

Case Report

JARCHO-LEVIN SYNDROME

A. K. Manicka Vasuki.

Assistant professor, Department of anatomy, PSG Institute of Medical Sciences and Research, Coimbatore, Tamil Nadu, India.

ABSTRACT

Jarcho-Levin syndrome or Spondylocostal Dysostosis is characterized by Short neck and stature, vertebra-costal segmentation defects, pulmonary hypoplasia, congenital diaphragmatic hernia and renal agenesis. During dissection of 23 weeks old female fetus in the department of Anatomy, we observed Short neck, pulmonary hypoplasia, left diaphragmatic hernia with right renal agenesis. X-Ray showed hemivertebra in the lower two thoracic vertebra. This is an autosomal recessive disorder and hence the risk of such anomalies in the subsequent pregnancy is also increased. Identification of chromosomal abnormality and genetic counseling is necessary to avoid recurrence of the condition and its complications in future pregnancies.

Congenital diaphragmatic hernias occur in 1/2000 to 1/4000 births. They are relatively common with a majority of them belongs to Bochdalek variety, hernia occurring posterolaterally thro' foramen of Bochdalek. This is more frequent on the left side (85 – 90% of cases). In the newborn period, it may present with severe respiratory distress. Eventhough it occur as a isolated entity, it is important to look for other congenital malformations like cardiac anomalies, lung agenesis or hypoplasia, renal agenesis, hydronephrosis, vertebral anomalies like hemivertebra, fused vertebra, lumbar lordosis, kyphoscoliosis, genital anomalies, cleft lip and palate and chromosomal anomalies such as Trisomy 21, 18 or 13 may coexist with Congenital diaphragmatic hernia. Familial inheritance is reported in 2% of the cases.

KEY WORDS: Costovertebral, diaphragmatic hernia, fetus, renal agenesis.

Address for Correspondence: Dr. A.K. Manicka Vasuki, Assistant Professor, Department of Anatomy, PSG Institute of Medical Sciences and Research, Coimbatore - 641004, Tamil Nadu, India.

E-Mail: vasukikalyan01@gmail.com

Access this Article online

Quick Response code



DOI: 10.16965/ijar.2015.344

Web site: International Journal of Anatomy and Research
ISSN 2321-4287
www.ijmhr.org/ijar.htm

Received: 18 Dec 2015 Accepted: 12 Jan 2016
Peer Review: 18 Dec 2015 Published (O): 31 Jan 2016
Revised: None Published (P): 31 Jan 2016

INTRODUCTION

Costovertebral syndromes are characterized by multiple vertebral segmentation defects of vertebrae and anomalies of the ribs. It may present as dominant or recessive form of inheritance. There are two subtypes 1. Spondylothoracic dysostosis characterized by crab like or fan like deformities of the ribs and neural tube defects. This may be fatal due to respiratory failure in infancy. Spondylocostal dysostosis-characterized by short neck, short trunk, mild scoliosis with vertebral anomalies including butterfly vertebrae, hemivertebrae and

fused hypoplastic vertebrae. This type may have normal life expectancy.

Congenital diaphragmatic hernias occur in 1/2000 to 1/4000 births. It accounts for 8% of all major congenital malformations and constitutes one of major surgical emergencies in the newborn. The most commontype of congenital diaphragmatic hernia is through posterolateral foramen of Bochdalek with 80% predominance on the left side. Only 1-5% of Congenital diaphragmatic hernia occur through Foramen of Morgagni, which is retrosternal at the

sternocostal triangle, known as Larey space. The Morgagni type occur commonly right sided in the anterior mediastinum.

Sometimes newborn may be born asymptomatic and are discovered accidentally from chest or abdominal X-Ray performed for other reasons. Rarely newborn may born with respiratory symptoms or complications such as strangulation or perforation of abdominal viscera, gastric volvulus or cardiac tamponade. Surgical correction is needed in all cases of Congenital diaphragmatic hernia to avoid the risk of complications, especially bowel strangulation.

CASE REPORT

Fig. 1: Showed Left congenital diaphragmatic Hernia (CDH), hypoplastic lungs on both sides and Horseshoe shaped Liver.

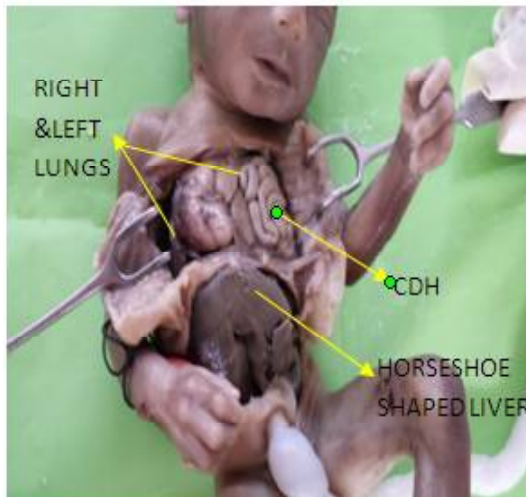
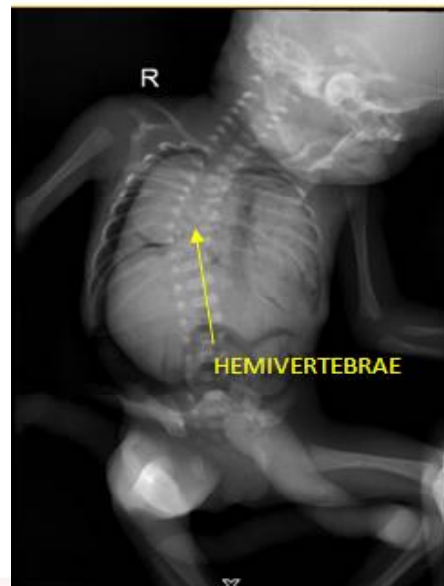


Fig. 2: Showed Empty right renal fossa.



Fig. 3: Showed Hemivertebrae in the last two thoracic vertebrae.



After obtaining ethical clearance from the parent, dissection of 23 weeks aborted male fetus in the department of Anatomy, PSG IMS&R, Coimbatore revealed left congenital diaphragmatic hernia with mediastinal shift, pulmonary hypoplasia and right renal agenesis. The liver was found to be horseshoe shaped and was enlarged. Fetogram showed hemivertebrae in the last two thoracic vertebrae.

Prenatal ultrasound of the fetus showed left congenital diaphragmatic hernia, right renal agenesis and hemivertebrae in the last two thoracic vertebrae.

DISCUSSION

Jarcho-Levin syndrome is a rare genetic disorder characterized by multiple vertebral and rib anomalies. Jarcho and Levin described this disorder in 1938 [1].

In 1968, Rimoin et al [2] suggested the term Spondylocostal dysplasia for this syndrome and has two types. Type I includes autosomal recessive inheritance and early death and type II is inherited as autosomal dominant pattern with less severe form and better life expectancy. Solomon et al (1978) [3] classified this into two groups, based on the presence or absence of rib malformations. Spondylocostal dysplasia has vertebral anomalies, intrinsic rib malformation and flaring of bone. Spondylothoracic dysplasia has vertebral anomalies and fanlike rib configuration without intrinsic rib malformation.

Vasanti Arole et al [4] observed short neck, left diaphragmatic hernia, talipes equinovarus, two meningocele sacs on the back of neck, ectopic anal opening and vertebral anomalies in 20 weeks old aborted fetus. Lumbar vertebrae was found to be articulating with the base of the skull. Thoracic vertebrae forming a ring of bones under the skull to which ribs were articulating. Sacrum was seen as a vertical strip of bone. A multilobulated horseshoe shaped kidney was seen just under the diaphragm.

Yangsoon Park et al [5] observed Cervicothoracic hemivertebrae, diminished right sided ribs, pulmonary hypoplasia with left diaphragmatic hernia, anomalous rotation of foregut, unfused pancreas and anomalous drainage of Superior venacava in 30 weeks preterm female fetus.

Ali Karaman et al [6] reported short neck and trunk, thoracolumbar lordosis, hemivertebrae and block vertebrae, imperforate anus with hydrocephalus in a 38 weeks newborn.

Olga Marikova et al [7] reported malsegmentation of T6 – T11 vertebrae, butterfly vertebrae T5 and scoliosis, 13 ribs on the left side with fusion of 4th & 5th ribs on that side with features of Osteogenesis imperfecta which consists of generalized osteoporosis, acetabular protrusion, helmet type of cranium and triangular facial bones.

Sandeep vijayan et al [8] reported Spondylocostal dysostosis with Sprengel's shoulder in 4 years old boy who was brought with complaints of left shoulder deformity.

Balanescu et al [9] reported Morgagni congenital diaphragmatic hernia in 7 months old boy who presented with respiratory distress.

In the present case, We found Congenital left diaphragmatic hernia, right renal agenesis, horseshoe shaped liver with hemivertebrae in the last two thoracic vertebra in 23 weeks fetus. The Spondylocostal dysostosis with Congenital diaphragmatic hernia and renal agenesis is the rarest presentation. Prenatal diagnosis is important to diagnose the condition and genetic counseling is necessary to prevent the recurrence.

CONCLUSION

Base on clinical and radiological evaluation of infants with multiple vertebral segmentation defects, two types of Jarcho-Levin syndrome have been identified. Spondylocostal dysostosis presents with minor vertebral and rib abnormalities without fanlike thoracic configuration. Many of the infants born with this disorder die due to respiratory failure. Eventhough this subtype is less life threatening, regular monitoring of the growth, respiratory function and spinal deformity is absolutely essential. Identification of Karyotype and mutant alleles help to provide genetic counseling in future pregnancies.

Hence awareness of the clinical and radiological features of this syndrome and genetic counseling for at risk pregnancies is necessary to identify the condition early and to have better care of these newborns.

Conflicts of Interests: None

REFERENCES

- [1]. Jones KL Smith's Recognizable patterns of Human Malformation, In: Jone S(Ed).JLS Miscellaneous Syndromes.6th ed.Philadelphia, Elsevier Saunders, 2006:690-691
- [2]. Rimoin DL, Fletche BD, McKusick VA: Spondylocostal dysplasia. Am J Med. 1968;45:948-953.
- [3]. Solomon L, Jimenez RB, Reiner L: Spondylothoracic dysostosis, Report of two cases and review of the literature. Med 1978;102:201-205.
- [4]. Vasanti Arole, Vatsalaswamy et al. Jarcho-Levin syndrome with multiple developmental anomalies, Ind.J.Basic and App.Med.Res. June2015;4(3):55-58
- [5]. Yangsoon Park, M.D.Gyungyub Gong et al. Jarcho-Levin syndrome – a report of an autopsy case with cytogenetic analysis, J. Korean Med.Sci, Dec 1993;8(6):471-475.
- [6]. Ali Karaman, Hasan Kahveci et al, Jarcho – Levin syndrome and Hydrocephalia: case report. Med.J.Bakirkoy. 2013;9:183-185
- [7]. Olga Marikova, Ivo Marik et al, Osteogenesis imperfect and Spondylocostal dysostosis in a male child, Magyar Radiologia 2004;78(4):190-192
- [8]. Sandeep vijayan, Hitesh Shah, Spondylocostal dysostosis with Sprengel's shoulder – report of a new association with Jarcho – Levin syndrome, Kerala J.Ortho, July 2012;25:103-104
- [9]. Balanescu, Ulici, Morgagni congenital diaphragmatic hernia in a 7 months old boy – a case report, JASI 2012:291-295.

How to cite this article: A. K. Manicka Vasuki. JARCHO-LEVIN SYNDROME: A CASE REPORT. Int J Anat Res 2016;4(1):1825-1827. DOI: 10.16965/ijar.2015.344