Case Report

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Sacral agenesis - A case report

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Abstract

Sacral agenesis is a rare congenital malformation of partial or complete absence of sacral or lower lumber region. We report 6 month old child with sacral agenesis.

Case history

Six month old female child presented with high spike fever found to have high C - reactive protein treated for acute pyelonephritis which needed IV antibiotics.

She was third born with two healthy siblings to a non-consanguineous parents and pregnancy was complicated with advance maternal (39 years) gestational diabetes mellitus which needed only diet control. Her anomaly scan was reported as normal and polyhydramnios was not noted.

She was born by elective caesarean section with the birth weight of 2.315kg APGAR was 10 and non-asphyxiated throughout.

She was found to have lower limb abnormality and dimples in back and suspected sacral agenesis at local Hospital where she deliver and which was confirmed with MRI tertiary hospital. She was seen by a paediatric neurologist paediatric nephrologist orthopaedic team as well as rehabilitation team. Her USS KUB was normal and decided to do a serial USS KUB and renal functions to delay the renal impairment and delay the development of neurogenic bladder. Her 2D ECHO is normal. She was referred to orthopaedic team for knee joint contractions to teaching Hospital Jaffna where she was underwent quadriceps plasty.

On examination she has few dysmorphic features with round face and low set ears. Anterior fontanelle was opened without bulging fontanels. Suture lines were normal. She had short flat of upper nose. She had thin lower lip. Upper limb examination was normal. Lower limb tone was increased. Power was 2. Club foot and bilateral knee joint contraction was noted. Reflexes exaggerated. Bilateral dimple sign was noted in back and able to found a bony defect in lower part of back. Cardiovascular examination abdominal examination and respiratory examination was normal. Her growth was adequate.

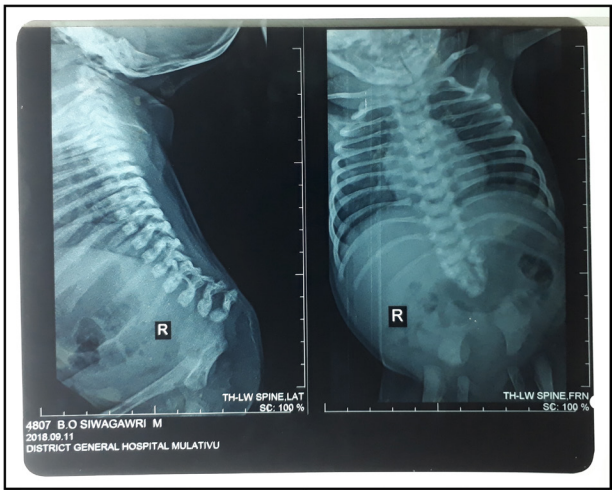

Figure1: x-ray spine

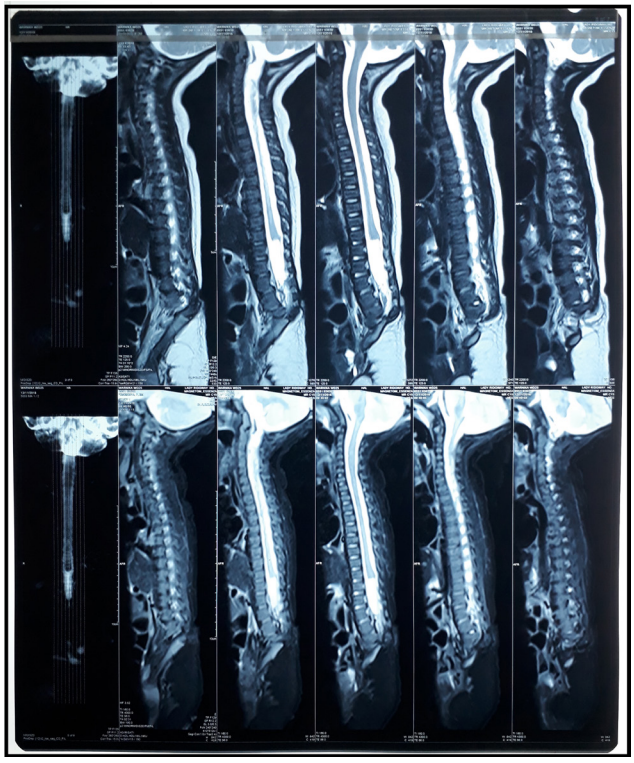

Figure2: MRI of spine

Discussion

Sacral agenesis is a rare disease. It occurs during the 3rd to 7th week of gestational period. It is due to low blood supply to lower limb during the development**.** It is due to maternal diabetics genetic factor and some teratogens are the known risk factors. but our child had only gestational diabetics which needed diet control only. They can present varies symptoms and signs depends on involvement. Increase frequency frequent stool passage lower limb abnormalities are the symptoms. Buttock dimpling postural abnormality such as sitting Buddha position limb and joint contractions are the sign.(1) Gastrointestinal disorder such as difficulty in bowel control, genitourinary disorder such as neurogenic bladder renal ectopic fused ureters and agenesis of one or both kidneys, lower extremities disorder such as knee joint contractions and Varus abnormalities, congenital cardiovascular anomalies respiratory complications and progressive complications are the known associations.(2)

Sacral agenesis is classified into 4 types. They are,

- Partial or total unilateral sacral agenesis.
- Partial sacral anomaly with bilateral symmetrical defect.
- Ilium articulating with the sides of the lowest vertebra present.
- Caudal end palate of vertebra resting above fused ilia or an iliac amphiarthrosis.

Diagnosis can be made by prenatally with MRI and USS, but diagnosis in early trimester is difficult due to incomplete sacral agenesis. (3) But short crown lump length is a clue. MRI of spine and x-rays will help to diagnose after birth. Identification of gene mutation is the definitive diagnosis.

We were unable to do the mutated gene. They can have club foot hip and knee contraction. Treatment involved spinal stabilization and correction of contractions by orthopaedic team. Limb Physiotherapy and rehabilitation has an important role. Urological and nephrological support will help to postpone renal complication. Cardiologist and Gastroenterologist will help to Continues Parental counseling and family support is most precise role in management find out the associated anomalies. (4)

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