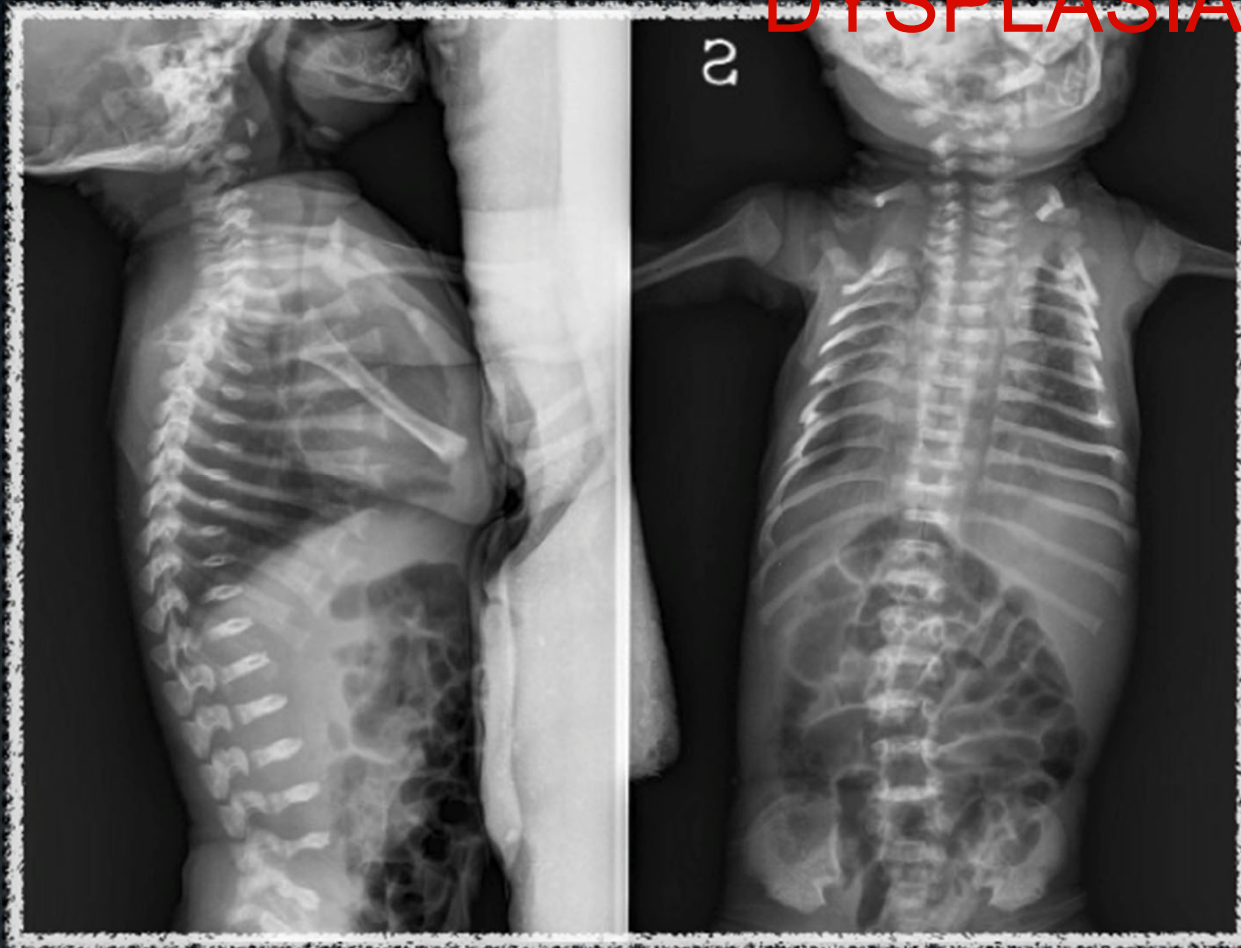


EARLY BRACE TREATMENT IN METATROPIC DYSPLASIA



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Introduction

- Metatropic Dysplasia is a rare disease in which as reported in literature Kyphoscoliosis mainly in lumbar and thoracolumbar area is progressively severe with poor results also with surgery correction. Thorax deformities, esophagites with refluxum, respiratory distress are often observed.
- Heterozygous missense mutations of transient receptor potential vanilloid 4 channel (TRPV4) cause a spectrum of skeletal disorders, including brachyolmia, spondylometaphyseal dysplasia Kozlowski type, metatropic dysplasia, parastremmatic dysplasia, and spondyloepimetaphyseal dysplasia Maroteaux type.
- The aim of the work is reporting on the result of brace treatment in five cases of Metatropic dysplasia with severe kyphoscoliosis treated from the age of six months up to four years.

Related works

- **TRPV4-associated skeletal dysplasias.** Nishimura G, Lausch E, Savarirayan R, Shiba M, Spranger J, Zabel B, Ikegawa S, Superti-Furga A, Unger S. Génétique Médicale, CHUV, Av. Decker 2, 1011 Lausanne, Switzerland. Am J Med Genet C Semin Med Genet. 2012 Aug 15;160C(3):190-204. doi: 10.1002/ajmg.c.31335. Epub 2012 Jul 12.
- **Metatropic dysplasia in four-year-old boy--physiotherapy and orthopaedic care problems--case report.** Kleszczyński J, Błoch M, Hofbauer K, Dominiak P. Division of Physiotherapy in Paediatrics and Neurology, Department of Physiotherapy in Musculoskeletal Dysfunctions, University of Physical Education in Wrocław. teofrastus@wp.pl Ortop Traumatol Rehabil. 2012 May-Jun;14(3):289-302.
- **TRPV4 related skeletal dysplasias: a phenotypic spectrum highlighted by clinical, radiographic, and molecular studies in 21 new families.** Andreucci E, Aftimos S, Alcausin M, Haan E, Hunter W, Kannu P, Kerr B, McGillivray G, McKinlay Gardner RJ, Patricelli MG, Sillence D, Thompson E, Zacharin M, Zankl A, Lamandé SR, Savarirayan R. Genetic Health Services Victoria and Murdoch Childrens Research Institute, Parkville, Victoria, Australia. elena.andreucci@unifi.it Orphanet J Rare Dis. 2011 Jun 9;6:37.
- **Metatropic dysplasia: clinical and radiographic findings in 11 patients demonstrating long-term natural history.** Kannu P, Aftimos S, Mayne V, Donnan L, Savarirayan R. Genetic Health Services Victoria, 10th Floor Royal Children's Hospital, Melbourne, Australia. Am J Med Genet A. 2007 Nov 1;143A(21):2512-22.

Methods

In the last three years we have followed four cases that arrived at our observation in the first months after births.

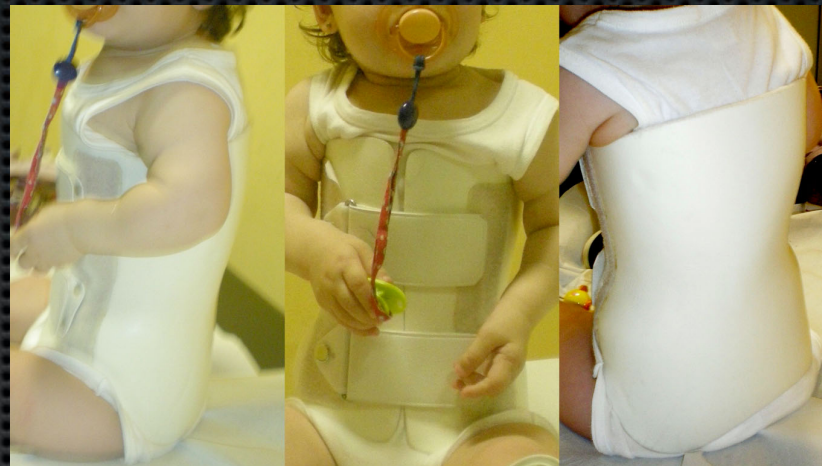
One of the three cases was extremely severe with triangular chest and a severe kyphoscoliosis.

The remaining thorax was less involved but the spinal deformity was more than 40 degrees at the first evaluation (about 4- 6 months).



Bracing

In all case we used a full time semisoft (Podialene ®) progressive brace correction modelled on traction (Tu-Ta Rizzoli ®) at the beginning with anterior opening then at eighteen months (mean age of walking 18 - 24 mths) with posterior opening. The brace had to be replaced every 6-8 months.



1st case

C.A

First evaluation 10 months

T12 - L4 50° SX

Lumbar Kyphosis: 55°

Full time treatment with Podialene Brace.

36 months

T12 - L3 42° SX

Lumbar Kyphosis: 27°

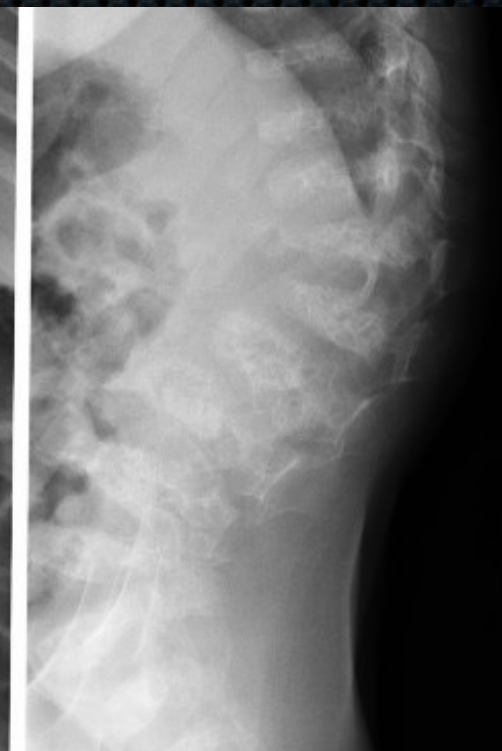
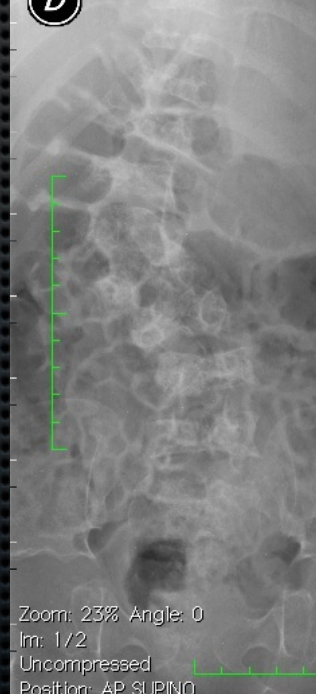
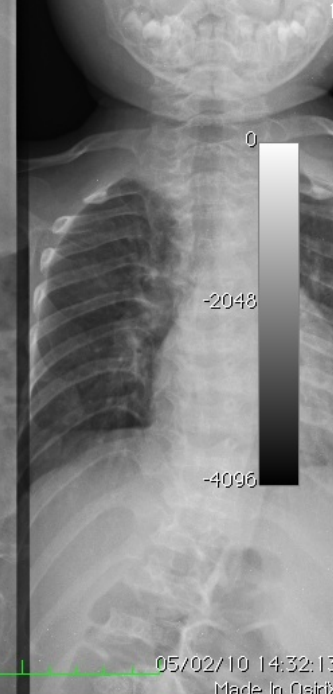


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View size: 478 x 584
W: -2048 WW: 4096



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Position: AP SUPINO

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RX RACHIDE LOIMBO-SACRALE — AP



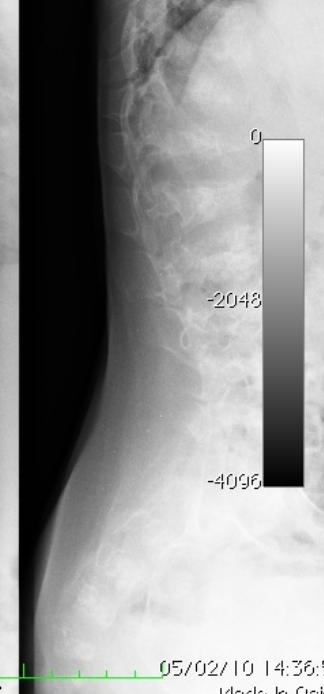
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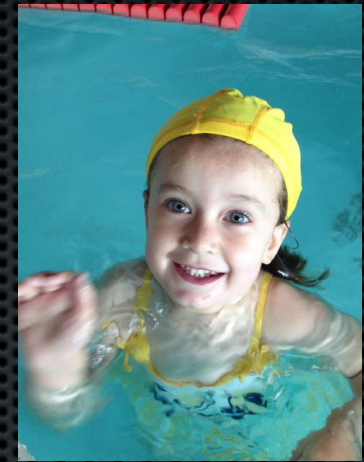
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Uncompressed
Position: LATERALE DECUBITUS

1328577 (1 y, 1 y)
RX RACHIDE DORSALE — LATERALE



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C.A.42 months



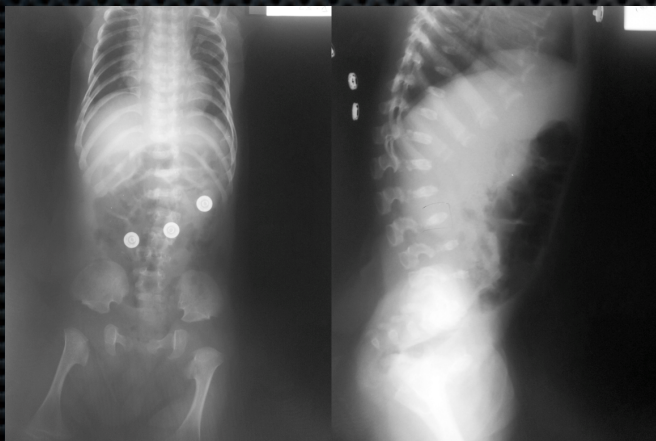
Doc, Is it
O.K.?

The girl is actually walking with good reduction in lumbar kyphosis and scoliosis, good alignment of knee and ankle, hip flexion but reduced hyperabduction and extrarotation.

2nd case T.K. Born October 2009



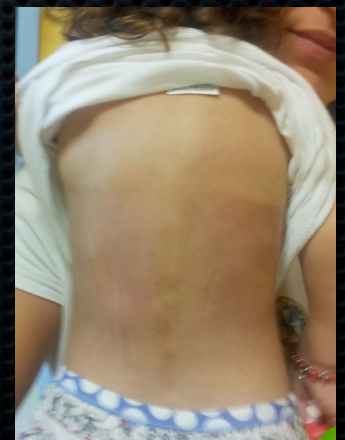
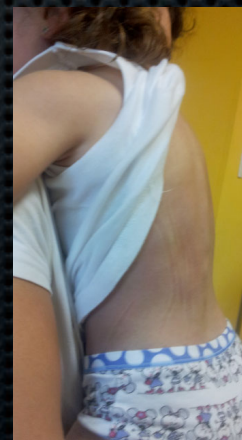
This girl arrived at first evaluation at 8th months. Bracing was well accepted by the baby and the family with a full time use (22 hrs/day). She actually have at 35 months a complete clinical correction of kyphosis and scoliosis.



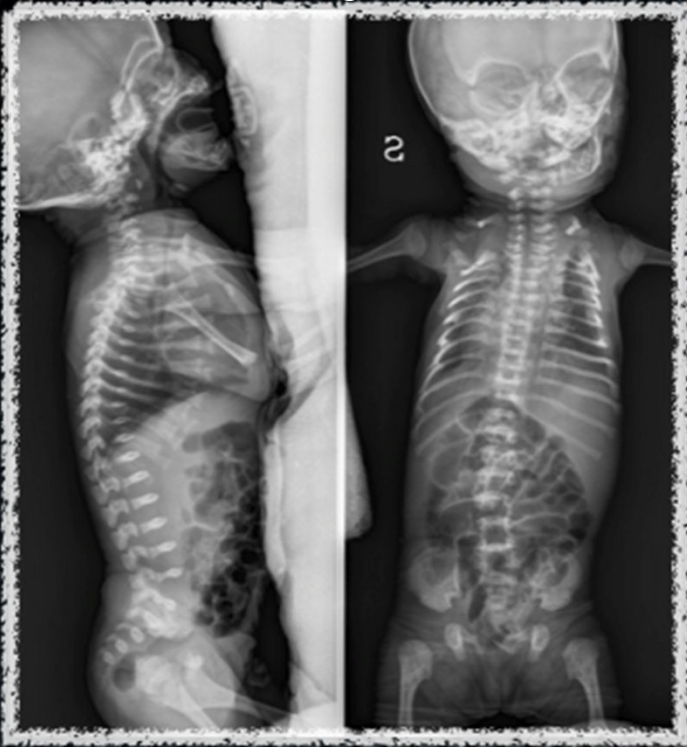
X-ray at 8 months



X-ray at 18 months



3rd case L.A. Born July 2009



9 months Thoracolumbar
Kyphosis with mild left
lumbar scoliosis

20 months: Thoracolumbar
kyphosis apex T10 50°
Lumbar Thoraco scoliosis
T10 L4 26° sx Podialene
Brace used 8 hours a day.

30 months: Clinical
evaluation: improvement of
Thoracolumbar Kyphosis -
Podialene Brace posterior
opening used 8 hours a day.



4th case B.N. born March 2010.



This little boy, born in Spain arrived at our observation by means of Italian Association for M.D. on F.B. The first brace was built in Italy on the guide of calibrated photos: after two years of treatment bracing was discontinued waiting for surgery decision.



Results

In two cases we obtained normalization on the lateral plane and reduction of scoliosis from 40° to 20°. In one case despite bracing we had a progression of the thoracolumbar kyphosis from 30° to 50° and an actual improvement to 40° when changing the opening of the brace from anterior to posterior. In the severe form bracing has been used only few hours a day and was ineffective on the progression but however had a positive respiratory response increasing diafragmatic breathing.

Conclusion

Use of a full time semisoft brace in metatropic dysplasia seems effective in reducing progression of kyphoscoliosis, improving and not reducing respiratory function.

All children and family agree on photo display

Doctor can you help me to solve the problem?

