High Type Imperforate Anus Without Associated Anomalies in a Nigerian Child: A Case Report

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ABSTRACT

Background: *The High type imperforate anus is usually associated with congenital anomaly in the other systems or organs. It is rare to have the above type without accompanying anomalies.*

Aim: *To report a case of high imperforate anus without associated anomalies of the other systems or organs in a Nigerian child who was presented at the University of Port Harcourt Teaching Hospital, Port Harcourt.*

Methods: *A case report of a 4-day old Nigerian male delivered to a 34-year old para 2^{+1} woman by spontaneous vertex delivery, presented with failure to pass meconium after 4 days. Clinical and imaging examinations revealed a high type imperforate anus without associated anomalies of the other systems. Relevant review of literature was also done.*

Conclusion: *This is a rare clinical entity. The role of radiological imaging in the management of this condition is highlighted.*

Key Words: *Imperforate anus, High type, Associated anomaly.*

Afrimedic Journal 2011;2(1):24-27

INTRODUCTION

Anorectal anomalies (imperforate anus) are a group of related anomalies of the hindgut. The incidence of imperforate anus in Nigeria is unknown, but in the developed countries, it is 1 in 5,000 live births.[1] Males are more affected than females with a ratio of 3:2. The aetiology of imperforate anus is unknown, but the most acceptable theory is faulty development of the primitive mesoderm at about 7th week of intrauterine life.[2, 3]

Anorectal anomalies (imperforate anus) are classified as high, intermediate and low. The intermediate and high anomalies in particular, are associated with fistulae into the urinary system.[4] The frequency of additional anomalies in patients with imperforate anus is nearly 50%. [5]

We report a rare case of high imperforate anus without associated anomalies in a 4-day old neonate.

CASE REPORT

Baby S.E is a 4-day-old male Nigerian, delivered to a 34-year-old para 2^{+1} woman. The patient presented at the surgery department of the University of Port Harcourt Teaching Hospital on the 6th May, 2010, with failure to pass meconium, progressive abdominal distension and refusal of feeds since birth. There was no associated projectile vomiting. The pregnancy was uneventful and delivery was by Spontaneous Vertex Delivery (SVD) at term at the University of Port Harcourt Teaching Hospital. The child cried immediately after delivery.

The Baby weighed 3.5kg at birth. There was no history of abnormal increase in abdominal girth or skin rash on the mother during pregnancy.

Physical examination revealed a neonate in respiratory distress but was not pale, anicteric or febrile, T = 36.4^0C.

The abdomen was distended, but there was no hepatoslenomegally or palpable mass. Rectal exam showed no visible patent anus, but dimple on the anal region was seen. The respiratory rate was 38/min. There was intercostal recession with flaring of alae nasi. Breath sounds were essentially normal.

An impression of imperforate anus was made. Plain abdominal x-ray (supine and cross table lateral) was done to determine whether it was a high or low type. Abdominal ultrasound was done to exclude associated congenital malformations of abdominal viscera. Abdominal radiograph showed dilated large and small bowels (See fig. I). Plain radiography of the pelvis and spine that were done to exclude associated vertebral and sacral abnormalities were normal. Plain cross table lateral radiograph of the abdomen demonstrated the upper level of the rectal gas. The estimated distance from the anal verge to gas filled rectum was 50mm (See fig. II).

Abdominal ultrasound scan showed distended bowel loops, but all other intra-abdominal viscera (kidneys, liver, gas bladder, spleen) were normal in positions and echopattern. Spinal ultrasonograhpy did not show any evidence of tethered cord or any other spinal anomalies.

Haematological indices were all normal with Packed Cell Volume of 0.40, White Blood Cell count = 7×10^3 mm^3, differential count of neutrophil 45%, lymphocytes 35%,

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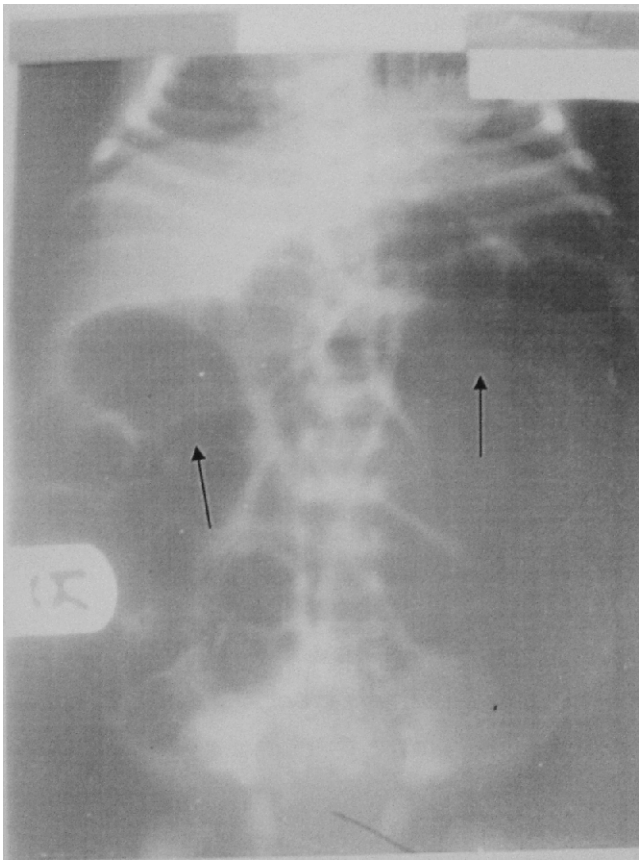
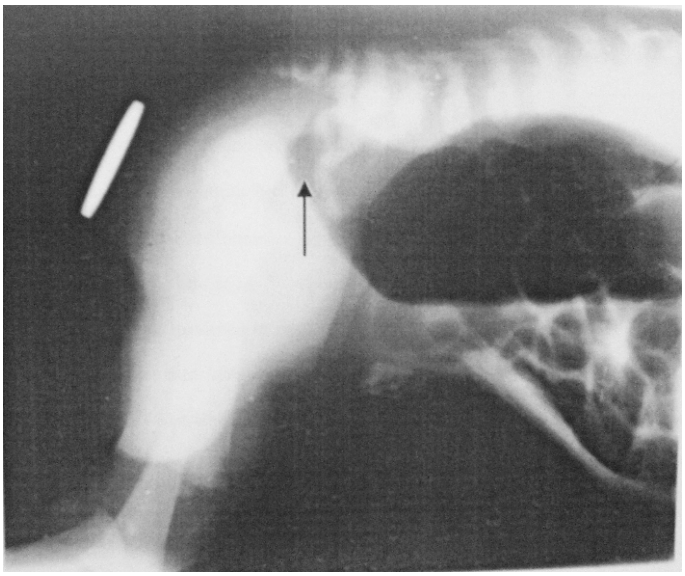
eosinophil 10% and basophil 1%. Blood urea and creatinine were within normal limits. Urinalysis was also normal. A nasogastric tube was successfully passed; thus, excluding oesophageal atresia.

Fig I: Plain abdominal radiograph showing dilatation of small and large bowel loops (arrows) with symmetrical abdominal distension.



The baby was referred to the surgical team and 3 days later, surgical operation involving divided sigmoid colostomy was performed successfully and patient was discharged on the 14th post operative day. The definitive surgery which is a pull through procedure was performed on our patient at the age of 14 months with satisfactory result.

Fig II: Plain cross table lateral radiograph demonstrating the upper level of the rectal gas (arrow) with rounded margin. The level is 50mm from the coin in the anal region, being the area of symphysis pubis, indicating the high type of imperforate anus.



DISCUSSION

Anorectal malformations include a wide spectrum of defects in the development of the lowest portion of the intestinal and urogenital tracts. Many children with these malformations are said to have an imperforate anus because they have no opening where the anus should be.

Anorectal malformation presents with a wide spectrum of defects, ranging from relatively low malformations to very complex cloacal anomalies.[6, 7] Reports of incidence of anorectal malformation (imperforate anus) range from 1 per 1,500 to 1 per 5,000 live births.[8, 9] Uba et al reported an average of 8 cases per year from Jos, although the overall incidence in Nigeria is unknown.[10] They also reported that males were effected more than females. Our patient is a male. Bhargava et al reported that imperforate anus occurred equally in males and females among Indian children.[11] Kim et al reported slightly increased incidence of this disease in females than in males, especially in low type.[12]

Although the precise embryologic defect that causes the spectrum of malformations described as imperforate anus has not been determined, defects in the formation of shape of the posterior uro-rectal septum account for many of the described anomalies of imperforate anus.[11] No clear risk factors predispose a person to have a child with imperforate anus, but a genetic linkage is sometimes present.[13] Most cases of imperforate anus, are sporadic, without a family history of the condition.[13]

Prenatal ultrasonographic examination findings are often normal, although the demonstration of polyhydramnious or intrabdominal cyst may suggest imperforate anus with associated foetal hydrocolpos or hydronephrosis. [11]

Physical exam may not show any external orifice but a dimple at the position of the anus may be seen.[7] There was a dimple in the position of the anus in our patient.

Other associated findings may include absence of an umbilical artery, abdominal masses (dilated kidney, bladder, hydrocolpos, ectopic kidney, duplication) and external fistulous tracts.[4] These were not seen on our patient. The remainder of physical examination is focused on associated malformations for example, cardiovascular, urogenital, gastrointestinal and musculoskeletal systems.

Barnes et al documented that the frequency of additional anomalies in patients with imperforate anus is nearly 50%.[5] They described non-random associations of anomalies with imperforate anus to include Vertebral and Tracheo-oesophageal, Radial and Renal anomalies (VATER). Other anomalies involving the heart (cardiovascular system) and limbs (muscular system) have been included (VACTERL).

Cardiovascular malformations occur in 12% - 22% of patients, the most common lesions being tetralogy of Fallot and ventricular septal defect.[14] Many gastrointestinal malformations that have been described include oesophageal fistula without atresia with and without fistula, which may be proximal or distal and trachea-oesophageal fistula without atresia.[5] Up to 10% of patients have trachea-eosophageal abnormalities of which the most common form is proximal oesophageal atresia that accounts for 82% of trachea-eosophageal abnormality.[5,14] Bello et al reported a case of high imperforate anus with associated trachea-esophageal fistula in a Nigerian child.[15]

Duodenal obstruction due to annular pancreas or duodenal atresia occurs in a small percentage of patients.[7] Malrotation with Ladd bands that causes obstruction has also been reported.[7, 8] Hirschsprung's disease has also been described in association with imperforate anus, although the incidence of this combined condition is unknown.[15]

The association of imperforate anus and vertebral anomalies has been recognized and patients with high type anorectal anomalies have an increased risk of this association. Lumbosacral anomalies predominate and occur in approximately one-third of patients with imperforate anus.[5] The frequency of spinal dysraphism increases with the severity of the lesion, that is, 17% in patients with low type and 46% in patients with cloacal anomalies.[17] The commonest type of dysraphism being tethered cord is present in 25% of cases. Cord lipomas and syringohydromelia are also common.[17] Lee et al described a triad of sacral defect, presacral mass and imperforate anus.[18] This finding emphasizes the importance of plain radiography of the sacral region and pelvic ultrasound. This triad was not seen in our patient. 50% of patients with imperforate anus have urological abnormalities, vesico-ureteric reflux, and renal-agenesis. Cryptorchidism has been reported to occur in 3%-19% of males.[19] In females, vaginal and uterine abnormalities are common.[20] It has been documented that bicornuate uterus and didelphys occur in 35% of female patients with imperforate anus.[20] Vaginal duplication and agenesis have been reported with vaginal agenesis being associated with ipsilateral dysplastic ovary and kidney.[20]

The radiologic modalities employed in imaging of imperforate anus are in inverted radiography, (which has been replaced by cross table lateral radiography), distal loopography, ultrasound, computerized tomography and magnetic resonance imaging (MRI). They are used to determine the level of the distal pouch, identify the presence of fistulas, and to diagnose any associated anomalies. Plain sacral radiography in two views, lateral and antero-posterior, are used to measure sacral ratios and look for defects and presacral masses. Cross table lateral radiography demonstrated a high type imperforate anus in our patient. The proximal rectal gas shadow seen was more than 1cm from the radio-opaque marker in the anal dimple. The puborectalis is the landmark for distinguishing low from high type of imperforate anus. This means that the descent of the rectum below the puborectalis sling indicates low type and above it indicates high type.[20, 21] Abdominal ultrasonography is used to examine the genito-urinary tract and to look for any other masses. Ultrasonography is usually performed before surgery and repeated after 72 hours because early ultrasonographic findings may be insufficient to exclude hydronephrosis due to vesico-ureteric reflux. [23] This is because vesico-ureteric reflux builds up over a period of time. It is also used to evaluate the type of imperforate anus. Itan et al demonstrated that infracoccygeal ultrasonography is an excellent diagnostic modality for demonstrating high and low imperforate anus.[23] Ultrasound displays directly the puborectalis muscle and demonstrates the relationship between the distal pouch to the puborectalis muscle.

Computed Tomography (CT) scan may demonstrate the presence of puborectalis muscle and external anal sphincter prior to surgery. It may also display the anatomic relationship between the pulled-through intestine and the levator sling in post-operative scans.[24] MRI would exclude associated malformations such as meningocoele or myelomeningocoele, teratoma or mixed lesions as well as demonstrate the presence of

the puborectalis muscle and anal sphincter prior to surgery.[23] Computed Tomography and MRI were not used on this patient because of lack of funds by the parents of the patient.

All patients who have anorectal malformation with no significant life threatening co-morbidity should survive. Therefore, prognosis is determined based on the probability of primary incontinence. Surgical complications worsen the chances for primary continence.[23]

CONCLUSION

A case of high imperforate anus without associated congenital anomalies in a 4 day-old neonate is reported. This clinical entity is rare. The role of radiology in the management of this condition is highlighted.

REFERENCES

1 Ogunbiyi TAJ. The Initial Management of Imperforate Anus. Nigeria Med J 1972; 2: 119-124.

2 Arsic D, Qi BQ Beasly SW. A Possible Explanation for the VATER association. J Paediatric Child Health 2001; 3117-121.

3 Maglner AD. Oesophageal Atresia and Hypertrophic Pyloric Stenosis: Sequential Co-existence of Disease. AJR 1989; 147: 324-330.

4 Cremin BJ. Radiological Assessment of Anorectal Anomalies. Clin Radiol. 1971; 126: 445-446.

5 Barnes JC, Smith WL. The VATER Association. Radiology 1978; 126:445-449.

6 Pathak K, Saifullah S. Congenital Anorectal Malformation: An Experience Based on 50 Cases. Indian J Paeditr 1909; 36: 370-379.

7 Pena A. Anorectal Malformations. Semin Peadiatr Surg 1995;4: 35-47.

8 Pena A. Current Management of Anorectal Malformations. Surg Clin North Am 1992; 72:1393 1416.

9 Gupta DK, Charles AR, Srinavas M. Paediatric Surgeries in India A Specialty Come of Age. Paediatr Surg 2002; 18; 649-652.

10 Uba AF, Chirdan BL, Ardill W, Edino ST. Anorectal Anormaly: A Review of 82 Cases seen at JUTH, Nigeria. Nig Postgrad Med J 2006; 13(1): 61 -65.

11 Bhargara P, Mahajan JK, Kumar A. Anorectal Malformations in Children. J Indian Assoc Peadiatr Surg. 2006; 11 136-139.

12 Kim Hl, Eow KW, Penner JG, Blair GK, Murphy JJ, Webber EM. Presentation of Low Anorectal Malformations Beyond Neonatal Period. Paediatrics 2000; (5): 68-70.

13. Shaul DB, Harrison EA. Classification of Anorectal Malformations Initial Approach, Diagnostic Tests, and Colostomy. Semin Paediatr Surg54120 1997; 6(4): 187 195.

14. Dennis M, David L. Oesophageal Atresia. In: Ravine and Putman (ed). Textbook of Diagnostic Imaging 2nd edition, WB Saunders Philadelphia. 2000; 737-750.

15. Bello TO, Fadeora SO. Imperforate Anus Associated with Oesophageal Atresia and Trachea-oesohageal Fistula: Sequential Co-existence of Disease. WAJR 2000; 7(1):9-11.

16. Flageole H, Fecteau A, Laberge JM, Guttman FM. Hirschsprung's Disease, Imperforate Anus and Down's Syndrome: A Case Report. J Paediatr Surg 1996; 31(6): 759-760.

17. Levitt MA, Patel M, Rodriguez G. The Tethered Spinal Cord in Patients with Anorectal Malformations. J Paediatr Surg. 1997; 32(3): 462-468.

18. Lee SC, Chun YS, Jung SE. Curriano Triad: Anorectal Malformation, Sacral Bony Abnormality and Presacral Mass: A Review of 11 cases. J Paediatr Surg. 1997; 32(1): 58-61.

19. Hung AR, Acuna MF, Pena A. Urologic Injuries Associated with Repair of Anorectal Malformations in Male Patients. J Paediatr Surg 2002; 37(3): 339- 344.

20. Levitt MA, Stein DM, Pena A, Gynaecologic Concerns in the Treatment of Teenagers with Cloaca. J Paediatr Surg, 1998;33(2): 188-193.

21. Rickham PP, Hecker WC, Prevot J. Radiological Diagnosis of Anorectal Malformations. Prog Paediatr Surg 1976; 9:77-84.

22. Narasimhara KL, Prasal GR, Katariya S, Yadar K, Mitra SK, Pathal K. Prone Cross-table Lateral View: An Alternative to the Invertogram in Imperforate anus. AJR 1983; 140(2): 227-229.

23. Han TD, Kim I, Kim SW. Imperforate Anus US Determination of the type with Infracoccygeal Approach. Radiology 2003;228: 226 229.

24. Kohda E, Fujioka M, Ikausa H, Yokoyama J. Congenital Anorectal Anomaly; CT Evaluation. Radiology 1985;157:349-352.

25. Pena A, Guadino K, Torila J. Bowel Management of Faecal Incontinence in Patients with Anorectal Malformations. J Paediatr Surg 1998;33(1):133-137.