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Currarino Syndrome in a Fetus, Infant, Child, and Adolescent: Spectrum of Clinical Presentations and Imaging Findings

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Abstract

In 1981, Currarino et al described a triad of findings that consist of partial sacral dysgenesis, presacral mass (anterior meningocele, enteric cyst, or presacral teratoma) and anorectal malformation. Currarino syndrome exhibits variable expressivity and the clinical presentation tends to vary with the age of the subject such as spinal anomaly detected in the fetus, imperforate anus in the newborn, and intractable constipation or neurologic symptoms in the infant and older child. At any age, meningitis can be the presenting symptom and imaging is required for proper investigation. Meningitis, sepsis, urinary tract infections, and, rarely, malignant transformation of a teratoma are serious potential complications. This pictorial review describes the imaging findings, clinical history, surgical interventions, and genetic background in 5 children with this syndrome who presented in our hospital in the interval of 1 year.

Résumé

En 1981, Currarino et coll. ont décrit une triade associant une dysgénésie partielle du sacrum, une masse pré-sacrée (méningocèle antérieure, kyste entérique ou tératome pré-sacré) et une malformation ano-rectale. L'expressivité et la présentation clinique du syndrome de Currarino tendent à varier selon l'âge du patient. Il peut s'agir d'une anomalie rachidienne chez le fœtus, d'une imperforation de l'anus chez le nouveau-né et d'une constipation réfractaire ou de symptômes neurologiques chez le nourrisson ou l'enfant plus âgé. À tout âge, le syndrome peut se manifester sous forme de méningite et doit faire l'objet d'une étude d'imagerie afin d'établir le diagnostic approprié. Il est par ailleurs susceptible d'entraîner des complications telles qu'une méningite, une sepsie, une infection des voies urinaires et, en de rares occasions, la transformation d'un tératome en tumeur maligne. Cet examen iconographique présente les constatations radiologiques, les dossiers cliniques, les interventions chirurgicales et les caractéristiques génétiques de cinq enfants atteints du syndrome de Currarino qui ont été suivis à notre hôpital au cours d'une période d'un an.
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Key Words: Currarino; Fetal; Imaging; Magnetic resonance imaging; Syndrome

In 1981, Currarino et al [1] described a triad that consists of partial sacral dysgenesis or hemisacrum, presacral mass (anterior meningocele, enteric cyst, or presacral teratoma) and anorectal malformation. This congenital syndrome is thought to be caused by malformation of the caudal notochord, which leads to aberrant secondary neurulation with incomplete separation of the ectodermal and endodermal layers in the developing embryo [2].

Most cases are related to an autosomal-dominant trait. In 1995, the underlying gene defect causing Currarino syndrome was localized in chromosome 7q36. Later, nearly all familial cases were found to be associated with a mutation of the gene HLXB9. Approximately 30% of sporadic cases also have HLXB9 mutation. No obvious genotype-phenotype correlation has been identified [3,4].

Currarino syndrome exhibits variable expressivity and may be diagnosed on prenatal ultrasound. It can present as an imperforate anus at birth, intractable constipation from anorectal stenosis or extrinsic compression from a presacral mass, or as acute meningitis [1,2,5−9].

This review describes the different clinical presentations, imaging findings, genetic profile, surgical management, and

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clinical outcome of 5 children with Currarino syndrome who attended our hospital in the interval of 1 year. Each patient presented at a different stage of life: a fetus (antenatal magnetic resonance [MR]), an infant, 2 siblings during their first years of life, and a teenager.

Imaging Cases

Case 1: The Fetus

The first patient is a 33 weeks gestational age fetus. A fetal MR imaging (MRI) was requested for a suspicious lumbar defect and possible presacral mass seen on prenatal ultrasound at 32 weeks. Fetal MR confirmed signs of low-lying cord, anterior sacral defect and a mass between the defect and the bladder (Figure 1, A-C). Absence of meconium in the rectum also raised the presence of an anorectal pathology.

The infant was born by normal vaginal delivery without complication and postnatal ultrasound (Figure 1, D-E) and MR confirmed the anomalies adding the finding of a didelphys uterus. A colostomy was performed at 1 month of age, release of tethered cord at 2 months of age, and anorectal malformation was corrected by sagittal anorectoplasty at 5 months of age. The closure of the colostomy was performed at 13 months of age. A double STING (subtrigonal teflon injection) procedure was performed at 33 months of age due to recurrent urinary tract infection (UTI) related to bilateral grade 2 reflux. Presacral teratoma resection was performed at 3 years of age.

Genetic consultation showed a de novo mutation secondary to HLXB9 mutation. The possibility of the parents of having a second child with this syndrome is $<1\%$. After 4 years and 8 months of follow-up, intermittent bladder catheterization is still necessary due to a neurogenic bladder and recurrent UTIs. The child is otherwise well.

Case 2: The Neonate

The second patient is a term baby, born by vaginal delivery at a community hospital after a normal pregnancy. At 9 days of life, he was brought to the emergency department for signs of dehydration. On examination, the infant had fever and bulging fontanel. Cranial and spinal ultrasound were performed and showed large ventricles with thick ependymal lining suggesting meningitis, low-lying cord and absence of normal sacrum with a presacral mass (Figure 2, A and B). MRI done on the following day confirmed these abnormalities (Figure 2C). A preoperative computed tomography scan of the pelvis was performed (Figure 2D).

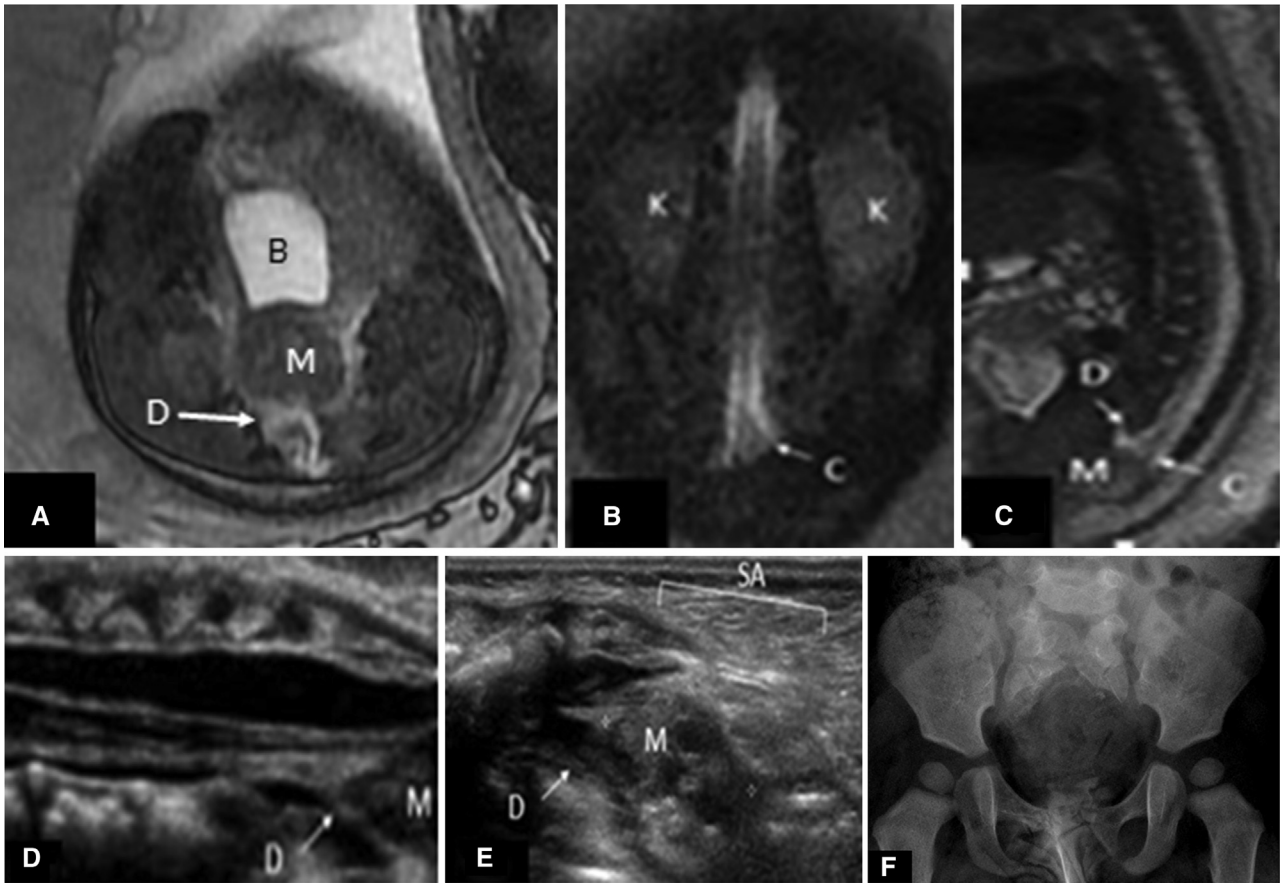

Figure 1. The fetus with prenatal ultrasound suggesting a sacral defect. Fetal magnetic resonance (A) axial steady-state free precession (SSFP), (B) coronal SSFP, (C) sagittal SSFP, (D, E) neonatal ultrasound, and (F) radiograph of the pelvis at 12 months. Fetal magnetic resonance imaging demonstrates an anterior spinal defect (D), sacral agenesis (SA), and a presacral mass (M) behind the bladder (B). The spinal cord extends below the level of the kidneys (K) with a broad-shaped conus (C) extending to the mass. Postnatal spinal ultrasound and radiography confirm these findings demonstrating fat signal within the mass suggestive of teratoma.

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