Presacral mature cystic teratoma associated with Currarino syndrome in an adolescent with androgen insensitivity: illustrative case

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BACKGROUND Currarino syndrome is a rare disorder that classically presents with the triad of presacral mass, anorectal malformation, and spinal dysraphism. The presacral mass is typically benign, although malignant transformation is possible. Surgical treatment of the mass and exploration and repair of associated dysraphism are indicated for diagnosis and symptom relief. There are no previous reports of Currarino syndrome in an androgen-insensitive patient.

OBSERVATIONS A 17-year-old female patient presented with lack of menarche. Physical examination and laboratory investigation identified complete androgen insensitivity. Imaging analysis revealed a presacral mass lesion, and the patient was taken to surgery for resection of the mass and spinal cord untethering. Intraoperative ultrasound revealed a fibrous stalk connecting the thecal sac to the presacral mass, which was disconnected without the need for intrathecal exploration. The presacral mass was then resected, and pathological analysis revealed a mature cystic teratoma. Postoperatively, the patient recovered without neurological or gastrointestinal sequelae.

LESSONS Diagnosis of incomplete Currarino syndrome may be difficult but can be identified via work-up of other disorders, such as androgen insensitivity. Intraoperative ultrasound is useful for surgical decision making and may obviate the need for intrathecal exploration during repair of dysraphism in the setting of Currarino syndrome.

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KEYWORDS Currarino syndrome; presacral mass; spinal dysraphism; untethering

The presacral region is complex, composed of tissue from each of the germ layers. Therefore, a variety of neoplasms, including teratomas, anterior meningoceles, lipomas, and dermoid cysts, may arise.1 Presacral masses have an incidence of 1 in 40,000 births, making them a rare pathology.2 These are typically benign lesions, with a small percentage demonstrating malignant transformation. There is a strong association with Currarino syndrome, which is a triad of presacral mass, anorectal malformation, and spinal dysraphism, although any of these findings can occur in isolation.3 Currarino syndrome has a female predominance (1.39:1) and has a familial autosomal dominant inheritance with associated MNX1 gene mutations.4 Associated mass lesions are identified on investigation of common symptoms, such as chronic constipation, sacrococcygeal/perianal pain, and urinary incontinence, but they may also be found incidentally on imaging. It is extremely rare to find a presacral mass in an adolescent or adult because most cases present in the first decade of life. In this report, we present a 17-year-old phenotypic female patient who has an XY karyotype with complete androgen insensitivity and was found to have a presacral cystic mass and sacral defect without an anorectal malformation. To our knowledge, there are no previously reported cases of an androgen-insensitive female with these findings.

Illustrative Case
A teenage female was initially evaluated at the age of 14 years for delayed menarche. She and her family elected to wait before...
pursuing further work-up. Further evaluation was pursued at the age of 17 due to continued lack of menarche. On examination, the patient was found to have a blind vaginal dimple and breast development with little pubic hair.

Laboratory evaluation revealed a 46,XY karyotype with positive SRY. Pertinent normal findings included inhibin, lactate dehydrogenase, alpha-fetoprotein, cancer antigen (CA) 125, CA 19-9, carcinoembryonic antigen, and prolactin. Abnormal laboratory findings included elevated testosterone (>500 ng/dL; normal range 9–58), elevated follicle-stimulating hormone (>14 mIU/mL; normal range 1.7–12.6), and estrogen (<20 pg/mL; normal range 21.4–259.0 pg/mL). The combination of blind vaginal pouch, phenotypic female characteristics, elevated testosterone, and XY karyotype led to the diagnosis of complete androgen insensitivity.

Ultrasound of the pelvis did not visualize a uterus, ovaries, or a cervix. The patient then underwent magnetic resonance imaging (MRI) of the pelvis without contrast, which confirmed an absent uterus and ovaries and revealed a nonenhancing, complex cystic mass in the pelvis measuring 4.5 × 6.0 × 4.0 cm in a presacral/right perirectal position (Fig. 1). Associated with this was a ventral sacral defect at S3 with fat extruding through the defect.

Currarino syndrome became a concern, based on the available evidence. However, no anorectal malformation was identified despite the presence of the sacral defect and presacral mass. The patient was evaluated by adult neurosurgery, pediatric neurosurgery, pediatric surgery, and gynecological surgery in consideration of surgical intervention due to the presence of a mass and concern for associated spinal dysraphism. Ultimately, complete neural axis imaging was obtained and showed no other abnormality. The patient was scheduled for surgery with neurosurgery and pediatric surgery with the aim of repairing the patient’s spinal dysraphism and resecting the mass during the same operation, as per previous description.5

We proceeded to the operating room for planned spinal cord untethering and mass lesion resection. The patient was positioned prone on an open Jackson table. Intraoperative fluoroscopy was used to identify pertinent anatomical landmarks, an incision was made, and the sacrum was dissected via a midline approach. A defect was noted at the inferior aspect of S3. A sacral laminectomy was completed, and ultrasound was used to identify a stalk connecting the thecal sac and the mass without evidence of fluid connectivity between the thecal sac and the cystic component of the mass (Fig. 2). Therefore, we elected to forgo intrathecal exploration for tethered cord, and instead the stalk was tagged with 2-0 silk ties and vascular clips and was sectioned, which separated the mass from the thecal sac and untethered the spinal cord. A small laminoplasty was then performed using a standard plating system. Pediatric surgery then scrubbed into the case and resected the mass from the rectum and anus. Gross total resection was...
achieved (Fig. 1). Following surgery, the patient was able to ambulate short distances without issue and was passing flatus. She was discharged on postoperative day 2 and was doing well at the outpatient follow-up without neurological or gastrointestinal deficits.

Hematoxylin and eosin staining analysis of the resection specimen revealed large areas of epidermoid cyst filled with keratin debris, adjacent proliferation of epithelioid cells with uniform round nuclei arranged in solid nests and cords, a small focus of columnar epithelium with scattered mucinous cells, and a small cluster of ductal structures consistent with sweat glands. Immunohistochemistry was performed on the epithelioid region for further characterization. The epithelioid cells were strongly positive for glial fibrillary acidic protein; positive for S-100 protein; variably positive for synaptophysin; and negative for chromogranin, cytokeratin AE1/AE3, smooth muscle actin, transcription termination factor 1, and CDX2. These findings altogether were consistent with a final diagnosis of mature cystic teratoma.

Discussion

Observations

Presacral masses are a set of rare tumors that may arise from any of the three germ layers. Commonly, these masses are due to a congenital disorder called “Currarino syndrome” that is characterized by a presacral mass, anorectal malformation, and spinal dysraphism. There is an estimated incidence of 1 in 40,000 births, with a slight female predominance of 1.39:1; however, the actual incidence may be more common because it is thought that many cases go undiagnosed.2,4 The pathogenesis of Currarino syndrome is commonly due to abnormal separation of the ectoderm and endoderm during development that causes a connection between the spinal column and bowel due to failed anterior vertebral fusion.6 In over half of cases, Currarino syndrome is caused by a mutation in the MNX1 gene on chromosome 7q36. Although the disorder is mostly hereditary, it can also arise in an acquired form with variable expression. Our patient is the first in her family to have the disorder, so she represents a sporadic case. It is reported that in more than 80% of cases, Currarino syndrome is diagnosed in the first 10 years of life; however, incomplete Currarino syndrome is typically diagnosed in adolescence and adulthood secondary to evaluation of symptoms such as constipation, sacrococcygeal pain, incontinence, and others.7 In this case, Currarino syndrome was incidentally identified via evaluation of androgen insensitivity syndrome, which is unlikely unrelated. The combination of an incomplete Currarino syndrome and an evaluation focused on lack of menarche made the ultimate diagnosis of Currarino syndrome more difficult in this case. However, diagnosis was likely made earlier than otherwise would have been possible for the reasons discussed.

Presacral masses as part of Currarino syndrome are most commonly anterior sacral meningoceles or teratomas; however, there are also reports of enteric cysts, dermoid cysts, epidermoid cysts, carcinoid tumors, lipomas, leiomyosarcomas, yolk sac tumors, and hamartomas.8 This report presents a case with mature cystic teratoma. Although benign, there have been rare malignant transformations of teratomas that have been described in the literature.9 Although diagnosis was made difficult due to competing evaluation for androgen insensitivity syndrome as discussed, the likely earlier identification of Currarino syndrome allowed appropriate surgical intervention and possible complication avoidance with the risk of malignant transformation of the presacral mass.

The most common sacrococcygeal defects are hemisacrum or scimitar sacrum, but there are other abnormalities that may occur.10 Anorectal stenosis is the most common anorectal malformation, followed by rectal stenosis/atresia, anal ectopia, and intestinal duplication. An anorectal malformation was absent in our patient, but she did present with a ventral sacral defect at S3 in addition to the teratoma.

Constipation is typically the sole symptom described with these patients because it is thought to be caused by mass effect with mechanical compression, a tethered spinal cord, sacral nerve root compression, or an anus in the anterior position.11 Other symptoms that may occur are urinary issues, perianal sepsis, and meningitis.8 In many cases, there may be an incomplete triad with only one or two anomalies. Because of the variability in presentation, the diagnosis of Currarino syndrome can often be missed.6 As discussed, evaluation for androgen insensitivity in this case likely led to earlier recognition of Currarino syndrome despite the diagnostic difficulty, which may be important for this patient in prevention of various complications related to local mass effect and of possible meningitis with connection between the presacral mass and the thecal sac.

Computed tomography and MRI can be useful to diagnose Currarino syndrome and to get full visualization of the spine and presacral mass. They can also help to visualize if there is a tethered cord. Tethered cord syndrome is important to identify because it is associated with an increased risk of meningitis, which is one of the lethal complications of Currarino syndrome.12 Another complication that may occur is pneumocephalus due to erosion of the presacral mass into the rectum, forming a neuenteric fistula.

Management of Currarino syndrome is dependent on the findings seen in each unique case. Timely identification and intervention are recommended for the best long-term outcomes of patients because increased morbidity and mortality can arise due to severe complications.8 Neurosurgical intervention is often required with symptomatic lesions, specifically if there is evidence of neurological deficits or bladder/bowel involvement.6 Surgical approaches to remove the presacral mass include sacral laminectomy, an anterior abdominal approach, and laparoscopic techniques; however, the posterior sagittal approach has been found to be the most effective.9,6,13 The posterior approach is frequently employed to repair associated spinal dysraphism in the same operation as the mass resection. A surgeon’s comfortability and knowledge will ultimately dictate the approach used. A posterior approach was used in this case due to the sacral defect, relatively midline position of the mass, and desire to explore the thecal sac and detether the patient. Notably, the use of intraoperative ultrasound obviated the need for intrathecal exploration, which has been routine in previous reports of the surgical management of Currarino syndrome.6 This is important due to the risks of intrathecal exploration, including infection, injury to nervous tissue, postoperative cerebrospinal fluid leak, and others. This case demonstrates that not all cases may require intrathecal exploration and that intraoperative ultrasound may be used to identify appropriate candidates for entirely extradural surgical management.

To the best of our knowledge, this is the first case presentation of an XY androgen-insensitive female with an incomplete Currarino syndrome. Diagnosis of Currarino syndrome in this case was difficult due to the rarity of the pathology, patient presentation with an incomplete clinical triad, the unique genetic findings in our patient, and the age at presentation. The description of this case and the
surgical technique may be useful for clinicians who encounter similar patients in the future.

Lessons
This case highlights the diagnosis and management of Currarino syndrome in a unique setting of androgen insensitivity. The syndrome’s presentation has been classically described as a triad, but all features may not be present in an individual patient, as was seen in this case. Androgen insensitivity and Currarino syndrome are likely unrelated, but evaluation of androgen insensitivity may result in earlier diagnosis of an incomplete Currarino syndrome despite diagnostic difficulty. This case also demonstrates the utility of intraoperative ultrasound in assisting with decision making on the surgical management of associated spinal dysraphism. We hope that this description will increase awareness of Currarino syndrome so that appropriate laboratory and imaging investigations can be completed in an efficient manner in patients with similar presentations, and we also hope that it will assist surgeons in surgical planning and management.

References

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