

A Very Rare Cause of Anal Atresia: Currarino Syndrome

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Abstract

Currarino syndrome (triad) is an extremely rare condition characterized by presacral mass, anorectal malformation, and sacral bone deformation. The complete form of this syndrome displays all three irregularities. Herein, we report a male case who was admitted to our hospital with symptoms of urinary system infection and persistent constipation 2 years after colostomy operation performed with the indication of rectovestibular fistula and anal atresia, diagnosed as Currarino syndrome based on imaging modalities. In a patient who was admitted because of the presence of anal atresia, in order to preclude potential complications, probable concomitancy of this syndrome should not be forgotten. Early diagnosis is important for the prevention of meningitis, urinary tract infections, and malignant change.

Keywords: Currarino syndrome; Presacral mass; Anorectal malformation; Sacral bone deformation; Urinary tract infections

Introduction

Currarino syndrome (CS) is a very rarely seen condition which is termed as Currarino triad. In fact, from 1981 on, nearly 300 cases have been published. It is a hereditary pathology which is characterized by a triad of sacrococcygeal bone defect, presacral mass, and anorectal malformation. Sacrococcygeal bone defect is always a component of this syndrome. When it is associated with all of these three anomalies, it is called complete form, while in the presence of one or two components, it is named incomplete form. Our case was a rarer complete form. *MNX1* gene (motor neuron and pancreas homeobox 1, HGNC ID: 4979) mutations have been reported in all cases with familial Currarino triad. The condition demonstrates fa-

miliar predisposition, and has a autosomal dominant inheritance pattern [1]. Nearly 30% of the cases with CS are sporadic [2]. This triad which was firstly described by Currarino et al is asymptomatic in more than 33% of the affected children. Symptoms such as intractable constipation, urinary retention, incontinence, and bowel obstruction in infancy are frequently associated with this presentation. Rarely association of Hirshsprung disease (HD) with CS has also been reported [3]. It can also accompany urogenital anomalies. Indeed, in more than 15% of the cases with Currarino triad, concomitant Mullerian duct anomalies are detected. General incidence of Mullerian duct anomalies has been reported as 7% [4]. Neonatal diagnosis is rarely established.

Cases with sacral agenesis should be investigated as for presacral masses and anorectal changes [5]. For cases with suspect CS, multidisciplinary approach at an early stage is important [6]. In the presence of anal atresia, prevention of potential complications is possible with raising awareness about this syndrome.

Case Report

A 2-year-old male infant was admitted to our hospital with inability to urinate and intractable constipation. From his medical history, it was learnt that 24 h after his birth, he had undergone colostomy operation with the indication of anal atresia and rectovestibular fistula. On physical examination, any abnormal finding apart from abdominal distension was not detected. Complete urinalysis revealed abundant leukocytes and erythrocytes in urine. The plain abdominal X-ray performed in the standing position demonstrated a defect at the right lower side of the sacrum, and a dysplastic image displayed deviation to the left (scimitar sacrum) (Fig. 1). On abdominopelvic ultrasound, a multilocular septated cystic lesion measuring 5.7 × 6 cm in the presacral area was detected (Fig. 2). On abdominal tomogram, hypoplastic appearance of the right sides of S3, and vertebrae inferior to S3, leftward deviation of sacrum (scimitar sacrum), and a presacral cystic lesion were detected (Fig. 3). On lumbosacral magnetic resonance imaging (MRI), widening of the right S3-S4 and S4-S5 neural foramina, and a multilocular, septated anterior sacral meningocele (ASM) which continued with sacral spinal canal anteriorly displaced bladder, and rectum were observed (Fig. 4).

We performed posterior sagittal anorectoplasty (PSARP). The patient was also operated in the Clinics of Neurosurgery with the indication of ASM.

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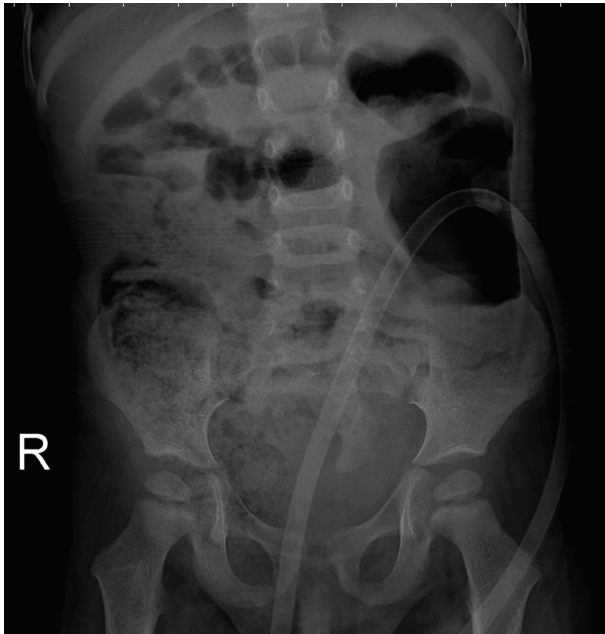


Figure 1. An abdominal radiograph: a defect at the right lower side of the sacrum, and a dysplastic image displaying deviation to the left (scimitar sacrum).

Discussion

This syndrome which was named after scientist Guldo Currarino who first discovered this triad is characterized by a sacral bone defect, a congenital hindgut anomaly, and a pre-sacral tumor. This syndrome has been reported to stem from the HLXB9 gene mutations on the chromosome 7. Therefore,

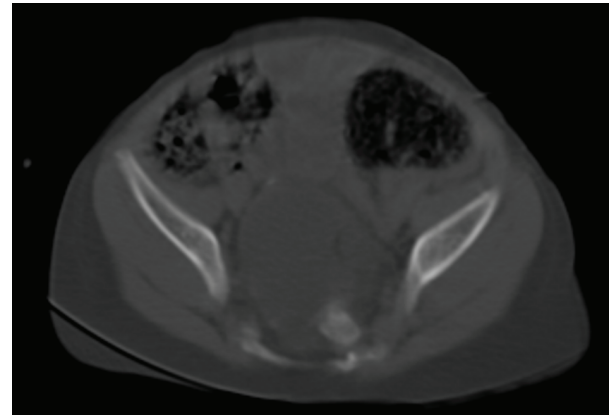


Figure 3. Abdominal tomogram: hypoplastic appearance of the right sides of S3, and vertebrae inferior to S3, leftward deviation of sacrum (scimitar sacrum), and a presacral cystic lesion.

presacral masses and sacral agenesis w/o anorectal anomalies are suggestive of possible familial CS [5].

Presacral mass which accompanies CS may consist of a teratoma, a hamartoma, a neuroenteric cyst, anterior meningocele or a combination of these four entities [7]. Sacral agenesis is defined as partial or complete congenital absence of sacrum. Most cases are asymptomatic. It has been reported that presacral mass results in symptoms such as intractable constipation, urinary incontinence, sacral anesthesia, paraesthesia of the lower extremities, disturbance of anal sphincter control, etc. Among them, constipation is the most commonly reported symptom [8]. Other symptoms are related to recurrent urinary tract infections, nausea, headache, and lumbar pain [8]. In our case, clinical symptoms of urinary infection were associated with constipation. Though rarely its association with



Figure 2. An ultrasound examination revealed a multilocular septated cystic lesion measuring 5.7 × 6 cm in the presacral area.

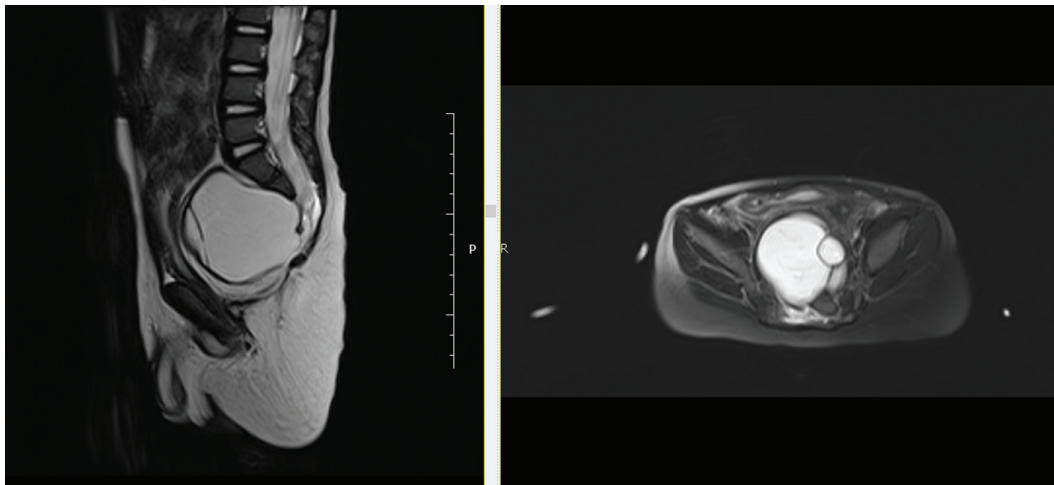


Figure 4. Lumbosacral MRI: widening of the right S3-S4 and S4-S5 neural foramina, and a multilocular, septated anterior sacral meningocele (ASM) which continued with sacral spinal canal anteriorly displaced bladder, and rectum were observed.

HD has been reported [9]. Still, presence of CS was reported in an adult case with chronic anal fistula [6]. In the diagnosis and follow-up of CS, radiological imaging modalities such as ultrasonography (US), computed tomography (CT), and MRI play a vital role [10].

Kassir et al reported MRI as a specific and sensitive non-invasive diagnostic tool in cases with anorectal malformation [6]. MRI can be used reliably in the diagnosis of this syndrome, associated mass lesions, and other pathologies, and also during post-treatment follow-up period of cases with CS [10].

Regardless of its anatomical characteristics, presacral mass should be completely removed during surgical resection. In our case, anterior meningocele was detected, and sacral laminectomy was applied through retroperitoneal approach. In these cases, in order to avoid serious neurological complications, the connection between spinal canal and tumor should not be overlooked. Though rarely, since its malignant transformation into teratoma has been reported, multidisciplinary approach is very important in cases with CS which can result in morbidities and mortality.

Conclusion

Anal atresia can be one of the components of Currarino triad. In a patient who was admitted because of the presence of anal atresia, in order to preclude potential complications, probable concomitancy of this syndrome should not be forgotten.

Source of Support

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Conflict of Interest

None.

Funding

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Author Contributions

Sevgi Buyukbese Sarsu has contributed to writing this paper. Sevgi Buyukbese Sarsu, Mehmet Ergun Parmaksiz, Esra Cabalar, Ali Karapur and Cihat Kaya have contributed to data collections.

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