Case Report

Caudal regression syndrome: A rare case report

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Abstract
Caudal Regression Syndrome (CRS) is resultant to anomalous development of caudal spinal cord and vertebral column segments during embryonic life. Although exact cause is not known multiple factors such as toxic, ischemic, infectious insults before 4th week of gestation are leading causes. CRS is frequently associated with maternal diabetes.

Keyword: Caudal Regression Syndrome

Introduction

Caudal Regression Syndrome (CRS) is a rare sporadic disorder occurring due to anomalous development of caudal ridge mesoderm resulting in absence of caudal cord and vertebral elements thus variable neurological affection of lower limbs and bladder bowel involvement. Associated anomalies of various system are frequently present. CRS is frequently associated with maternal diabetes. Prenatal ultrasound can be used for screening whereas MRI of spine is used in children and adults for diagnosis. Here we present a rare case report of CRS in 2year old female child.

Case report

A 2-year-old female child presented to us with complaints of dribbling of urine and fecal incontinence since birth. She was an offspring of a non-consanguineous marriage, born by normal vaginal delivery at home by trained birth attendant. No prenatal ultrasounds were done. She cried at birth and no obvious anomalies were noted at birth. Her mother was a known diabetic. On examination she was active and alert in satisfactory general condition and has passed normal developmental milestone as per her age. She had small buttocks with flat perineum and loss of lumbar lordosis, weak anal wink reflex, no anal stenosis was found. Surprisingly she was able to walk on her own and sensation over bilateral lower limbs were found to be normal. Other systems were clinically normal.

X-ray of lower segment indicated absence of all sacral vertebrae. Ultrasound abdomen didn't reveal any abnormality. Magnetic Resonance Imaging of spine revealed S1 hemivertebrae with absent rest of sacrococcygeal vertebrae, cord end at L1 level, no other cord related abnormalities were seen.

Prognosis and natural course of disease were explained to the parents, they denied any surgical intervention. Perineal muscle strengthening exercises and regular follow-up was advised, the child is under regular follow up but parents have refused any further investigation or intervention.

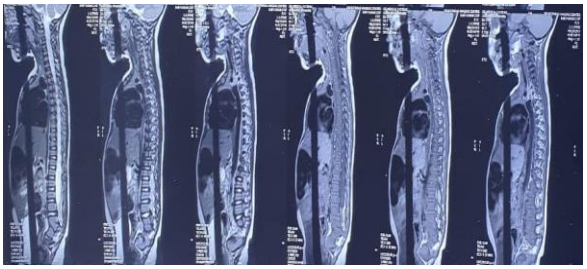

Fig1. Sagittal section MRI showing absence of caudal sacrococcygeal vertebral segments, cord ending at level

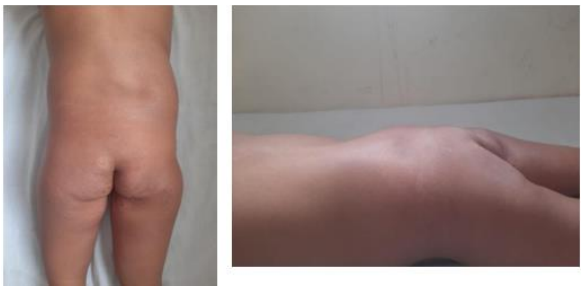

Fig 2. Clinical pictures of patient depicting flat buttocks, loss of lumbar lordosis, nappy rashes due to chronic fecal incontinence

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Discussion

Caudal Regression Syndrome (CRS) is resultant to anomalous development of caudal spinal cord and vertebral column segments during embryonic life. Although exact cause is not known multiple factors such as toxic, ischemic, infectious insults before 4th week of gestation are leading cause[1].

There is anomalous development of caudal vertebrae, spinal cord, urogenital, gastrointestinal organs, and hind limbs variably all of these structures originate from caudal eminence mesoderm[2].

the most severe clinical form of disorder is sirenomelia or mermaid syndrome, in this the lower limbs are fused together[3]. CRS is a rare sporadic disorder with incidence of about 0.1 to 0.4 per 10,000 live births[4]. Estimated sex ratio male: female is about 3:1[5]. 20-60% cases of CRS are associated with maternal diabetes as in our case, maternal diabetes increases risk of CRS by 200 folds[6]. Other than diabetes many drugs are found to induce CRS, in a study Retinoic acid in high dosage induced CRS in mice fetuses[7]. Other drugs such as Minoxidil and trimethoprim-sulphamethoxazole are found to be teratogenic[8].

Associated Anomalies in CRS[9]

Primary structural defect	Secondary effects
Agenesis/incomplete development of sacral /lumbar/thoracic vertebrae	Disruption of the appropriate length of distal spinal cord; flat buttocks, short natal cleft, and dimples in the buttocks
Spinal cord disruption	Neurological deficits affecting lower limbs (sensory and motor) and bladder and bowel continence; severely hypoplastic lower segment
Lack of movement in lower limbs	Lack of growth in affected parts; flexion and abduction at the hips; popliteal webs
Musculoskeletal system	Equinovarus and calcaneo-varus, flexion contractures of hips and knees, hip dislocation, pelvic deformity, absent fibula, scoliosis, kyphoscoliosis, absence of ribs, bifid/fused ribs, syndactyly, polydactyly, hypoplastic/absent radii, Pierre-Robin syndrome
Gastrointestinal system	Anorectal malformation, tracheoesophageal fistula, abdominal wall defects, malrotation of gut, duodenal/ colonic atresia, inguinal hernia
Genitourinary system	Renal agenesis and dysplasia, nonspecific hydronephrosis, dilated ureters, ectopic ureters, vesicoureteral reflux, fused kidneys, absent bladder, vesical/cloacal exstrophy, rectovaginal and rectourethral fistulae, transposition of external genitalia, hypospadias, urethral atresia, Mullerian duct agenesis

Renshaw classified the spectrum of CRS into 5 types based on type of defect and articulation between bones. Type 1 has total or partial unilateral sacral agenesis. Type II has variable lumbar and total sacral agenesis and the ilia articulates with the sides of the lowest vertebra. Type III: has variable lumbar and total sacral agenesis and the caudal end plate of the lowest vertebra rests above fused ilia or an iliac amphiarthrosis. Type IV: has fusion of soft tissues in both lower limbs; and type V, also known as sirenomelia, has fused bones of lower limbs

Diagnosis

Prenatal screening with ultrasonography is possible, in 1st trimester caudal regression will lead to reduced cranio-caudal ratio, which can be further confirmed in 2nd trimester by non-visualisation of sacrum and/or lumbar vertebrae with abrupt interruption of the image of the rachis more or less extended during the study of the spine. The lower limbs, generally immobile, present a characteristic position: in "frog", characteristic[10]. Foetal MRI makes it possible to confirm the diagnosis and to determine the level of the terminal medullary cone, which will be a major prognostic factor.

Treatment options

The treatment is challenging as the pathology is irreversible a multimodality team-based approach with paediatrician urologist paediatric surgeon orthopaedic surgeon is usually needed. Usually, orthopaedic deformities bladder bowel incontinence and renal functions requires specialist attention to improve the quality of life of affected individual.

Prognosis

Prognosis is fair, most patients survive and lead a normal life except for neurological deficits and bladder bowel affections. However, bladder affection may lead to chronic renal disease and its complications, thus long-term urological intervention is needed in such patients.

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