

Currarino Triad: A Rare Combination of Anomalies and Justification for a Review of Phenotypic Classification

Aiwanlehi Eighemhenrio^{1,2*} , Williams Akerele^{1,2}, Samuel Egbele¹, Anthony Owolabi^{3,4}, Folorunso Omosofe^{5,6}, Lillian Akerele^{7,8}

¹Department of Surgery, Irrua Specialist Teaching Hospital, Irrua, Edo State, Nigeria.

²Department of Surgery, College of Medicine, Ambrose Alli University, Ekpoma, Nigeria.

³Department of Radiology, Irrua Specialist Teaching Hospital, Irrua, Edo State, Nigeria.

⁴Department of Radiology, College of Medicine, Ambrose Alli University, Ekpoma, Nigeria.

⁵Department of Anaesthesia, Irrua Specialist Teaching Hospital, Irrua, Edo State, Nigeria.

⁶Department of Anaesthesia, College of Medicine, Ambrose Alli University, Ekpoma, Nigeria.

⁷Department of Paediatrics, Irrua Specialist Teaching Hospital, Irrua, Edo State, Nigeria.

⁸Department of Paediatrics, College of Medicine, Ambrose Alli University, Ekpoma, Nigeria.

Correspondence to: Aiwanlehi Eighemhenrio, Department of Surgery, Irrua Specialist Teaching Hospital, Irrua, Nigeria. E-mail: eighemhenrioaiwanlehi@gmail.com

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ABSTRACT

Introduction: Currarino Triad or Syndrome (CS) is a rare congenital disease characterized by the triad of anorectal malformations (ARMs), presacral mass and sacral anomalies with an estimated prevalence of approximately 1 in 100,000 live births worldwide. Due of its rarity, global incidence data is based on case reports rather than widespread epidemiological screening, and only about 300 cases reported worldwide so far. The disorder is an autosomal dominant genetic trait caused by a mutation in the MNX1 homeobox gene. In 2000, the first large series of Currarino cases was genetically screened for MNX1 mutations and it was shown that the gene is specifically causative for the syndrome. The index patient in this article had the complete triad of features in addition to a rare association of situs inversus. The patient had persistent cloaca which is the extreme of anorectal malformation in which there is a common channel for the urinary, vagina as well as rectal opening.

X-ray showed non visualization of the sacrum and a presacral mass measuring about 8 cm by 4 cm. Currarino triad as seen in this patient occurred with abdominal ultrasound findings of situs inversus.

Aim: The aim of this study is to report a case of this very rare congenital anomaly (Currarino triad) seen at the Paediatric Surgery Unit of Irrua Specialist Teaching Hospital Irrua, Edo State Nigeria.

The additional abdominal findings have not been previously reported in literature hence the need for a reclassification of this rare congenital condition.

Case Report: IB is a 2 month old infant who was delivered at term to 24 year old woman. At presentation she was observed not to have an anal opening and the external genitalia was poorly developed and was passing both urine and faeces from a single opening in the external genitalia since birth. She had an umbilical defect measuring about 8 cm in its longest diameter. On further examination she has a sacral mass measuring about 8 cm by 4 cm. A lumbosacral x-ray was done which revealed sacral agenesis. A diagnosis of Currarino syndrome (triad) was made from the combination of the findings of anorectal malformation (Persistent Cloaca), sacral mass and sacral agenesis. She had an abdominopelvic ultrasound scan which revealed situs inversus.

Discussion: Currarino syndrome is a rare, it is definitively diagnosed when a patient exhibits two or more of the following: Anorectal Malformations, Sacral Bone Defects and Presacral Mass. This index patient had the extreme of anorectal malformation which is persistent cloaca. Cloacal malformations are rare; they only occur in approximately 1 in 50,000 live births. The coexistence of this triad with situs inversus (the mirror-image reversal of chest and abdominal organs) in this patient is extremely rare and a literature search did not reveal any reported case. The triad is most widely classified phenotypically by clinicians into three forms based on severity. The association with situs inversus in this patient calls for a review of the phenotypic classification.

Conclusion: The patient in this article had a combination of Currarino triad and situs inversus which is an extremely rare combination. Such had not been previously documented. A review of phenotypic classification is needed to incorporate such combination as the fourth form.

Keywords:

Currarino Syndrome (CS), Ultrasound, Persistent cloaca, Presacral mass.

Introduction

Currarino Syndrome (CS) is a rare congenital disease characterized by the triad of Anorectal Malformations (ARMs), presacral mass and sacral anomalies. It occurs in approximately 1 in 100,000 people [1]. It is caused by mutations in the MNX1 gene. The MNX1 gene (Motor Neuron and Pancreas Homeobox 1), located on chromosome 7q36.3, encodes a transcription factor critical for embryonic development, specifically in spinal cord motor neuron specification, pancreatic beta cell differentiation and hindgut development. Mutations lead to Currarino syndrome (autosomal dominant) and severe neonatal diabetes. Anterior sacral meningocele is the most common presacral mass in patients with Currarino syndrome, occurring in 60% of cases. Its presence may significantly affect the surgical management of these patients [2,3]. Other potential presacral masses include presacral teratoma and enteric cyst. Presacral teratoma usually is considered to be a variant of sacrococcygeal teratoma. However, the presacral teratoma that is characteristic of the Currarino syndrome may be a distinct kind [4]. Teratomas in CS are often benign however, a malignant transformation has been observed, even in childhood [5-7].

Incidence

Currarino triad (or syndrome) is an extremely rare congenital condition, with an estimated prevalence of approximately 1 in 100,000 live births worldwide. Because of its rarity, global incidence data is based on case reports rather than widespread epidemiological screening and only about 300 cases reported worldwide so far [8]. The incidence of Currarino triad (or Currarino syndrome) in Africa is not specifically quantified in epidemiological studies, but it is generally recognized as an extremely rare condition, similar to global trends.

Genetics

The disorder is an autosomal dominant genetic trait [5] caused by a mutation in the MNX1 homeobox gene. In 2000 the first large series of Currarino cases was genetically screened for MNX1 mutations and it was shown that the gene is specifically causative for the syndrome, but not for other forms of sacral agenesis. The study was published in the American Journal of Human Genetics [9]. Among its related pathways of action of MNX1 is neural stem cells and Lineage-specific markers. Gene Ontology (GO) annotations related to this gene include DNA-binding transcription factor activity and sequence-specific DNA binding. The locus for CS was initially mapped to chromosome 7q36 [10] and here, the MNX1 gene (earlier termed HLXB9), encoding motor neuron and pancreas homeobox protein 1 (MNX1, earlier termed HB9) was first described by Harrison et al [11].

Aim

The aim of this study is to report a case of this very rare congenital anomaly (Currarino triad) seen at the Paediatric Surgery Unit of Irrua Specialist Teaching Hospital Irrua which is tertiary hospital located in Southern Nigeria. This study also attempts to introduce a new classification of this condition

based on the peculiar findings in this case report.

Case Report

IB is a 2 month old infant who was delivered at term to a 24 year old woman at a peripheral delivery home about 30 km to our tertiary hospital. At presentation she was observed not to have an anal opening and the external genitalia was poorly developed and was passing both urine and faeces from a single opening in the external genitalia since birth. She had an umbilical defect measuring about 8 cm in its longest diameter (Figure 1). A diagnosis of persistent cloaca was entertained. She had been on breast milk until three days prior to presentation when she was introduced to formula feeds. She was severely constipated, not tolerating the formula feeds occasioned by vomiting, gross abdominal distension and visible peristalsis seen on the anterior abdominal wall necessitating her presentation to our centre.



Figure 1: Single perineal opening (persistent cloaca).

On further examination she has a sacral mass measuring about 8 cm by 4 cm. The mass was soft, not tender, not mobile, fluctuant and transilluminates poorly (Figure 2 and 3).



Figure 2: Sacral mass with single perineal opening.



Figure 3: Presacral mass with flaccid limbs.

The tone on both lower limbs were reduced, though it was difficult to assess the power on the limbs but the muscle bulk appeared reduced. A lumbosacral x-ray was done (Figure 4) which revealed sacral agenesis (Figure 5).



Figure 4: Spine x-ray.

Invest./Radiology Result

SACRUM / LUMBOSACRAL

THERE IS A SOFT TISSUE MASS IN THE SACROCOCCYGEAL REGION DEVOID OF LUCENCIES AND CALCIFICATIONS

THERE IS NON VISUALIZATION OF THE SACRUM SUGGESTIVE OF SACRAL AGENESIS

IMPRESSION: SACRAL MASS WITH NON VISUALISATION OF THE SACRUM FOR WHICH CONSIDERATION IS NEURAL TUBE DEFECT

MRI IS RECOMMENDED

DRS AMHAKHIAN / OGBEBOR / EJODAMEN

Figure 5: Report of the spine x-ray.

A diagnosis of Currarino syndrome (triad) was made from the combination of the findings of Anorectal Malformation (Persistent Cloaca), Presacral mass and Sacral agenesis in a patient presenting with severe constipation. She had an abdominopelvic ultrasound scan (Figure 6) which revealed situs inversus, see ultrasound scan report (Figure 7).



Figure 6: X-ray of the chest and abdomen.

Discussion

Currarino syndrome is a rare, hereditary congenital disorder characterized by a classic triad of anomalies: Anorectal malformations (such as anal stenosis), sacral bone defects, and a presacral mass (often an anterior sacral meningocele or teratoma). The most common clinical presentation in patients is severe chronic constipation. This Clinical triad occurs in roughly

ABDOMINOPELVIC SCAN

There is extensive intra-abdominal gas limiting the visualization of some intra-abdominal structures.

The liver is demonstrable in the left hypochondrial region but however appears normal in size outline and parenchymal echotexture.

The gallbladder was contracted probably due to post-prandial effect.

The spleen was demonstrable in the right hypochondrial region and appear normal in size, outline and parenchymal echotexture.

The pancreas was not demonstrable, obscured by bowel gas.

There are no demonstrable hepatic mass lesions, cysts or collection.

Both kidneys measure (4.7 x 2.5) cm and (4.5 x 2.8) cm for the right and left respectively and show normal cortical echogenicity and cortico-medullary differentiation.

There are no demonstrable renal stones, cysts or hydronephrosis.

Minimal right sided pleural effusion noted.

There are no demonstrable para-aortic lymphadenopathy or intra-abdominal masses.

The visualized bowel loops are not dilated and show normal peristaltic activity.

The urinary bladder was underfilled for comment on pelvic structure.

Impression: Sonographic findings of situs inversus.

DRS.
AREGBOKHAI/OTUOKPAIKHIAN/EJODAMEN

Figure 7: Abdominal ultrasound scan report.

20% of cases, Currarino syndrome is definitively diagnosed when a patient exhibits two or more of the following: Anorectal malformations, Sacral bone defects and Presacral mass typically includes anal stenosis, anal atresia (imperforate anus), or rectoperineal fistulas. This index patient had the extreme of anorectal malformation which is persistent cloaca. Persistent cloaca is a complex congenital birth defect in which a female infant's rectum, vagina and urinary tract are fused into a single, common channel. Cloacas appear in a wide spectrum of variation in females born with the malformation. The severity of the condition depends on where the three tracts have joined

together. The single orifice, called a common channel, may occur varying in length from 1 to 10 cm [12,13]. A common channel less than 3 cm in length usually has good sphincter muscles and a well-developed sacrum [14]. This type of malformation happens when the embryonic anorectal and urogenital channels fail to separate during the sixth and seventh weeks of gestational development. Cloacal malformations are rare; they only occur in approximately 1 in 50,000 live births [11,12]. Due to their complexity, cloacal malformations require multidisciplinary treatment from pediatric surgeons, urologists and gynecologists. In persistent cloaca Colostomy is performed shortly after birth to allow stool to empty safely into a stoma bag, preventing infection and distension. This is also considered the first step in the management of patients with Currarino syndrome. Treating persistent cloaca within Currarino syndrome requires a highly specialized, multidisciplinary approach. Management is always staged, beginning with fecal diversion (colostomy), followed by complex anatomical reconstruction and simultaneous or staged removal of the associated presacral mass. Managing this condition is a long-term process. While many patients eventually attain urinary and fecal continence, some require ongoing bowel management (like enemas) or catheterization. Secondary surgeries may also be required during adolescence to address gynecological functions.

Anterior sacral meningocele is the most common presacral mass in patients with Currarino syndrome, occurring in 60% of cases. Its presence may significantly affect the surgical management of these patients [2,3]. Other potential presacral masses include presacral teratoma and enteric cyst. Presacral teratoma usually is considered to be a variant of sacrococcygeal teratoma. However, the presacral teratoma that is characteristic of the Currarino syndrome may be a distinct kind [4]. The presacral mass in this patient was soft, not tender, not mobile, fluctuant and transilluminates poorly. The surface was smooth and the mass measured about 8 cm by 4 cm.

Treatment for a presacral mass in Currarino triad (Currarino syndrome) primarily involves surgical excision to prevent complications like infection, bowel obstruction, or malignant transformation. The choice of approach depends on the size and location of the mass, as well as the presence of an anterior sacral meningocele. The Kraken procedure also known as posterior procedure is preferred for tumors under 10 cm in diameter. It avoids entering the peritoneal cavity, reducing the risk of abdominal adhesion and allows for the simultaneous closure of any meningocele. The Anterior/Trans abdominal approach is often required for very large masses (over 10 cm) or when the meningocele neck is too wide for transducer ligation via a posterior approach alone. The posterior approach will be most suited for this index patient.

Sacral bone defects in Currarino triad could present in the form of a partial absence or severe malformation of the sacrum or coccyx. The first sacral vertebra (S1) usually remains intact, creating a characteristic "scimitar" (sickle-shaped) sacrum on imaging. A "scimitar sacrum" (also called hemisacral agenesis or sickle-shaped sacrum) is a unilateral crescent-shaped defect in the lateral sacrum often associated with Currarino triad. In this patient there was non visualization of the sacrum which might represent an agenesis the bone.

Rare combination of currarino triad with situs inversus

The coexistence of this triad with situs inversus (the mirror-image reversal of chest and abdominal organs) is extremely rare and a literature search did not reveal any reported case. This combination introduces multiple layers of clinical complexity. In this patient the liver was demonstrated in the left hypochondrion and the spleen in the right hypochondrion. Situs inversus is sometimes associated with Primary Ciliary Dyskinesia (PCD)/Kartagener's syndrome. If PCD is also present, it can cause chronic respiratory issues that complicate anesthesia and postoperative recovery. Situs inversus has two main subtypes: dextrocardia and levocardia. A person is said to have dextrocardia if the point of the heart is on the right side. A person is said to have levocardia if the heart is on the left side, yet the other organs are flipped. Levocardia is rarer and often associated with other cardiac abnormalities. Another subtype is situs ambiguous. This occurs when several organs are in abnormal positions, but not in an easily defined pattern. Though this patient did not have a chest X-ray to demonstrate the heart but an abdominal ultrasound scan demonstrated flipping of the internal organs.

Justification for a new classification of currarino triad

The triad is most widely classified phenotypically by clinicians into three forms based on severity:

1. Complete currarino syndrome: Patients present with all three classic features:

- Anorectal Malformation (ARM)
- Sacral osseous defect (partial sacral agenesis, commonly a sickle-shaped/scimitar sacrum, while typically sparing the first sacral vertebra S1)
- Presacral mass (often an anterior sacral meningocele, teratoma, or developmental cyst)

2. Mild currarino syndrome: Patients present with the sacral bony defect plus only one of the other two classic anomalies (sacral defect+ARM or sacral defect+presacral mass).

3. Minimal currarino syndrome: Patients present with only the characteristic sacral bone defect (usually a hemispheric) without the other two anomalies.

An additional phenotypic category of Currarino triad becomes necessary based on the rare occurrence with situs inversus necessitating the addition of a fourth classification thus;

4. Complex (Irrua) Currarino triad: Patients presenting with the complete triad of classic features co-existing with situs inversus.

Patients with Currarino triad heavily present with severe, intractable constipation. Those who also have situs inversus may not have worsening constipation but the transposition of organs is clinically important for diagnosing conditions like primary ciliary dyskinesia or avoiding surgical complication though the association may not affect the management protocol.

Conclusion

Currarino triad is a rare congenital condition characterizes by a trio of congenital features with an estimated prevalence of approximately 1 in 100,000 live birth. Patients with Currarino

triad heavily present with severe, intractable constipation. The patient in this article had a combination of Currarino triad and situs inversus which is an extremely rare combination. Such had not been previously documented. A review of phenotypic classification is needed to incorporate such combination as the fourth form.

Conflict of Interests

The authors declare no conflict of interest.

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