Prenatal diagnosis of Jarcho-Levin syndrome

Diagnosticul prenatal al sindromului Jarcho-Levin

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Abstract

Jarcho-Levin syndrome includes a range of anomalies of the spine and ribs resulting in a short thorax and is frequently referred to in the literature as spondylocostal or spondylothoracic dysplasia. Our case was diagnosed prenatally using ultrasound and confirmed postnatally by the clinical evolution and the imagistic data (X-ray, magnetic resonance and computer tomography). Due to the rarity of this condition, there are a limited number of reports in the scientific literature and very few are diagnosed prenatal.
Keywords: *prenatal diagnosis, spondylocostal dysplasia, Jarcho-Levin syndrome, abnormal vertebrae*

Rezumat

Sindromul Jarcho-Levin include un grup de anomalii ale coloanei și coastelor ce duc la un torace scurt și este frecvent descris ca displazie spondilocostală sau spondilotoracică. Cazul nostru a fost diagnosticat ecografic prenatal și confirmat postnatal de evoluția clinică și de rezultatele explorărilor imagistice (radiografie, tomografie computerizată, rezonanță magnetică nucleară). Datorită raritățiii acestei patologii, există un număr limitat de raportări în literatura de specialitate și foarte puține cazuri sunt diagnosticate prenatal.
Cuvinte-cheie: *diagnostic prenatal, displazie spondilocostală, sindrom Jarcho-Levin, anomalie vertebrală*

In 1938, Saul Jarcho and Paul Levin described a syndrome that grouped severe anomalies of the spine and anomalies of the ribs resulting in a short thorax[(1)]. Children with severe forms had frequent pulmonary complications (generated by the thoracic restriction) and had a high mortality in the first years of life. What clinicians presently include in Jarcho-Levin syndrome encompasses a range of diagnoses from spondylocostal dysplasia, spondylothoracic dysplasia to bizarre vertebral anomalies and multiple hemivertaebre[(2,3)]. The disease is inheritable with both autosomal recessive pattern and autosomal dominant pattern. Due to the rarity of this condition very few cases are described prenatally, and from our knowledge no prenatal diagnosis has been reported in our country.

A patient of 23 years old with no prior obstetrical history presented in our department at 20 weeks for the sonographic anomaly screening. We noticed an abnormal shape of the spine, with abnormal vertebrae and an abnormal position of the spinal cord in the medullar canal. We did not notice any ribs anomaly and the cardio-thoracic ratio was normal. No additional anomaly except a single umbilical artery was noticed. Our diagnosis at this moment was: hemivertaebrae; suspicion of spina bifida oculta. We recommended kariotyping, but the patient refused the procedure for personal reasons. The patient came back at 30 weeks and the ultrasound revealed the same aspect of the spine we noticed at 20 weeks. Though no obvious abnormally shaped ribs were seen a shorter and slightly narrower thorax was noticed. After a careful review of existing literature we decided it was a form of Jarcho-Levin syndrome. Family was informed of the possibility that the child might have a rare disease with potential inherited component; hence it was important to know if any similar cases were ever recorded in the family history - an attentive anamnesis was conducted but none came up. Our recommendations were delivery in a tertiary center, MRI and genetic counseling postnatal.

The baby was extracted by C-section at 39 weeks for an obstetrical indication unrelated to the baby's condition. It was small for gestational age (2460 g), a condition frequently associated with single umbilical artery. The postnatal X-ray confirmed our suspicion that it was a form of spondylocostal dysplasia. In spite of the thoracic restriction, baby adapted well and was discharged after 6 days. Patient was referred to a radiology center where at the age of 13 months both CT and MRI were performed.They showed sever disorder of the vertebrae and ribs and abnormal position of the spinal cord in the medullar canal. The final diagnosis was spondylocostal dysplasia (Jarcho-Levin syndrome).

Discussion

Jarcho-Levin syndrome includes a large spectrum of spondylothoracic and spondylocostal dysplasias. Though the true frequency is no really known, about 400 cases have been reported in the literature[(4)]. The groups' characteristics include anomalies of the vertebrae and ribs - the anomalies of the vertebrae frequently include hemivertebrae and fusion of the vertebrae generating scoliosis and/or kyphosis aspect that we met in our case as well. The spine and ribs anomaly generate a thorax with limited expansion and have frequent respiratory problems[(5,6,7)]. With postnatal growth, the need for a larger respiratory volume leads to a protrusion of the diaphragm towards the abdominal cavity and consecutively an increased abdominal pressure. As a result of this, patients frequently develop inguinal hernia - our patient also deve-

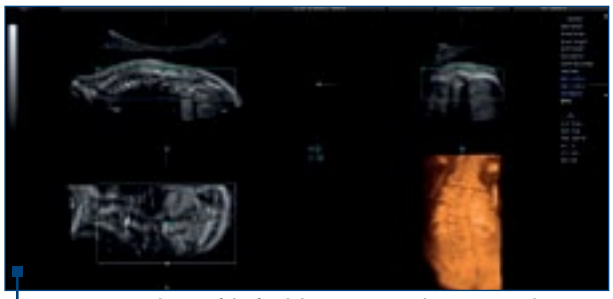

Figure 1. 3D rendering of the fetal thorax at 20 weeks (gestational age) - notice the abnormal shape of the spine, abnormal vertebrae and the abnormal position of the spinal cord in the medullar canal

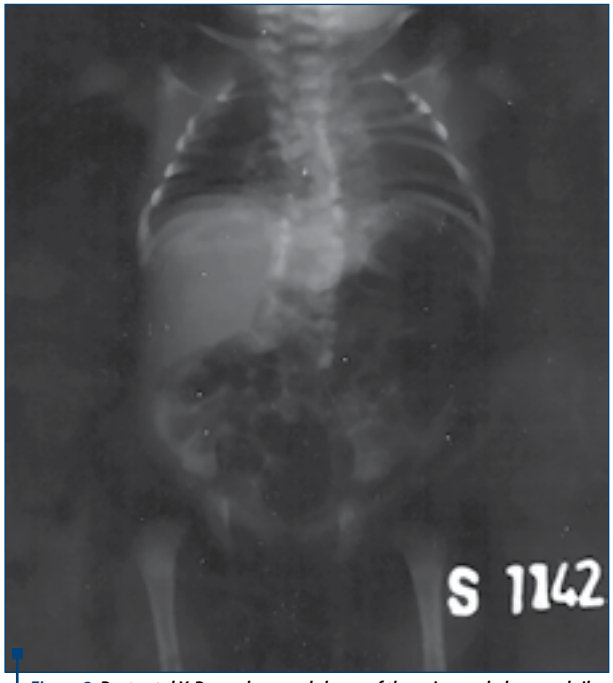

Figure 2. Postnatal X-Ray - abnormal shape of the spine and abnormal rib cage (small thorax)

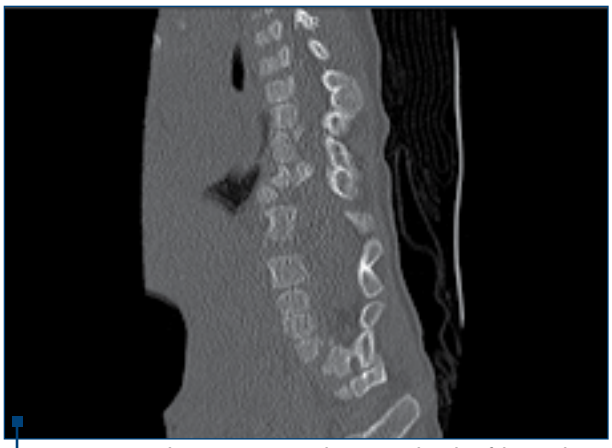

Figure 3. Postnatal CT scan (at 13 months) - severe disorder of the vertebrae

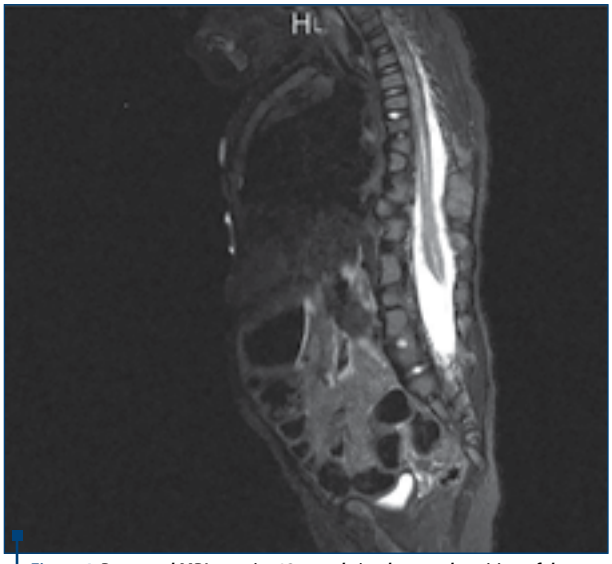

Figure 4. Postnatal MRI scan (at 13 months) - abnormal position of the spinal cord in the medullar canal

loped inguinal hernia that was operated on at nine months postnatal. To this moment four genes involved in spondylocostal dysplasias were discovered: DLL3, MESP2, LFNG and HES7 - they explain 25% of the diagnosed cases[8,9]. The mechanism described is the alteration of the Notch signaling pathway which takes part in the somite segmentation[10]. The result is malformation and fusion of the vertebrae and ribs as described by Jarcho and Levin. Prenatal diagnosis helps in counseling the parents and referring the case to tertiary centers for delivery. A special mention is deserved for the use of 3D ultrasound both for the diagnosis (as it offers a multi-angle approach of the pathology) and for the patient who finds the images more suggestive. ■

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