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Original Research Paper

Paediatrics

CAUDAL REGRESSION SYNDROME-A CASE REPORT

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ABSTRACT

BACKGROUND: Caudal Regression Syndrome(CRS) is a spectrum of congenital malformation which consists of anomalies of the rectum ,genitourinary system,lumbosacral spine and the lower limbs.

CASE CHARACTERISTICS: A 5yrs 8months female child with sacral agenesis,hypoplasia of lower limb with neurogenic bladder with chronic kidney disease(CKD).

OBSERVATION: CRS usually present with variety of other associated anomalies.therefore thorough examination should be done to rule out other anomalies

MESSAGE: Proper diagnosis helps in therapeutic and may improve the outcome. CIC helps in prevention or delay in occurrence of CKD.

KEYWORDS : Caudal Regression Syndrome, Chronic Kidney Disease, Continous Intermittent Catheterization.

INTRODUCTION:

CRS is a rare congenital anomaly characterized by distal vertebral malformations, usually co-existing with spinal cord malformations, neurological deficit in lower limb & neurogenic bladder.

The incidence is 1 in 40,000-60,000 live births. The syndrome occurs more frequently in offsprings of diabetic than nondiabetic mothers[1].The Incidence is about 1 in 350 infants of diabetic mothers which is about 200fold increase in diabetic patients than in general population[2].

CRS may range from absent coccyx as an isolated finding without neurological sequelae, to sacral or lumbosacral agenesis[2]. It can affect the lower extremities, the lumbar and coccygeal vertebrae and corresponding segment of the spinal cord. The neurological orthopedic, gastrointestinal, genitourinary and cardiac anomalies,imperforate anus malformed genitalia and renal dysplasia/aplasia and congenital heart defects are commonly seen[3].

CASE REPORT:

A 5yrs 8months old girl, the only child of non-consanguinous healthy parents. According to the mother's report, she had gestational diabetics and TIFFA scan was not done. Antenatal USG'S were done and were told normal to parents. The child was delivered preterm with 1kg birth weight. Postnatal examination showed hypoplastic lower limb with abducted hips with hypoplastic gluteal muscles.

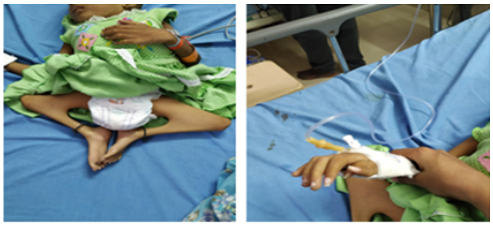

FIG 1A: showing picture of baby with Caudal Regression Syndrome

FIG 1B :showing Polydactyly

X-Ray &MRI of the lumbosacral region suggests of CRS/sacral agenesis. Neuro sonogram was normal; also had b/l contractures of knee with right femur fracture.

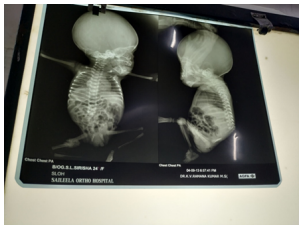

FIG 2: showing x-ray of baby with Caudal Regression Syndrome

USG abdomen revealed right pelvic kidney with b/l mild hydro uretro nephrosis, TFT's Normal, and all other routine blood investigations were normal.

At the age of 4yrs,due to acute pyelonephritis, the child was taken to higher center and evaluated. USG abdomen suggestive of right pelvic kidney,b/l hydrouretronephrosis, trabeculations & diverticular & post-void residue in urinary bladder indicative of neurogenic bladder.MCUG was done suggestive of neurogenic bladder with b/l VUR.

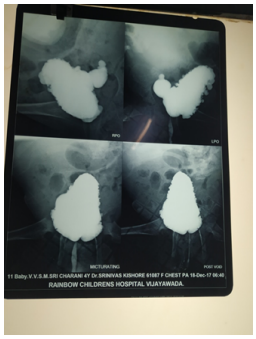

FIG 3: showing Micturating Cystourethrogram

Routine blood investigations were done, and renal function tests were slightly above the baseline.

Antibacterial prophylaxis with Cefixime was started, and CIC was taught to the parents and was advised to do it regularly. But the child had lost to follow up for the next two years and at the age of 5yrs 8months she was referred to our hospital because of deterioration of renal function.
USG abdomen and pelvis showed progression of b/l hydronephrosis and dilatation of ureters.

TREATMENT:
Thorough disease history taken from his mother revealed self-discontinuation of therapy at least 1 ½ years ago. Therefore therapy was restarted and diagnosis of CRS with CKD with anemia with neurogenic bladder was made.

The child was kept on i.v antibiotics and nephrologist and urologist consultation was taken, and inj Ceftriaxone, inj sodium bicarbonate,inj erythropoietin 2000IU s.c every weekly and tab amlodipine and continuous bladder drainage were advised.

Currently, the girl is under ambulatory nephrological care and her CKD is stable.

DISCUSSION:
CRS is a result of neural tube defects that appear at an early embryonic stage. Its etiopathogenesis is still unclear. Several etiological factors were suggested including genetic predisposition vascular hypoperfusion Vitamin A intoxication exposure to organic fat solvents, radiation, use of lithium salts, amphetamine or alcohol[4]. The incidence of CRS is significantly higher in children of diabetic mothers[1,3,4].

In some patients with CRS, specific musculoskeletal manifestations may be observed including shortening of lower limbs due to lack of their growth, flexed and abducted hips, flexion contractures of hips, knees and pelvic deformity. In those patients, CRS may be suspected prenatally during routine USG examination, but it should be emphasized that the final diagnosis can be made exclusively by performing MRI of the lumbosacral spinal cord.

In the majority of the patients with CRS, different forms of lower urinary tract neurogenic dysfunctions were observed including neurogenic bladder, lack of voluntary sphincter control. Some of them cannot be recognized before toilet training.

Patients who are suffering from CRS seem to represent the population at high risk of CKD development so that urological and nephrological examinations, including a urodynamic study are necessary for all children with CRS for identifying urinary system disorders.

CKD may be prevented or delayed in patients with CRS by the adequate treatment that usually includes anticholinergic agents, alpha-adrenergic antagonists, antibacterial prophylaxis and CIC[1,5].

The course of the disease in our patient showed importance of such therapy. Its discontinuation leads to a progression of b/l hydro uretronephrosis and significant lowering of eGFR.

In conclusion, there is no doubt that proper diagnosis helps in therapeutic decisions and may improve the outcome.

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