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Full Length Research Paper

Full currarino syndrome associated to elhers danlos. one patient report at the hospital para El niño poblano Mexico

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The Currarino triad described in 1981 involves the association of partial sacral agenesis with intact first sacral vertebra (sickle-shaped sacrum), a presacral mass, and anorectal malformation. Gene map locus 7q36, some cases of this syndrome are caused by mutation in the HLXB9 homeobox gene and Dominant inheritance has been observed. The patient in this study was diagnosed as Full Currarino syndrome, with the clinical features, (sacral agenesis, presacral mass, and anorectal malformation). The diagnosis is usually made in children (80%) and adults (20%). In 2007, it was found that prenatal diagnosis is possible. The patient had skin and bones articulation hyperlaxitud, where elastic fibers alteration was confirmed by skin biopsy. It has evidence of autosomal dominant inheritance. In 1995, it was associated to chromosome 7q36 and only hemisacrum is reported in 1998 by mutations in the HLXB9 gene. In 1994, it was described as association with partial trisomy of 13q and 20p. The caryotipe of the patient in this study was normal 46 X.

Key words: Sacral agenesis, presacral mass, teratoma, anorectal malformation, homeobox genes, chromosome.

INTRODUCTION

The Currarino triad involves the association of partial sacral agenesis with intact first sacral vertebra (sickle-shaped sacrum), a presacral mass, and anorectal malformation reported by Currarino et al., 1981. The specific sacral anomaly is distinct to this syndrome. O'Riordain et al. (1991) reported an affected family, where out of ten affected members, only four were symptomatic, three of these had the full Currarino syndrome (sacral agenesis, presacral mass, and anorectal malformation), and the fourth with an anterior meningocele with no anorectal alterations. However all the affected members had clinical and X-ray partial sacral agenesis. Ascending infection resulting in E. coli bacterial meningitis has also been reported. Associated urinary and genital malformations have been observed including rectovaginal fistula, du-plex ureter, hydronephrosis, vesicoureteral reflux, bicornuate uterus, and neurogenic bladder.

Malignant degeneration of the presacral teratoma has also been reported by Ashcraft and Holder (1974) and Yates et al. (1983). Belloni et al. (2000) and Cretolle et al. (2000) presented evidence that of the 5 specific categories

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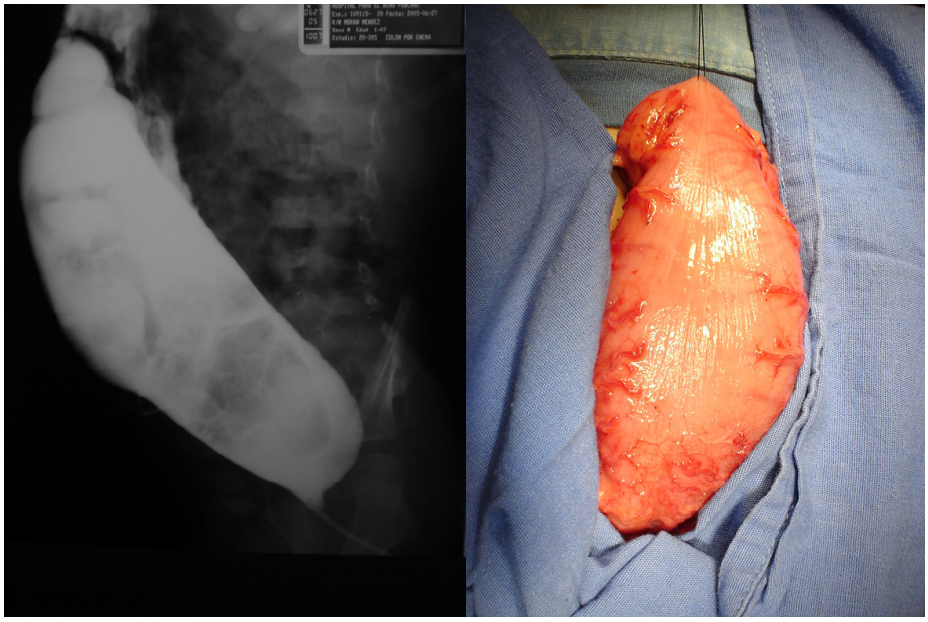

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Figure 1. Mega-rectosigmoides **A.** Gatroenterological study and **B.** surgery shows the dilated sigmoid colon.

of sacrococcygeal anomalies as Kalitzki (1965), Pang (1993) and Capra et al. (1996) also did only hemisacrum is caused by mutations in the HLXB9 gene. Lynch et al. (2000) provided a review of the clinical features and molecular basis of Currarino syndrome (Kim et al., 2007) reported two families with Currarino syndrome. The proband in the first family was a 25-year-old woman with chronic constipation, presacral meningocele, and rectal dilatation. Two of her sisters and a nephew were also affected. In the second family, a 6-month-old girl have a presacral mature cystic teratoma with an extension of the spinal canal associated with possible cord tethering to the mass, anterior bony defect of distal sacrum, and left hydroureteronephrosis. Her father had chronic constipation and an anal fistula, and her paternal grandmother had a megacolon. Where the genetic background was variable for the disease.

Genetic analysis identified two different HLXB9 mutations that segregated with the phenotype in each family; there were a total of four asymptomatic mutation carriers in the two families, indicating reduced penetrance. And genetic heterogeneity or undetected somatic mosaicism in sporadic cases has been also reported.

MATERIALS AND METHODS

We describe a two years old male patient with the clinical triada asso-ciation, he was studied at the Pediatric Hospital, the first clinical symp-toms were skin and bone articular hyperlaxitud, it was thought at the beginning to be an Elher-Danlos syndrome that was later confirmed by hystophatological biopsy. Chronic constipation, and abdominal distention, anorectal malformation Figure 1 and perianal fistula was diagnosed later.

Mega-rectosigmoides and sacral agenesis was confirmed on X-ray and IRM Figures 2 and 4. Presacral mass, immature teratoma Figure 3 at sacral region was confirmed by tumor biopsy. A normal caryotipe was reported where no chromosomal aberrations were observed, 46 XY Figure 5, normal male.

RESULTS AND DISCUSSION

The Currarino triad is a hereditary transmitted syndrome, originally defined by Currarino et al., 1981 as ASP-association, consisting of an anorectal malformation, a sacral bony defect and a presacral mass (Ashcraft and Holder, 1974).

In most cases autosomal dominant transmission is suggested. In family members one or two features of the syndrome may be missing, indicating an incomplete form of this complex However, Ashcraft and Holder reported in 1974 evidence of autosomal dominant inheritance. In 1995 it was associated to chromosome 7q36 and only hemisacrum is reported in 1998 by mutations in the HLXB9 gene. Nagai in 1994 described association with partial trisomy of 13q and 20p. The caryotipe of the patient in this study was normal 46 XY where no deletion or translocation was observed Figure 5.

A review of the literature up to 1991 revealed 48 patients with the clinical triada association. In more than 80% of cases, this complex is diagnosed in the first decade, whereas incomplete Currarino syndrome is diagnosed predominantly in adults. Most frequently the

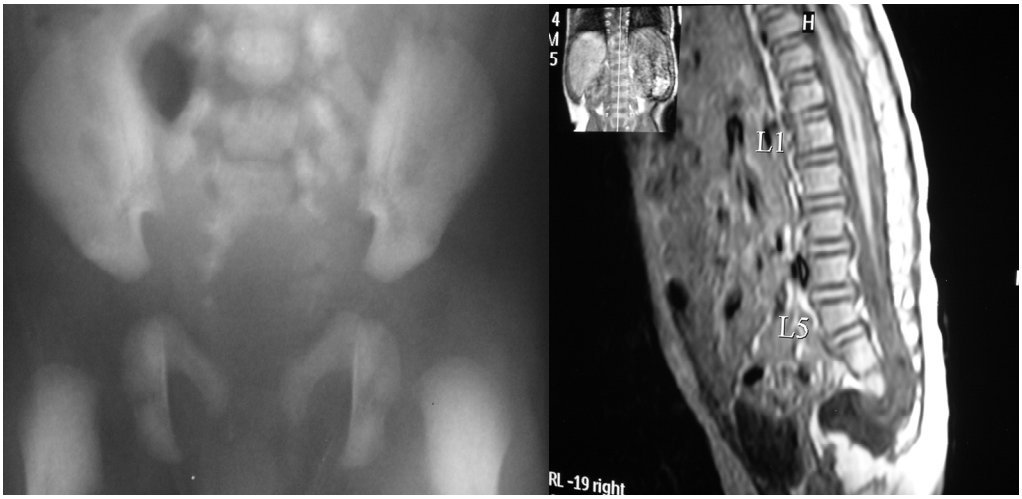

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Figure 2. Pre-sacral Teratoma. **A.** Pelvic X ray and **B.** MRI for tumor localization and clinical diagnosis. Both studies are showing a hemisacrum with a right sided defect.

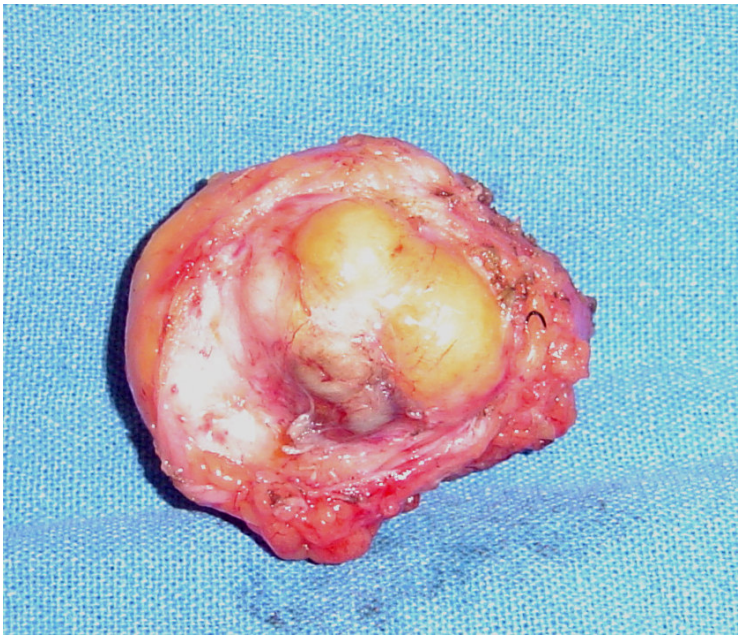

Figure 3. Presacral tumor diagnosed by histophatological studies.

presacral mass in this syndrome was reported to be an anterior meningocele (47%) and a benign teratoma (40%). The number of patients with Currarino syndrome has been underestimated so far. It has been recommend anorectal examination, pelvic ultrasound and pelvic x-rays in all patients with a history of chronic constipation since early childhood, in order to avoid diagnosis failure.

Therefore positive findings should lead to further investigations such as barium enema, MRI, myelography and family screening studies.

The patient in this study was diagnosed as Full Currarino syndrome, with the clinical features reported by Guido Currarino in 1981, (sacral agenesis Figure 4, pre-sacral mass Figures 2 and 3, and anorectal malformation Figure 1).

The diagnosis is usually made in children (80%) and adults (20%).Cretolle in 2007 reported that prenatal diagnosis is possible. The patient had as mentioned before, skin and bones articulation hyperlaxitud, where elastic fibers alteration was confirmed by skin biopsy and

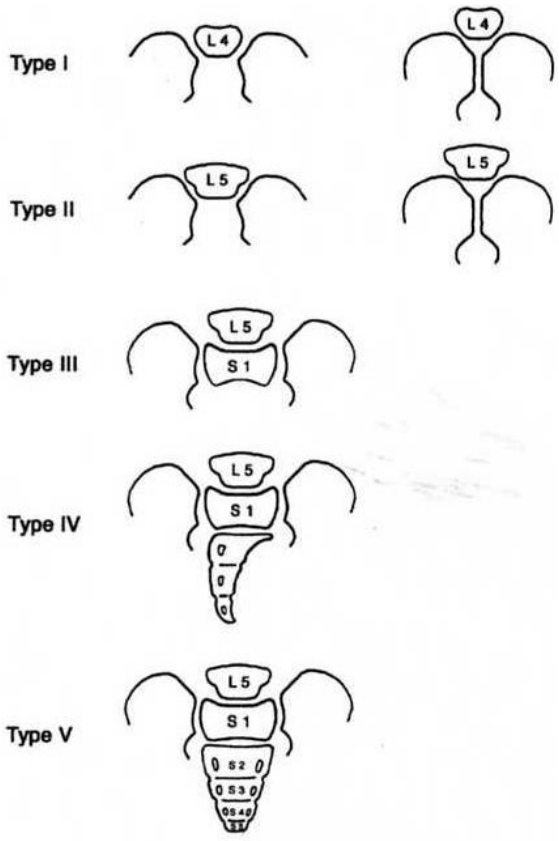

Figure 4. Sacral anomalies according to Kalitzki, 1965; Pang, 1993 and Capra et al., 1996 classification.

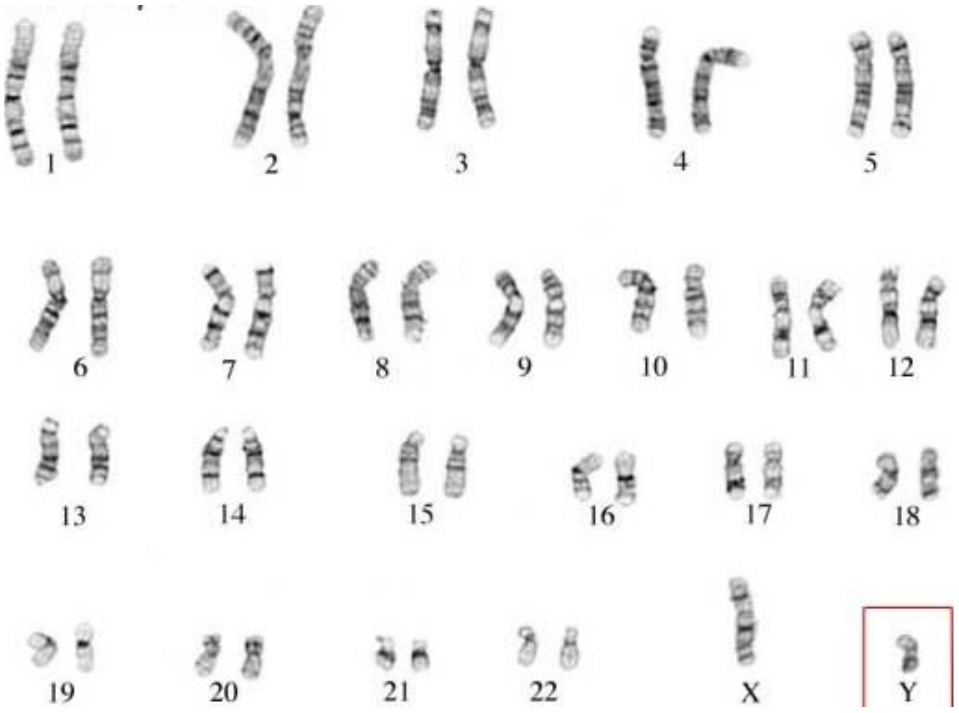

Figure 5. Normal caryotipe was reported, where no chromosomal aberrations in number or structure were observed.

diagnosed as Elhers Danlos syndrome. This association between Currarino and Elhers Danlos syndrome is a rare finding not reported at the literature.

A multidisciplinary medical study among genetics, pediatrics, pediatric surgery and neurosurgery is required to ensure adequate surgical treatment, considering both the risk of malignant degeneration as well as the risk of intraoperative nerve damage. And it is very important to have an early diagnosis for patients with the Currarino syndrome for a better treatment a quality of life.

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