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LETTER TO EDITOR (VIEWERS CHOICE)

ANORECTAL MALFORMATIONS: BEYOND THE IMPERFORATE ANUS

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Anorectal malformations (ARMs) constitute a significant portion of congenital anomalies in pediatric surgery (1/2000-5000 live births).[1,2,3]

ARMs encompass a broad range of defects[1,4], varying from relatively minor ones, which can be easily treated and carry an excellent functional prognosis, to complex cases that are challenging to manage and often accompanied by other anomalies, resulting in a less optimistic prognosis.[4]

In 50% of children with ARMs, associated anomalies have been reported: genitourinary anomalies (40-50%), cardiac anomalies (30-35%), spinal anomalies (25-30%), gastrointestinal problems (5-10%) and VACTERL anomalies (4-9%).[1,5,6,7]

A retrospective descriptive study was performed, involving the review of clinical files of neonates with ARMs admitted to a Neonatal Intensive Care Unit of a portuguese level II hospital over a 5-year period (January 31, 2017 to January 31, 2022).

7 neonates were diagnosed with ARMs accounting for 0.09% of all births (1/1163 live births), a higher incidence than reported in the literature.[1,2,3] We found a predominance in females (4), contrary to what was reported.[2,3,5]

As described[8], we also reported that vestibular fistula (2) was the most frequent ARM in female patients. We encountered one case of a complex cloaca spectrum malformation, which was previously deemed rare but now ranks as the third most common defect in females.[2] In our sample we had 2 newborns with colonic atresia (CA), a rare entity and the least common type among cases of intestinal atresia.[9,10] In fact, only 9 cases of CA associated to ARM were described in literature. Additionally, one of our cases presented a combination of CA and VACTERL with ARM, an exceedingly rare incidence, with only a single similar case described. Furthermore, we identified 3 cases of VACTERL association, an exceptionally rare condition.[11]

As expected, since it remains infrequent and occurs in merely up to 16% of cases[12], none of our cases had a confirmed prenatal diagnosis of ARM. However we had some findings on prenatal ultrasound (hydramnios, hydronephrosis and intestinal distention) that should leave us alert to that possibility.

As described, in most of our cases (6/7), the anus remains unperforated and the lower intestinal segment may terminate blindly or form a connection via a fistula with the urinary or genital tract or perineum.[11]

Regarding associated anomalies, our incidence (87.7%) was higher than what is typically reported. The genitourinary and cardiac anomalies were, as expected, the most common.

A multi-step approach was performed with a protective colostomy in the early postnatal, followed by a delayed repair (5 to 19 months later) using a posterior sagittal anorectoplasty, which provided in most cases a good functional result. The less favorable outcomes resulted mainly from the other associated anomalies and not exactly from the ARM/surgical repair.

We are aware that children with an ARM face the potential for gross motor function issues, particularly when accompanied by major comorbidities[1,13], but we only detect that in one case.

Given that our study reports only the cases in the last 5 years, we still do not have the ideal follow-up time to realize all the outcomes that may arise.

Compliance with Ethical Standards

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Conflict of Interest None

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