Abnormal Ribs, Hemivertebrae and Sacral Agenesis: A Rare Skeletal Deformity in Infant of a Diabetic Mother: A Case Report

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Abstract

A good weight male baby of 4 kg delivered at 40 weeks gestation on 28th January, 2016 at Darul Sehat Hospital, Karachi. At birth he had Apgar score of 6/1, 7/5 and admitted in Neonatal Intensive Care Unit for blood glucose monitoring and basic laboratory investigations which included complete blood count, C-reactive protein, blood culture and sensitivity and chest x-ray. An incidental finding on chest x-ray was present as abnormal rib cage with fractured ribs followed by infantogram which showed hemi-vertebrae and agenesis of sacral bones which is a part of caudal regression syndrome, this is a very rare finding of skeletal deformity seen in infant of diabetic mother. Hence this case is being reported, as local literature is scarce.

Keywords: Diabetic mother, fetal abnormality and structural deformity

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Introduction

Caudal Regression Syndrome (CRS) represents a spectrum of structural defects of the caudal region. Malformations vary from isolated partial agenesis of the coccyx to the lumbosacral agenesis[1].

This finding is rare as estimated incidence is 1:7500 - 100,000 and there is no gender predilection. The vast majority of cases are sporadic however familial cases occasionally occur as well[2,3].

In some cases, caudal regression syndrome may be associated with mutations of the VANGL1 gene located on the short arm (p) of chromosome 1 (1p13). The exact role this gene mutation plays in the development of caudal regression syndrome is unknown. This mutation is inherited as an autosomal dominant trait. Genetic diseases are determined by the combination of genes for a particular trait that are on the chromosomes received from the father and the mother. Dominant genetic disorders occur when only a single copy of an abnormal gene is necessary for the appearance of the disease. The abnormal gene can be inherited from either parent, or can be the result of a new mutation (gene change) in the affected individual. The risk of passing the abnormal gene from affected parent to offspring is 50 percent for each pregnancy regardless of the sex of the resulting child[4-6].

Severe cases are usually identified in utero or at birth. Mild cases may not be identified until adulthood. Caudal Regression Syndrome has different associations: 20% cases are associated with

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maternal diabetes (Type-I or Type-II). It occurs in up to 1% of pregnancies of women having Diabetes Mellitus[7].

Here we present a case of an Infant of Diabetic Mother with skeletal deformity which is a very rare presentation.

Case Report

This patient was born to a 32 years old mother, who was diagnosed to be diabetic one year back and was using regular insulin with poor glycaemic control. She was a booked case; Gravida 6 Para 3+3, other antenatal history was unremarkable. She gave birth to a full term, male baby via normal vaginal delivery with good Apgar score. The new born was 4 kilogram in weight, 47 centimeter in length and 36 centimeter head circumference, all within normal ranges. Baby was admitted in Neonatal Intensive Care Unit for sugar monitoring and for basic laboratory investigations as an infant of a diabetic mother.

On examination he was pink and had no dysmorphic features; vitally stable with normal primary reflexes and there were no visible congenital abnormalities.

Chest was clear, cardiovascular system examination revealed first and second heart sounds were audible with no added sounds; abdomen was soft and central nervous system / loco motor examination was normal with good strength and normal reflexes. Rest of the systemic examination was found normal.

His Blood Investigations were found to be normal except for Hypomagnesaemia which was corrected by giving Magnesium Sulphate infusion.

Chest x-ray showed abnormal rib cage with fractured ribs (Fig.1) followed by Infantogram which showed Hemi-vertebrae (Lumbar region) and sacral agenesis (Fig. 2).

Echocardiography, Ultrasound KUB were also normal, blood sugars were also normal throughout the admission. Baby was treated supportively and parents were councelled and advised to follow up in orthopaedic and neurosurgical clinic.

Discussion

Hemi-vertebrae and sacral agenesis is a congenital abnormality. It is a rare disease that occurs in 1/25000 live births to 1-2/100,000newborns. It occurs most commonly in infants of diabetic mothers, not gestational, at a rate of one in 350, making

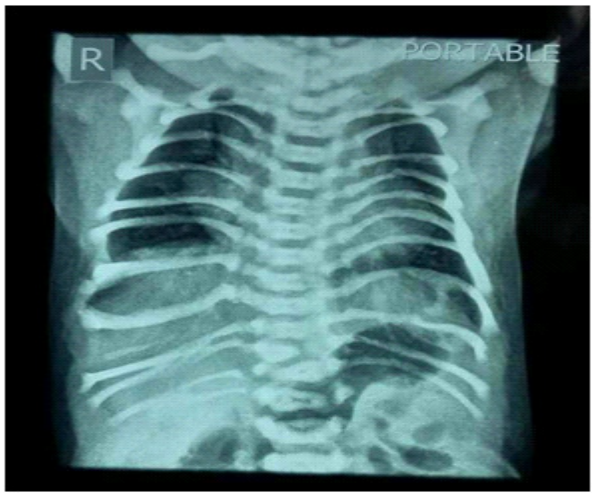

Fig 1. Chest X ray showing abnormal rib cage with fractured ribs

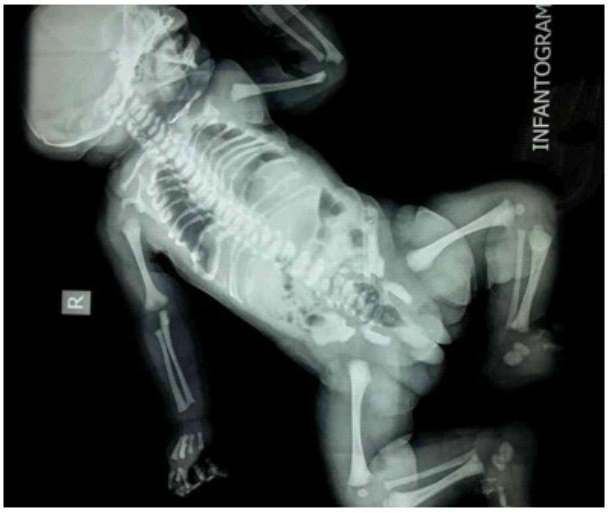

Fig 2. Infantogram showing abnormal rib cage with fractured ribs and Hemivertebrae

caudal regression syndrome as the most characteristic fetal abnormality of diabetic embryopathy although this estimation seems to be high, not all cases of sacral agenesis were born to diabetic mothers. Only 8-22% of them were born to those pregnancies[8].

The exact cause of caudal regression syndrome is unknown. Researchers believe that both environmental and genetic factors may play a role in the development of the disorder. Most cases appear to occur sporadically, which suggests environmental factors or a new mutation. Most likely, caudal regression syndrome is multifactorial. In addition, different genetic factors may contribute to the disorder in different people (genetic heterogeneity)[9].

Most important aetiological factor contributing to this condition is hyperglycaemia where the risk of congenital regression syndrome increases when blood sugar is poorly controlled, as in our case.

The environmental factors that play a role in the development of caudal regression syndrome are unknown, although numerous different potential factors have been suggested including alcohol, retinoic acid, lack of oxygen (hypoxia) and amino acid imbalances. More research is necessary to determine what environmental factors play a role in the development of caudal regression syndrome[10].

Some infants with caudal regression syndrome may have a genetic predisposition to developing the disorder.

Most cases are sporadic but multiple genetic factors play a role in determining the risk of developing this abnormality. Congenital regression syndrome is associated with multiple congenital abnormalities. Central nervous system anomalies like Hydrocephalus and Myelomeningocoele are not rare but was not encountered in our patient.

Orthopaedic anomalies like scoliosis,limb contracture and club feet with rib hypoplasia are common findings which not all but few were present in our patient. There may be imperforate anus and intestinal malrotation which was not the case here.

The most common and most significant anomalies found are of genitourinary system where the neurogenic bladder is the most common. Renal defects include unilateral renal agenesis, horseshoe kidneys, ureteralduplication and bladder extrophy. Renal ultrasound in our patient was normal[11].

Management requires a multidisciplinary approach by neurosurgeons, urologists, nephrologists, physical therapists and psychologists. Surgical interventions such as transureteroureterostomy and cutaneous vesicostomy, clean intermittent catheterization and/or anticholinergic drug administration are generally required to treat urological disorders. Colostomy is performed to treat an imperforate anus. Depending on the severity of the syndrome, orthopedic interventions may also be required. Treatment is only supportive because the primary pathology is irreversible.

Prognosis is poor. Early neonatal death in the severe forms occurs from cardiac, renal and respiratory complications. Surviving infants usually have normal mental function.

Careful diabetic control in the preconceptional period and the first eight weeks of pregnancy may lower the chances of congenital anomalies[13].

Genetic and pre-pregnancy counseling and screening provides the most hope for prevention of this debilitating disease. If possible, counseling should be initiated in diabetic mothers before their pregnancy. High correlation between this defect and the diabetic mother indicates a need for strict glycaemic control prior to the embryonic organogenesis period. Because CRS is thought to arise from an embryologic defect early in pregnancy, strict pre-pregnancy control should be used as a preventive strategy. Prenatal diagnosis should focus efforts on discerning the extent of dysgenesis so that parental decisions can be made to continue the pregnancy and postnatal interventions to treat the congenital anomalies associated with CRS can be planned[14,15].

Conclusion

Adequate control of hyperglycaemia in diabetic mothers is important to avoid congenital regression syndrome. Prognosis depends upon the severity of the case and associated anomalies.

Conflict of Interest

Authors have no conflict of interests and no grant/ funding from any organization for this study.

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