

## CURRICULUM VITAE



### INFORMAZIONI PERSONALI

|                         |                                                                                                                                                                                                                                                               |
|-------------------------|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <b>Cognome e Nome</b>   | <b>GASPARINI PAOLO</b>                                                                                                                                                                                                                                        |
| <b>Qualifica</b>        | <b>ABILITATO ALLA PROFESSIONE DI MEDICO-CHIRURGO, PROFESSORE ORDINARIO DI GENETICA MEDICA</b>                                                                                                                                                                 |
| <b>Incarico attuale</b> | <b>PROFESSORE ORDINARIO, DIRETTORE DI DIPARTIMENTO (DIPARTIMENTO DEI SERVIZI E DIAGNOSTICA AVANZATA), DIRETTORE DI STRUTTURA COMPLESSA (GENETICA MEDICA), DIRETTORE DI STRUTTURA COMPLESSA AD INTERIM (LABORATORIO DI DIAGNOSTICA AVANZATA TRASLAZIONALE)</b> |
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### 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

• **Esperienze professionali (incarichi ricoperti)**

**2005-oggi**

**Università degli Studi di Trieste, Trieste.**

*Dipartimento Universitario Clinico di Scienze Mediche Chirurgiche e della Salute.*

- Direttore Vicario del 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.
- Direttore della IScuola di Specializzazione in Genetica Medica dell'Università degli Studi di Trieste.
- Coordinatore del Dottorato in Medicina Personalizzata e Terapie Innovative dell'università degli Studi di Trieste.
- 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).
- Direttore di Struttura complessa ad Interim: Laboratorio di Diagnostica Avanzata Traslazionale.

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 Viadella 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 (Committee for Advanced Therapies) dell'EMA (European Medicines Agency) (per nove anni sino al febbraio 2021).
- 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)
- Nel 2023 è stato nominato delegato italiano del CHMP (Committee for Medicinal Products for Human Use) dell'EMA
- Nel corso del 2022/2023 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

## **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'Università 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) presso l'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'Università 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 bacino 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 terapia con interferon e/o chemioterapia e/o trapianto di midollo osseo.
- f) caratterizzazione e sviluppo di sonde molecolari ipervariabili da utilizzarsi a scopi di 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 diversi congressi Nazionali ed Internazionali.

### Gennaio 1984 - Gennaio 1987

Clinica Medica A dell'Università 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 è 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 Istocompatibilità 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, Gai 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, Ivanson 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 chloride 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*

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#### **MONOGRAFIE E/O VOLUMI**

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#### **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" per la 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.

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