ISSN (P) : 2589-9120 / (O) : 2589-9139
PubMed-National Library of Medicine - ID: 101773527

International Journal of Medical Science and Applied Research (IJMSAR)

Available Online at: https://www.ijmsar.com

Volume – 4, Issue –3, May – 2021, Page No. : 19 – 25

Spondylocostal Dysostosis or Jarcho Levin Syndrome, A Rare Cause of Respiratory Distress in Infants: A Case Report

[1]Dr. Karima Larbi Ouassou, Department of pediatrics- Mohammed V Military Instruction Hospital – Rabat, Morocco

[2]Dr. Amal Hassani, Department of pediatrics- Mohammed V Military Instruction Hospital – Rabat, Morocco

[3]Dr. Anas Ayad, Department of pediatrics- Mohammed V Military Instruction Hospital – Rabat, Morocco

[4]Dr. Abdelilah Radi, Department of pediatrics- Mohammed V Military Instruction Hospital – Rabat, Morocco

[5]Dr. Rachid Abilkassem, Department of pediatrics- Mohammed V Military Instruction Hospital – Rabat, Morocco

[6]Dr. Aomar Agadr, Department of pediatrics- Mohammed V Military Instruction Hospital – Rabat, Morocco

Citation of this Article: Dr. Karima Larbi Ouassou, Dr. Amal Hassani, Dr. Anas Ayad, Dr. Abdelilah Radi, Dr. Rachid Abilkassem, Dr. Aomar Agadr," Spondylocostal Dysostosis or Jarcho Levin Syndrome, A Rare Cause of Respiratory Distress in Infants: A Case Report", IJMSAR – May – 2021, Vol. – 4, Issue - 3, P. No.19-25.

Corresponding Author: Dr. Karima Larbi Ouassou, Department of pediatrics- Mohammed V Military Instruction Hospital – Rabat, Morocco

Type of Publication: A Case Report

Conflicts of Interest: Nil

Abstract

Autosomal recessive spondylocostal dysostosis is a rare condition, which can manifest as simple scoliosis, or in more severe cases as life-threatening respiratory complications, as described in this work.

Keywords

Spondylocostal, dysplasia, Jarcho-Levin, costo-vertebral, respiratory distress.

Introduction

Spondylocostal dysplasia or Jarcho-Levin syndrome is a rare autosomal recessive disease characterized by severe abnormalities of the spine and ribs, involving the respiratory prognosis, which may be associated with non - skeletal manifestations.

We report in this work the case of a patient from Morocco, diagnosed in the pediatric department of the military hospital in Rabat.

Case report and results

Amira, 18 month old infant, female, 2nd in a sibling of 2 with a 7 years old brother followed for unexplored scoliosis, from a 3rd degree consanguineous marriage. The pregnancy was well monitored and carried to term, the delivery was via the high route for borderline pelvis, with the notion of neonatal respiratory distress for which she was intubated

ventilated for 11 days with good progress. The patient presented two episodes of respiratory discomfort at 10 and 17 months old respectively, managed by nebulization of ventoline and by oral corticosteroids. At 18 months old, the infant is admitted to our training for the management of afebrile respiratory distress. On admission, the infant presented a dysmorphic face (bulging forehead, flattened base of the nose)(Figure 1). She was also pale, afebrile, polypneic with perioral cyanosis, signs of respiratory struggle, a thorax deformed into a keel, snoring rales and discrete bilateral sibilations. The osteo-articular examination revealed the presence of dorso-lumbar scoliosis. A delay in psychomotor acquisitions was also noted (standing position possible with assistance, walking impossible without support). the rest of the physical examination was normal.

A chest X-ray revealed a globular thorax, a right alveolar syndrome with heterogeneous opacities (Figure 2); Thoracic CT revealed ventilation disorders with alveolar foci, complex costo-vertebral malformations in stages with intrathoracic ascent of the 2 kidneys and of segment IV of the liver (renal ectopia)(Figure 3) . Cytomegalovirus serology as well as cytobacterilogic study of sputum came back negative. In addition, a cerebral MRI revealed an aspect of leukodystrophy with damage to the corticospinal tracts, with white matter abnormalities (Figure 4). The metabolic balance returned to normal; the diagnosis of autosomal recessive spondylocostal dysostosis was made based on all clinical and radiological data. The genetic confirmation of the diagnosis by the sequencing of the exom was planned but not made for lack of means.

From a therapeutic standpoint, the patient was put on oxygen therapy, nebulization of β2 mimetics, IV corticosteroid therapy and antibiotic therapy (C3G and Aminoglycosides); the course was marked by regression of respiratory discomfort with dependence on oxygen. The patient died a month later at home from respiratory complications.

Discussion

Jarcho Levin syndrome refers to the set of clinical abnormalities first described in two patients published in 1938 by Saul Jarcho and Paul Levin at Johns Hopkins Hospital in Baltimore, who presented with respiratory failure secondary to abnormalities costo-vertebral (1). Over time this pathology was named Jacho-Levin syndrome, and is used to describe several congenital costo-vertebral anomalies. (2)

At birth this syndrome is suspected due to a small size, a short neck and trunk (2,3), with low implantation of the scalp (4, 5), with an aspect of the "symmetrical crab thorax" (6)

Radiologically, rib abnormalities are observed, including irregularity in size and shape, absence of one or more ribs, or posterior fusion ribs. Vertebral malformations are described as a type of vertebrae fused together as a block or poorly formed into tiered hemi-vertebrae forming a puzzle-like appearance, most often associated with scoliosis (2,3,7). Our patient indeed presents all of these malformations as described on her standard x-ray.

For some patients, other skeletal abnormalities have been described, such as a winged scapula, a malformation of the odontoid process, an absence of atlas, an irregular clavicle, a hypoplastic humerus, or enlarged iliac wings (3).

In addition, apart from costo-vertebral anomalies, other extra-skeletal anomalies can be observed, in particular urogenital malformations (horseshoe kidneys, renal agenesis and hypoplasia,

bladder and genital duplication) (6), spina bifida, anal malformations (atresia of the anal canal, imperforation of a ventral anus), inguinal hernias, Meckel's diverticulum and polydactyly (8,9). For our patient, we observed the presence of leukodystrophy with cortico-spinal bundles, with white matter abnormalities, as well as bilateral renal ectopia and intra-thoracic segment IV of the liver.

In Jarcho-Levin syndrome two phenotypes have been described: Spondylocostal dysplasia (SCD) associating costal and vertebral numerical and structural abnormalities, and spondylothoracic dysplasia (STD) also called Lavy-Moseley syndrome, which is not characterized only by the deformation of the rib cage (10, 11)(Table 1).

Table 1: Clinical and genetic findings that distinguish spondylocostal dysostosis (SCD) from spondylothoracic dysostosis (STD)

	SCD	STD
Initial description	Jarcho and Levin (1938)1	Lavy, Palmer, Merritt (1966)2
Location of 1st report	Baltimore	Indianapolis
Ethnicity	colored", name Anglo-Saxon	Puerto Rico
Rib anomalies	Yes; overgrown, fused, missing	Yes; fused posteriorly
Spine anomalies	Yes; fusion, hemi, and block vertebrae	Yes; Cervciothoracic fusion, fusion of occiput and C1
Distribution	Worldwide	Over 50% from Puerto Rico
Clinical course	Variable thoracic insufficiency; most survive	Severe thoracic insufficiency; high infant mortality, 25% or more survive to adulthood
Associated genes	DLL3	MESP2
Surgical treatment	Expansion thoracoplasty possible	Expansion thoracoplasty not commonly used

Spondylocostal dysostosis is the phenotype described for our patient, following an autosomal recessive mode of inheritance since her parents did not present any apparent malformation. In some families, autosomal dominant inheritance has nevertheless been described (5).

Several genes involved in the Notch signaling pathway have been implicated in SCD, including a mutation in one of the 4 genes - DLL3, MESP2, LFNG and HES7 (12) located on chromosome 19p13 (11,13). were associated with a significant reduction in the levels of the Pax1 and Pax9 proteins expressed in the chondrocytes of the spine (3,14); So genetic tests such as Exom sequencing can make it possible to identify these mutations for affected patients and their families. (Table 1 article1)

Due to the asymmetry of the thoracic abnormalities described in STD impeding lung capacity, SCD remains a better evolutionary prognosis (15). However, in children with SCD, exaggeration of scoliosis with bone growth may worsen the restrictive syndrome requiring urgent orthopedic surgical management (16).

However, the majority of children with Jarcho-Levin syndrome die at an early age, following

respiratory infections in a field of respiratory failure. (7).This was the fate of our patient, who had died at home from infectious respiratory distress despite ventilatory assistance.

Conclusion

Jarcho Levin syndrome remains a rare cause of respiratory distress in newborns and infants; Due to the importance of costo-vertebral malformations and the intensity of thoraco-lumbar scoliosis, the majority of patients with this pathology die at an early age despite the advances made in respiratory resuscitation and orthopedic surgery (7).

This is a rare pathology, of course, but which can be diagnosed early by antenatal screening which would be possible from the 12th week of gestation by a morphological ultrasound (17).

References

1. Walter E. et .al. Clinical and radiological distinction between spondylothoracic dysostosis (Lavy-Moseley syndrome) and spondylocostal dysostosis (Jarcho-Levin syndrome) , Published online: 22 December 2010
2. Chandra N, Kumar S, Raj V, Vishwakarma PK, Sinha S, Saha RP; Jarcho-Levin syndrome with splenic herniation: a rare presentation. - Am J Case Rep. 2016;17:745–8.
3. Heilbronner DM, VA C, Renshaw TS. Spondylothoracic dysplasia .A case report. J Bone Joint Surg 1984;66A:302–3.
4. Campbell RM Jr (2009) Spine deformities in rare congenital syndromes. Spine 34:1815–1827
5. Turnpenny PD, Alman B, Cornier AS et al (2007) Abnormal vertebral segmentation and the notch signaling pathway in man. Dev Dyn 236:1456–1474
6. Kazuharu Takikawa et.al. Spine and Rib Abnormalities and Stature in Spondylocostal Dysostosis SPINE Volume 31, Number 7, pp E192–E197 ©2006
7. Karnes PS, Day D, Barry SA, et al. Jarcho-Levin syndrome: Four new cases and classification of subtypes. J Med Genet 1991;40:264–70.
8. Martinez-Frias ML, Bermejo E, Paisàn L, et al. Severe spondylocostal dysostosis associated with other congenital anomalies. A clinical/epidemiologic analysis and description of ten cases from the Spanish registry. J Med Genet 1994;51:203–12.
9. Rimoin DL, Fletcher BD, McKusick VA. Spondylocostal dysplasia: A dominantly inherited form of short-trunked dwarfism. J Med Genet 1968;45:948–53
10. Roberts AP, Conner AN, Tolmie JL, et al. Spondylothoracic and spondylocostal dysostosis. J Bone Joint Surg Br 1988;70:123–6.
11. Solomon L, Jimenez B, Rainer L. Spondylothoracic dysostosis. Arch Pathol Lab Med 1978;102:201–5.
12. Beine O, Bolland J, Verloes A, Lebrun FR, Khamis J, Muller C. Spondylocostal dysostosis: a rare genetic disease. Rev Med Liege. 2004;59:513–6.
13. Aburakawa K, Harada M, Otake S. Clinical evaluations of the treatment of congenital scoliosis. Orthop Surg & Trauma 1996;39:55–62
14. McMaster MJ. Congenital scoliosis. In: Westein SL, ed. The Pediatric Spine. New York, NY: Raven Press; 1994:227–44.
15. Cornier AS, Ramirez N, Arroyo S et al (2004) Phenotype characterization and natural history of spondylothoracic dysplasia syndrome: a series of 27 new cases. Am J Med Genet A 128A:120–126

16. Mortier GR, Lachman RS, Bocian M, et al. Multiple vertebral segmentation defects: Analysis of 26 new patients and review of the literature. J Med Genet 1996;61:310–9.

17. Farley FA, Loder RT, Nolan BT, et al. Mouse model for thoracic congenital scoliosis. J Pediatr Orthop 2001;21:537–40

Figures

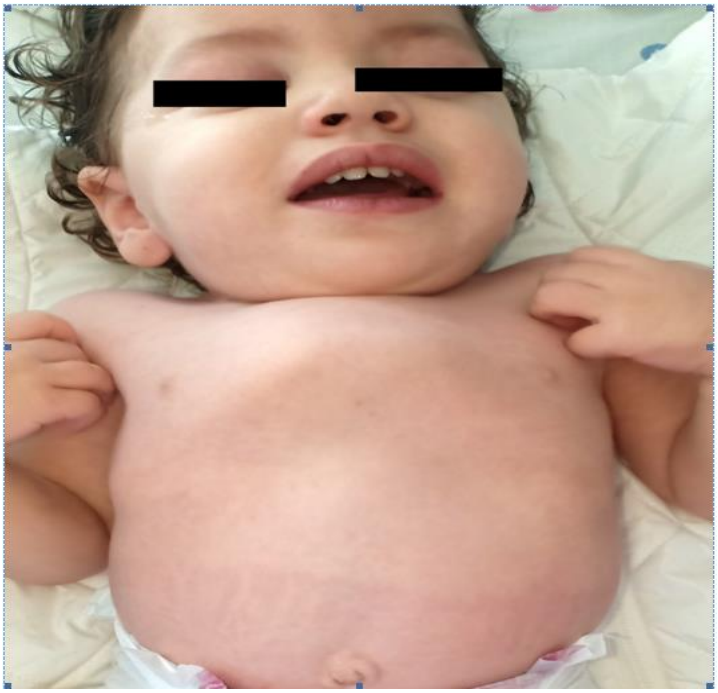
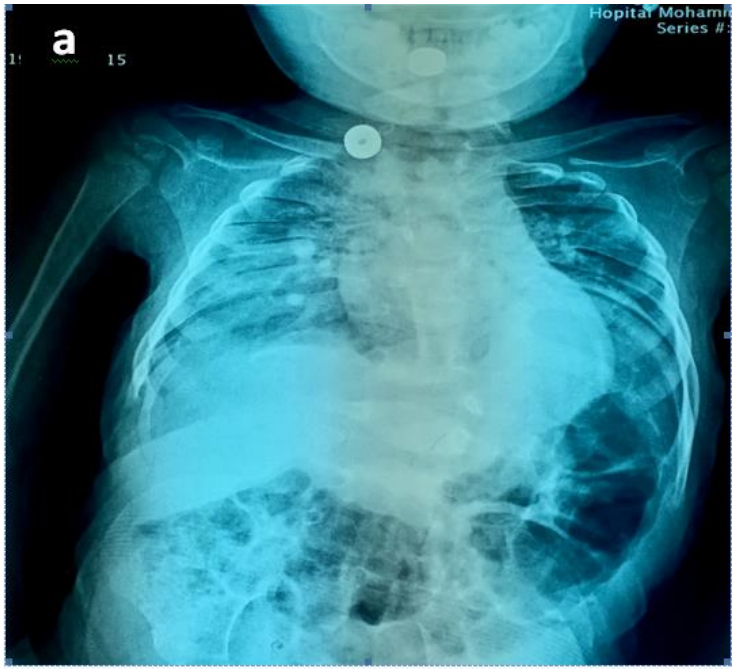


Figure 1: Photo taken in the supine position of our patient showing the keeled thorax with dysmorphic syndrome



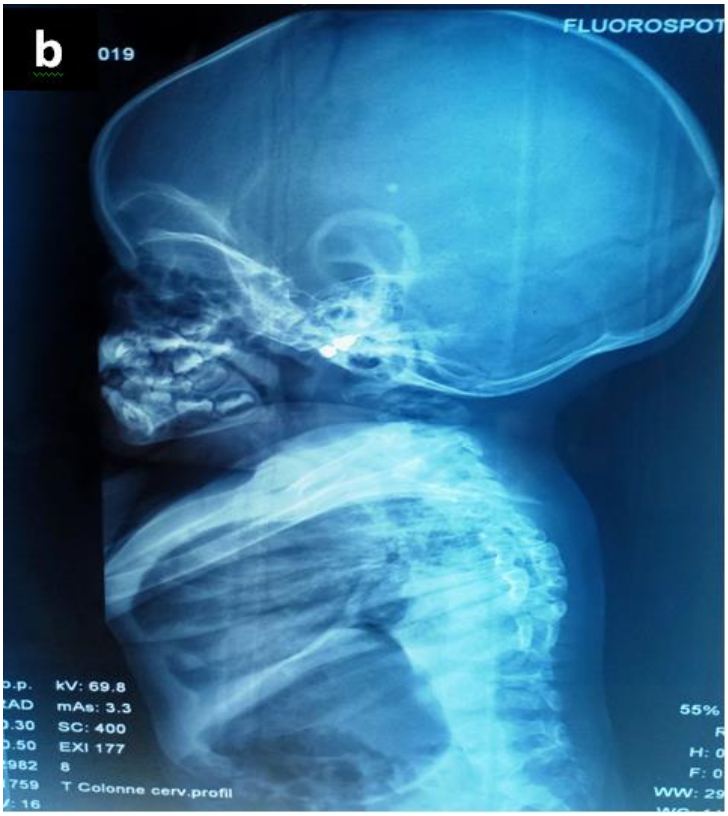


Figure 2: chest x-ray face (a) and profile (b) of our patient showing the keeled thorax and costo-vertebral anomalies

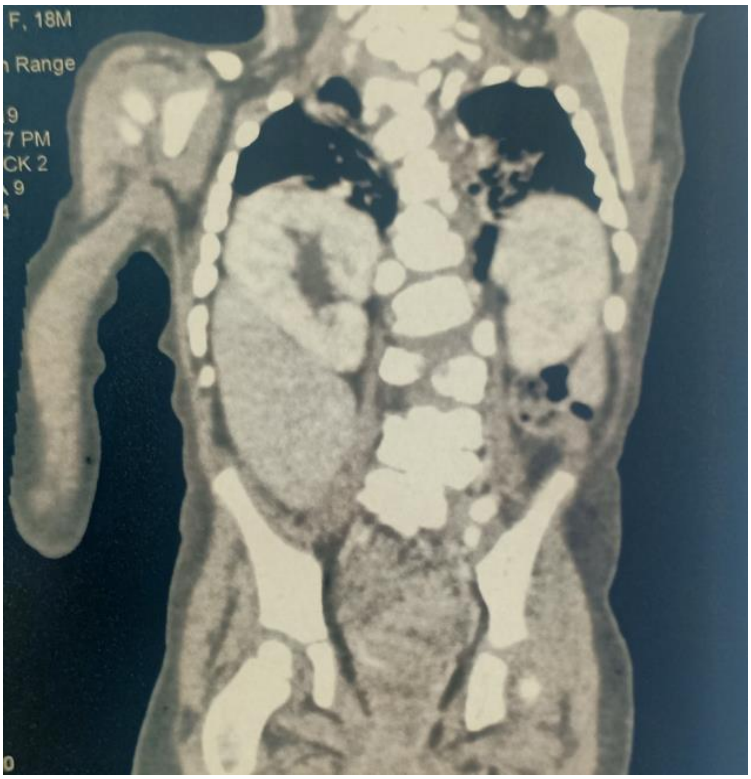


Figure 3: Thoraco-abdominal CT scan of our patient in frontal section showing the various costo-vertebral anomalies, as well as renal and hepatic ectopy