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Research Article

A CASE OF JARCHO- LEVIN SYNDROME

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ABSTRACT

Jarcho- Levin syndrome or costovertebral dysplasia or spondylocostal dysplasia or spondylocostal dystosis is a rare genetic disorder that represents a spectrum of short trunk skeletal dysplasia with variable involvement of the vertebrae and ribs1. Jarcho and Levin, in 1983 described a pattern of vertebral and costal anomalies distinctly different from the well known Klippel- feil syndrome2. The subtypes are spondylothoracic dysplasia predominant vertebral defects and spondylocostal dysplasia describes the variant with vertebral and instrinsic costal anomalies3. Despite being rare, this disorder has been focus in recent times due to controversies surrounding it4. The purpose of reporting this case is to bring clinical understanding of this rare disorder and review the emerging current knowledge about it. In this article, we report a case of spondylocostal dystosis in 2 month old female from Doda, Jammu.

INTRODUCTION

Jarcho- Levin syndrome is a type of short trunk skeletal dysplasia with vertebral and ribs anomalies. It has been both autosomal dominant and recessive modes of inheritance. The vertebral anomalies seen are – hemivertebrae, absent vertebrae, fused vertebrae, block/ wedge vertebrae, sickle shaped vertebrae due to segmentation and formation defects (pebble-beach appearance). The costal defects noted are crab– like or fan like appearance of the thorax due to crowded ribs, posterior fusion of the ribs and absent or irregular of bifid ribs [3,5,6,7].

Epidemiology

Nearly 400 cases have been described in the world literature and about 15 in the Indian literature. The disorder has been noted in consanguineous and non- consanguineous families. Most cases reported follow the pattern of autosomal recessive inheritance. In southern India, consanguinity is common. Despite this face, the disorder is seen uncommonly and reported very rarely from this part of the country [3,5,6,7]. At conception, each sib of an affected individual has a 25% chance of being affected, a 50% chance of being an asymptomatic carrier, and a 25% chance of being unaffected and not a carrier[8].

Classification

There are two major types of Jarcho-levin syndrome. These are:

***Spondylothoracic Dystosis*:** It is having vertebral body malformation and ribs with flare in a fan like pattern but which are not significantly malformed. It is and autosomal recessive trait and patients have higher mortality rate and greater incidence of neural tube defects[7].

***Spondylocostal Dysplasia*:** It represents a group of very rare genetic disorder, characterized by vertebral and costal segmentation defects, sometimes accompanied by visceral malformation[8]. It is inherited in an autosomal recessive manner and is caused by a mutation in one of four genes DLL3, MESP2, LFNG, HES7. Rarely spondylocostal dysplasia can be inherited in an autosomal dominant manner by one gene namely TBX6 mutation. A mutation may lead to somitogenesis defects, with segmentation defects of axial skeleton and deformation[9]. Intrinsic rib anomalies like broadening, bifurcation and asymmetrical fusion are noted. The survival rate in spondylocostal dysplasia is high after the age of six months (56%) [10]. Almost all the patients have normal intelligence and neurological abnormalities are infrequent[4].

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Clinical Features

The main features of spondylocostal dystosis are abnormally fused bone of the spine, abnormal side- to- side curvature of the spine (scoliosis), rib bones are fused or missing, short, rigid neck and short torsos as a result they are having short bodies but normal length of arms and legs called short trunk, dwarfism, spine and rib abnormally causes short chest that leading to life threatening breathing problem[11], inguinal hernia particularly in males[12].

Repeated respiratory infections, complex congenital heart disease, atrial septal defects, anomalous pulmonary venous return[13], an obstruction of the bladder may occasionally cause a swollen stomach and pelvis in an infant, undescended testicles, absent external genitalia, a double uterus, closed or absent anal and bladder opening or a single umbilical artery[14].

Distinctive facial features and mild cognitive deficits may sometimes occur, birth defects may also affects the central nervous system, webbed (syndactyl), elongated and permanently bent (camptodactyl) fingers are common, a broad head, wide nasal bridge, nostrils that tip forward, upwardly stranded eyelids and an enlarged posterior skull are other symptoms that may be evident[13]

Clinical Diagnosis

Diagnosis is clinical and may be supported by ultrasonography, genetic analysis, and spinal radiographs. Prenatal diagnosis is possible using fetal ultrasound. Prenatal diagnosis by ultrasound can be done as early as 16 weeks of gestation after conception. Ultrasound criteria for diagnosis are unpaired or poorly formed vertebrae, indistinct or fused posterior ribs, irregular short "Pebble-like" appearance of the spine, short trunk, protuberant abdomen, hernias, normal amniotic fluid, normal limb length and normal biparietal diameter [15].

Case Report

A 2 months old female baby came to Shri Maharaja Gulab Singh (SMGS) Hospital, Shalamar, Jammu with chief complaints of breathing difficulty, fever, excessive crying and bluish discoloration of the body during crying. She was admitted with the same complaints in the same hospital 2 months back. General and systemic examination of the baby revealed the following symptoms such as Scoliosis, Dusky lips, Pallor, Short neck, S_1 and S_2 present , Louder S_2, Liver 3cm below rib costal margin and spleen palpable.

The result of serological examinations was normal. The patient was afebrile with unstable vitals (respiratory and heart rate) and fair GCS. Ultrasonography of head and abdomen was normal. X- ray chest revealed abnormal side- to- side curvature of the spine (scoliosis) with fused rib bones. ECHO revealed that client is having cyanotic congenital heart disease, trans-positioning of great arteries with moderate Pulmonary Stenosis.

Management

The treatment is directed towards the specific symptoms that are apparent in each individual. Treatment may require the coordinated efforts of a team of specialists.

- Infants who experience breathing difficulties can require respiratory support by oxygen therapy or mechanical ventilation
- Some infants may require intensive care, which involves constant monitoring in a hospital
- Surgery is required to repair an inguinal hernia
- Antibiotics may be required to treat recurrent respiratory infections
- The surgical treatment for thoracic insufficiency include vertical expandable prosthetic titanium ribs (VEPTR) and chest wall reconstruction with latissimus dorsis flap transfers or polyprophylene mesh16.
- At present, the symptomatic treatment is provided to the client including:
- Propped up position
- Oxygen inhalation
- Tablet Ciplar 10mg – it is a non selective beta blocker medicine which was given orally twice a day. The side effects of this medicine are breathing difficulty, irregular heart beat, skin blistering, dizziness etc.

The client was receiving efficient symptomatic treatment at Shri Maharaja Gulab Singh hospital from health care team members as they were putting great efforts for the survival of client. Despite all resuscitative measures, the client died due to respiratory failure.

DISCUSSION

It is a rare genetic disorder characterized majorly by defects of the bones of the spine and abnormalities of the ribs. Counseling of the affected family is not a simple task. The exact clinic-radiological diagnosis with molecular diagnosis is essential for accurate genetic counseling and prognostication of each individual case. The treatment of the client is based on manifestation including intensive medical care, bone surgery and orthopedic treatment[17].

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