

Case Report

A case of diabetic fetopathy: caudal regression syndrome and associated anomalies

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Received: 06 April 2019

Accepted: 02 May 2019

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ABSTRACT

Uncontrolled periconceptional diabetes in mother can give rise to severe malformation in the offspring. Author describe a case of diabetic fetopathy presenting as a case of Caudal regression syndrome along with cardiovascular, gastrointestinal and genitourinary anomaly. Though most cases of Caudal regression syndrome are sporadic, it is the most specific lesion of diabetic fetopathy. Clinical presentation varies depending on the severity of the neurological lesion along with the presence of other anomalies, most commonly genitourinary malformation. The index case described here had a type IV lumbosacral agenesis with severe bilateral motor and sensory deficits along with D-Transposition of great arteries and Pyloric atresia. Such cases arise due to inadequate prenatal care of diabetic mother, which is not uncommon in developing country like India.

Keywords: Caudal regression syndrome, Diabetes, Fetopathy, Lumbosacral agenesis

INTRODUCTION

Lumbosacral agenesis or caudal regression syndrome (CRS) is a rare congenital malformation, characterized by the absence of sacrum and variable portion of lumbar vertebrae with abnormal development of the lower segment of spinal cord. Incidence of CRS varies between 1 per 25000 live birth to 2.5 per 100000 live birth in different birth defect registry.¹ It usually presents with bilateral severe motor paralysis of both lower limbs along with sensory deficits in a newborn, however mild cases can escape recognition until childhood. Though it is the most specific lesion of diabetic fetopathy, not all cases of CRS are born to diabetic mother.² In a developing country, like India where level of antenatal care is still suboptimal, pregnant women can go unnoticed with uncontrolled pre gestational diabetes mellitus with subsequent birth of a severely malformed baby. Authors report a case of a baby with caudal regression syndrome

and other congenital malformations, born to a diabetic mother with poor prenatal care.

CASE REPORT

A 30 year old, third gravida mother presented at 34 weeks of gestation with spontaneous preterm labour. She was unbooked with history of spontaneous abortions and still birth in her previous pregnancies with no living issue. She had been suffering from diabetes for last 5 years, which was inadequately controlled with oral hypoglycemics only. No prenatal investigation reports or anomaly scan was available. No HbA1C report was available. She gave birth to an asphyxiated girl baby of birth weight 2050 gm, length 42 cm and head circumference 32 cm. After resuscitation baby was shifted to NICU and ventilated. On clinical examination, both lower limbs were noted to be hypoplastic with severe flexion and adduction of hip, flexion of knees and

bilateral club feet (Figure 1). Face and upper limb were normal.



Figure 1: Hypoplastic lower limb with flexion and adduction of hip, flexion of knee and bilateral club feet.



Figure 2: A dimple at the lumbosacral area (horizontal arrow) with patulous anus (vertical arrow).

A dimple was noted on the skin at the lumbosacral region along with patulous anus (Figure 2). On further examination, sacrum was not palpable.

X ray spine revealed absent lumbar vertebrae from L2 downwards with absence of sacrum and apposition of both ilium with formation of a midline cleft (Figure 3). Diagnosis of lumbosacral agenesis was made.

Abdominal sonography revealed bilateral hydronephrosis with prominent bladder compatible with neurogenic bladder. Cardiovascular examination was unremarkable at birth with normal abdominal examination. Baby passed meconium and urine within 24 hours of life. Though baby was able to maintain oxygen saturation more than 90% on day 1 of life, baby started having progressive cyanosis from day 3. Echocardiography revealed D- transposition of great arteries with restrictive ASD and closing PDA

and she was immediately started on Prostaglandin to maintain ductal patency. Baby had bilious vomiting and bilious aspirate since day 3 of life. Abdominal radiography revealed single large gastric shadow with absence of distal air (Figure 3) and provisional diagnosis of pyloric atresia was made. Neurosonography was normal. Spinal sonography revealed absent sacrum and lower lumbar vertebrae with the gap filled with fibrofatty tissues (Figure 4).

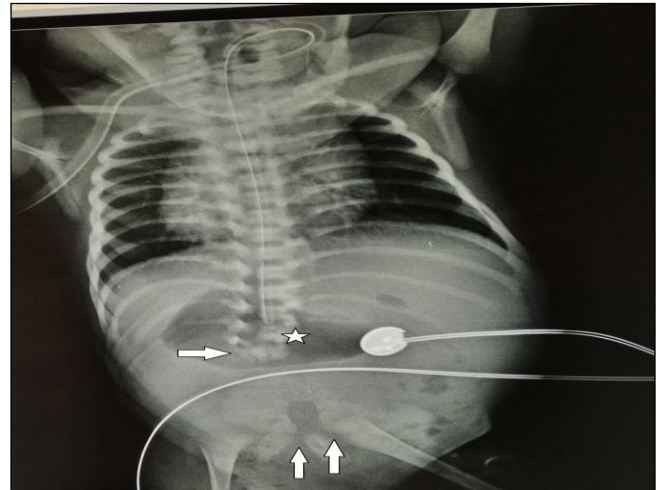


Figure 3: Absence of the lumbosacral vertebra (horizontal arrow) with apposition of ilium of both sides (vertical arrow). Single large gastric shadow shown with asterisk.

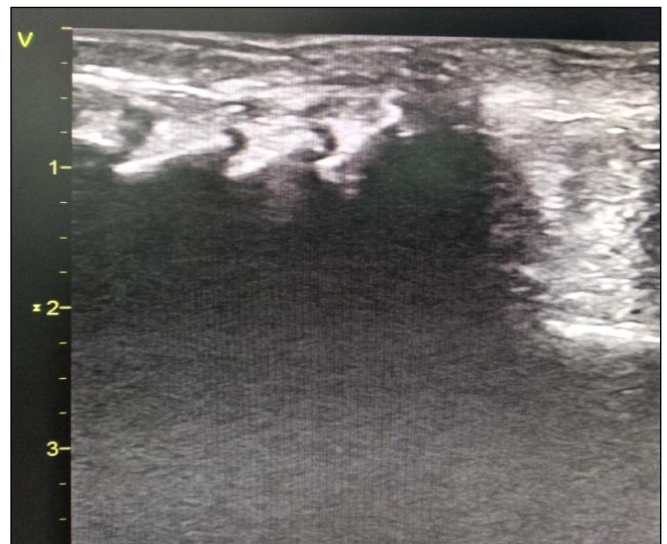


Figure 4: Spinal USG showing absent sacrum and lower lumbar vertebrae with the gap filled with fibrofatty tissues.

Micturating cystourethrography and spinal MRI was planned but could not be materialized due to difficulties in shifting a ventilated baby to a distant radiology unit. Baby's condition gradually deteriorated, and the baby died at the later part of the first week of life.

DISCUSSION

CRS usually results from an abnormal development of the caudal aspect of the spinal cord and vertebral column. Although precise etiology is not known, teratogenic, ischemic and infectious etiologies before the fourth week of gestation are thought to be contributory.³ Majority of the cases are sporadic, but multiple genetic factors are increasingly being discovered, such as a mutation in the HLXB9 gene can present as Currarino syndrome with sacral agenesis, sacrococcygeal teratoma and anorectal malformation.⁴ There is an association with poorly controlled pre gestational diabetes and adequate control of diabetes before conception and during the first trimester is associated with decreased incidence of this syndrome.⁵ However, history of maternal diabetes is present only 16% to 22% of cases.⁶ Most of the cases are diagnosed antenatally through demonstration of an abrupt termination of the lumbar spine and hypoplastic lower limbs. CRS is associated with multiple other anomalies, most significant of which are orthopedic deformities and genitourinary anomalies. Renshaw studied 23 individuals with CRS between 1959-1978 and classified the defect into 4 types, of which type 4 is the most severe variety.⁷ Type I describes CRS with total or partial unilateral sacral agenesis, while type II deformity includes partial sacral agenesis with partial but bilaterally symmetrical defect and a stable articulation between ilia and a normal or hypoplastic first sacral vertebra. Both these types can remain undiagnosed until later in childhood. Type III presents with variable lumbar and total sacral agenesis with ilia articulating with the sides of the lowest vertebra present. Type IV defect, as in this case, presents with variable lumbar and total sacral agenesis with the lowest vertebra resting above fused ilia.

In this case, there is associated cardiovascular, genitourinary and gastrointestinal defects along with CRS, all of which are well recognized complications of poor periconceptional diabetic control. There is general agreement that malformation rates are not increased in women with gestational diabetes. However, among women with pre gestational diabetes, those with insulin nondependent and insulin dependent, should be considered separately. Soler et al, reported similar incidence of congenital malformation between diabetic women on oral hypoglycemic drugs and insulin (8.7% vs 8.1%) which was significantly higher than non-diabetic women(1.7%).⁸ Women with insulin dependent diabetes have significantly more chance of having malformed baby with earlier onset and longer duration carrying more risk. Using a developmental morphologic approach, Mills et al showed that all the anomalies occur before 7th weeks of gestation.⁹ In Kucera's review of literature, relative risk of CRS in infants of diabetic mother was over 200.¹⁰ Data from collaborative perinatal project estimated relative risk of 3.1 of congenital heart diseases among infants of diabetic mother.¹¹ Hyperglycemia is considered teratogenic during the period of organogenesis

and periconceptional diabetic control is crucial for preventing malformation.

CONCLUSION

Adequate control of hyperglycaemia during the periconceptional period is crucial to avoid diabetic fetopathy. A malformed infant of diabetic mother should have thorough clinical and radiological examinations to rule out anomalies of major organ systems.

Funding: No funding sources

Conflict of interest: None declared

Ethical approval: Not required

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Cite this article as: Pal S, Sardar SK. A case of diabetic fetopathy: caudal regression syndrome and associated anomalies. *Int J Contemp Pediatr* 2019;6:1764-6.