The Currarino Syndrome Presenting as Neonatal Intestinal Obstruction in a Nigerian Male Child

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ABSTRACT

We describe the first documented radiologic findings of complete Currarino triad presenting as neonatal intestinal obstruction in a Nigerian male child. A 1-week-old male neonate was admitted for rectal agenesis and had a divided colostomy during which an anterior sacral meningocele was discovered intraoperatively. The imaging evaluation revealed further, the triad of a partial sacral defect, anorectal atresia, and anterior sacral meningocele. These radiologic findings define the rare association known as the Currarino syndrome (CS). The CS expresses a wide phenotypic expression, and so its diagnosis may be elusive. This case emphasizes the need for physicians to be cognizant of the clinical presentation and radiologic findings of CS. It is a triad to remember in infants with anorectal malformations and even older children or adults presenting with chronic constipation.

Key words: Currarino; intestinal obstruction; male; neonate; Nigerian

Introduction

A constellation of congenital anomalies may very rarely coexist as a triad involving the sacral bone, anorectal canal, and neural tube. Radiologic imaging is key in making the correct presurgical diagnosis. In 1838, Byrant was the first to observe a distinct type of sacral bony defect. Later in 1926, Kennedy reported the cooccurrence of sacral meningocele and rectal atresia. Finally, in 1981, Currarino described the triad of anorectal atresia, partial sacral agenesis, and a presacral mass. The presacral mass can be an anterior sacral meningocele, enteric cystic or a presacral teratoma.[1] The Currarino triad is a dominantly inherited condition. The causative gene has been located on chromosome 7q36.[2-4] This triad shows a familial tendency in 50–60% of cases.[5] The phenotypic expression associated with the mutation of this gene varies widely from mild asymptomatic clinical presentation to severe clinical presentation as seen in our patient with a complete triad. Notably, the sole manifestation in most symptomatic patients is chronic constipation. The etiology of constipation in these patients could be multifactorial such as mechanical compression, anorectal stenosis, atresia, or tethered cord. In the neonatal period, Currarino syndrome (CS) poses both diagnostic and therapeutic challenges. Because of these complexities, optimal management approach of these patients should be multidisciplinary.

To the best of our knowledge, only about 300 cases have been reported in literature.[6-8] Until now, there has being no reported case of CS in a Nigerian neonate.

Case Report

A male neonate presented to our pediatric unit at 1 week of age with failure to pass meconium since birth, absent anus, progressive abdominal distension, and difficulty in breathing. He was delivered to a 21-year-old multigravida, through

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an unsupervised vaginal birth at home after a full term uneventful pregnancy. His parents were nonconsanguinous, and there was no family history of similar or any other congenital abnormality. His mother and elder brother however had a history of chronic constipation for which they had never been evaluated. He had no symptoms referable to the genitourinary or cardiopulmonary systems.

On examination, he was acutely ill-looking, not pale, anicteric, and afebrile. He weighed 3 kg. His abdomen was however grossly distended with visible peristalsis. He had normal male external genitalia. His anal dimple ended blindly about 1 cm from the anal verge. There was no perineal fistula and no other obvious anomalies. His initial diagnosis was neonatal intestinal obstruction from rectal atresia. He was subsequently worked up, and he had divided sigmoid colostomy. He did well with the initial colostomy, and at 30 months old, he had posterior sagittal anorectoplasty for the correction of the anorectal malformation (ARM). At surgery, an anterior sacral meningocele was discovered, and the sac ruptured inadvertently. The sac was repaired, and the procedure abandoned to allow for further radiologic reevaluation.

Plain pelvic radiographs revealed the scimitar configuration of a hemisacral defect and an intact first and second sacral bones [Figure 1]. The lateral film of the distal loop cologram confirmed the rectal atresia. A widened presacral space below S2 and beaking of the anterior rectal blind stump below this level was also noted [Figure 2].

On magnetic resonance imaging (MRI) of the lumbo sacral spine, a multiseptated cystic mass was seen anterior to the sacrum and continuous with an expanded thecal sac which was consistent with an anterior sacral meningocele. Figure 3a and b, there was no spinal cord tethering on MRI.

One month later, a combined pediatric and neurosurgical team simultaneously excised the meningocele and performed an anorectoplasty through the posterior sagittal approach [Figure 4]. The colostomy was closed 3 weeks later. He presently has fecal and urine continence with intermittent constipation and fecal soiling. Is continent of feces and urine but has intermittent constipation and fecal soiling.

Discussion

There has only being about 300 cases have been reported of CS in literature.[6-8]

ARM occurs in 1:5000 live births, and it represents a spectrum which ranges from imperforate anal membrane to complete caudal regression.[9]

In the surgical practice of our hospital, ARM is the leading cause of neonatal intestinal obstruction accounting for 68.9% of cases in a series, followed by Hirschsprung's disease (7.3%).[10] Often times, it is associated with other VACTERL anomalies; however, triad of the abnormalities in the CS has not been previously documented. The CS is a form of caudal regression syndrome in which malformations occur caudal to the posterior neuropore. Its true incidence still remains unknown, although it is seen more frequently in females[11] (unlike in the case presented), with 80% of the cases diagnosed before 12 years of age. The most common presentation is chronic constipation in the neonatal period or since childhood. Some patients are diagnosed in adulthood, heralded by complications such as fistula in ano or anorectal abscess[6] while others remain asymptomatic and are only discovered during family screening of patients with CS.[5,11]

In the index patient, the triad was complete, and the sacral findings describe a true CS, which typically spares the first sacral bone. Sacral abnormalities reported in a series by Lynch *et al*.[11] include scimitar sacrum in 75% of the patients as was seen in this patient. Bifid sacrum with a central defect was seen in 22% while <5% of the patients had more subtle

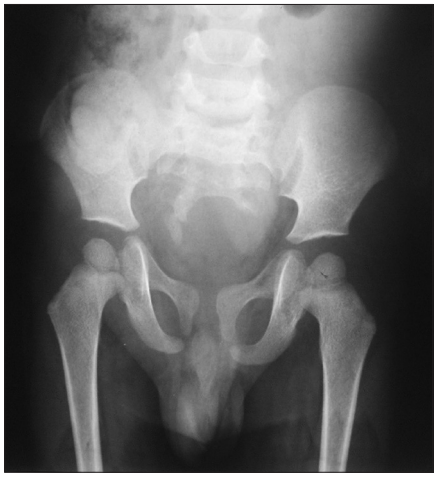

Figure 1: Plain pelvic X-ray (anterior - posterior view) showing a hemisacral defect and an intact first and second sacral bones

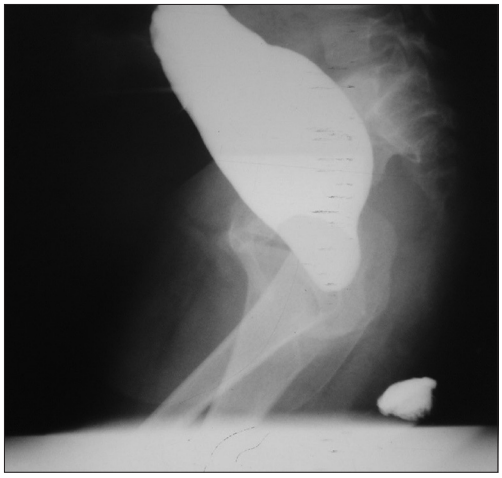

Figure 2: Distal loop cologram which shows a blind ending atretic rectum

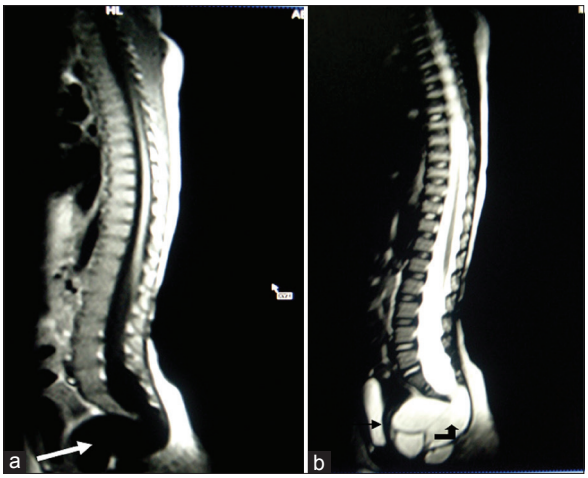

Figure 3: Sagittal views of lumbo sacral magnetic resonance imaging. T1-weighted (a) and T2-weighted (b) an anterior sacral cystic mass which is a meningocele (white arrow). It is continuous superiorly (black arrow) with a widened thecal canal, and there is compression of the urinary bladder (black arrowhead)

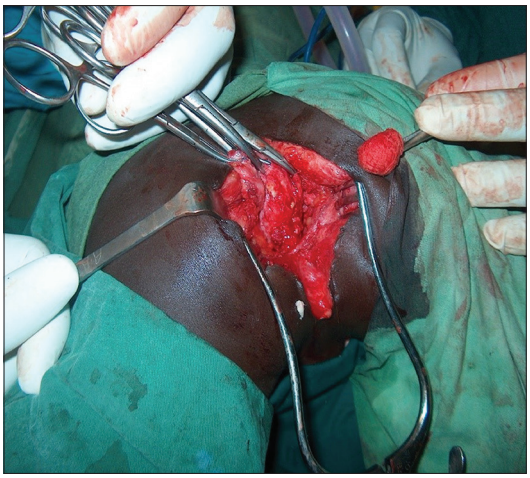

Figure 4: Correction of the anorectal malformation through a posterior sagittal anorectoplasty

abnormalities such as pepper pot sacrum. In the incomplete forms of CS, one or two of the abnormalities may be absent. Apart from a presacral mass which could be an anterior sacral meningocele (60%), a teratoma (25%), or an enteric cyst,[5] other frequently associated spinal abnormalities include tethered cord, lipoma, or a lipomeningocele.

Urogenital malformations are commonly associated in female patients with CS, and this was not present in this case.

Radiologic investigation is central to making the diagnosis of CS, especially in our locality where genetic analysis is not readily available. Antenatal diagnosis may pose a dilemma and could still be a challenge postnatally.[12] Plain X-rays of the pelvis provide the index imaging finding that characterizes the syndrome. Although it is considered the preliminary investigation of choice in the neonatal period, gaseous dilation may obscure the radiographic findings of sacral defect. Hence, subsequent pelvic radiograph after irrigating the distal loop may be more revealing. Pelvic ultrasound could also serve as a screening tool in the prenatal and postnatal period to exclude spinal dysraphism, pelvic cystic masses, and also to assess the kidneys for urogenital anomalies. Pelvic ultrasound involves nonionizing radiation and it is an especially valuable diagnostic imaging modality in our locality because it is more readily available and cheaper relative to other diagnostic imaging modalities. Computerized tomography (CT) and MRI are both useful when planning surgical resection and to detect complications such as meningitis which could occur as a result of fistula between the spinal canal and the rectum or anus. CT would provide better osseous detail; however, MRI has the added advantage of detecting occult spinal masses and spinal cord tethering which is a known association of this condition in (18%) and can be easily missed.[13]

Contrast studies of the gastrointestinal tract will reveal ARMs, which could be an atresia or a stenosis. In patients with Hirschsprung's disease, pathological confirmation is necessary as it is a diagnostic mimic of CS or an association.[14-16]

Although patients with milder symptoms may be managed conservatively. Surgical intervention involving a multidisciplinary team approach is required in patients with severe ARM and also in patients with presacral mass as was the case in this patient.

Early diagnosis and treatment are necessary to avoid complications which may be associated with CS such as meningitis, malignant degeneration of a presacral teratoma, or sphincteric dysfunction.

Conclusion

In areas of the world with limited radiologic diagnostic modalities, the availability of relatively inexpensive initial imaging methods despite its drawback would allow for early detection of CS only where there is a heightened level of awareness and high index of clinical suspicion. Furthermore, we recommend that routine screening should be performed on the first-degree relatives of patients with ARM or neural tube defects as well as first-degree relatives of proven cases of CS.

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Conflicts of interest
There are no conflicts of interest.

References

1. Currarino G, Coln D, Votteler T. Triad of anorectal, sacral, and presacral anomalies. AJR Am J Roentgenol 1981;137:395-8.
2. Hagan DM, Ross AJ, Strachan T, Lynch SA, Ruiz-Perez V, Wang YM, *et al.* Mutation analysis and embryonic expression of the HLXB9 Currarino syndrome gene. Am J Hum Genet 2000;66:1504-15.
3. Belloni E, Martucciello G, Verderio D, Ponti E, Seri M, Jasonni V, *et al*. Involvement of the HLXB9 homeobox gene in Currarino syndrome. Am J Hum Genet 2000;66:312-9.
4. Lynch SA, Bond PM, Copp AJ, Kirwan WO, Nour S, Balling R, *et al.* A gene for autosomal dominant sacral agenesis maps to the holoprosencephaly region at 7q36. Nat Genet 1995;11:93-5.
5. Köchling J, Pistor G, Märzhäuser Brands S, Nasir R, Lanksch WR. The Currarino syndrome – Hereditary transmitted syndrome of anorectal, sacral and presacral anomalies. Case report and review of the literature. Eur J Pediatr Surg 1996;6:114-9.
6. Shoji M, Nojima N, Yoshikawa A, Fukushima W, Kadoya N, Hirosawa H, *et al.* Currarino syndrome in an adult presenting with a presacral abscess: A case report. J Med Case Rep 2014;8:77.
7. Urioste M, Garcia-Andrade Mdel C, Valle L, Robledo M, González-Palacios F, Méndez R, *et al.* Malignant degeneration of presacral teratoma in the Currarino anomaly. Am J Med Genet A 2004;128A: 299-304.
8. Isik N, Elmaci I, Gokben B, Balak N, Tosyali N. Currarino triad: Surgical management and follow-up results of four [correction of three] cases. Pediatr Neurosurg 2010;46:110-9.
9. Levitt MA, Pena A. Anorectal malformations. Orphanet J Rare Dis 2012;7:98.
10. Ameh EA, Chirdan LB. Neonatal intestinal obstruction in Zaria, Nigeria. East Afr Med J 2000;77:510-3.
11. Lynch SA, Wang Y, Strachan T, Burn J, Lindsay S. Autosomal dominant sacral agenesis: Currarino syndrome. J Med Genet 2000;37:561-6.
12. Patel RV, De Coppi P, Kiely E, Pierro A. Currarino's syndrome in twins presenting as neonatal intestinal obstruction – Identical presentation in non-identical twins. BMJ Case Rep 2014;2014. pii: bcr2014204276.
13. Lee SC, Chun YS, Jung SE, Park KW, Kim WK. Currarino triad: Anorectal malformation, sacral bony abnormality, and presacral mass – A review of 11 cases. J Pediatr Surg 1997;32:58-61.
14. Saberi H, Habibi Z, Adhami A. Currarino's syndrome misinterpreted as Hirschsprung's disease for 17 years: A case report. Cases J 2009;2:118.
15. Ohno K, Nakamura T, Azuma T, Nakaoka T, Takama Y, Hayashi H, *et al.* Familial Currarino syndrome associated with Hirschsprung disease: Two cases of a mother and daughter. J Pediatr Surg 2013;48:233-8.
16. Patel RV, Shepherd G, Kumar H, Patwardhan N. Neonatal Currarino's syndrome presenting as intestinal obstruction. BMJ Case Rep 2013;2013. pii: bcr2013200310.