EUROPEAN JOURNAL OF BIOMEDICAL AND PHARMACEUTICAL SCIENCES
http://www.ejbps.com

ISSN 2349-8870
Volume: 3
Issue: 2
331-333
Year: 2016

CASE REPORT OF CAUDAL REGRESSION SYNDROME; A RARE ENTITY.

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Article Received on 23/11/2015 **Article Revised on 14/12/2015** **Article Accepted on 03/01/2016**

ABSTRACT
Caudal regression syndrome (agenesis of the lumbar spine, sacrum, and coccyx, and hypoplasia of the lower extremities) is a well-established congenital anomaly associated with maternal diabetes mellitus. We present a case report of a neonate, with external pelvic and leg deformities, diagnosed as lumbosacral agenesis on infantogram, born to diabetic mother. Maternal diabetes, genetic predisposition and vascular hypo perfusion have been suggested as possible causative factors. In this presentation we emphasize clinical and radiological findings in this rare abnormality and also necessity of educating rural population for early screening of diabetic pregnant women.

KEYWORDS: Caudal regression, sirenomelia, lumbosacral dysgenesis, sacral agenesis.

INTRODUCTION
Caudal regression is rare syndrome which has a spectrum of congenital malformations ranging from simple anal atresia to absence of sacral, lumbar and possibly lowers thoracic vertebrae, to the most severe form which is known as sirenomelia.[1] The etiology of this syndrome is not well known. There are four levels (or "types") of malformation. First level is partial deformation (unilateral) of the sacrum. The second level is bilateral (uniform) deformation. The most severe types involve a total absence of the sacrum. Depending on the type of sacral agenesis, bowel or urinary bladder deficiencies may be present. Severe forms are commonly associated with cardiac, renal and respiratory problems, which are responsible for early neonatal death.[2]

Pathology being from an insult in early pregnancy (<4th week of gestation). Hyperglycemia, infection, toxic and ischemic insults have been implicated, resulting in disturbance of the primary neurulation process and followed by derailment of the process of degeneration and differentiation of an initially normally developed primary and secondary neural tube

Sacral agenesis or caudal dysplasia is an anomaly found more often in offspring of diabetic women. However familial cases occasionally occur. Severe cases are usually identified in-utero or at birth. Mild cases may not be identified until adulthood. There is no gender predilection. Maternal diabetes, Trauma, nutritional problems, toxic agents and genetic are the factors suggested in the etiology.[3]

CASE REPORT
A 23-year-old primi gravid woman from remote rural area with seven months of amenorrhea came to the hospital in early labor with the complaints of swelling of feet and head ache since two weeks. She is a known case of diabetes since one year. Her present history and clinical examination and bio chemical tests revealed diabetes and pregnancy induced hypertension. On examination bilateral pedal edema, abdominal wall edema was present and blood pressure was 150/100 mmHg. Her fasting blood glucose and postprandial blood glucose levels were 124 mg & 186 mg. She had no previous antenatal scans. Immediately after admission into hospital she landed in normal labor and delivered a female baby with birth weight 1.5 kg and immediate Apgar score 6-7. On external examination, both lower limbs were in persistent abduction and external rotation with bilateral club foot (Fig 1a &1b). . indentation on the skin of the lower back (sacral dimple) and flattening of the buttocks noted (fig 2).Due to low Apgar score baby was admitted in pediatric ICCU.

Baby was sent to radiology department for further evaluation. Ultrasound of abdomen Showed bilateral renal pyelectasis. Baby radiographs were taken for

skeletal survey which showed complete absence of lower lumbar vertebra (L3, 4 & 5) and sacrum with normal iliac bones (Fig 3a&3b). Both hip joints were in external rotation and abduction with bilateral congenital dislocation of hips. With these imaging findings we confirmed it as caudal regression syndrome with bilateral renal pyelectasis.

LEGENDARIES

Fig 1a & 1b: Photograph of baby showing persistent abduction and external rotation of lower limbs with bilateral club foot.

Fig 2. indentation on the skin of the lower back (sacral dimple) and flattening of the buttocks.

Fig 3a & 3b: Fetal Radiographs showing complete absence of lower lumbar vertebra (L3, 4 & 5) and sacrum with normal iliac bones.

Fig. 1 a.

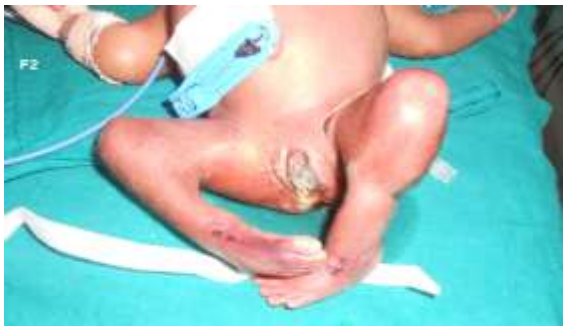

Fig. 1 b

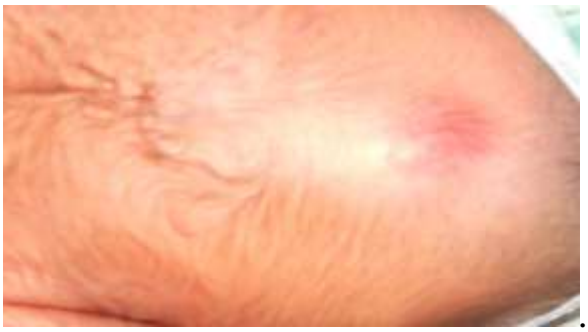

Fig. 2

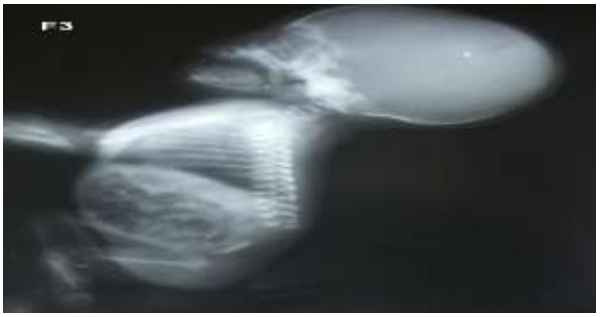

Fig. 3a

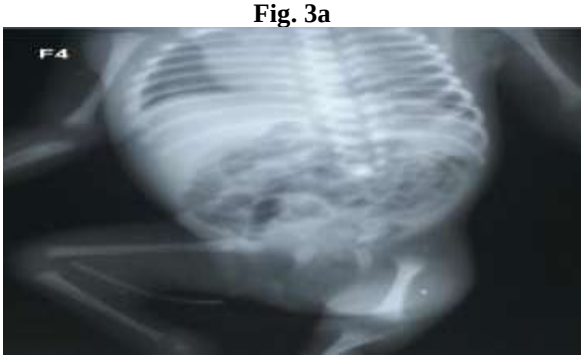

Fig. 3b.

DISCUSSION

Caudal regression syndrome is an uncommon malformation seen in 0.1-0.25: 10,000 of normal pregnancies. However it occurs in about one in 350 infants of diabetic mothers, representing an increase of about 200 times. Present scenario due to recent advances in antenatal ultrasound techniques, full blown syndromes is rarely seen. Associated skeletal, gastrointestinal, genitourinary and cardiac anomalies are commonly seen with this condition.[4]

Caudal regression syndrome with lumbo-sacral agenesis may range from absent coccyx, or as an isolated finding without neurologic sequale to sacral or lumbosacral agenesis. Damage to the nerves can also cause abnormalities of the lower limbs. The associated anomalies may range from deformities of feet, flexion contractures of hip and knees, dislocation of hips, pelvic deformity, kyphoscoliosis and absence of ribs.[5] Other anomalies include imperforate anus, malformed genitalia, renal dysplasia or aplasia and sirenomelia (fused lower extremities). In our case it is severe degree with total absence of lumbar vertebra and sacrum, associated with renal pyelectasis and pelvic girdle deformities with frog leg appearance and bilateral club foot.

The syndrome has been shown to occur more frequently in the offspring of diabetic mothers. Pinter E et. al., have demonstrated that hyperglycemia induced alterations in neural tube closure include disordered cells, decreased mitosis and changes indicating premature maturation.[6] Altered oxidative metabolism from maternal diabetes may cause increased production of free oxygen radicals in the developing embryo which may be teratogenic.

Segmental spinal dysgenesis (SSD) is a rare congenital abnormality in which a segment of the spine and spinal cord fails to develop properly. SSD and caudal regression syndrome probably represent two faces of a single spectrum of segmental malformations of the spine and spinal cord.

Diagnosis can be made in the first trimester by noting the short crown-rump length.[7] Sonography in second and third trimester can also demonstrate the absence of sacrum and shortened femurs. Sonography may detect associated urinary anomalies, such as renal agenesis, cystic dysplasia, calyectasis and gastro-intestinal anomalies such as duodenal atresia.[8] Early detection of caudal regression syndrome at 11 weeks of gestational age by transvaginal ultrasound scanning was reported.[9] It is unfortunate in our case no antenatal scan done from beginning due to unknown reason indicating the necessity of educating diabetic mothers for early scans.

If detected early, parents can have time to make decision regarding termination of pregnancy which at a later gestational age may not be possible. Early screening for anomalies during organogenesis is mandatory in high-risk patients, such as those with pregestational diabetes and poor glycemic control. Standard prenatal care is not altered if patient is not willing for termination.

The prognosis for children with caudal regression syndrome depends on the severity of the lesion and the presence of associated anomalies. Surviving infants have usually a normal mental function and they require extensive urologic and orthopedic assistance. Extensive surgery in tertiary center is usually needed to repair the defects.

In conclusion Caudal regression syndrome is a broad term for a rare complex disorder, characterized by abnormal development of the lower (caudal) end of the spine is well-established associated with maternal diabetes mellitus .early antenatal screening mandatory in all diabetes mellitus pregnant woman to detect fetal anomalies and for proper counseling of diabetic mother.

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