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Isolated imperforate anus in dizygotic twins

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ABSTRACT

The authors report an isolated imperforate anus case in a dizygotic twin, which is the first reported case in the literature, the other reported cases were in monozygotic twins. The literature review points to a multifactorial etiology in which environmental triggers interact with genetic susceptibility; in most of these cases, the defect was low in monozygotic twins, as in this dizygotic twin's case. Numerous documented familial cases pointed to an underlying genetic characteristic that is still mostly unknown, but all these cases presented as monozygotic twins, which supports the genetic factor. Our case was the first reported case in the literature of a dizygotic twin with an isolated imperforate anus who has an older sibling who presented at birth with anal stenosis.

1. Introduction

Imperforate anus (IA), also known as the congenital absence of the normal anus, is one of the anorectal malformation (ARM) [1–4], which occur in around 8 and 12 weeks of embryonic development as a result of the hindgut's incomplete formation [5]. The incidence of ARM is 1 per 2500–5000 live births [6]. Isolated ARM represents one-third of all ARM [7]. The etiology of the imperforate anus is unclear till now; a combination of environmental factors and genetics appeared to be critical. Maternal health is the most important environmental factor, in which nicotine, alcohol, multiple pregnancies, gestational diabetes, obesity, and assisted reproductive techniques are all found to be somewhat consistently linked to ARM [8]. Approximately 1.4% of instances have a positive family history [5]. We report an exceptional case of dizygotic twins concordant with isolated imperforate anus. No other well-documented cases of dizygotic twins with isolated anorectal malformations have been reported. On the other hand, these dizygotic twins have a brother who was born with anal stenosis, which is another type of ARM.

2. Case presentation

Here is a brief case study of late-term male twins born at 36 + 6 weeks of gestation to a previously healthy non-smoker 28-year-old woman, gravida 6 para 3 abortion 2, by emergency cesarean delivery due to previous cesarean sections is presented here. The mother's body mass index during pregnancy was 31. Her past medical and surgical histories were insignificant except for two missed abortions in the first trimester, which were managed by evacuation and curettage. The mother received 2 doses of dexamethasone at a gestational age of 30. Nuchal translucency and triple test were not performed, oral glucose tolerance test readings were 76,113 and 94 mg/dl. Diagnostic ultrasonography during pregnancy revealed diamniotic dichorionic twins with an echogenic focus in the left ventricle of the first twin; otherwise, the pregnancy was uneventful with no history of exposure to teratogenic drugs. The parents were

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first cousins once removed. Furthermore, the twin's older brother was born with another form of ARM, which is anal stenosis; otherwise, there was no family history of congenital abnormalities on either the paternal or maternal sides.

The birth weight of baby 1 was 2.375 kg, Apgar score 8/9, and the cord pH was 7.31. Baby 2 had a birth weight of 2.357 kg, Apgar score 8/9, and the cord pH was 7.29. On physical examination, both were active, well, crying, and pink in color. The central nervous system examination showed good Moro and sucking reflexes, respiratory and cardiovascular systems examinations were normal except for a systolic murmur of grade II and mild respiratory distress, so they were kept on continuous positive airway pressure, chest X-ray showed mild hyaline membrane disease. The gastrointestinal system examination showed a lax abdomen, but abdominal distention developed after one day. For that, patients were NPO, nasogastric tube was immediately inserted, and it passed smoothly toward the stomach. An abdominal X-ray was done and showed no coiling. On perineal examination, it was noticed that both had an absent anal opening and passed meconium from a small fistula in the raphe. A trial of anal dilation was done and failed. Otherwise, both had normal male external genitalia. The diagnosis was made on clinical grounds; based on the presence of the congenital malformations outlined above, there were no other congenital abnormalities or associated vertebral defects.

They were admitted to our neonatal intensive care unit immediately for further evaluation. An invertogram was done for both infants 24 hours after birth. This revealed a distance of more than 1 cm from the surface; this was consistent with a low-type anorectal malformation. Ultrasound to the pelvis and abdomen for the twins was normal. Echocardiography was normal too. Renal ultrasonography shows no abnormality apart from mild right-sided hydronephrosis in the second baby. X-rays of the spine revealed no associated vertebral defects. After consulting with the pediatric surgery team, it was decided to perform an urgent double barrel colostomy. Both babies underwent an urgent double barrel colostomy on day 2, which confirmed the diagnosis of congenital atresia and stenosis of the anus with a small raphe fistula. The postoperative period was uneventful, and both babies were discharged after one week following the colostomy in good general condition. Later on, a follow-up session will be held after 3 months of discharge in order to set a date for the posterior sagittal anorectoplasty surgery and the colostomy closure afterward.

3. Discussion

3.1 The international classification of ARM was formulated by the Pediatric Surgical Congress, and it divided ARM into high, mid and low types in relation to the levator ani muscles [9]. On radiographic examinations and anatomical sections, they realized that the pubococcygeal line, which runs from the top of the os pubis to the bottom of the os coccyx, corresponds with the location of the levator ani muscles' attachment to the pelvic wall, dividing high-type malformations from intermediate and low forms of ARM below this anatomic line [10]. In addition, in healthy people, the “I-point,” or the lowest point of the ischial tuberosity, corresponds to the deepest point of the funnel formed by the levator ani muscles [10]. Every blind rectal pouch that was located between the pubococcygeal line and the I-point was categorized as an intermediate abnormality [10]. This classification considerably impacted the surgical strategy because of these anatomical relationships and became the most recommended one for surgical treatment [9].

3.2 Herein, we describe a case of isolated IA that developed in dizygotic twins. The literature identifies 6 well-documented reported cases of isolated IA in monozygotic twins [7,11]. All are of the low type and associated with perineal fistulae [7,11]. There are no previously reported cases of dizygotic twins, making our case to be the first reported one. However, cases of ARM in monozygotic and dizygotic twins with other abnormalities like Down syndrome and Vertebral deformities, Anal atresia, Cardiac defects, Tracheo-Esophageal fistula, Renal anomalies, and Limb abnormalities (VACTERL-association) have been recorded [12,13].

3.3 ARM could be isolated or associated with other anomalies (syndromatic), based on a genetic point of view. It is debatable if ARM that is isolated has a distinct pathogenesis from syndromatic ARM [14]. A retrospective study of 168 cases with ARM concluded that high-type ARM is less common than low-type ARM but is more associated with fetal deaths and other anomalies [3,14]. The existence of ARM in twins raises a question about this condition's heredity. A review of the literature regarding familial syndromic low-type ARM suggests multiple patterns of inheritance, including an autosomal-recessive and autosomal-dominant trait with variable inheritance expression [15]. Regarding familial isolated low-type ARM, a review study on 10 articles suggests multiple inheritance patterns, including autosomal dominant, autosomal-recessive, and heterogeneous patterns of inheritance [15]. Reported cases of unclassified ARM in 3 families suggest a sex-linked recessive pattern of inheritance for this anomaly [16]. However, for all reported cases of ARM in twins, the exact pattern of inheritance is not fully understood. Thus, further investigations are needed to clarify the heritability of this condition.

3.4 To the best of our knowledge, of the six previously reported cases of ARM in monozygotic twins, there was no reported affected sibling or positive family history, except for one case of a female twin with an affected maternal aunt. In our case, there is an older brother who has been found to have anal stenosis; otherwise, there is no other family history of ARM.

4. Conclusion

In summary, ARM in twins is rare. As far as we can tell, our case is the first reported case of ARM in dizygotic twins. We recommend that further studies are needed to fully understand how genetics may play a role in the etiology of this illness. It will be easier to understand the precise pattern of inheritance in various types of abnormalities with additional reporting on comprehensive familial ARM cases.

Patient consent

Patient consent was obtained from the parents.

Authorship

All authors attest that they meet the current ICMJE criteria for Authorship.

Author contributions

DA and HS contributed equally in writing the bulk of the article and made a significant contribution to the work. AY and MT assist in writing and communication with the patient's parents. MA participated in the coordination and acquisition of data. All authors contributed to manuscript revisions, and all authors approved the final version of the manuscript and agreed to be held accountable for its content.

Data availability statement

The data used to support the findings of this study are available from the corresponding author upon reasonable request.

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Declaration of competing interest

The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

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