal Ferro, M., Stolfo, D., Gandin, I., Medo, K., Graw, .S, Gigli, M., Gagno, G., Zaffalon, D., Castrichini, M., Masè, M., Cannatà A, Brun F, Storm G, Severini GM, Lenarduzzi S, Girotto G, Gasparini P, Bortolotti F, Giacca M, Zacchigna S, Merlo M, Taylor MRG, Mestroni L, Sinagra G. *Prognostic Prediction of Genotype vs Phenotype in Genetic Cardiomyopathies*. **J Am Coll Cardiol**. 2022 Nov 22;80(21):1981-1994. doi: 10.1016/j.jacc.2022.08.804. PMID: 36396199.

435. Sy MR, Chauhan J, Prescott K, Imam A, Kraus A, Beleza A, Salkeld L, Hosdurga S, Parker M, Vasudevan P, Islam L, Goel H, Bain N, Park SM, Mohammed S, Dieterich K, Coutton C, Satre V, Vieville G, Donaldson A, Beneteau C, Ghoumid J, Van Den Bogaert K, Boogaerts A, Boudry E, Vanlerberghe C, Petit F, Bernardini L, Torres B, Mattina T, Carli D, Mandrile G, Pinelli M, Brunetti-Pierri N, Neas K, Beddow R, Tørring PM, Faletra F, Spedicati B, Gasparini P, Mussa A, Ferrero GB, Lampe A, Lam W, Bi W, Bacino CA, Kuwahara A, Bush JO, Zhao X, Luna PN, Shaw CA, Rosenfeld JA, Scott DA. *Exome sequencing efficacy and phenotypic expansions involving esophageal atresia/tracheoesophageal fistula plus.* **Am J Med Genet A.** 2022 Dec;188(12):3492-3504. doi: 10.1002/ajmg.a.62976. Epub 2022 Sep 22. PMID: 36135330; PMCID: PMC9669235.

436. Grati FR, Bestetti I, De Siero D, Malvestiti F, Villa N, Sala E,

Crosti F, Parisi V, Nardone AM, Di Giacomo G, Pettinari A, Tortora G, Montaldi A, Calò A, Saccilotto D, Zanchetti S, Celli P, Guerneri S, Silipigni R, Cardarelli L, Lippi E, Cavani S, Malacarne M, Genesio R, Beltrami N, Pittalis MC, Desiderio L, Gentile M, Ficarella R, Recalcati MP, Catusi I, Garzo M, Miele L, Corti C, Ghezzo S, Bertini V, Cambi F, Valetto A, Facchinetti B, Bernardini L, Capalbo A, Balducci F, Pelo E, Minuti B, Pescucci C, Giuliani C, Renieri A, Longo I, Tita R, Castello G, Casalone R, Righi R, Raso B, Civolani A, Muzi MC, di Natale M, Varriale L, Gasperini D, Nuzzi MC, Cellamare A, Casieri P, Busuito R, Ceccarini C, Cesarano C, Privitera O, Melani D, Menozzi C, Falcinelli C, Calabrese O, Battaglia P, Tanzariello A, Stampalija T, Ardisia C, Gasparini P, Benn P, Novelli A. *Positive predictive values and outcomes for uninformative cell-free DNA tests: An Italian multicentric Cytogenetic and cytogenomic Audit of diagnOstic testing (ICARO study).* **Prenat Diagn.** 2022 Dec;42(13):1575-1586. doi: 10.1002/pd.6271. Epub 2022 Nov 30. PMID: 36403097.

437. Dattilo V, Ulivi S, Minelli A, La Bianca M, Giacopuzzi E, Bortolomasi M, Bignotti S, Gennarelli M, Gasparini P, Concas MP. *Genome-wide association studies on Northern Italy isolated populations provide further support concerning genetic susceptibility for major depressive disorder.* **World J Biol Psychiatry.** 2023 Feb;24(2):135-148. doi: 10.1080/15622975.2022.2082523. Epub 2022 Jun 20. PMID: 35615967.

438. Bakker MK, Kanning JP, Abraham G, et al. including Gasparini, P. *Genetic Risk Score for Intracranial Aneurysms: Prediction of Subarachnoid Hemorrhage and Role in Clinical Heterogeneity.* **Stroke**. 2023 Mar;54(3):810-818. doi: 10.1161/STROKEAHA.122.040715. Epub 2023 Jan 19. PMID: 36655558; PMCID: PMC9951795.

439. Crudele F, Bianchi N, Terrazzan A, Ancona P, Frassoldati A, Gasparini P, D'Adamo AP, Papaioannou D, Garzon R, Wójcicka A, Gaj P, Jażdżewski K, Palatini J, Volinia S. *Circular RNAs Could Encode Unique Proteins and Affect Cancer Pathways.* **Biology** (Basel). 2023 Mar 24;12(4):493. doi: 10.3390/biology12040493. PMID: 37106694; PMCID: PMC10135897.

440. Reurink J, Weisschuh N, Garanto A, Dockery A, van den Born LI, Fajardy I, Haer-Wigman L, Kohl S, Wissinger B, Farrar GJ, Ben-Yosef T, Pfiffner FK, Berger W, Weener ME, Dudakova L, Liskova P, Sharon D, Salameh M, Offenheim A, Heon E, Girotto G, Gasparini P, Morgan A, Bergen AA, Ten Brink JB, Klaver CCW, Tranebjærg L, Rendtorff ND, Vermeer S, Smits JJ, Pennings RJE, Aben M, Oostrik J, Astuti GDN, Corominas Galbany J, Kroes HY, Phan M, van Zelst-Stams WAG, Thiadens AAHJ, Verheij JBGM, van Schooneveld MJ, de Bruijn SE, Li CHZ, Hoyng CB, Gilissen C, Vissers LELM, Cremers FPM, Kremer H, van Wijk E, Roosing S. *Whole genome sequencing for USH2A-associated disease reveals several pathogenic deep-intronic variants that are amenable to splice correction.* **HGG Adv.** 2023 Jan 18;4(2):100181. doi: 10.1016/j.xhgg.2023.100181. PMID: 36785559; PMCID: PMC9918427.

441. Graham CA, Spedicati B, Pelliccione G, Gasparini P, Concas MP. *Regulator of G-Protein Signalling 9: A New Candidate Gene for Sweet Food Liking?* **Foods.** 2023 Apr 22;12(9):1739. doi: 10.3390/foods12091739. PMID: 37174278; PMCID: PMC10178705.

442. Lenarduzzi S, Spedicati B, Alessandrini B, Tesolin P, Paldino A, Gigli M, Sinagra G, Gasparini P, Ferro MD, Girotto G. *Whole-exome sequencing: Clinical characterization of pediatric and adult Italian patients affected by different forms of hereditary cardiovascular diseases*. **Mol Genet Genomic Med.** 2023 May;11(5):e2143. doi: 10.1002/mgg3.2143. Epub 2023 Feb 14. PMID: 36788754; PMCID: PMC10178800.

443. Jonard L, Brotto D, Moreno-Pelayo MA, Del Castillo I, Kremer H, Pennings R, Caria H, Fialho G, Boudewyns A, Van Camp G, Ołdak M, Oziębło D, Deggouj N, De Siati RD, Gasparini P, Girotto G, Verstreken M, Dossena S, Roesch S, Battelino S, Trebušak Podkrajšek K, Warnecke A, Lenarz T, Lesinski-Schiedat A, Mondain M, Roux AF, Denoyelle F, Loundon N, Serey Gaut M, Trevisi P, Rubinato E, Martini A, Marlin S. *Genetic Evaluation of Prelingual Hearing Impairment: Recommendations of an European Network for Genetic Hearing Impairment.* **Audiol Res.** 2023 May 10;13(3):341-346. doi: 10.3390/audiolres13030029. PMID: 37218840; PMCID: PMC10204519.

444. Magri C, Marchina E, Sansone E, D'Adamo AP, Cappellani S, Bonfanti C, Terlenghi L, Biasiotto G, Zanella I, Sala E, Caruso A, Lombardo M, Gasparini P, De Palma G, Gennarelli M. *Genome-wide association studies of response and side effects to the BNT162b2 vaccine in Italian healthcare workers: Increased antibody levels and side effects in carriers of the HLA-A*03:01 allele*. **HLA**. 2023 Dec;102(6):707-719. doi: 10.1111/tan.15157. Epub 2023 Jul 19. PMID: 37469131.

445. Ganji-Arjenaki M, Kamali Z; International Consortium of Blood Pressure; Sardari S, de Borst M, Snieder H,Vaez A. *Prioritization of Kidney Cell Types Highlights Myofibroblast Cells in Regulating Human Blood Pressure.* **Kidney Int Rep**. 2024 Mar 13;9(6):1849-1859. doi: 10.1016/j.ekir.2024.03.001. eCollection 2024 Jun. PMID: 38899223.

446. Stevens H, Graham CA, Concas MP, Piluso F, Mavrommatis Y, King A, Pilic L, Gasparini P. *TAS1R2 rs35874116 Associations with Taste, Diet, and Health in an Italian Population*. **Nutrients**. 2025 Jan 17;17(2):329. doi: 10.3390/nu17020329. PMID: 39861458; PMCID: PMC11769509.

447. Menghi L, Fontana L, Camarda S, Endrizzi I, Concas MP, Gasparini P, Gasperi F. *Gender Moderates the Associations Between Responsiveness to Alarming Oral Sensations, Depressive Symptoms, and Dietary Habits in Adolescents.* **Nutrients**. 2025 May 12;17(10):1653. doi: 10.3390/nu17101653. PMID: 40431393; PMCID: PMC12113894.

448. Spedicati B, Pasquetti D, Santin A, Zampieri S, Morgan A, Lenarduzzi S, Nardone GG, Paccagnella E, Cappellani S, Diplotti L, Pensiero S, Parentin F, Gasparini P, Battaglia Parodi M, Girotto G. *Hidden in the Genome: The First Italian Family with North*

Carolina Macular Dystrophy Carrying a Novel PRDM13 and CCNC Duplication. **Biomedicines**. 2025 Aug 5;13(8):1904. doi: 10.3390/biomedicines13081904. PMID: 40868158; PMCID: PMC12383978.

449. Nardone GG, Andrioletti V, Santin A, Morgan A, Spedicati B, Concas MP, Gasparini P, Girotto G, Limongelli I. *A Hitchhiker Guide to Structural Variant Calling: A Comprehensive Benchmark Through Different Sequencing Technologies*. **Biomedicines**. 2025 Aug 9;13(8):1949. doi: 10.3390/biomedicines13081949. PMID: 40868203; PMCID: PMC12383524.

450. Perotto M, Paldino A, Mazzarotto F, Barbati G, Stroeks SLVM, Verdonschot JAJ, Akhtar M, Elliott P, Ochoa JP, Garcia-Pavia P, de Frutos F, Sepp R, Hategan L, Prasad S, Yazdani M, Morris-Rosendahl D, Palinkas ED, Girolami F, Olivotto I, Parikh VN, Fatkin D, Lakdawala N, McKenna WJ, Stolfo D, Gigli M, Brun F, Collesi C, Giacca M, Zacchigna S, Severini GM, Lenarduzzi S, Spedicati B, Santin A, Girotto G, Gasparini P, Taylor MRG, Mestroni L, Merlo M, Sinagra G, Dal Ferro M. *Genetic and Phenotypic Characterization of Nexilin (NEXN)-Related Cardiomyopathy: Results From a Multicentric Study.* **JACC Heart Fail**. 2025 Sep;13(9):102529. doi: 10.1016/j.jchf.2025.102529. Epub 2025 Jul 18. PMID: 40680702; PMCID: PMC12405182.

451. Fortunato F, Fiocco L, Margutti A, Neri M, D'Amico A, Bertini E, Ricci E, Mercuri EM, Pane M, Massa R, Greco G, Berardinelli AL, Cereda C, Pini A, Merlini L, Fusco C, Rodolico C, Messina S, Fiorillo C, Bruno C, Pedemonte M, Traverso M, Moroni I, Maggi L, Gibertini S, Pegoraro E, Picillo E, Politano L, Scutifero M, Vercellino F, Massaro F, Filosto M, Gasparini P, Ricci F, Mongini TE, Selvatici R, Ferlini A, Gualandi F. *Landscape Analysis of COL6A1, COL6A2, and COL6A3 Pathogenic Variants in a Large Italian Cohort Presenting with Collagen VI-Related Myopathies: A Nationwide Report.* **Biomolecules**. 2025 Oct 8;15(10):1426. doi: 10.3390/biom15101426. PMID: 41154655; PMCID: PMC12564197.

452. Spedicati B, Pianigiani G, Santin A, Gasparini VR, Falcomer I, Feresin A, Bonati MT, Mazzà D, Paccagnella E, Pasquetti D, Rubinato E, Granata C, Murru FM, Pinamonti M, Bussani R, Fantasia I, Stampalija T, Gasparini P, Zampieri S, Girotto G. *Beyond the Curtains: Identification of the Genetic Cause of Foetal Developmental Abnormalities Through the Application of Molecular Autopsy*. **Genes** (Basel). 2025 Oct 2;16(10):1167. doi: 10.3390/genes16101167. PMID: 41153384; PMCID: PMC12562548.

453. Piluso F, Graham CA, Stevens H, Camarda S, King A, Pilic L, Mavrommatis Y, Gasparini P, Concas MP. *Associations of CD36 rs1761667 with Fat and Umami Food Perception, Diet Quality, and BMI in Two European Studies*. **Foods**. 2025 Nov 7;14(22):3813. doi: 10.3390/foods14223813. PMID: 41299972; PMCID: PMC12651780.

454. Graham CA, Stevens H, Piluso F, Camarda S, Mavrommatis Y, Gasparini P, Concas MP. *Differences in smell and taste*

performance and food liking between patients with stroke and healthy controls. **Sci Rep**. 2025 Nov 27. doi: 10.1038/s41598-025-27469-w. Epub ahead of print. PMID: 41310363.

455. Causio FA, Farina S, Maio A, Beccia F, Russo L, Baccolini V, Chiara M, Cicchetti A, Colombo GI, Comandé G, Coviello D, De Maria R, Delledonne M, De Vito C, Galeone D, Gasparini P, Horner D, Martinelli G, Marzuillo C, Palazzani L, Pitini E, Sanguinetti M, Scarpa A, Tartaglia M, Tiziano FD, Tonon G, Dallapiccola B, Villari P, Calabrò GE, Boccia S. *The italian national genomic strategy: current status, challenges, and future perspectives in clinical practice and public health.* **J Community Genet**. 2025 Dec;16(6):699-703. doi: 10.1007/s12687-025-00828-w. Epub 2025 Oct 13. PMID: 41082076; PMCID: PMC12569334.

456. Modenini G, Mercuri G, Abondio P, Nardone GG, Santin A, Tesolin P, Spedicati B, Pecori A, Pianigiani G, Concas MP, Girotto G, Gasparini P, Boattini A, Mezzavilla M. *Variability of transposable elements in six genetic isolates from North-Eastern Italy and their relationship with alcohol consumption, tobacco use and BMI*. **BMC Genomics**. 2025 Nov 11;26(1):1027. doi: 10.1186/s12864-025-12225-1. PMID: 41219721; PMCID: PMC12607106.

PUBBLICAZIONI SU RIVISTE SCIENTIFICHE NAZIONALI

1. P.Gasparini. *Meccanismo di leucemogenesi nell'uomo: traslocazione dell'oncogene 5' myc all' "Enhancer" di una catena immunoglobulinica pesante nel DNA tumorale di una leucemia linfoblastica acuta (L3)* - ottobre 1985 (Tesi di laurea con assegnazione della dignita' di stampa) (allegato solamente fotocopia dichiarazione di dignita' di stampa)
2. P.Gasparini, A.Savoia, P.Bertazzon, F.Bertoldo, C.Morandi, P.F.Pignatti. *A comparative analysis of different membranes in DNA transfer and hybridization procedures*. **Chimica Oggi/Chemistry Today**11: 43-45, 1988
3. P.Gasparini, A.Savoia, D.De Leo, M.Marigo, PF.Pignatti. *L'uso di sonde molecolari di DNA nella determinazione della paternita.* **Rivista Italiana di Medicina Legale**11: 158-166, 1989
4. E.Bellone, P.Mandich, F.De Stefano, F.Sangiuolo, G.Novelli, P.Gasparini, A.Savoia, E.Trabetti. *Frequenze di marcatori polimorfici ipervariabili nella popolazione italiana*. **Il DNA in Medicina Legale**, collana Studi di Medicina Legale, Scienze Criminologiche e Sociali, Edizioni Colosseum, p. 29-35, 1989
5. P.Gasparini, A.Savoia, PF.Pignatti, C.Morandi, P.Mezzelani, G.DeSandre. *Search of HIV DNA by polymerase chain reaction in the urine sediments od seropositive individuals*. **Recenti Progressi in Medicina/ Italian Journal of Internal Medicine**81: 35-36, 1990
6. PF.Pignatti, P.Gasparini. *DNA e Medicina*. **Exacta Medica**3: 70-75, 1990
7. P.Gasparini, A.Bonizzato, M.Dognini, A.Savoia, P.F.Pignatti,

G.Borgo, G.Cabrini, G.Mastella. *La Fibrosi Cistica*. **Medico e Bambino** 9:16-23, 1991

8. C. Camaschella, A. Roetto, L. Sbaiz, I. Dianzani, G. De Sandre, A Totaro, A. Piperno, P. Gasparini. *Molecular Genetics of Hemochromatosis*. **Bulletin of Molecular Biology and Medicine**20:117-119, 1995
9. P.Gasparini, L.Bisceglia, J. Calonge, V. Nunes, M. Palacin, E. Beccia, M Gallucci, L. Zelante. *Progress in the Analysis of the Gene Responsable for Cystinuria*. **Bulletin of Molecular Biology and Medicine** 20:127-128, 1995
10. S.Calvano, L.Bisceglia, E.Memeo, L.Zelante, P.Gasparini. *Utility of a nucleotide tail into PCR primers for a large variety of molecular techniques*. **Minerva Biotecnologica**7: 25-28, 1995
11. G.Lama, AM.Aurino, P.Gasparini, L.Zelante. *Cistinuria*. **Rivista Italiana di Pediatria (IJP)**21, 69-74, 1995
12. P.Gasparini. "Introduzione alla biologia molecolare" **Prospettive in Pediatria** 26:199-201, 1996
13. P.Gasparini, G.Camera, L.Zelante. *Malattie dello scheletro da mutazioni nei geni per i recettori dei fattori di crescita (FGFR)*. **Prospettive in Pediatria**26, 65-74, 1996
14. A.Notarangelo, R. Latino, P. Gasparini, L. Zelante, V. M. Fazio, M. Rinaldi. *Efficient Transfection of Adherent Cells Using Cell Suspensions*. **Focus** 19 num.3:58-59, 1997
15. S.Melchionda, P.Fortina, X.Estivill, V.Volpini, N.Govea, M.Milà, S.Surrey, A.Totaro, P.Stanziale, L.Zelante, P.Gasparini. *Analysis of microsatellites markers for linkage studies of genetic deafness*. **Minerva Biotecnologica**9: 93-97, 1997
16. P.Gasparini, L.Bisceglia. *Cistinuria*. **Enciclopedia Medica Italiana** (Aggiornamento II, Tomo I, 1221-1223), UTES Editore, Firenze 1998
17. Carella AM, Bianco G, Carella M, Gasparini P. *Diagnosis of hereditary hemochromatosis with molecular analysis of DNA in patients with anti-HCV positive liver cirrhosis. Clinical case*. **Minerva Medica**89:323-7, 1998
18. P.Gasparini, S. Melchionda, E Di Iorio, A Grifa, L. Zelante. *Genetica delle sordita' non sindromiche*. **Prospettive in Pediatria**30:357-362, 2000
19. P.Gasparini. *Ruolo della Biologia Molecolare nella Diagnostica Medica*. **Il Policlinico**– Sez. Pratica 107:457-465, 2000

MONOGRAFIE E/O VOLUMI

1. F.Simonelli, A.Ciccodicola, P.Gasparini. "Occhio ed orecchio tra pediatria e genetica". Collana Pediatria Politematica, Casa Editrice UTET Periodici Scientifici, Milano, 2000
2. M.Palacin, V.Nunes, P.Gasparini, R.Goodyer. Cystinuria. In "The Metabolic and Molecular Bases of Inherited Diseases", 8° ed, Scriver et al Eds, .Mac-Graw-Hill, vol III, p 4909-4932, 2000
3. Paolo Gasparini. "Connexins". In "Genetic Hearing Loss" Edited by Patric J. Willems, 14, Page 207-222, 2003.
4. Paolo Gasparini, Salvatore Melchionda, Enzo Di Iorio, Anna Grifa,

Leopoldo Zelante. "Sordità" su Malattie Genetiche Molecole e Geni , A. Cao, B. Dallapiccola, L.D. Notarangelo, Editore Piccin, p. 759-773, 2004

5. Morgan A, Gasparini P, Girotto G. "Hearing loss" in "Clinical DNA Variant Interpretation: Theory and Practice" edited by Conxi Lázaro Jordan Lerner-Ellis, Amanda Spurdle, a book in the series "Translational and Applied Genomics". Elsevier/Academic Press (2020).

Premi e riconoscimenti per l'attività scientifica, inclusa l'affiliazione ad accademie di riconosciuto prestigio nel settore

- 2021 Premio Medicina Italia per la Ricerca Scientifica.
- 2011 Premio Grande Ippocrate per la Ricerca Scientifica quale migliore ricercatore/divulgatore.
- 2010 Vincitore di "QNRF award for biomedicine" al convegno annuale della Qatar National Research Foundation per la migliore presentazione nel settore biomedico.
- 2003 Vincitore di "The European Society of Human Genetics/Nature Publishing Group Award" per l'articolo più citato nel 2001.
- 2000 Vincitore di "International Journal of Molecular Medicine Award**"** perla ricerca scientifica.
- 1999 Premio Augustale 99 per la Ricerca Scientifica.
- 1994 Premio SIBIoC 94 per la comunicazione "Rivelazione di mutazioni mediante RNA single strand conformation polymorphism (rSSCP).
- 1991 AIRH Junior Award per la genetica medica per il contributo dato allo sviluppo della ricerca in Genetica Medica Molecolare ed in particolare nel campo delle malattie ereditarie ad insorgenza infantile come la Fibrosi Cistica.
- 1988 Socrea - Premio Sirtori per la genetica medica per il migliore comunicazione scientifica nel campo della Genetica Medica a tema: "Diagnosi prenatale di fibrosi cistica in 24 ore mediante l'uso della Polymerase Chain Reaction" tenuta al 3° Congresso Nazionale F.I.S.M.E.
- Iscritto all'Ordine dei Medici di Verona.