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Currarino Syndrome – Autopsy Features of a Rare Case

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Abstract: Currarino syndrome is a triad of congenital malformations of the hindgut characterised by presacral mass, sacral bone agenesis and anorectal malformation. HLXBg gene has been identified as the major causative gene in Currarino syndrome. The condition is very rare and has a familial predisposition with an autosomal dominant inheritance .We report autopsy features of Currarino syndrome in an 18 weeks fetus, to highlight its clinical features and to provide accurate information for further genetic counselling. Early diagnosis and multidisciplinary approach are essential which help to reduce the morbidity and mortality related to Currarino syndrome.
Keywords: Currarino syndrome, sacral agenesis, presacral mass, anorectal malformation.

INTRODUCTION

Currarino syndrome is an extremely rare condition characterised by presacral mass, agenesis of sacral bone and anorectal malformation. Sacrococcygeal bone defect is always a component of this syndrome. The syndrome is in complete form when associated with all of these three defects, while in presence of one or two components it is referred as incomplete form.The condition represents a familial predisposition with an autosomal dominant inheritance [1].

Urogenital anomalies are frequently associated with this syndrome [2]. We present autopsy features of a rare case of Currarino syndrome to raise the awareness about this syndrome.

CASE REPORT

A 26 years old lady presented to antenatal clinic with 18 weeks pregnancy. She was a third gravid. Her first pregnancy resulted in spontaneous abortion with congenital malformations. The second pregnancy was without any complications. There was no history of consanguineous marriage and no history of Diabetes Mellitus. Her antenatal ultrasonography revealed a single intrauterine fetus of average gestational age 18 weeks with absent cardiac activity and evidence of cystic lesion in the presacral region ? Sacrococcygeal teratoma (fig-1) .

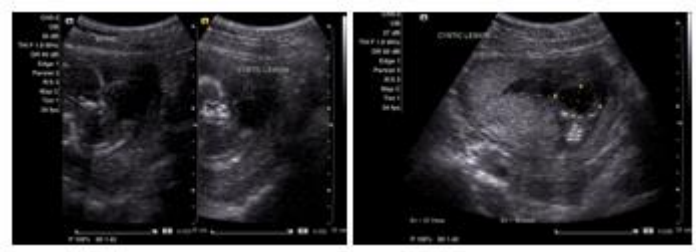

Fig-1: Antenatal USG – Cystic lesion in presacral area

Her complete blood count and urine examination were normal and seromarkers for HIV, HBsAg and VDRL were nonreactive. The termination of pregnancy was done as the baby was dead and the fetus was sent for neonatal autopsy.

AUTOPSY FINDINGS

A premature fetus with weight of 180 grams was received for autopsy. External examination revealed distention of lower abdominal wall (fig 2). There was imperforate anus and external genitalia were not formed (fig 3). In situ examination of thoracic cavity revealed situssolitus , while abdominal cavity

revealed complete agenesis of sacral bone , with a huge cyst measuring 12 x 8 cm at the presacral region. There was agenesis of rectum and anus (fig 4). There was cystic renal dysplasia. External surface and cut surface of both kidneys revealed multiple cysts, the microscopy of which showed premature glomeruli, multiple cysts lined by flat epithelium. Interstitium showed undifferentiated mesenchyme (fig 5). There was agenesis of bladder as well as of external and internal genital organs.

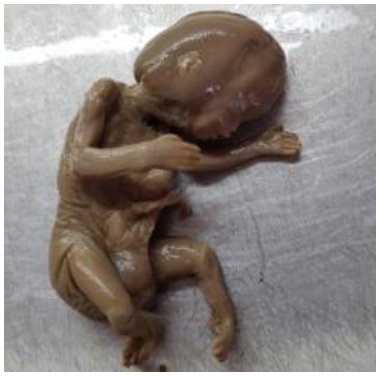

Fig 2:-External examination–Distention of lower abdominal wall

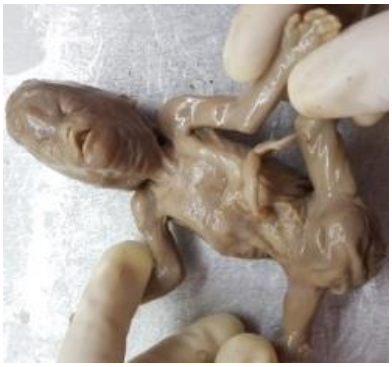

Fig 3:- Imperforate anus, absent external genitalia

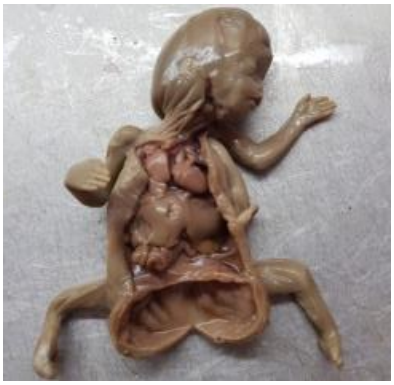

Fig 4:- Sacral agenesis with enteric cyst

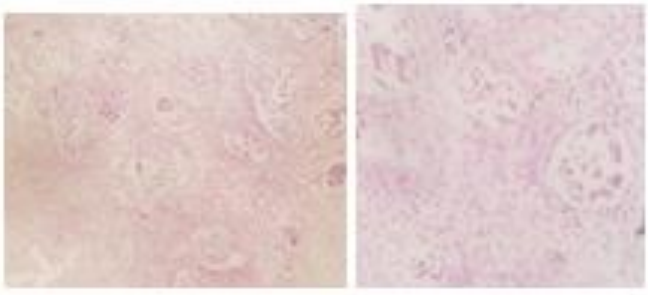

Fig 5:- Cystic renal dysplasia – microscopic features (100X HE)

Considering all these features, the condition was diagnosed as a case of Currarino syndrome with its complete form.

DISCUSSION

Currarino syndrome classically consists of a triad of sacrococcygeal bone defect, presacral mass and anorectal malformation. The complete form of this syndrome reveals all of these three defects. The syndrome is a rare entity with reported incidence as 1 to 9 cases per 1, 00,000 populations [3, 4]. About two thirds of the reported cases have familial inheritance with an equal gender distribution, while female predominance is seen in sporadic cases [5-8]. The genetic defect has been traced to the HL XBg gene on chromosome 7q36 [4]. Presacral mass may consist of a teratoma, a hamartoma, enteric cyst, anterior meningocele or a combination of these [8]. Sacral agenesis is defined as partial or complete absence of sacrum. Isolated sacral agenesis can be seen as a consequence of diabetic embryopathy which is referred as caudal regression syndrome [9, 10]. The other syndromes with sacral agenesis are VATER and OEIS syndrome. Maternal diabetes was not seen in our case. The gender of the fetus could not be determined as external and internal genital organs were not formed.

Our case revealed complete form of Currarino syndrome as there was sacral agenesis, enteric cyst in the presacral region and anorectal agenesis. The syndrome may be associated with genitourinary defects and rectovaginal fistula [4, 7, 11]. This case had cystic renal dysplasia and absence of genital organs.

Most cases are asymptomatic. Presacral mass results in symptoms such as constipation, urinary incontinence, sacral anaesthesia, paraesthesia of lower extremities, and disturbance of control of anal sphincter, recurrent urinary tract infections, nausea, headache and lumbar pain [11].

Radiological imaging modalities like prenatal ultrasonography, computed tomography and MRI play a vital role for making the diagnosis of Currarino syndrome. But in our case antenatal USG could not help to arrive a definite diagnosis and the definite diagnosis was done with neonatal autopsy. This fact highlights the importance of neonatal autopsy in making the diagnosis of such rare syndromes. In all cases surgical management is needed [12]. DE functioning sigmoid colostomy followed by excision of presacral mass and repair of anorectal malformation in the same or next to setting is the definitive surgical management [13]. As teratoma has a malignant potential, complete histopathological examination and follow up of these cases for recurrence is needed. All first degree relatives of the cases should be referred for pelvic X-ray. Relatives with abnormal X-ray should be referred for surgical management. In our case, the mother was adviced genetic study and counselling.

CONCLUSION

Antenatal USG plays an important role in the early diagnosis of Currrarino syndrome. However definitive diagnosis is done with neonatal autopsy , in case of fetal death. Genetic counselling as well as awareness of the hereditary nature of Currarino syndrome ensures the identification of asymptomatic heterozygotes. Prompt diagnosis and multidisciplinary approach can help to minimise the morbidity related to this syndrome.

REFRENCES

1. Markljung E, Adamovic T, Cao J, Naji H, Kaiser S, Wester T, Nordenskjold A. Novel mutations in the MNX1 gene in two families with Currarino syndrome and variable phenotype. Gene. 2012;507(1):50-53 .
2. Robbins JB, Broadwell C, Chow LC, Parry JP, Sadowski EA. Müllerian duct anomalies: embryological development, classification, and MRI assessment. Journal of Magnetic Resonance Imaging. 2015 Jan 1;41(1):1-2.
3. Monclair T, Lundar T, Smevik B, Holm I. CurrarinossyndromvedRikshospitalet 1961–2012. TidsskrNorLegeforen nr. 22 2013;133:2364–8.
4. Ross AJ, Ruiz-Perez V, Wang Y, Hagan DM, Scherer S, Lynch SA, Lindsay S, Custard E, Belloni E, Wilson DI, Wadey R. A homeobox gene, HLXB9, is the major locus for dominantly inherited sacral agenesis. Nature genetics. 1998 Dec;20(4):358.
5. Gupta R, Sharma SB, Mathur P, Goyal RB. Variants of Currarino Syndrome: Embryological Association and Review of Pertinent Literature. International Journal of Embryology. 2014 Nov 9;2014.
6. Lynch SA, Wang Y, Strachan T, Burn J, Lindsay S. Autosomal dominant sacral agenesis: currarino syndrome. J Med Genet 2000;37(8):561–6.
7. Liu Y, Sugiyama F, Yagami K, Ohkawa H. Sharing of the same embryogenic pathway in anorectal malformations and anterior sacral myelomeningocele formation. PediatSurgInt 2003;19(3):152–6.
8. Crétolle C, Zérah M, Jaubert F, Sarnacki S, Révillon Y, Lyonnet S, Nihoul-Fékété C. New clinical and therapeutic perspectives in Currarino syndrome (study of 29 cases). Journal of pediatric surgery. 2006 Jan 1;41(1):126-31.
9. Alles AJ, Sulik KK. A review of caudal dysgenesis and its pathogenesis as illustrated in an animal model. Birth defects original article series. 1993;29(1):83-102.
10. Harlow CL, Partington MD, Thieme GA. Lumbosacral agenesis: clinical characteristics, imaging, and embryogenesis. Pediatric neurosurgery. 1995;23(3):140-7.

11. Baek SK, Hwang GS, Vinci A, Jafari MD, Jafari F, Moghadamyeghaneh Z, Pigazzi A. Retrorectal tumors: a comprehensive literature review. World journal of surgery. 2016 Aug 1;40(8):2001-15.
12. Lee KS, Gower DJ, Mcwhorter JM, Albertson DA. The role of MR imaging in the diagnosis and treatment of anterior sacral meningocele: report of two cases. Journal of neurosurgery. 1988 Oct;69(4):628-31.
13. Lee SC, Chun YS, Jung SE, Park KW, Kim WK. Currarino triad: anorectal malformation, sacral bony abnormality, and presacral mass—a review of 11 cases. Journal of pediatric surgery. 1997 Jan 1;32(1):58-61.