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Delayed diagnosis of anorectal malformations: a call for standardization of the current definitions

Felix Oyania ,[1] Kara Faktor,[2] Sarah Ullrich,[3] Doruk Ozgediz,[2] Meera Kotagal[3]

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[1]Mbarara University of Science and Technology Faculty of Medicine, Mbarara, Uganda
[2]University of California San Francisco, San Francisco, California, USA
[3]Cincinnati Children's Hospital Medical Center, Cincinnati, Ohio, USA

Correspondence to
Dr Felix Oyania; oyafel@icloud.com

Delayed diagnosis of children with anorectal malformations (ARMs) remains a challenge across low-, middle-, and high-income countries. However, definitions of what constitutes a delayed or late presentation and diagnosis of ARM vary.

We encountered a significant challenge in defining the term 'delayed diagnosis of ARM' during our foundational research. Therefore, we feel that there is an urgent need to clearly define the term to standardize care for ARM across the globe and minimize morbidity and mortality. Consensus about the definition(s) of delayed presentation of this congenital condition would facilitate more reliable metrics of health system performance in managing the condition and provide a foundation for interventions to reduce delays, morbidity, and mortality.

Different authors and regions have defined delayed diagnosis of ARMs relative to their context. However, these definitions may not be clinically obvious depending on the type of malformation as described by the Krickenbeck anatomic classification of ARMs.[1] For example, children with complete obstruction due to no external fistula for passage of stool present as neonatal surgical emergencies, whereas those with incomplete obstruction due to a visible external fistula may be misdiagnosed or present late in life. Additionally, children with complex malformations such as cloaca in female babies may present late if there are no associated life-threatening cardiac or respiratory conditions or complications like hydrocolpos.

Definitions for a delayed diagnosis of ARM vary significantly, ranging from hours to weeks after birth (table 1). Many studies define delayed diagnosis as greater than 24,[2–6] 48,[7–11] or 72[12–15] hours of life. Others extend this to 7 days after birth[16 17] and even beyond the neonatal period (after 28 days)[18–20]

Most importantly, some have defined delayed diagnosis of ARM as a diagnosis made at birth versus after discharge from the birth facility.[13–15 21]

Before discharge from the birth facility, we recommend any diagnosis suspicious of ARM requires postnatal examination by a pediatric surgeon to confirm or exclude ARM diagnosis. For facilities without pediatric surgeons, such as in low- and middle-income countries, consultations using available means of communication, including sharing photos after obtaining patient consent, are advised to minimize the delays in diagnosis.

FUTURE DIRECTION AND RECOMMENDATION

The key to ARM recognition is early perineal examination, specifically—inspection and palpation of the perineum in the neonate soon after birth. Newborn perineal examinations should be incorporated into all newborn guidelines/protocols for birth personnel and documented in postnatal charts. Creating a consistent definition could incentivize early examination and diagnosis of these children in the hours to days after birth, reducing life-threatening complications. Furthermore, standardization of the definition will enable all pediatric surgeons across the world to speak the same language when caring for these children, allowing for clearer delineation of diagnostic patterns.

We agree with the authors who defined the delayed diagnosis of ARM as a diagnosis made after discharge from the health facility following birth.[13–15] Identification of ARM at the time of birth and early pediatric surgery referral should be the standard of care for children with these anomalies. This would facilitate more reliable metrics of health system performance in managing the condition and provide a foundation for interventions to reduce delays, morbidity, and mortality. Additionally, we recommend a postnatal pediatric surgeon examination before discharge to confirm or exclude ARM diagnosis.

Table 1 The definitions of delayed diagnosis of ARMs

Article	Definition of delayed diagnosis of ARM
Davidson *et al*.[14] Murthi *et al*.[15] Long *et al*.[13] Aldeiri *et al*.[21]	At birth/after discharge from the birth facility
Lindley *et al*.[6] Govender *et al*.[5] Kruger *et al*.[3] Soeselo *et al*.[2] Galley *et al*.[4]	>24 hours
Eltayeb[9] Wilson *et al*.[11] Karakus *et al*.[10] Kayima *et al*.[8] Reddy *et al*.[7]	>48 hours
Sohail *et al*.[12] Davidson *et al*.[14] Murthi *et al*.[15] Long *et al*.[13]	>72 hours
Divya *et al*.[16] Sinha *et al*.[17]	>7 days
Kim *et al*.[19] Sharma *et al*.[20] Statovci *et al*.[18]	Beyond neonatal period
Sinha *et al*.[17] Sharma *et al*.[20]	Beyond 3 months

ARM, anorectal malformation.

ARM is a clinical diagnosis, and the physical examination of all newborns is an essential tool for identifying ARM, it takes only a few minutes to complete. Emphasis on newborn perineal examinations and possible visible anomalies that may be identified during postnatal care is recommended to ensure timely intervention.

X Felix Oyania @Felix Oyania

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ORCID iD

Felix Oyania http://orcid.org/0000-0003-4773-7790

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