# The Currarino Syndrome: A Case Report of an Uncommon Presentation: Differential Diagnosis

## Farklı Klinik Özellikler Gösteren Currarino Sendromu Olgusu

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**ABSTRACT** The Currarino triad is a complex anomaly consisting of an anorectal malformation, a sacral bone defect and a presacral mass. It was first described in 1981 and approximately 250 cases have been reported to date. We report a case of Currarino syndrome in a 12-year-old girl. The diagnosis was established with anal atresia with rectovestibular fistula, sacral bone hypoplasia, infrasacral midline mass (tricholemmal cyst), spina bifida, and agenesis of the right 12th rib. This case was reported to contribute to the literature since the case contains rare findings (costal agenesis, spina bifida and tricholemmal cyst) in addition to the known features of the syndrome.

**Key Words:** Anus, imperforate; sacrococcygeal region; congenital abnormalities

**ÖZET** Currarino triadı, anorektal malformasyon, sakral kemik defekti ve presakral kitleden oluşan bir kompleks anomalidir. İlk defa 1981 yılında tanımlanmış ve günümüze kadar yaklaşık 250 olgu bildirilmiştir. Bu makalede 12 yaşında Currarino sendromu tanısı alan bir kız olgu sunulmaktadır. Tanı rektovestibüler fistüllü anal atrezi, sakrum hipoplazisi, infrasakral kitle (trikolemmal kist), spina bifida ve sağ 12. kot agenezi ile konmuştur. Bu olgu sendromun bilinen özelliklerine ek olarak seyrek rastlanan bu bulguların vurgulanması amacıyla olgu sunulmuştur.

**Anahtar Kelimeler:** İmperfore anüs; sakrokoksigeal anomali; konjenital anomali

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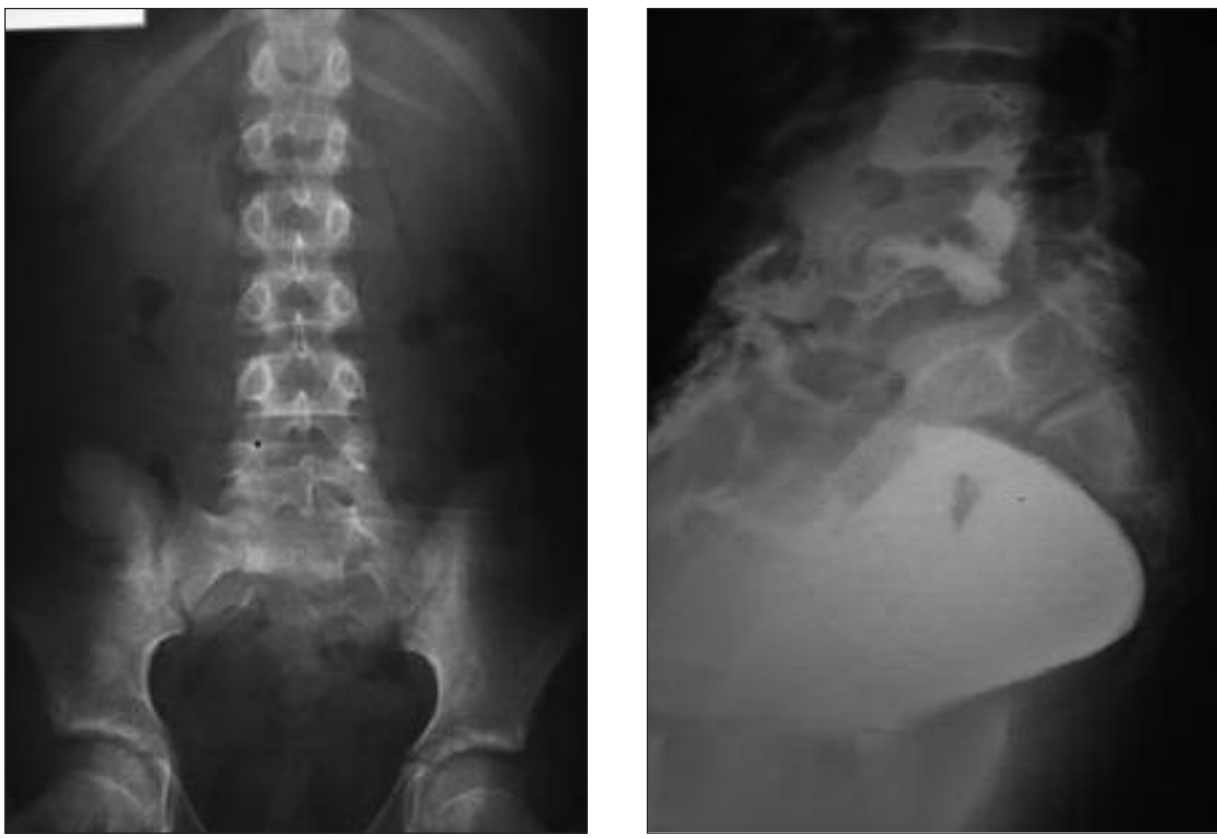
The Currarino syndrome is a relatively rare inherited disorder and consists of an anorectal malformation, sacral bone anomalies and a presacral mass.[1] The syndrome may be complete if all 3 anomalies are present, or incomplete if the triad is associated with only 1 or 2 defects.[2,3] A positive family history is supportive of the diagnosis; however, if the family history is negative for the Currarino triad, the diagnosis cannot be excluded.[4] Any variation of a sacral bony defect may confirm the diagnosis. We reported a case of Currarino syndrome in a 12-year-old girl. The diagnosis was established with anal atresia with rectovestibular fistula, sacral bone hypoplasia, infrasacral midline mass (tricholemmal cyst), spina bifida, and agenesis of the right 12th rib. This case was reported to contribute to the literature since the patient had rare features in addition to the known components of the syndrome.

## CASE REPORT

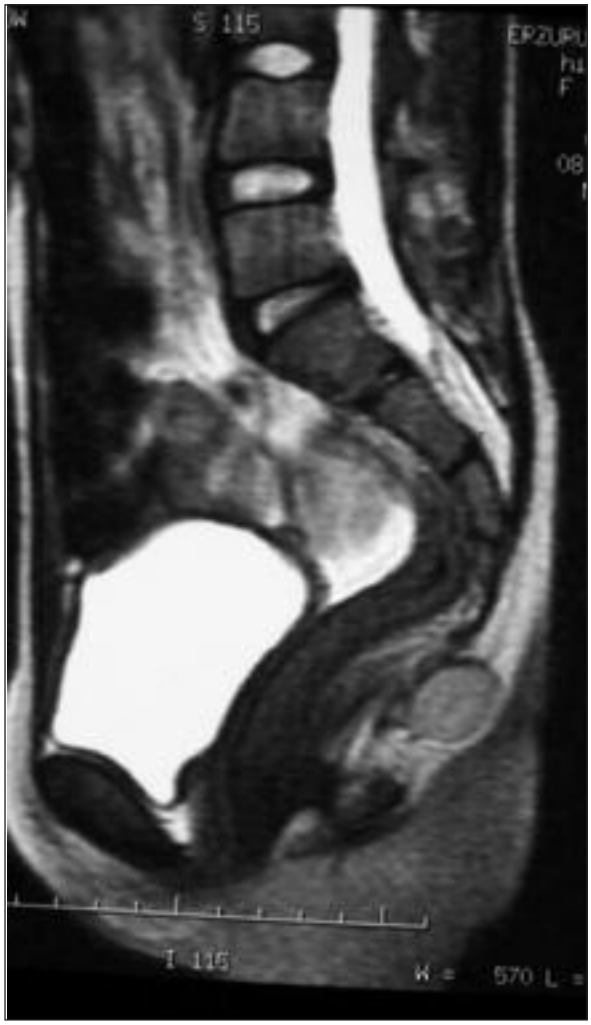
A 12-year-old girl presented to the outpatient clinic of pediatric surgery with rectovestibular fistula. The patient had received the diagnosis during the newborn period. The vestibule fistula was dilated at that time; she occasionally had constipation, defecation from the fistula and urinary tract infection once. The definitive treatment of the patient had been postponed untill that age by the parents since the patient had no major complaint and was from a family with low socioeconomic level. The physical examination revealedand a partially mobile 3x2 cm infrasacral mass bimanually palpable. The patient and her family were not aware of the mass. The laboratory results of the case were normal and the diagnosis of Currarino syndrome was made. On the abdominopelvic roentgenograms, skeletal anomalies were present such as the absence of 12$^{th}$ rib on the right, spina bifida at the 1$^{st}$ sacral vertebra, and agenesis of the 4$^{th}$ sacral vertebrae and the coccyx (Figure 1). Retrograde non-ionic contrast medium was administration of for colonic dilatation; anal canal and rectum were visualized and were evaluated as normal. In addition to the above described pathological findings an isolated, well-circumscribed 2 x 2 x 2 cm mass of cystic nature was detected in MRI examination at the midline inferior to sacral vertebrae and posterior to anorectal junction in the fatty tissue which was isointense with the muscle tissue in T1 weighted sequences, hyperintense in T2 weighted sequences and non-supressed in fat-supressed sequences (Figure 2). Cytogenetic analysis revealed no quantitative or structural anomaly.

The diagnosis of Currarino syndrome was made with the findings of physical and radiographic

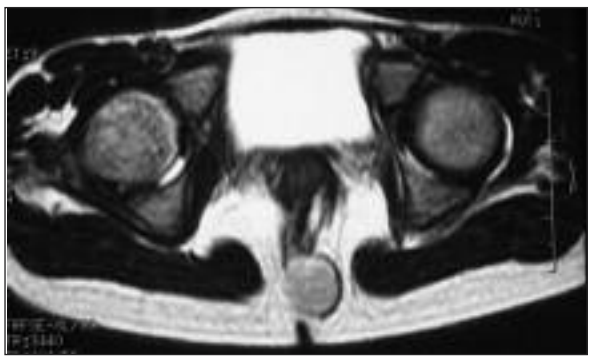


**(A)** **(B)**

**FIGURE 1: (A)** PA plain abdominal roentgenogram shows agenesis of the 12$^{th}$ rib on the right side, lumbalization and spina bifida at the 1$^{st}$ sacral vertebra. **(B)** Lateral sacral roentgenogram shows agenesis of the 4$^{th}$ sacral vertebrae and coccyx.



(A)



(B)

**FIGURE 2:** Pelvic MRI of the patient. Sagittal **(A)** and axial **(B)** SE T2 WI show agenesis of the coccyx and a well-defined, thin-walled cystic mass with a 2 cm diameter at the midline inferior to sacral vertebra and posterior to anorectal junction.

examination. The infrasacral mass was excised and posterior sagittal anorectoplasty were performed in the same session; the patient was well at one year folow-up. The histopathologic examination of the infrasacral cyst revealed a diagnosis of tricholemmal cyst of benign nature which was composed of epidermis and fibrous stroma, lacking neural elements and circumscribed with squamous epithelium.

## CONCLUSION

Currarino syndrome is an autosomal dominant disease of which the diagnosis is made by the triad of anorectal malformation, presacral mass and sacral anomaly.[1-5] The findings demonstrate differences in some cases.[2,3] The diagnosis of incomplete cases may be delayed until adulthood.[2,3] A positive family history is supportive of the diagnosis; however, if the family history is negative for the Currarino triad, the diagnosis cannot be excluded.[2,3] Hence, screening of parents and siblings of the patient revealed no symptoms or findings indicative of the syndrome and sacral x-rays of the family members were normal. The caryotype of our patient was also normal. Celia et al were not able to detect HLXB9 mutation in 5 cases including a series of 29 patients.[2]

Clinical manifestations of the Currarino syndrome are extremely variable and not all components of the classic triad need to be present. Its very first manifestation realized at birth is frequently anorectal malformation. The most frequent type of anorectal anomalies is the rectoperineal fistula. Other anal malformations include anal atresia, anorectal stenosis, anteriorly placed anus, rectal duplication, and fistulae (rectourethral, rectovaginal, rectovesicular).[1-5] Neonatal-onset bowel obstruction, chronic constipation, renal/urinary tract anomalies, gynecological anomalies, and tethered spinal cord are the common manifestations of the condition.[2-5]

The presacral mass may be frequently anterior meningocel or presacral teratoma, rarely neuroenteric cyst, dermoid-epidermoid cyst or very rarely another benign tumoral lesion (tricholemmal cyst) like in our case.[1-5] Presacral mass is not always presented in every Currarino syndrome case or it may be localized anywhere neighboring the sacrum. Although masses neighboring the sacrum

which do not produce any pressure effect may persist asymptomatically until adult ages, some cases which become overt with pressure findings and have malignant transformation have also been reported.[2,4] Therefore, Currarino syndrome should be investigated in children with anorectal malformation and/or constipation.

Different types of sacral agenesis have been described and classified into the following categories:[5]

I) Total sacral agenesis with normal or short transverse pelvic diameter and associated with agenesis of some lumbar vertebrae.

II) Total sacral agenesis without involvement of lumbar vertebrae.

III) Subtotal sacral agenesis or sacral hypodevelopment (with S1 present).

IV) Hemisacrum.

V) Coccygeal agenesis.

Type III sacral agenesis was present in our case according to this classification. In addition to sacral anomalies reported in the literature in Currarino syndrome, costal anomalies were reported very rarely. The costal anomalies were stated as costal fusion, supernumerary ribs and accessory ribs in the literature.[2,3,5] and it was characterized as unilateral costal agenesis in our case.

In conclusion, costal agenesis, spina bifida and tricholemmal cyst which we thought not to be reported previously were presented in a girl with Currarino syndrome in this article. In the diagnosis and investigation of Currarino syndrome, plain radiographs should be the first line examination method to detect sacral anomalies and other associated skeletal anomalies in the early diagnosis.

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