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Case Report

VACTERL Association with High-Type Imperforate Anus, Crossed Fused Renal Ectopia, and Hexadactyly in a Neonate: A Case Report

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ABSTRACT

Background: Anorectal malformations (ARMs) are congenital anomalies of the distal gastrointestinal tract and are frequently associated with abnormalities of other organ systems, particularly the genitourinary tract. The association of a high-type imperforate anus with crossed fused renal ectopia is rare and may suggest an underlying syndromic pattern such as VACTERL association.

Case Presentation: We report a term male neonate delivered by caesarean section due to breech presentation and oligohydramnios. Antenatal ultrasonography demonstrated dilated pelvic bowel loops with mixed echogenic intraluminal contents and non-visualization of the right kidney in its normal position, raising suspicion of distal bowel obstruction and crossed fused renal ectopia. Postnatally, the neonate failed to pass meconium and developed abdominal distension. Clinical examination confirmed the absence of an anal opening. Imaging revealed a high-positioned distal rectal pouch consistent with high-type imperforate anus, associated intestinal obstruction, and crossed fused renal ectopia. A preaxial limb anomaly was also noted, while echocardiography showed no cardiac defects. These findings were suggestive of partial VACTERL association. The neonate underwent a sigmoid loop colostomy on the second day of life with an uneventful postoperative course.

Conclusion: This case report highlights a rare combination of anomalies within the VACTERL spectrum and emphasizes the importance of antenatal imaging, comprehensive postnatal evaluation, and timely surgical intervention in neonates with anorectal malformations.

Keywords: *Anorectal malformation, Imperforate anus, Crossed fused renal ectopia, VACTERL association, Neonate.*

INTRODUCTION

Anorectal malformations (ARMs) comprise a heterogeneous group of congenital anomalies of the distal gastrointestinal tract, with a reported incidence of approximately 1 in 2,000 to 1 in 5,000 live births. These defects range from minor anomalies to severe forms such as high-type imperforate anus, which are frequently associated with additional systemic abnormalities, most commonly involving the genitourinary and renal systems. The coexistence of anorectal malformations with renal anomalies and other structural defects raises suspicion for VACTERL association, a non-random constellation of vertebral, anorectal, cardiac, tracheoesophageal, renal, and limb anomalies.

Early antenatal detection of suggestive imaging findings is crucial for anticipating postnatal complications and facilitating timely multidisciplinary management. Reports of anorectal malformation associated with crossed fused renal ectopia are rare, particularly when accompanied by limb anomalies suggestive of partial VACTERL association. We present a rare case of a term neonate with high-type imperforate anus, crossed fused renal ectopia, and intestinal obstruction, successfully managed with early surgical intervention.

PATIENT DEMOGRAPHICS

The patient was a term male neonate born at 38 weeks and 2 days of gestation to a 31-year-old second gravida mother (G2A1). Delivery was performed by caesarean section due to breech presentation and oligohydramnios identified on antenatal ultrasound. There was no history of parental consanguinity. The birth weight was 2.8 kg; Apgar scores were not available in medical records.

CASE REPORT

A 31-year-old second gravida (G2A1) underwent regular antenatal care. Serial antenatal ultrasound examinations demonstrated breech presentation with progressive oligohydramnios, with the amniotic fluid index decreasing to 4 cm on the final scan. At 34 weeks' gestation, ultrasound revealed a mildly dilated foetal bowel loop in the pelvis containing heterogeneous intraluminal echogenic material, as shown in **Figure 1a and 1b**, raising suspicion of an anorectal malformation with possible intestinal obstruction. Additionally, the right foetal kidney was not visualized in its normal anatomical location, with findings suggestive of crossed fused renal ectopia, as shown in **Figure 2a and 2b**. These antenatal findings indicated a complex congenital anomaly, possibly within the VACTERL spectrum. Quadruple marker screening was negative, and there was no history of parental consanguinity.

The neonate was delivered at 38 weeks and 2 days of gestation by caesarean section due to breech presentation and oligohydramnios. The infant was male, with a birth weight of 2.8 kg. In the immediate postnatal period, the neonate failed to pass meconium and developed progressive abdominal distension. Physical examination revealed absence of an anal opening, as shown in **Figure 4.** Preaxial hexadactyly involving the right upper limb was also noted, as shown in **Figure 3**.

Prenatal

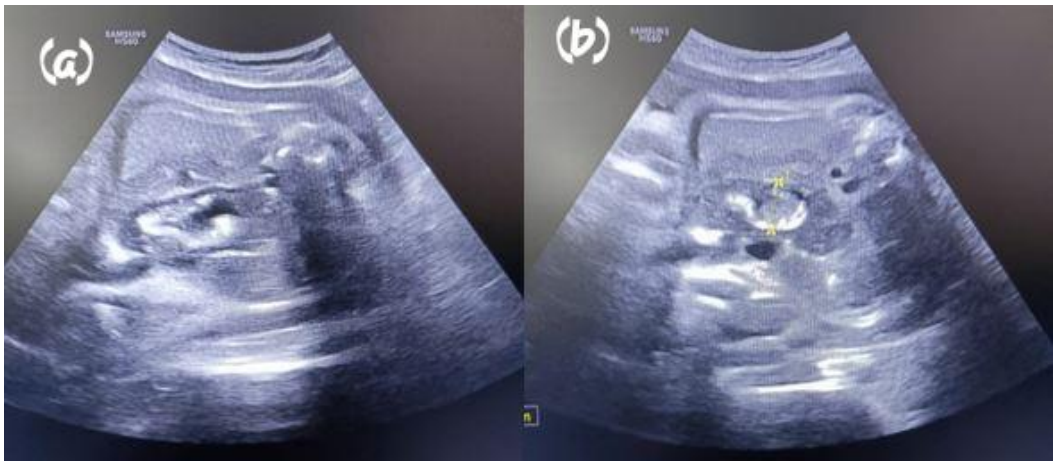

Figure 1a and 1b shows Antenatal ultrasound images at 34 weeks' gestation showing a mildly dilated fetal bowel loop in the pelvis containing heterogeneous mixed echogenic intraluminal contents, raising suspicion of anorectal malformation with possible intestinal obstruction.

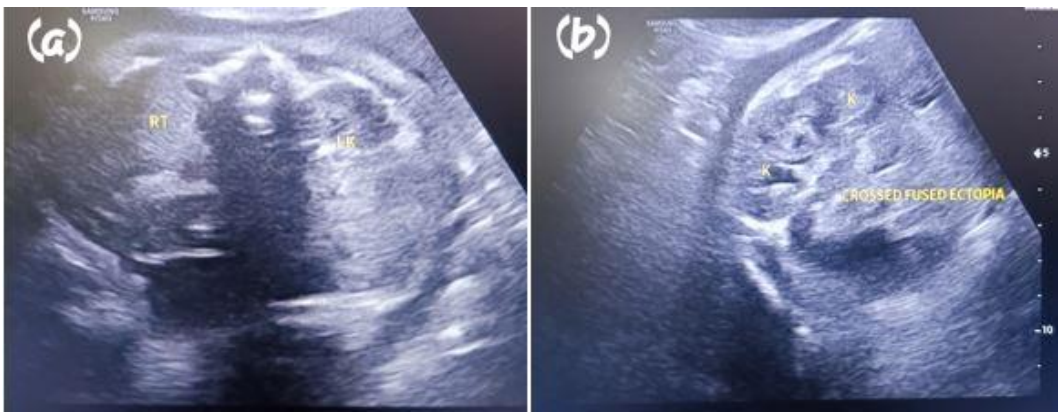

Figure 2a and 2b shows Antenatal ultrasound images demonstrating an empty right renal fossa (a) with a crossed fused renal ectopia identified on the left side (b).

Postnatal
Day 1

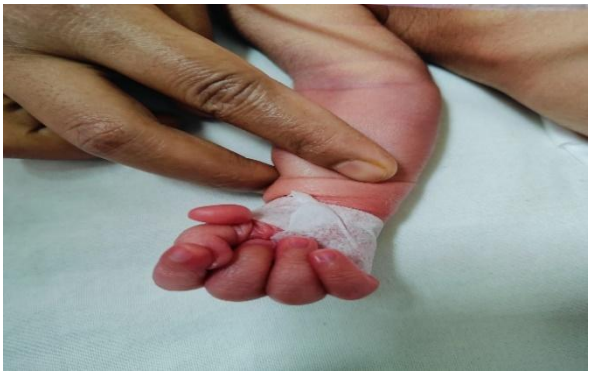

Figure.3 shows Clinical photograph obtained on day 1 of life showing preaxial hexadactyly involving the right upper limb.

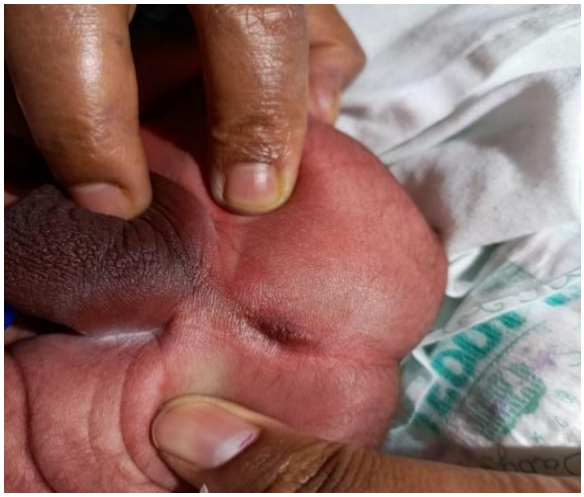

Figure 4: Clinical photograph showing absence of an anal opening, consistent with imperforate anus.

Clinical Presentation
On physical examination, the anal opening was absent, confirming imperforate anus. No visible vertebral anomalies or dysmorphic features were observed. Preaxial hexadactyly involving right upper limb was also noted. The abdomen was distended, and there were no other remarkable findings on general examination.

Investigations
Postnatal transabdominal ultrasonography performed on day 1 demonstrated a rectosigmoid pouch in the midline on longitudinal section, as shown in **Figure 5**, along with dilated bowel loops containing heterogeneous echogenic intraluminal contents, as shown in **Figure 6a–c**, suggestive of intestinal obstruction. Renal ultrasonography confirmed crossed fused renal ectopia, with the right kidney fused to the left side, corroborating the antenatal findings, as shown in

Figure 7.
An erect abdominal radiograph demonstrated excessive gas-filled, mildly dilated bowel loops. A lateral invertogram performed on day 2 showed a high-positioned distal rectal pouch, consistent with a high-type imperforate anus, as shown in **Figure 8a and 8b.**

Two-dimensional echocardiography revealed levocardia, situs solitus, intact atrial and ventricular septae, normal atrioventricular and ventriculoarterial connections, and no evidence of congenital cardiac anomalies.

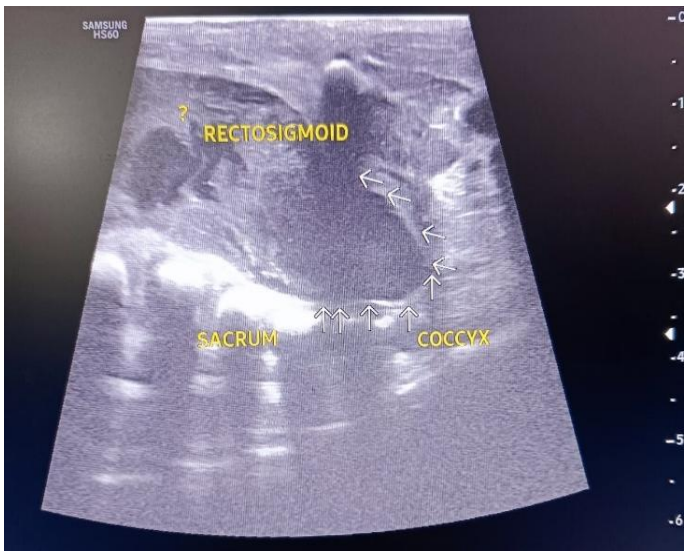

Figure.5 shows Postnatal transabdominal ultrasound performed on day 1 of life showing a rectosigmoid pouch located in the midline on longitudinal section.

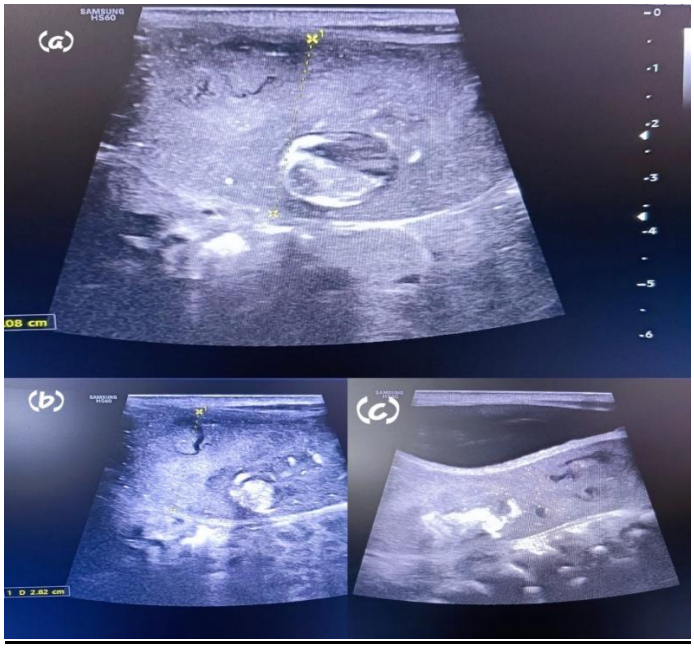

Figure.6a, 6b and 6c shows Postnatal transabdominal ultrasound images obtained on day 1 of life demonstrating dilated bowel loops with heterogeneous mixed echogenic intraluminal contents, consistent with intestinal obstruction.

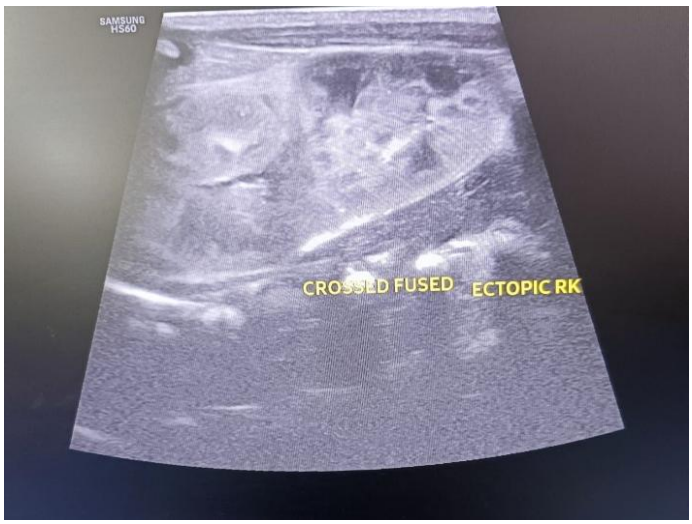

Figure.7 shows Postnatal abdominal ultrasound confirming crossed fused renal ectopia, with the right kidney fused to the left side, corroborating the antenatal findings.

Day 2

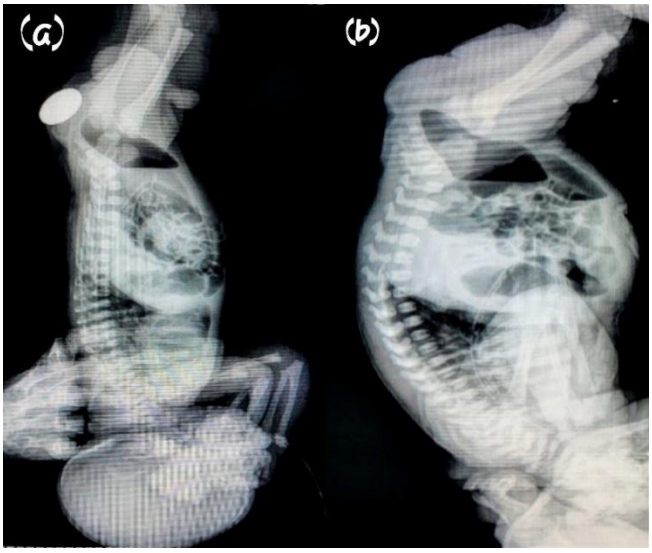

Figure 8a & 8b Lateral invertogram showing dilated bowel loops and high-positioned distal rectal pouch, consistent with high-type imperforate anus.

Management

In view of the confirmed diagnosis of high-type imperforate anus with associated intestinal obstruction, the neonate underwent surgical intervention on the second day of life. A sigmoid loop colostomy was performed under general anaesthesia. The procedure was uneventful.

Postoperative Course and Outcome

Postoperatively, the neonate was managed in the neonatal intensive care unit and remained clinically stable. The postoperative course was uneventful. Plans were made for further evaluation and staged definitive anorectal reconstruction.

DISCUSSION

Anorectal malformations (ARMs) represent a spectrum of congenital anomalies involving the terminal portion of the gastrointestinal tract. Their incidence is estimated to range between 1 in 2,000 and 1 in 5,000 live births worldwide.[1,2] These malformations frequently coexist with anomalies in other organ systems, particularly the urogenital tract. In the present case, a full-term male neonate was diagnosed with a high-type imperforate anus accompanied by crossed fused renal ectopia and signs of intestinal obstruction an unusual yet clinically significant triad that aligns with complex variants of ARM.

The frequent occurrence of associated anomalies in children with ARM is well documented. In a series by Bălănescu et al., approximately two-thirds (66.6%) of patients with ARM exhibited concurrent anomalies, with urogenital tract malformations being the most common.[6] Mittal et al. similarly identified urogenital anomalies in 43.8% of their cases, emphasizing the necessity of thorough screening in all neonates diagnosed with ARM.[7] Common renal anomalies include agenesis, ectopic kidneys, and fusion anomalies. Crossed fused renal ectopia, as identified in our patient, is an uncommon congenital abnormality with an estimated incidence of 1 in 1,300 to 7,500 live births.[8] These anomalies may go undetected without appropriate prenatal imaging or postnatal evaluation.

Prenatal ultrasound plays a crucial role in the early detection of ARM and associated structural anomalies. In our case, dilated bowel loops in pelvis showing mixed echogenicity contents. On antenatal ultrasound prompted early suspicion. Yin et al. have described the "line sign"(The "line sign" in the context of imperforate anus refers to a sonographic finding where a linear echogenic structure, rather than the normal "target sign," is observed in the fetal perineum during prenatal ultrasound. This sign can indicate a high-type imperforate anus, where the rectum ends above the levator ani muscles) as a helpful sonographic marker for diagnosing imperforate anus in utero.[9] Furthermore, Rohrer et al. have demonstrated that the diagnostic yield improves substantially when foetal MRI is combined with ultrasound during the antenatal period.[10]

The presence of multiple congenital anomalies, including imperforate anus, renal malformation, and limb abnormality (hexadactyly), raises clinical suspicion for a syndromic or association pattern. One such recognized entity is **VACTERL association**, which refers to a non-random constellation of congenital defects involving vertebral anomalies, anal atresia, cardiac defects, tracheoesophageal fistula, renal anomalies, and limb abnormalities. Diagnosis is considered when at least three of these six features are present. In our patient, the presence of anal atresia, a renal anomaly (crossed fused ectopic kidney), and polydactyly satisfies the minimum diagnostic criteria for partial VACTERL association. Similar variable

presentations have been documented, such as in the case reported by Pariza et al., reinforcing the concept of partial phenotypic expression within this association.[5]

While VACTERL association remains the most plausible clinical diagnosis, other syndromic differentials should be considered:

- **Townes-Brocks Syndrome** is a rare autosomal dominant disorder caused by mutations in the *SALL1* gene. It is characterized by anal atresia, preaxial limb anomalies including polydactyly or triphalangeal thumbs, renal malformations, and external ear anomalies, often with hearing impairment. Although ear abnormalities were not present in our patient, the overlapping gastrointestinal, renal, and limb features warrant consideration of this syndrome and support genetic evaluation.

- **Fanconi Anaemia** is another important differential. This inherited bone marrow failure syndrome can present with radial ray abnormalities, polydactyly, renal anomalies including ectopia, and, less frequently, anorectal malformations. Due to its hematologic manifestations, early screening with a complete blood count and chromosomal breakage testing is essential, particularly if there is a suggestive family history or clinical suspicion.

- **Bardet-Biedl Syndrome** is a ciliopathy that typically presents with postaxial polydactyly, renal anomalies (including structural abnormalities and progressive renal dysfunction), central obesity, hypogonadism, retinal dystrophy, and intellectual disability. While anorectal anomalies are not considered core features, they have been sporadically reported and should not be entirely excluded, especially in patients with overlapping renal and limb features.

Given the phenotypic overlap of these conditions, a comprehensive postnatal evaluation is essential. This includes detailed physical examination focusing on the vertebral column, cardiac evaluation with echocardiography, skeletal radiographs, spinal ultrasonography, and abdominal imaging. Genetic counselling and testing, including chromosomal microarray analysis and targeted gene panels (e.g., *SALL1*, *BBS*, and Fanconi anaemia-related genes), should be pursued to confirm the diagnosis and facilitate appropriate family guidance.

The high frequency of urogenital anomalies in ARM patients has been further supported by studies from India and other regions. Srivastava et al. reported urogenital anomalies in 33% of their ARM cohort, with renal anomalies such as agenesis being the most common.[11] Similarly, Mirshemirani et al. highlighted that early detection of urogenital tract abnormalities in ARM patients is crucial to prevent long-term renal damage and ensure appropriate surgical planning.[12]

CONCLUSION

Our case revealed the importance of detailed antenatal evaluation and early postnatal assessment in newborns with suspected congenital anomalies. The combination of a high-type imperforate anus and crossed fused renal ectopia, although rare, falls within the spectrum of urogenital abnormalities often seen with anorectal malformations. Early detection allowed for timely surgical management and prevented potential complications. This case also reminds us that even when all features of VACTERL association are not present, the possibility of related anomalies should always be considered. Careful follow-up is essential to monitor the child's growth, renal function, and plan for definitive anorectal reconstruction.

Declaration:
Conflicts of interests: The authors declare no conflicts of interest.
Author contribution: All authors have contributed in the manuscript.
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