

# Spondylocostal dysplasia in a neonate - An unusual association with maternal diabetes

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## Abstract

A female neonate who was born as large for gestational age baby to a Diabetic mother had a continuous murmur in the left infraclavicular area with scoliosis toward right and deficient ribs in the right side of her chest wall. She had a sacral dimple and a skin tag in the right lower back. Radiograph revealed hemivertebrae from T1-T6 level with absent third and seventh ribs and fused fourth, fifth and sixth ribs on the right side and a bifid right eighth rib. Echocardiogram revealed patent ductus arteriosus (PDA). Based on the above features she was diagnosed to have spondylocostal dysplasia. She was asymptomatic at the time of discharge.

Key words: Spondylocostal dysplasia, maternal diabetes, bifid rib, fused rib, hemivertebrae

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## Introduction

Spondylocostal dysostosis is a rare disorder of vertebral segmentation often inherited and some times sporadic associated with vertebral and rib anomalies, congenital heart disease and neural tube defects [1]. The recessively inherited form is usually severe causing death in infancy and is also referred to as spondylothoracic dysplasia. The dominantly inherited form is usually mild and runs a benign course and is referred to as spondylocostal dysplasia [2]. We report here a neonate born to gestational diabetes mother with clinical features consistent with spondylocostal dysplasia.

## Case Report

A term female neonate was born to nonconsanguineous parents by caesarean section. The mother was 37 year old, sixth gravida with gestational diabetes. The baby had an Apgar of 8 and 9 at one and five minutes respectively and a weight of 3.76 kg. Her length was 48 cms and head circumference 34.5 cms. She was found to have continuous murmur at the left second intercostal space and deficient ribs on the posterior aspect of right hemithorax. There was mild scoliosis with concavity towards right. She also had a skin tag in the lower back (Fig. 1) on the right side and a sacral dimple. There was no obvious facial dysmorphism or plethoric look.

The rest of the examination was essentially normal. The mother was diagnosed to have gestational diabetes during the second trimester and was started on insulin at 20 weeks of gestation. Apart from gestational diabetes, the mother was HBSAg positive too. The mother's blood group was O RHpositive.

The presentation was vertex, liquor was adequate and clear. The mother was not on any other drugs or known teratogens. The mother has one female child with three spontaneous abortions between the second and third month and one still birth at ninth month of gestation due to placental abruption.

The baby was started on breast feeds immediately after birth and kept by mother's side. The blood sugar levels were normal. Chest radiograph revealed hemivertebrae from T1 to T6 levels with absent 3rd and 7th rib and fused 4th, 5th and 6th ribs and bifid eighth rib on the right side (Fig. 2).

There was no evidence of spinal dysraphism on ultra-sound examination. Echocardiogram revealed a small PDA. Ultrasound cranium and abdomen were normal. Karyotype was normal. The baby received first dose of hepatitis B vaccine and immunoglobulin. She was also given BCG and oral polio vaccines. She was discharged on the 10th day of life and advised follow up.



Figure 1. Showing skin tag over the lower back of the baby.

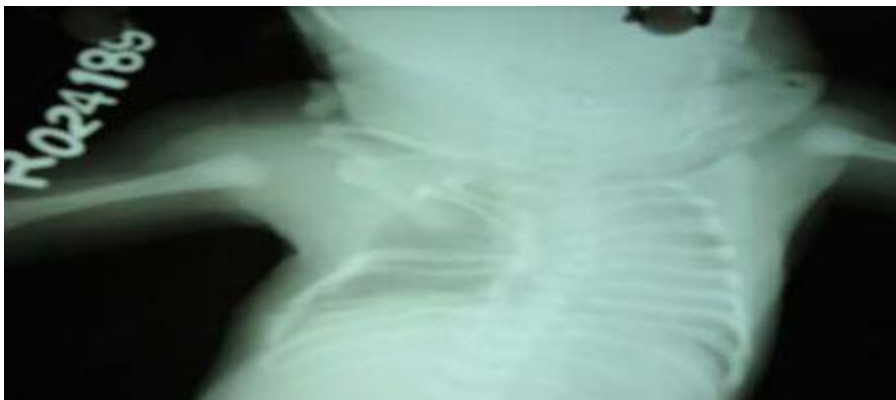


Figure 2. Chest radiograph showing absent 3rd and 7th rib along with with fused 4th,5th and 6th ribs and bifid 8th rib on the right side.

## Discussion

Spondylocostal dysostosis is a disorder of vertebral segmentation characterized by block or hemivertebrae and absent or fused ribs with scoliosis. The severe form, also known as spondylothoracic dysplasia or Jarcho Levin syndrome(1) is inherited recessively. Spondylocostal dysplasia is dominantly inherited and has a mild course and may even present as asymptomatic adults. The defect in vertebral segmentation typically occurs early in embryogenesis around 4 to 6 weeks. The rib cage may show varying defects from absent ribs, fused ribs or bifurcated ribs. Extrathoracic manifestations are more common in spondylocostal dysplasia. Congenital heart disease (ventricular septal defect and patent ductus arteriosus) has been reported [2]. Neural tube defects, renal anomalies and diaphragmatic hernia may be associated findings. In spondylothoracic dysplasia, the deformities are typically confined to the vertebral column and rib cage with typical short asymmetric rib cage which is described as crab like. Extrathoracic malformations are not very common. Spondylothoracic dysplasia is usually lethal in infancy with progressive respiratory insufficiency. The disorder can be identified antenatally by 3D ultrasound examination of the fetus [3].

The gene involved in the pathogenesis of this disorder has been mapped to dll 3 gene in mouse models which encodes for a ligand in the notched gene signal pathways and mutations in these pathways interfere with somite alignment and development [4]. The human gene that is syntenic to mouse dll gene is located at the 19q13.2-13.3 locus [5] and was found during mapping of 6 arab-israeli families with spondylothoracic dysplasia. Similarly protein truncating and missense mutations were noted in three families with spondylocostal dysplasia. A similar disorder with vertebral segmentation defect, namely alagille syndrome has mutation in the JAG1 gene, a ligand of notched receptor [6].

Apart from genetic mutations, thoracic vertebral malformations can arise due to maternal exposure to carbon monoxide as early as ninth day of gestation [7]. Rats exposed to boric acid also have been found to develop only 6 cervical vertebrae rather than the usual seven [8]. Rats exposed to class III antiarrhythmic agents in utero also develop similar vertebral malformations [9]. Other teratogens being valproic acid, phenytoin, maternal diabetes mellitus and hyperthermia. Differential diagnosis for this disorder includes Klippel Feil syndrome, Goldenhar syndrome, Alagille syndrome, VACTERL and VATER associations.

Diabetic embryopathy results from exposure of embryo to poorly controlled maternal diabetes. The features include caudal regression, situs inversus, arthrogyrosis, spinal and vertebral anomalies (polydactyly, sacral agenesis, sirenomelia, neural tube defects) and disorders of genitor-urinary system and anomalies of heart and blood vessels. Although vertebral and skeletal anomalies are more common in diabetic embryopathy, spondylocostal dysplasia as a distinct entity has not been commonly described.

Treatment consists of arresting the spinal cord curvature progression to scoliosis and prevention of restrictive lung disease from developing. This can be achieved by performing multiple expansion thoracoplasties, chest wall reconstruction using latissimus dorsi flaps and posterior spinal fusion for scoliosis. Chest physiotherapy and conservative therapies are helpful in the early stages [10].

In our case the neonate was born to a diabetic mother with poorly controlled diabetes beginning in the second trimester. The marriage was non consanguineous and there was no family history of similar disorder in other family members indicating possibly a teratogenic effect or a sporadic mutation. The parents were counseled about the long term effects of this disorder and motivated for further treatment and followup. The neonate was asymptomatic at the time of discharge.

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