Fondazione IRCCS Ca' Granda
Ospedale Maggiore Policlinico
Sistema Socio Sanitario
Regione Lombardia

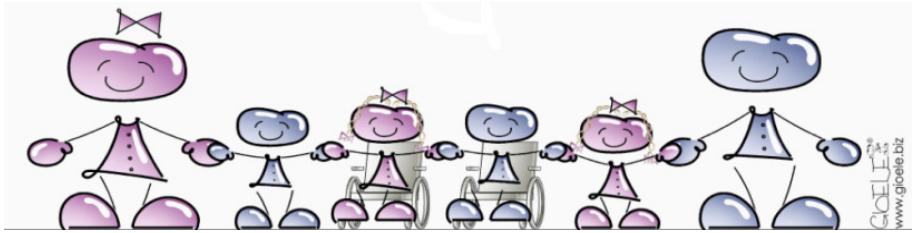
DIPARTIMENTO DI NEUROSCIENZE E DI SALUTE MENTALE

U.O. Neuropsichiatria dell'Infanzia e dell'Adolescenza - U.O.N.P.I.A

Sapre
Settore
Abilitazione Precoce
Genitori

"Approccio pratico ai problemi: posturazione, manipolazione e alimentazione nella SMA"

Chiara Mastella – Terapista della Riabilitazione
Counselor Sistemico Sanitario



Trieste, 16 Ottobre 2017

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SMA 1
Le forme più note

- **SMA 1 forma A - grave**
  - Esordio neonatale-1 mese di vita
- **SMA 1 forma B - comparsa precoce**
  - Esordio precoce sintomi 3-5 mesi
- **SMA 1 forma C - comparsa tardiva**
  - Esordio "tardivo" 5/6 mesi

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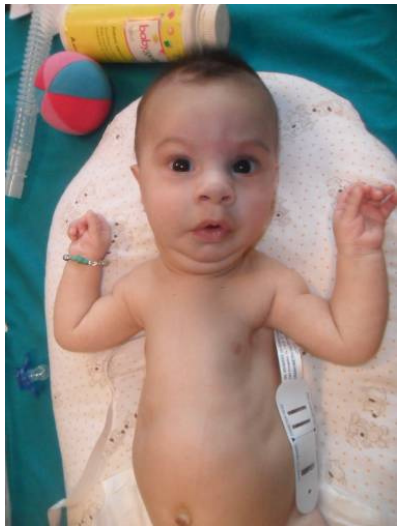
SMA 1A grave
Esordio neonatale
Di quali sintomi stiamo parlando?

- **Dalla nascita al 1° mese di vita**
  - Sintomi nel periodo perinatale
  - Nessun movimento antigravitario o molto limitato
  - Insufficienza respiratoria
    - FR aumentata
  - Perdita progressiva suzione autonoma 2° mese
  - Aspettativa vita spontanea < 3-6 mesi



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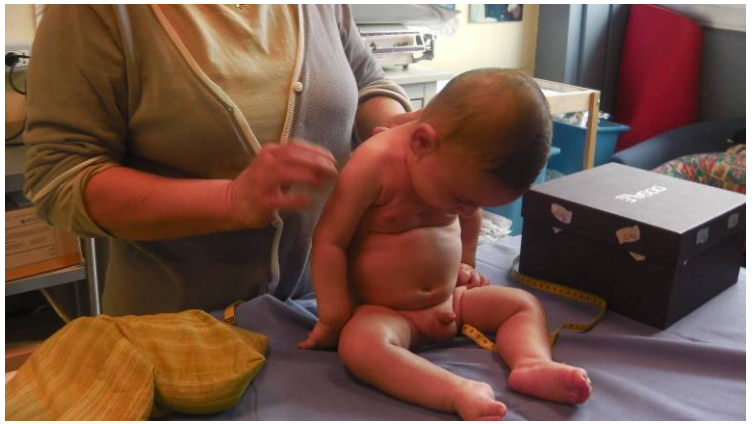
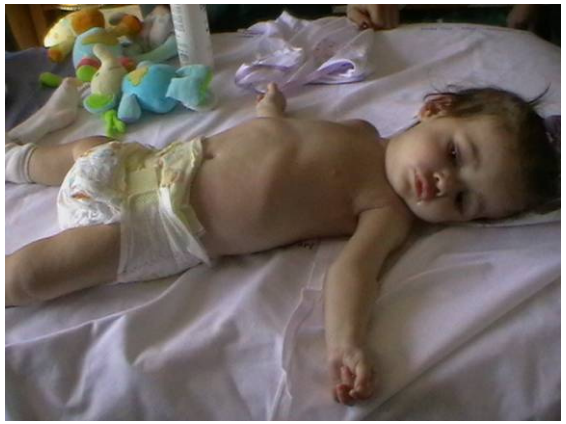
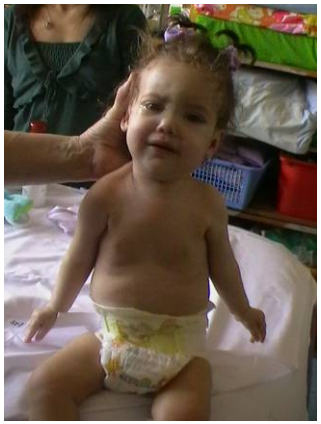
SMA 1B
Sintomi 3-6 mesi
Di quali sintomi stiamo parlando?

- **SMA 1 forma B**
  - Assenza controllo del capo
  - Movimenti distali presenti
    - Iniziale benessere
  - Progressiva insufficienza deglutizione
    - Sng 6-7 mesi
    - Aumento salivazione
  - Insufficienza respiratoria
    - Necessità ventilazione meccanica 8-10 mesi/Aspettativa vita spontanena 12-15 mesi di vita
  - Comunicazione: solo movimenti oculare o mugolii

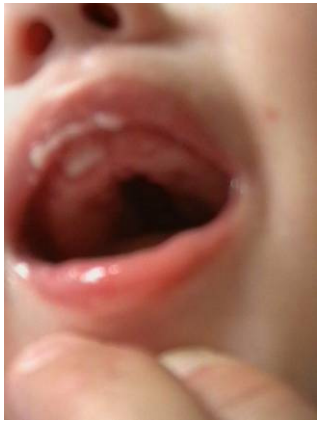


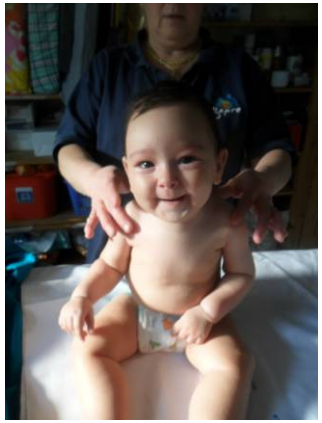


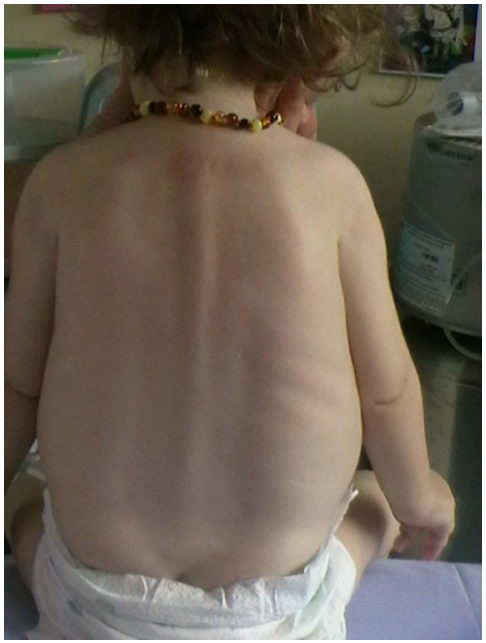


SMA 1C
Forma "tardiva"
Di quali sintomi stiamo parlando?

- **SMA 1 forma C - comparsa tardiva 6-7 mesi**
  - Iniziale controllo del capo,
    - Permane fino 8-10 mesi
  - Alimentazione orale anche oltre 12 mese vita
  - Aspettativa vita senza ventilazione < 18-24 mesi
  - Sopravvivenza oltre 3°/a vita con supporto ventilazione ed alimentazione
  - Comunicazione: parole singole comprensibili solo dai famigliari

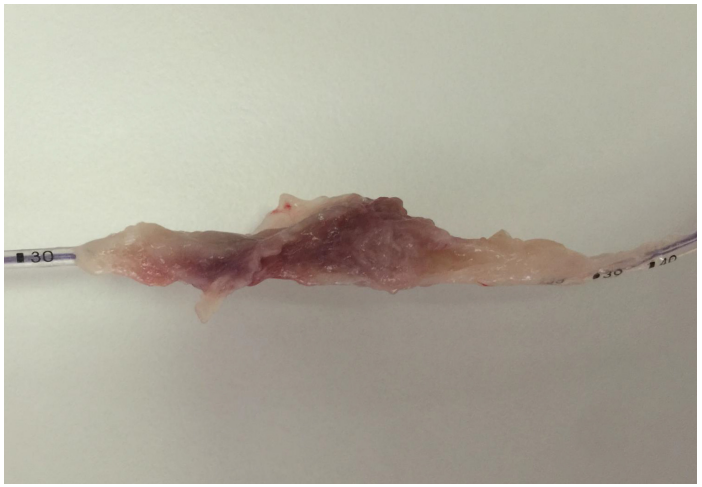


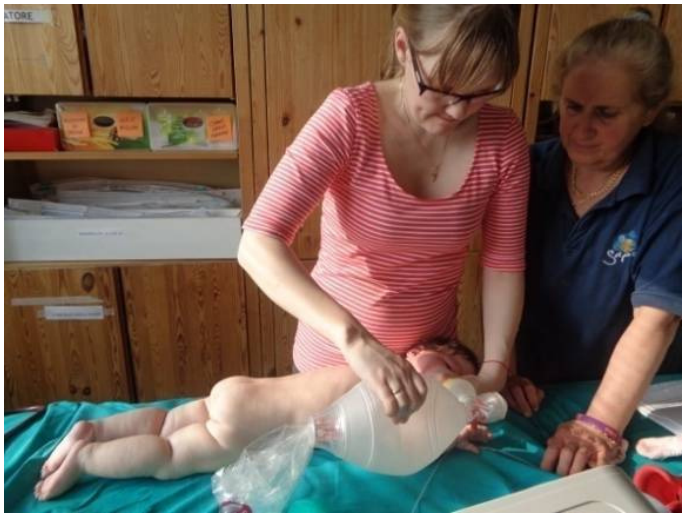


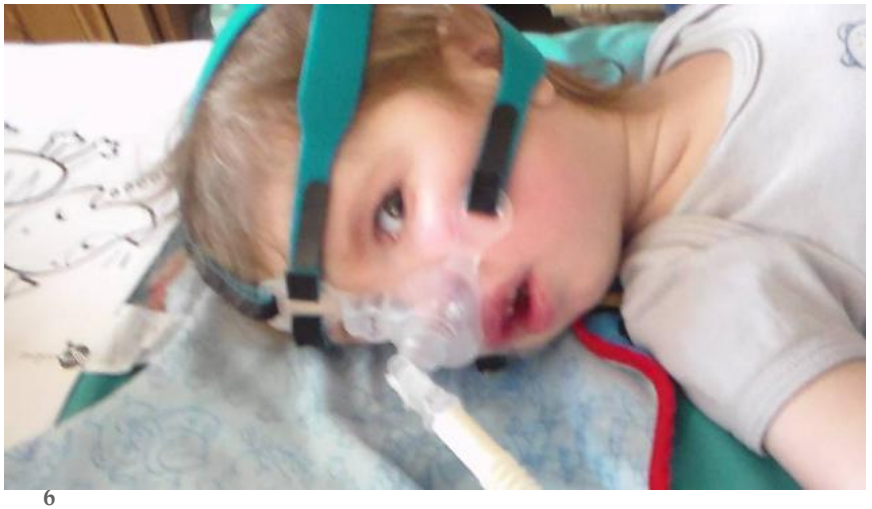


PROBLEMI RESPIRATORI
Di quali complicanze stiamo parlando?

- Insufficienza respiratoria cronica
- Ventilazione meccanica
  - in maschera, in tracheo
- Tosse debole o inefficace
- Catarro

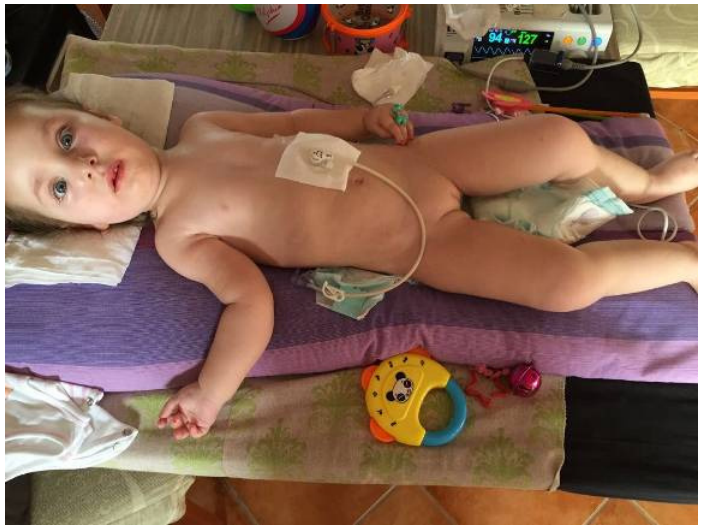


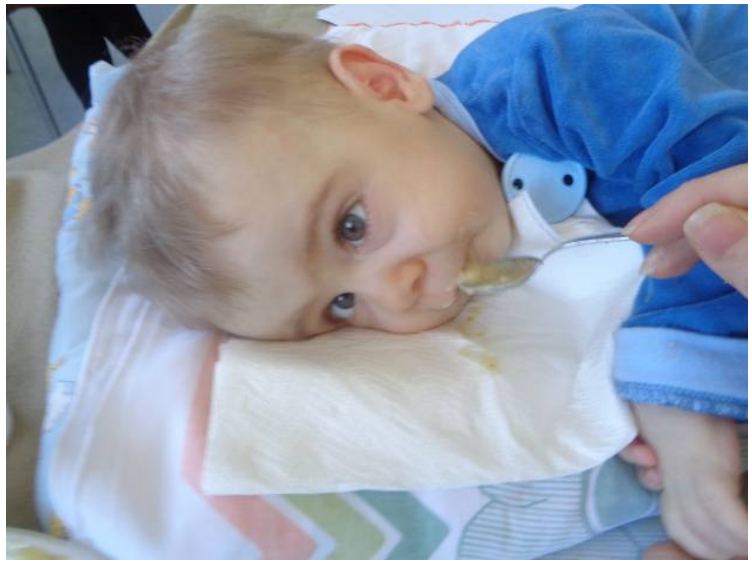




PROBLEMI DI DEGLUTIZIONE
Di quali complicanze stiamo parlando?

- Scialorrea
- Insufficiente capacità di alimentarsi
- Riduzione crescita
- Sondino naso gastrico
- Nissen-Peg





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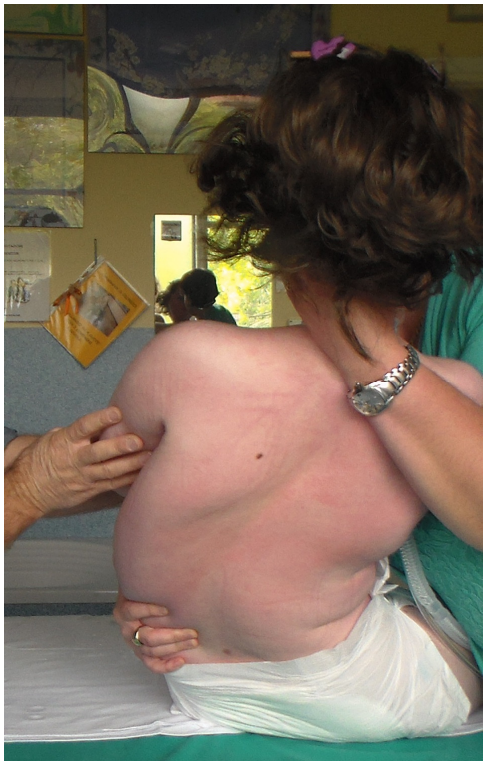
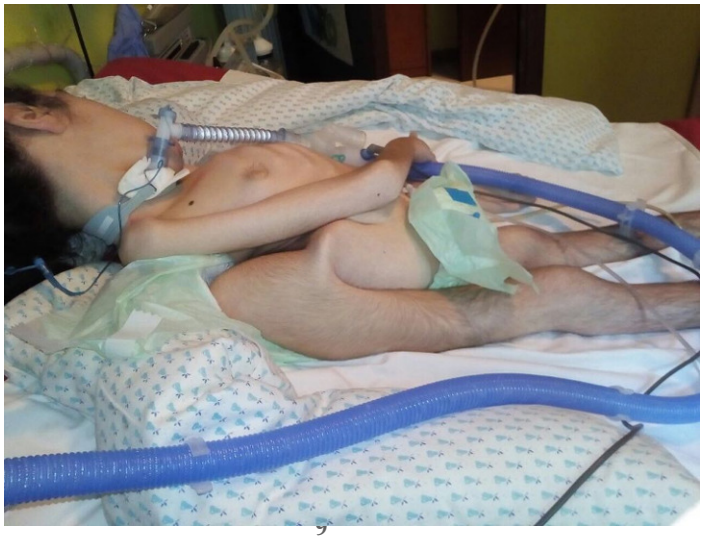
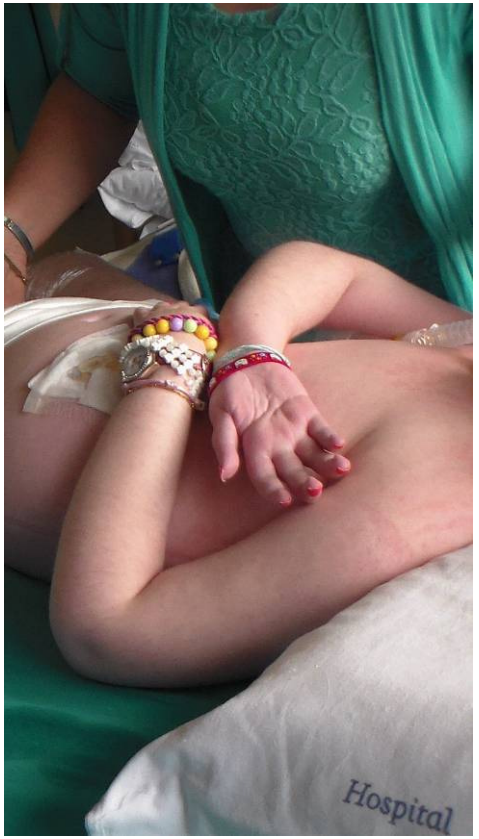
PROBLEMI POSTURALI
Di quali complicanze stiamo parlando?

- Posizione supina testa girata di lato
- Ricoveri ripetuti
- Deformità ortopediche
  - Scoliosi
  - Retrazioni
  - Dolore



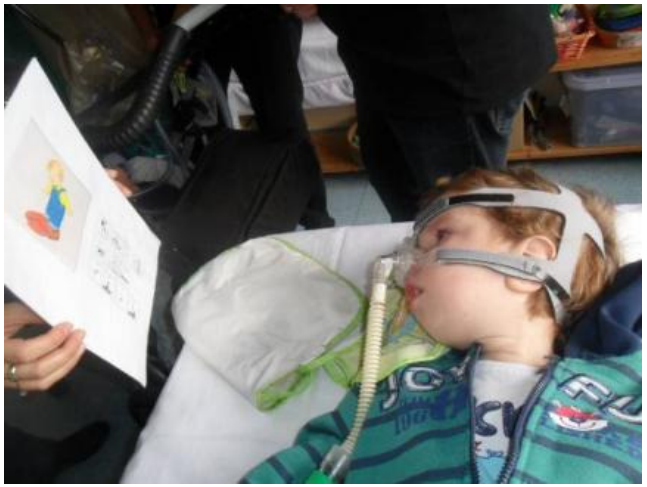
PROBLEMI DI DEFORMITA'





PROBLEMI DI COMUNICAZIONE
Di quali complicanze stiamo parlando?

- La maggior parte de bambini SMA 1 non parla
- Pochi parlano con linguaggio comprensibile solo alle persone di riferimento



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PROBLEMI DI DIPENDENZA
bambini ad alto livello assistenziale

Intanto la ricerca va avanti...

Roche

avexis

ISIS PHARMACEUTICALS

NOVARTIS

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Sistema Socio Sanitario Regione Lombardia

Sciroppo Trial Roche SMA 1 (RG7916)

Sapre Settore Abilitazione Precoce Genitori

Sciroppo Trial Novartis (LMI070)

NOVARTIS PHARMACEUTICALS

Ed ora le infusioni EAP Nusinersen diventato SPINRAZA da qualche settimana

AIFA Agenzia Italiana del Farmaco

Settembre 2017 SPINRAZA approvato per tutte le SMA.

SPINRAZA™ (nusinersen) injection 12 mg/5 mL

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Cosa fare con i bambini SMA, ora che la loro storia naturale sta cambiando?

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SMA Standards of Care

2007 – 5 aree principali

1. Diagnostica/Nuovi interventi
2. Clinica Pneumologica
3. Clinica Gastrointestinale - Nutrizione
4. Clinica Ortopedica/Riabilitazione
5. Cure Palliative

2016 – 9 aree principali

:

1. Diagnosis and genetics
2. Nutrition, Growth and Bone Health Care
3. Pulmonary Care
4. Orthopedic Care
5. Physical Therapy and Rehabilitation
6. Other organ system involvement
7. Acute care in the hospital setting
8. Medication
9. Ethics and palliative care

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Lo strumento di misurazione: la CHOP

Un aiuto per la ricerca degli obiettivi per il trattamento riabilitativo

CHOP INTEND

CHILDREN'S HOSPITAL *of* PHILADELPHIA INFANT TEST OF NEUROMUSCULAR DISORDERS

Name: Diagnosis:
MR: Gestational age:
DOE: Time of evaluation: Time since last feeding:
DOB: Current health: URI☐Gtube ☐ BIPAP☐ HRS/Day___ HRSoff BIPAP at testing___

Item	Position	Test Procedure	Graded Response		Score	
1 Spontaneous movement (Upper extremity)	Supine	Observe throughout testing May unweight limb or stimulate infant to facilitate response	Antigravity shoulder movement (achieves elbow off surface)	4	L	Best side:
			Antigravity elbow movement (achieves hand and forearm off surface)	3		
			Wrist movement	2	R	State:
			Finger movement	1		
			No movement of limbs	0		
2 Spontaneous movement (Lower extremity)	Supine	Observe throughout testing May unweight limb or stimulate infant to facilitate response	Antigravity hip movement (achieves feet and knees off surface)	4	L	Best side:
			Antigravity hip adduction/internal rotation (knees off surface)	3		
			Active gravity eliminated knee movement	2	R	State:
			Ankle movement	1		
			No movement of limbs	0		
3 Hand grip	Supine	Grip strength: place finger in palm and lift until shoulder comes off surface observe when infant looses grasp May use toy of similar diameter for older children	Maintains hand grip with shoulder off bed	4	L	Best side:
			Maintains grip with elbow off surface (shoulders on surface)	3		
			Maintains grip with forearm off surface (elbow supported on surface)	2	R	State:
			Maintains grip only with no traction	1		
			No attempt to maintain grasp	0		
4 Head in midline with visual stimulation*	Supine head midline	Visual stimulation is given with toy. *If head is maintained in midline for 5 seconds:* Place head in maximum available rotation and provide visual stimulation to encourage midline	Rotates from maximum rotation to midline	4	L>R	Best side:
			Turns head part way back to midline	3		
			Maintains midline for 5 or more seconds	2	R>L	State:
			Maintains midline, less than 5 seconds	1		
			Head falls to side, no attempts to regain midline	0		
5 Hip adductors	Supine, no diaper	Hips flexed and adducted Feet hip width apart and thighs parallel, knees slightly apart	Keeps knee off surface of bed > 5 sec or lifts foot off surface	4	L	Best side:
			Keeps knees off surface of bed 1-5 sec	2	R	State:
			No attempt to maintain knees off surface	0		
6 Rolling: elicited from legs*	Supine (arms at side) Keep side tested up roll away from the Side tested	1. Holding infant's lower thigh, flex hip and knee and adduct across midline bringing pelvis vertical maintain traction and *pause in this position*. 2. If infant rolls to side apply traction at a 45° diagonal to body and pause to allow infant to attempt to derotate body	When traction is applied at the end of the maneuver, rolls to prone with lateral head righting	4	To R	Best side:
			Rolls through side lying into prone without lateral head righting, clears weight-bearing arm to complete roll	3		
			Pelvis, trunk and arm lift from support surface, head turns and rolls onto side, arm comes thru to front of body	2	To L	State:
			Pelvis and trunk lift from support surface and head turns to side. Arm remains behind trunk	1		
			Pelvis lifted passively off support surface.	0		
7 Rolling: elicited from arms*	Supine (arms at side) Keep side tested up roll away from the Side tested	1. Hold infant at the elbow move toward opposite shoulder maintain traction on limb and *pause with the shoulders vertical* allow infant to derotate 2.if the pelvis achieves vertical continue to provide traction	Rolls to prone with lateral head righting	4	To R	Best side:
			Rolls into prone without lateral head righting; must clear weight-bearing arm completely to finish roll	3		
			Rolls onto side, leg comes thru and adducts, bringing the pelvis vertical	2	To L	State:
			Head turns to side and shoulder and trunk lift from surface	1		
			Head turns to side; body remains limp or shoulder lifts passively	0		
8 Shoulder and elbow flexion And horizontal abduction	Side-lying with upper arm at 30° of shoulder extension and elbow flexion and supported on body (restrain lower arm if needed)	Prompt reach for a toy presented at arms length at shoulder level (may provide stimulation and *observe spontaneous movement*)	Clears hand from surface with antigravity arm movement	4	L	Best side:
			Able to flex shoulder to 45 degrees, without antigravity arm movement	3		
			Flexes elbow after arm comes off body	2		
			Able to get arm off body	1	R	State:
			No attempt	0		
9 Shoulder flexion & Elbow flexion	Sitting in lap or on mat with head and trunk support (20° recline)	Present stimulus at midline and at shoulder level at arms length (may provide stimulation and *observe spontaneous movement*)	Abducts or flexes shoulder to 60 degrees	4	L	Best side:
			Abducts or flexes shoulder to 30 degrees	3		
			Any shoulder flexion or abduction	2		
			Flexes elbow only	1	R	State:
			No attempt to lift arm	0		
10 Knee extension	Sitting in lap or over edge of mat with head and trunk support (20° recline) thigh horizontal to ground	Tickle plantar surface of foot Or gently pinch toe	Extends knee to > 45 degrees	4	L	Best side:
			Extends knee 15 to 45 degrees	2		
			Any visible knee extension	1	R	State:
			No visible knee extension	0		
11 Hip flexion and foot dorsiflexion	Hold infant against your body with legs free, facing outward. Support at the abdomen with the child's head resting between your arm and thorax	Stroke the foot or pinch the toe	Hip flexion or knee flexion > 30°	4	L	Best side:
			Any hip flexion or knee flexion	3		
			Ankle dorsiflexion only	2	R	State:
			No active hip, knee or ankle motion	0		
12 Head control*	Sitting with support at the shoulders and trunk erect	Place the infant in ring sit with head erect and assistance given at the shoulders (front and back). (*may delay scoring a grade of 1 and 4 until end of test*)	Attains head upright from flexion and turns head side to side	4		Score:
			Maintains head upright for >15 sec (for bobbing head control score a 2)	3		
			Maintains head in midline for >5 sec. with the head tipped in up to 30° of forward flexion or extension	2		
			Actively lifts or rontes head twice from flexion within 15 seconds (do not credit if movement is in time with breathing)	1		State:
			No response, head hangs	0		
13 Elbow flexion Score with item 14	Supine	**Traction response:** pull to sit extend arms at 45 degree angle, to point of nearly lifting head off surface	Flexes elbow	4	L	Best side:
			Visible biceps contraction without elbow flexion	2		
			No visible contraction	0	R	State:
14 Neck Flexion Score with item 13	Supine	**Traction response:** hold in neutral proximal to wrist and shoulder at 45°. *to point of nearly lifting head off surface*	Lifts head off bed	4		Score:
			Visible muscle contraction of SCM	2		
			No muscle contraction	0		State:
15 Head/Neck Extension (Landau)	Ventral suspension: Prone, held in one hand upper abdomen	Stoke along spine from neck to sacrum. The coronal axis of the head when parallel to the bed surface = 0 degrees (horizontal)	Extends head *to* horizontal plane or above	4		Score:
			Extends head partially, but not to horizontal	2		
			No head extension	0		State:
16 Spinal Incurvation (Galant)	Ventral suspension: Prone, held in one hand upper abdomen	Stroke Right then Left thoracolumbar parasipinals or tickle abdomen or foot or tilt in infants with integrated Galant For infant over 10 kg knees and head may touch	Twists pelvis towards stimulus off axis	4	L	Best side:
			Visible paraspinal muscle contraction	2		
			No response	0	R	State:
Total score, best score on each side for each item (maximum 64 points):						

* Adapted from the Test of Infant Motor Performance, Campbell, SK; et al 2001.

Contractures:
L☐R☐Knee flexion
L☐R☐Ankle plantar flexion (Present < 20 degrees knee extended)
L☐R☐Hip adductor L☐R☐ ITB contracture (Note if leg cannot abduct and ext. rot. to contact surface in supine)
L☐R☐Shoulder protraction
L☐R☐Elbow flexion
L☐R☐Neck rotation
L☐R☐Neck lateral flexion
☐ Plagiocephaly
☐ Fixed spinal curve

Behavioral State: (Brazelton, TB.Neonatal Behavioral Assessment Scale, 2nd ed.,1984)
State 1 Deep sleep State 2 Light sleep
State 3 Drowsy or semi-dozing State 4 Alert, with bright look
State 5 Eyes open, considerable activity State 6 Crying

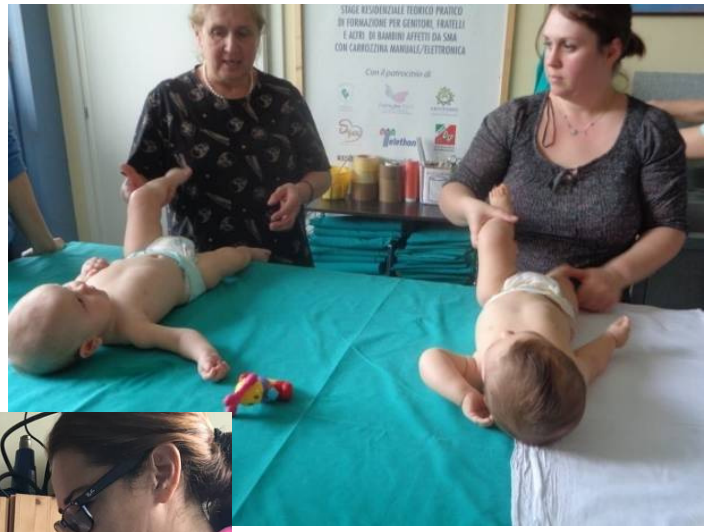
Testing environment:
Ideally test first thing in the AM or same time of day about 1 hour after feeding
Test on a firm padded mat
Diaper /onesie only unless the infant is cold
Test with red wool ball on ring to encourage participation
May use pacifier only if needed to maintain state 4 or 5 (see definition).
Mark as CNT (could not test) if patient could not be tested DO NOT MARK 0

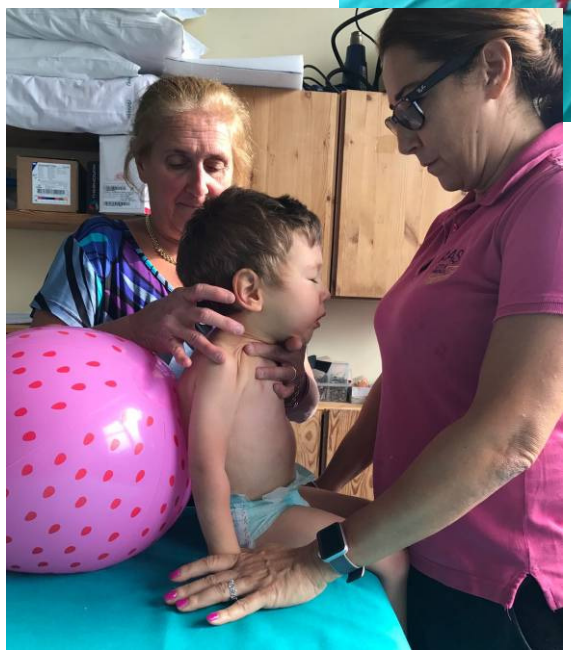
La Fisioterapia quotidiana

OBIETTIVI:

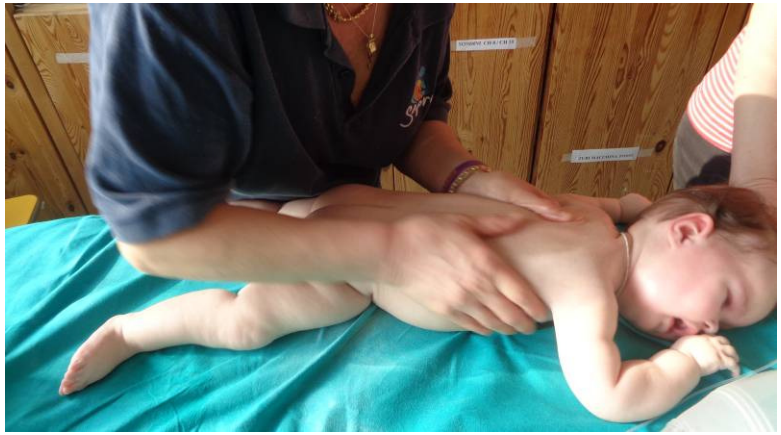
- **Cercare modi per aumentare**
- **abilità motorie residue ed acquisibili del bambino**
- **Mantenimento**
  - Peso
  - Tono muscolare
  - Massa muscolare
- **Prevenzione**
- **Protezione deformità potenziali**

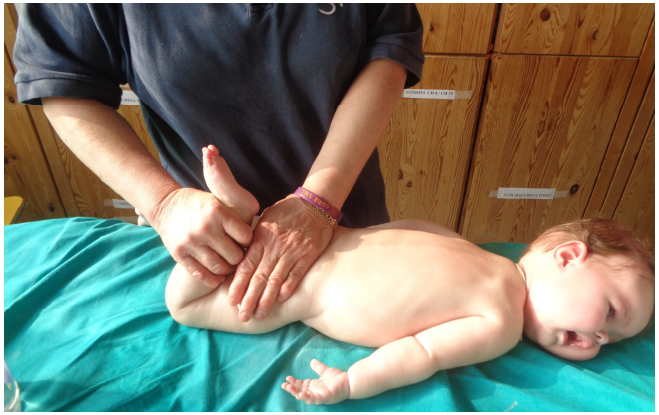
5. Physical Therapy and Rehabilitation

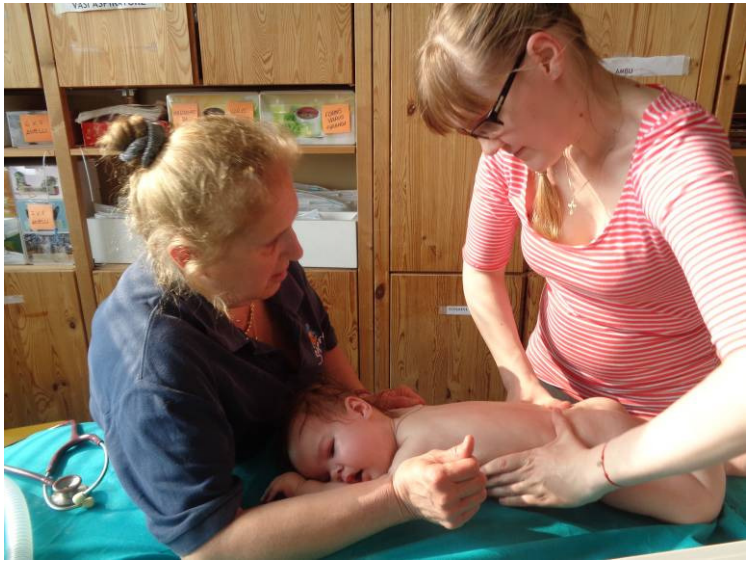


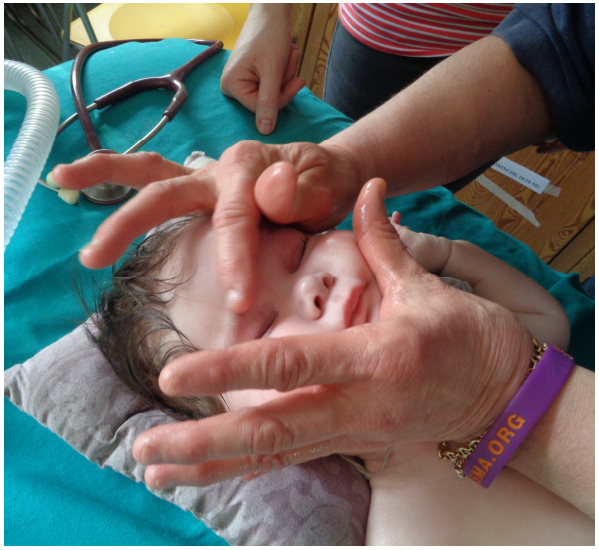


Il massaggio









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L'Allineamento del Corpo attraverso la cultura del salame di miglio

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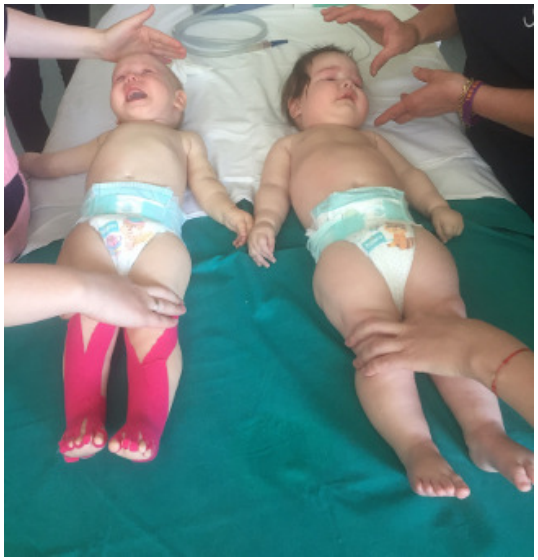
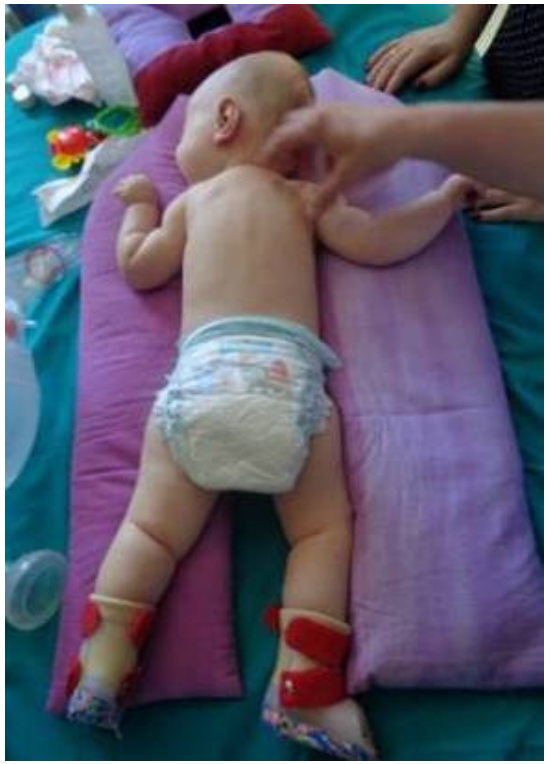
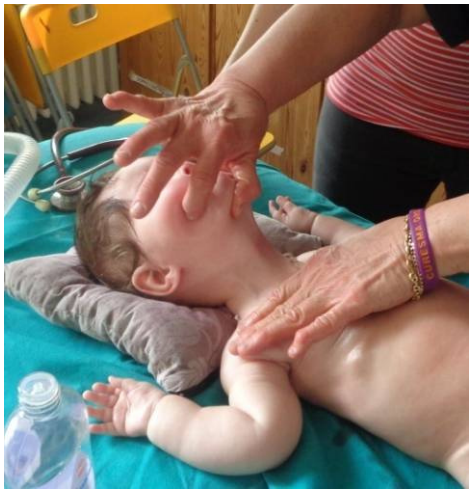
4. Orthopedic Care

6. Other organ system involvement



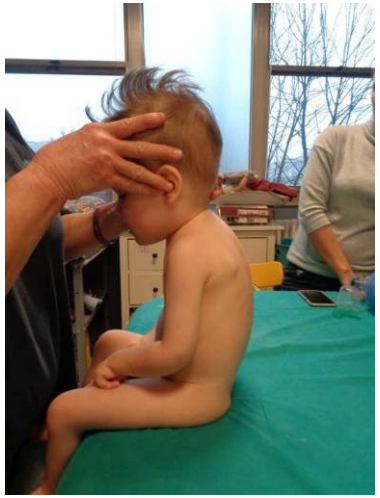


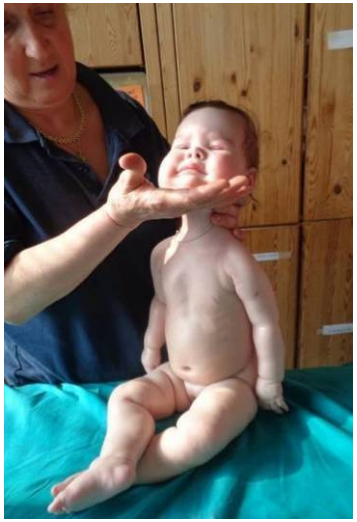
TESTA - COLLO
Allungamento muscoli collo
CHOP Sezioni Nr. 4 – 12- 14

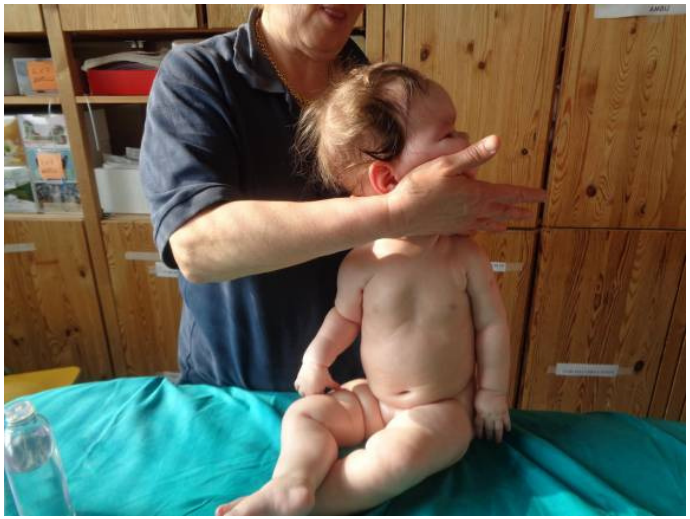


Allungamento collo

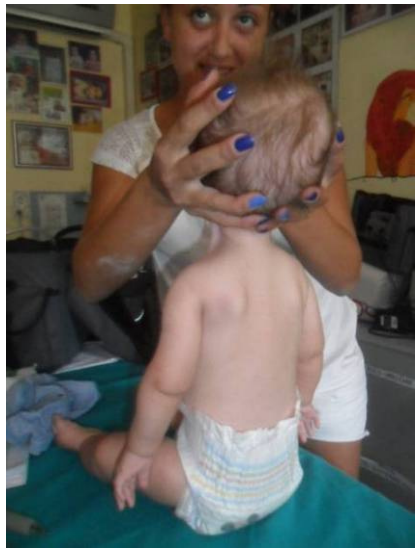
- Contrastare effetto gravità
- Proteggere colonna
- Scaricare peso testa
- Liberare vie aeree superiori







5. Physical Therapy and Rehabilitation



Il tiracollo

4. Orthopedic Care

5. Physical Therapy and Rehabilitation

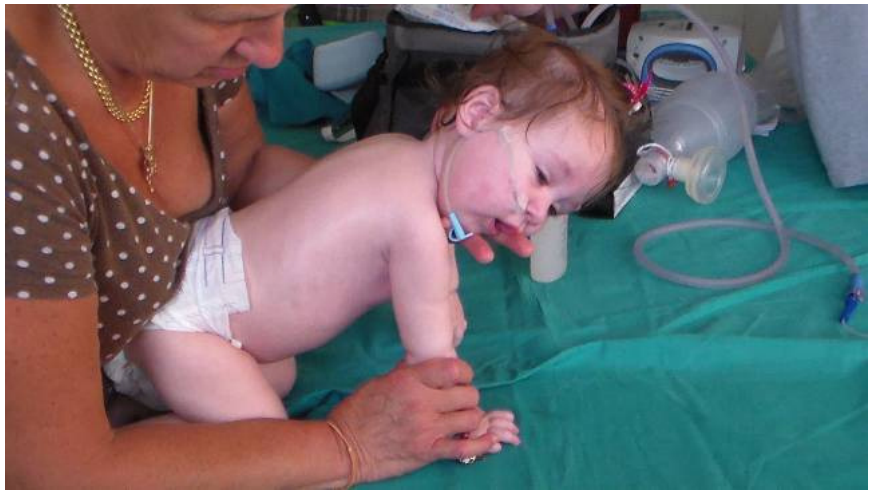
Fondazione IRCCS Ca' Granda Ospedale Maggiore Policlinico

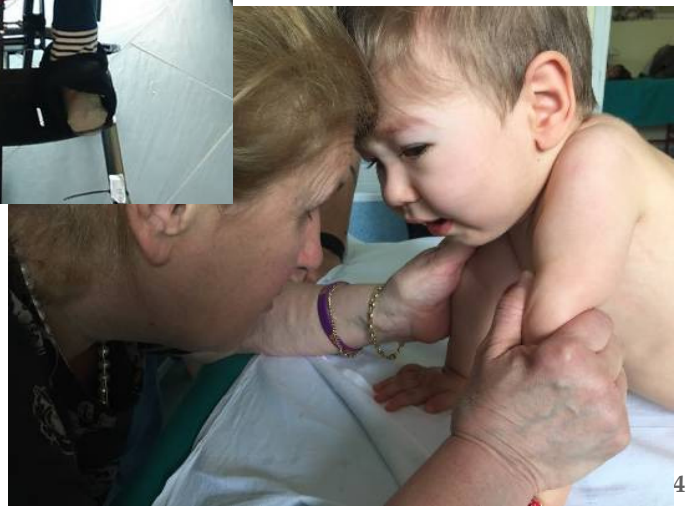
Sistema Socio Sanitario

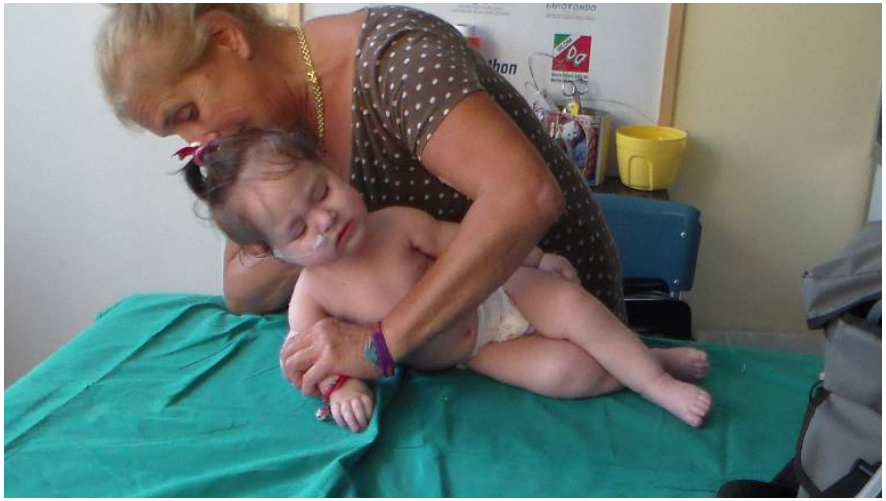
Regione Lombardia

Settore Abilitazione Precoce Genitori

Arti superiori
IN CARICO e SCARICO

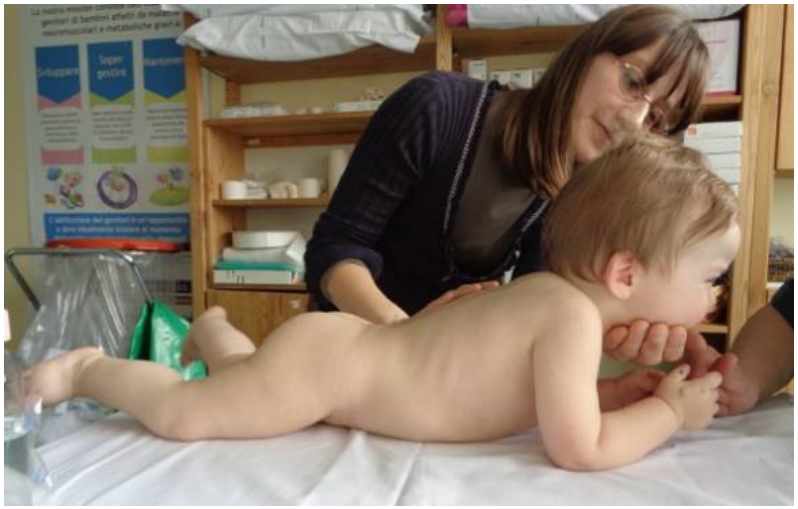


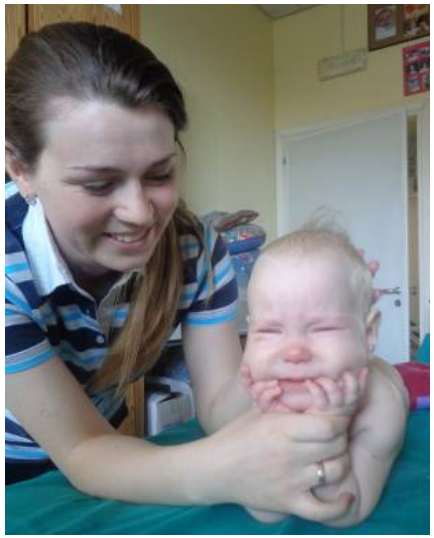




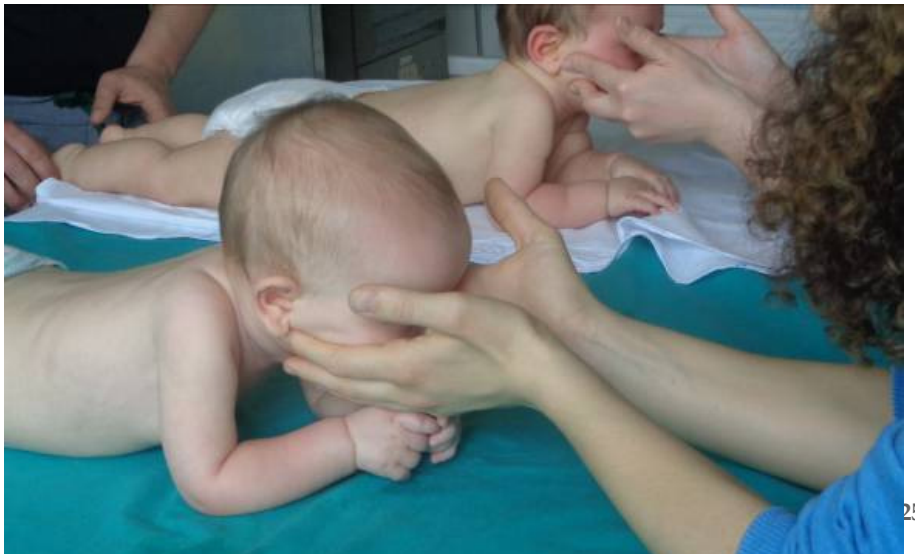
5. Physical Therapy and Rehabilitation

GOMITI



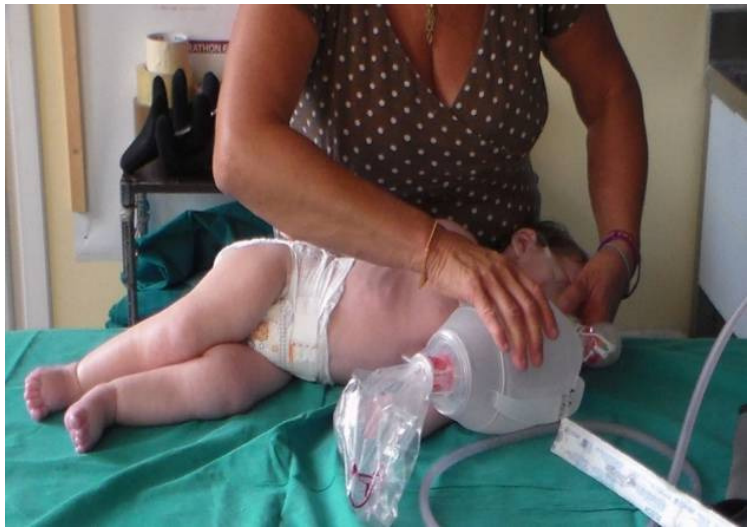


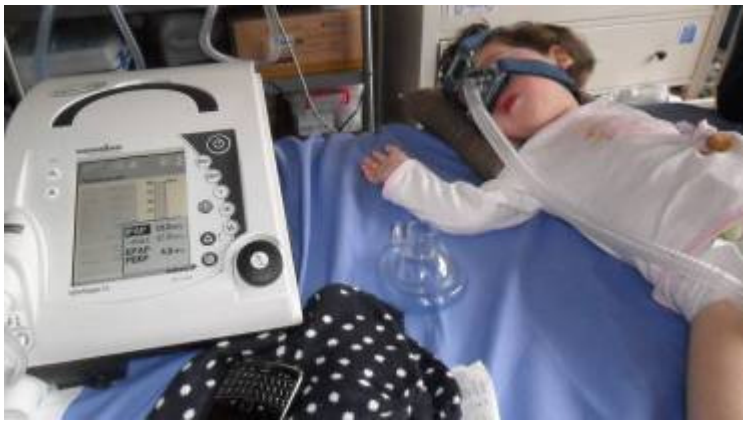
5. Physical Therapy and Rehabilitation

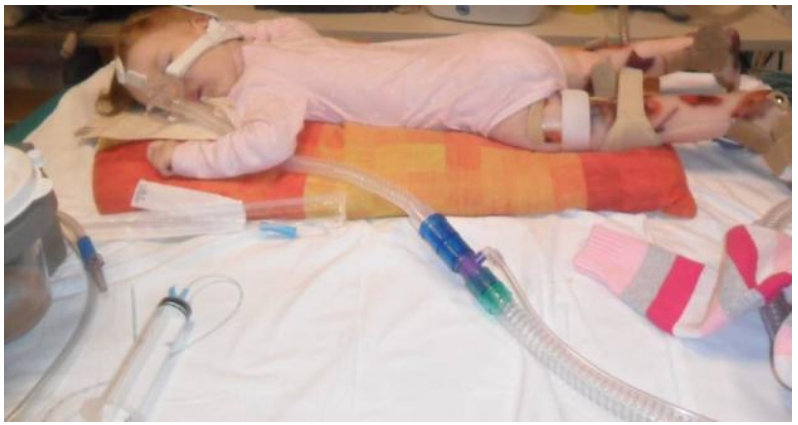


PROBLEMI RESPIRATORI
Saper usare Ambu, Macchina della tosse e Ventilatore, come esercizio e in emergenza

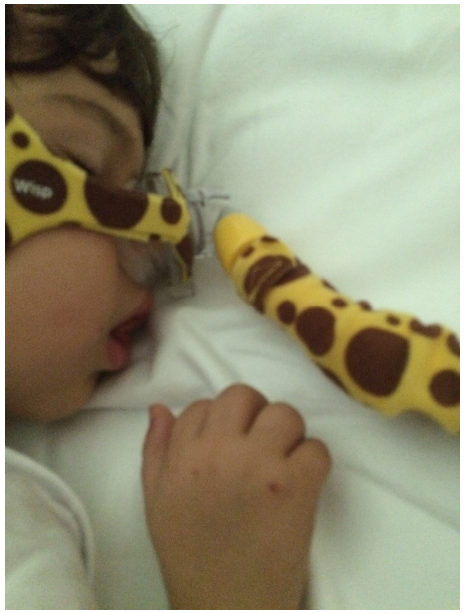
3. Pulmonary Care

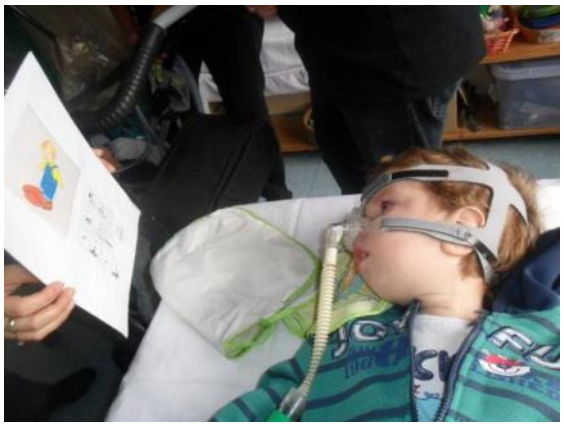




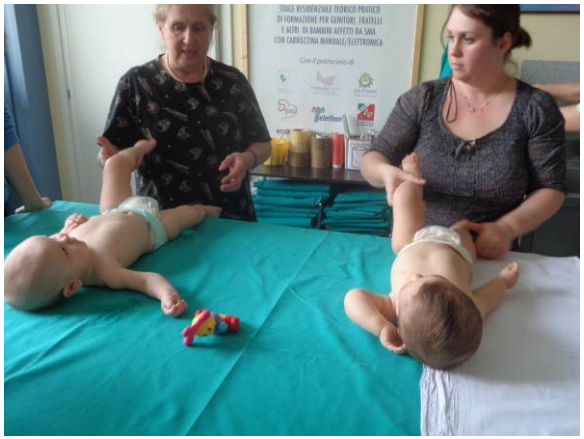


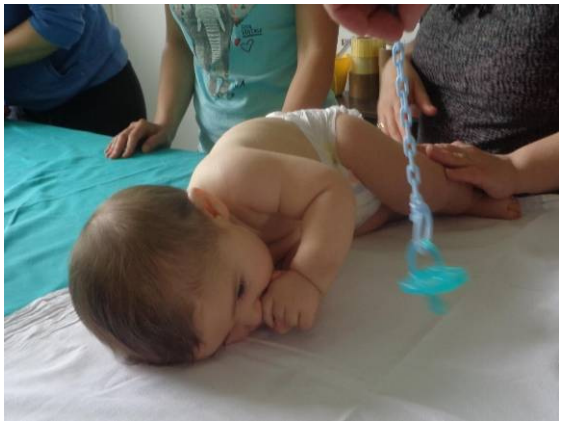
MASCHERE NIV

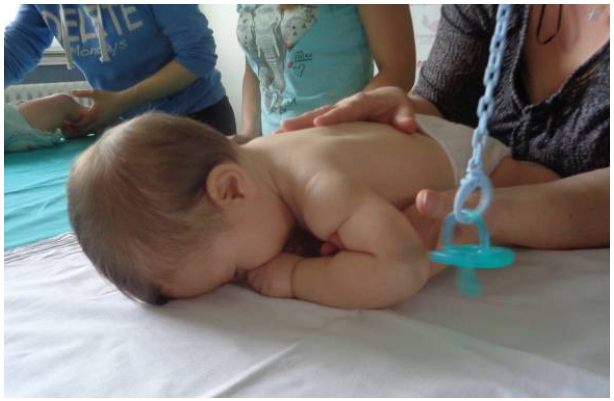




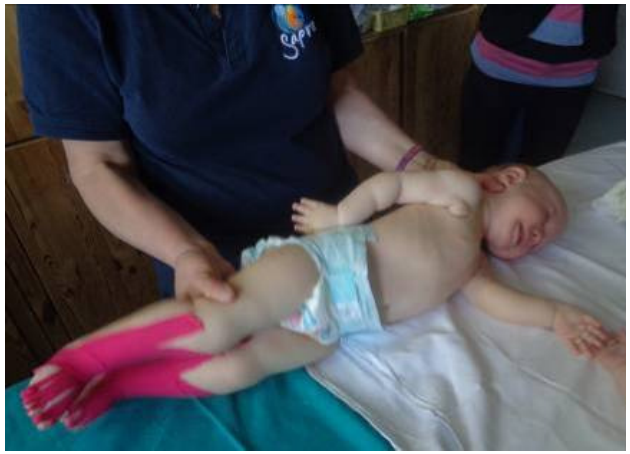
ROTOLAMENTO

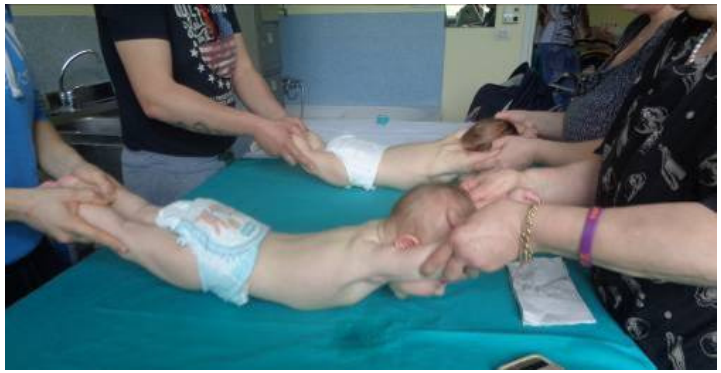




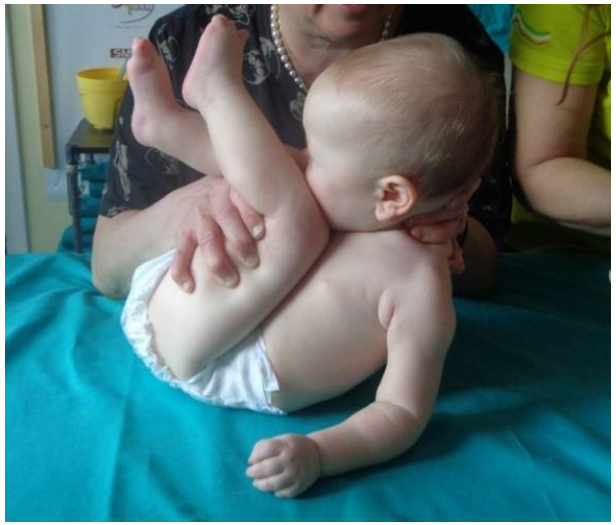


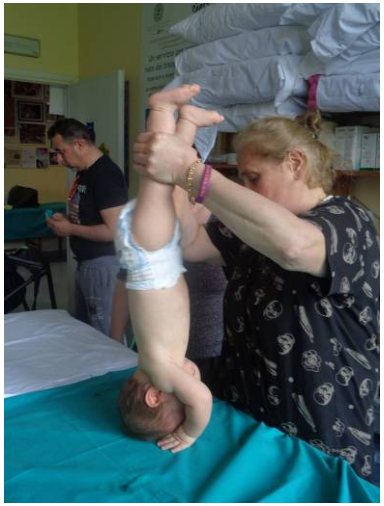

COLONNA



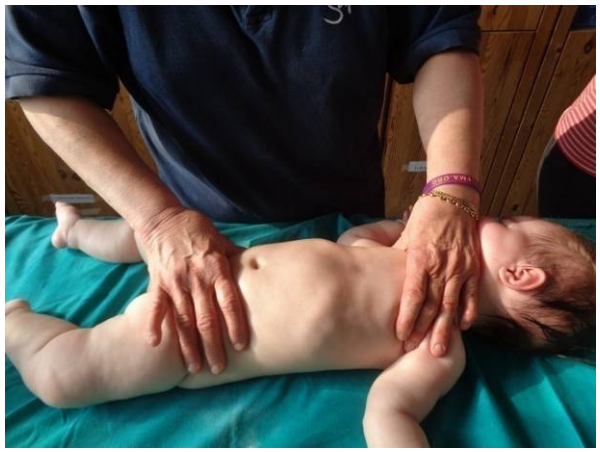
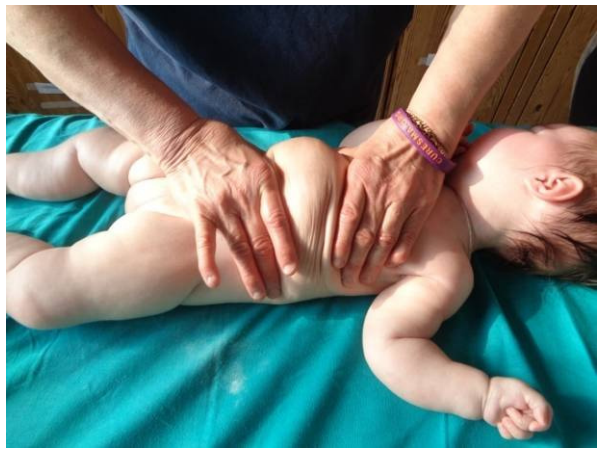
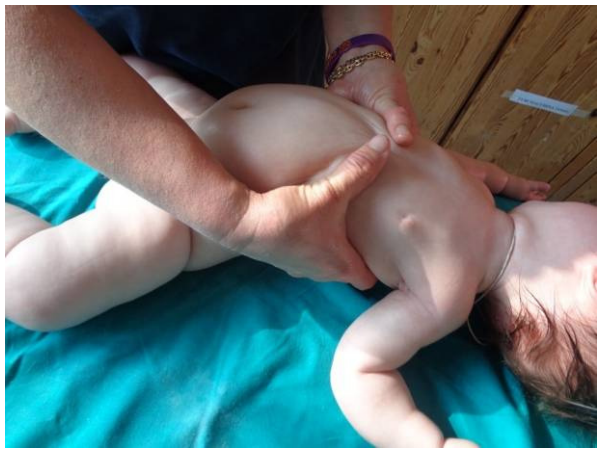


5. Physical Therapy and Rehabilitation

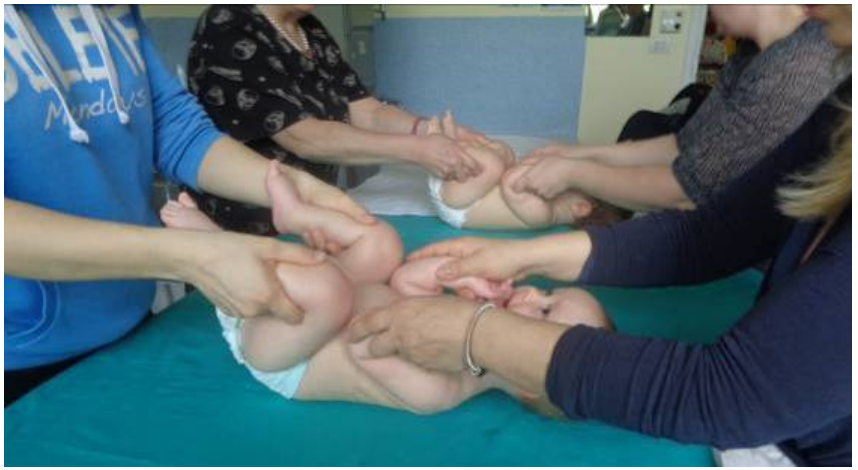


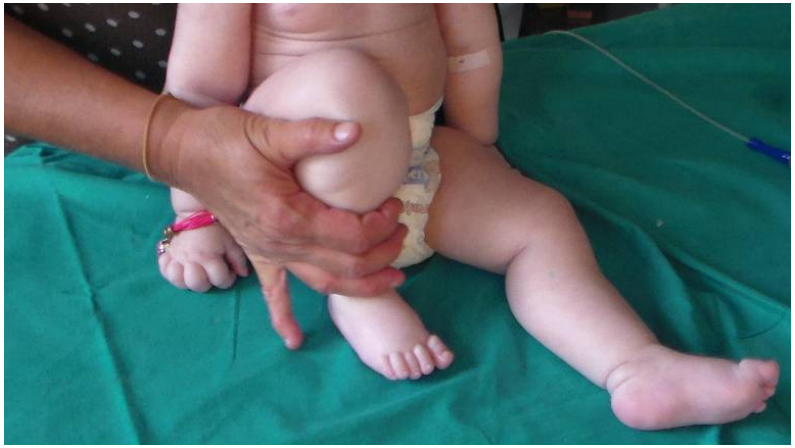


Manipolazione della cassa toracica

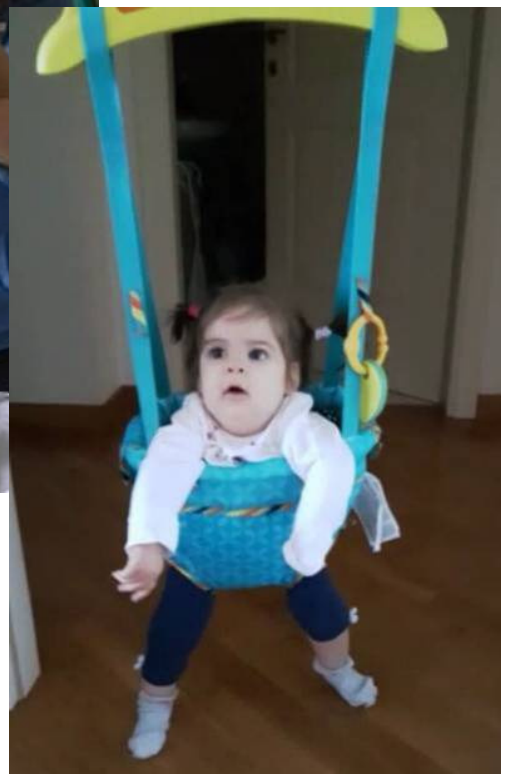

Arti inferiori
IN SCARICO





5. Physical Therapy and Rehabilitation

Arti inferiori
IN CARICO



5. Physical Therapy and Rehabilitation