CASE REPORT

SPONDYLOCOSTAL DYSPLASIA: A RADIOLOGICAL INSIGHT INTO A RARE SKELETAL ANOMALY

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ABSTRACT

Spondylocostal dysplasia is a rare genetic disease. It is also known as Jarcho-Levin syndrome. It is characterized by defects in vertebrae and abnormalities of ribs. It is inherited in both autosomal dominant and recessive pattern but the latter one is more common. It is of two types: Spondylocostal dysplasia type I (SCD- I) and spondylocostal dysplasia type II (SCD- II). The severity of disease varies among affected one. Though itis considered as less severe but respiratory compromise may pose severe threat. The diagnosis is based on clinical and radiological findings. With treatment, affected individual may survive to adult hood.

Key words: Jarcho-Levin syndrome, Spondylocostal dysplasia (SCD) and spondylothoracicdysplasia (STD), radiology, syndrome.

Introduction

Spondylocostal dysplasia is first described by Jarcho and Levin in 1938. The other synonyms are costo-vertebral dysplasia and Jarcho Levin syndrome. The latter name is obsolete now. It is a rare congenital anomaly of thoracic cage and spine.[1] The purpose of this case is to highlight the clinical and radiographic presentation.

Case Presentation

We present a case of eight years old female child with presenting complaint of short height, protuberant abdomen and recurrent chest infection. She is a product of consanguineous marriage. She born normal via spontaneous vaginal delivery. Antenatal sonogram was unremarkable. On clinical examination, she had short trunk dwarfism with normal limb span. She was intellectually normal. But her athletic activity was reduced. No neurological deficit was noted. She had three other siblings, two of them are male and entirely normal and one sister who had similar appearance like our patient. She also underwent ultrasound abdomen examination that was grossly normal. For curvature abnormality she underwent skeletal survey.

Discussion

Spondylocostal dysplasia is a rare congenital malformation. It follows two pattern of inheritance; autosomal dominant and autosomal recessive. Spondylocostal dysplasia is of two types; SCD type 1 and SCD type 2. The former one SCD type 1 is severe and it is diagnosed in utero. It follows autosomal recessive pattern. The latter one, SCD type 2 is milder form,

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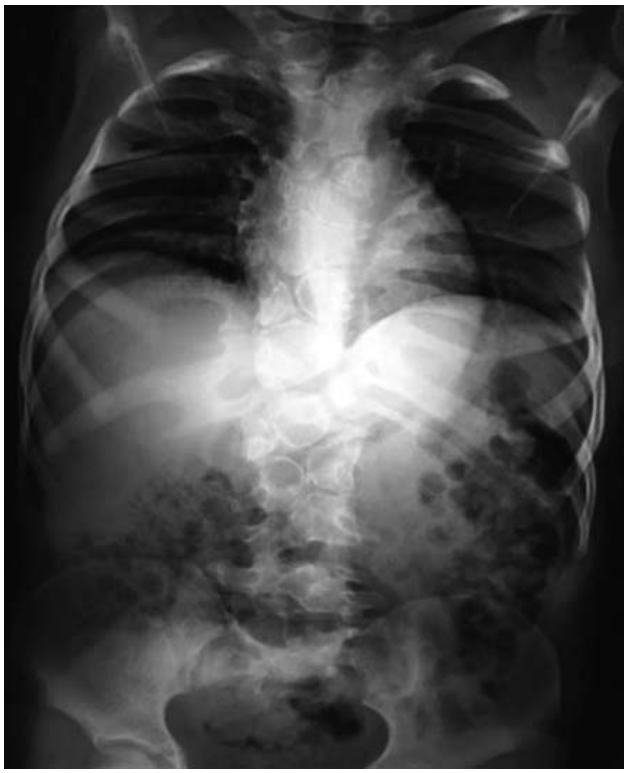

Figure 1: Radiograph of chest frontal projection demonstrates 9 ribs on right side and 10 ribs on left side. Note the rib synostosis on either side as well.
Failure of formation vertebral anomaly including hemi-vertebra and butterfly vertebrae involving dorsal spine are also noted.

and cannot be diagnosed in utero. This type follows autosomal dominant pattern.[2]

Spondylocostal dysplasia is caused by the mutation in one of the five gene: DLL3, MESP2, LFNG, HES7 and TBX6 gene. The exact prevalence of this disease is unknown; however, its reported incidence in one of the article is 1 per 40,000 births.[3] It affects both gender with female preponderance.[4]

Affected individual has abnormality in the spine and ribs. These patients usually have webbed neck, short height due to disproportionate shorter trunk, curvature abnormality of spine but normal limb size. In some individual thoracic insufficiency syndrome may occur due to mal developed rib cage.

Affected individual may show segmental or failure of formation vertebral anomaly. Vertebrae may be absent. Spine may show curvature abnormality/scoliosis as well. The ribs may be missing, bifid, malaligned, broadened, fused or may give fan-like or crab-like appearance of rib cage due to posterior fusion and flaring of anterior ends.[3-4]

It is a clinico-radiological diagnosis based upon characteristic symptoms, detailed patient and family history, clinical evaluation and specialized test/ X-ray spine that shows characteristic abnormality of spine and ribs. It can be confirmed by molecular testing which is only available at diagnostic service at specialized laboratories. Prenatal diagnosis may be possible by fetal ultrasound using high frequency probe which may detect associated anomalies of spondylocostal dysplasia.[5]

Kulkarni et al demonstrated that in their case associated anomaly of neural tube defect and genitourinary involvement.[6] Similarly, Banu Dane et all demonstrated association of neural tube defect as prominent finding with spondylocostal dysostosis.[7] Park WH et all showed another association of thoraco-abdominal wall hernia and imperforate anus.[8] Hatakeyama K, bring into light an association of heart disease and heterotaxy syndrome with this dysostosis and patent ductus arteriosus.[9-10] In one of the study,[11] splenic herniation was also found along with diastematomyelia in such cases. Diaphragmatic hernia has also been reported in literature.[12]

In our case, there is crab or fan like abnormality of rib cage demonstrating rib fusion and absent ribs as well. Dorsal spine showed only failure of formation vertebral anomaly. No such above mention associated gastro intestinal, genito-urinary, abdomino-thoracic wall hernia or neural tube defect like diastematomyelia or myelomeningocele were found.

There are numerous disorder that involve spine and ribs just like spondylocostal dysplasia such as Alagille syndrome, Klippel-Feil syndrome and VACTERL syndrome.

Treatment is directed towards specific symptoms and indulges the role of cardiologist, orthopedics, surgeon and pulmonologist. Such patient s entire family should be recommended for genetic counselling and psychological support as well.[12]

Such cases may undergo complication including recurrent chest infection like in our case, breathing difficulties sometime it may be severe enough to become a fatal condition, pulmonary hypertension. Neurological complications are rare. Intelligence remains unaffected.

Though the prognosis of this spondylocostal dysostosis is poor but not lethal if prompt diagnostic methods are used for early detection of pulmonary complication

like recurrent pneumonia, pulmonary hypertension and congestive heart failure.[13]

Conclusion

Any patient with short trunk dwarfism should raise the high index of suspicion for spondylocostal dysplasia. It is clinic-radiological diagnosis. Antenatal ultrasound may also diagnose the case and depict associated other system anomaly. Management should be directed towards the specific symptoms with specific attention to the genetic counselling and psychological support of family.

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