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Anal atresia associated with cutaneous melanocytic naevi and single umbilical artery: A rare neonatal case report

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Abstract
Anorectal malformations are a heterogeneous group of congenital anomalies frequently associated with defects involving other organ systems. Single umbilical artery is a common umbilical cord abnormality and is recognized as an important antenatal marker for congenital anomalies, including gastrointestinal malformations. Congenital melanocytic naevi are usually isolated dermatological findings; however, their association with structural congenital anomalies is uncommon and may suggest shared embryological mechanisms. We report a rare neonatal case of anal atresia associated with multiple cutaneous melanocytic naevi and antenatally detected single umbilical artery. A female neonate was born at term to a 27-year-old mother by lower-segment cesarean section. Antenatal ultrasonography detected a single umbilical artery and poor visualization of the perianal muscular complex, raising suspicion of anorectal malformation. Postnatal examination confirmed anal atresia, along with multiple melanocytic naevi distributed over the trunk, face, and extremities, and an associated umbilical hernia. The neonate was managed in the neonatal intensive care unit and evaluated by a multidisciplinary team, with a staged surgical approach planned. This case emphasizes the diagnostic value of antenatal markers such as single umbilical artery, the need for comprehensive evaluation in anorectal malformations, and the importance of reporting rare associations to enhance understanding of congenital anomaly patterns.

Keywords: Anal atresia, anorectal malformation, congenital melanocytic naevi, single umbilical artery, neonatal congenital anomalies, antenatal diagnosis

Introduction
Anorectal malformations (ARMs) represent a heterogeneous group of congenital anomalies characterized by abnormal development of the distal anus and rectum, with an incidence ranging from 1 in 4, 000 to 5, 000 live births worldwide [1]. These anomalies vary from low perineal fistulas to complex cloacal malformations and are frequently associated with anomalies involving the genitourinary, cardiovascular, spinal, and musculoskeletal systems. The presence of associated anomalies significantly influences both surgical management and long-term prognosis, making comprehensive evaluation essential in affected neonates [2].
Single umbilical artery (SUA) is one of the most common umbilical cord abnormalities, occurring in approximately 0.5-1% of singleton pregnancies and up to 5% of twin gestations [3]. While SUA may be an isolated finding, it is well recognized as an antenatal marker for congenital anomalies, particularly involving the cardiovascular, renal, gastrointestinal, and musculoskeletal systems. Gastrointestinal anomalies, including anorectal malformations, have been reported with increased frequency in neonates with SUA, highlighting the importance of detailed antenatal and postnatal evaluation when this finding is identified [4].
Advances in antenatal ultrasonography have improved the prenatal detection of structural anomalies, including SUA and indirect markers of anorectal malformations. Although direct visualization of the anal opening is often difficult, especially in late gestation, findings such as absence of the perianal muscular complex, dilated distal bowel, or associated anomalies may raise suspicion of anal atresia [5]. Early antenatal recognition allows planned delivery at tertiary centers and timely postnatal surgical intervention, thereby reducing morbidity.
Cutaneous melanocytic naevi are benign proliferations of melanocytes and are relatively common in the neonatal period. However, the presence of multiple melanocytic naevi, particularly when associated with congenital structural anomalies, raises the possibility of underlying syndromic or neurocutaneous associations [6]. Although cutaneous findings are not classically included in the VACTERL spectrum commonly associated with anorectal malformations they may provide important clinical clues to broader developmental

disturbances occurring during early embryogenesis [7].
The coexistence of anorectal malformation, single umbilical artery, and multiple melanocytic naevi is rarely reported in the literature. This unusual combination underscores the complex interplay of embryological events during early fetal development and highlights the need for meticulous systemic evaluation and long-term follow-up in such cases. Reporting rare associations contributes to better understanding of possible etiological links, aids clinicians in early recognition of associated anomalies, and enriches the existing literature on congenital malformations [8].
In this report, we describe a rare neonatal case of anal atresia associated with cutaneous melanocytic naevi and antenatally detected single umbilical artery, emphasizing its clinical relevance, diagnostic approach, and implications for multidisciplinary management.

Case Presentation; A female neonate was born to a 27-year-old G_3 P_1 L_1 mother by lower-segment cesarean section (LSCS) at a tertiary care teaching hospital. The current pregnancy was spontaneously conceived and supervised with regular antenatal visits. The mother had no history of fever, rash, teratogenic drug intake, radiation exposure, or substance abuse during pregnancy. There was no consanguinity. Her past obstetric history included one medical termination of pregnancy at three months ’gestation and one previous term LSCS resulting in a healthy female child.

Antenatal course and investigations
Routine antenatal investigations were within normal limits. The mother was blood group B positive, serology for HIV, HBsAg, and VDRL was non-reactive, and thyroid function tests were normal. Antenatal ultrasonography performed during the late second trimester (around 29 weeks of gestation) detected fetal anomaly in the form of a single umbilical artery (SUA). Subsequent follow-up ultrasonography and fetal Doppler studies confirmed the presence of SUA with normal umbilical artery and middle cerebral artery Doppler indices.
A third-trimester targeted ultrasound performed at approximately 33 weeks ’gestation reported difficulty in satisfactory visualization of the perianal muscular complex, raising the suspicion of anorectal malformation, specifically anal atresia. Fetal biometry corresponded to gestational age, amniotic fluid index was within normal limits, and no major cardiac, renal, spinal, or central nervous system anomalies were identified. Fetal echocardiography and Doppler parameters remained reassuring throughout pregnancy. Table 1 shows the maternal and antenatal characteristics.

Table 1: Maternal and Antenatal Characteristics

Variable	Details
Maternal age	27 years
Gravida / Parity	G3 P1 L1
Consanguinity	Absent
Mode of conception	Spontaneous
Antenatal care	Regular
Medical disorders during pregnancy	None reported
Teratogenic drug exposure	Absent
Antenatal infections (TORCH, fever, rash)	Not reported
Key antenatal USG finding	Single umbilical artery
Suspicion of fetal anomaly	Poor visualization of perianal muscular complex
Amniotic fluid status	Normal
Doppler studies	Normal
Other major anomalies on antenatal scan	Not detected
Mode of delivery	Lower-segment cesarean section
Indication for LSCS	Previous cesarean section

Delivery and immediate neonatal period: The baby was delivered at term gestation by elective LSCS due to previous cesarean section. The birth weight was 2.8 kg, appropriate for gestational age. The neonate cried immediately after birth and did not require resuscitation beyond routine suctioning, drying, and warming. Delayed cord clamping was performed. Apgar scores were 8 at one minute and 9 at five minutes. The neonate maintained oxygen saturation on room air and was hemodynamically stable. Table 2 shows the neonatal clinical profile and examination findings.

Table 2: Neonatal Clinical Profile and Examination Findings

Parameter	Observation
Sex	Female
Gestational age	Term
Birth weight	Appropriate for gestational age
Apgar score (1 min / 5 min)	Satisfactory
Cry at birth	Immediate
Resuscitation required	No
NICU admission	Yes
General condition	Stable
Cutaneous findings	Multiple melanocytic naevi over abdomen and extremities
Umbilical findings	Umbilical hernia present
Perineal examination	Absent anal opening (anal atresia)
Passage of meconium	Absent
Abdominal examination	Soft, non-distended
Cardiovascular examination	Normal
Respiratory examination	Normal
Neurological examination	Normal

Clinical examination; On general examination, the neonate was active, normothermic, and euglycemic, with stable vital parameters. Anthropometric measurements were appropriate for gestational age. There were multiple cutaneous melanocytic naevi, varying in size, distributed over the abdomen, chest, face, and extremities. The naevi were well-circumscribed, pigmented lesions without ulceration or discharge. An umbilical hernia was also noted.

Perineal examination revealed absence of a normal anal opening, consistent with imperforate anus (anal atresia). There was no passage of meconium per rectum. Abdominal examination was soft, non-distended, and non-tender, with no signs of acute intestinal obstruction at presentation. Cardiovascular, respiratory, and neurological examinations were within normal limits for a term neonate.

Postnatal investigations

Baseline hematological and biochemical investigations were within normal neonatal reference ranges. Abdominal ultrasonography was performed as part of the postnatal evaluation to screen for associated anomalies and revealed no gross renal or urinary tract abnormalities. Antenatal imaging records documenting single umbilical artery were reviewed and correlated with postnatal findings. Given the known association of anorectal malformations with multisystem anomalies, the neonate was planned for a structured evaluation protocol, including assessment of the genitourinary system, spine, and cardiovascular system, in coordination with pediatric surgery and neonatology teams.

Management and hospital course

The neonate was admitted to the Neonatal Intensive Care Unit (NICU) for close monitoring and further management. Enteral feeds were initially withheld, and the baby was started on intravenous fluids. A pediatric surgical consultation confirmed the diagnosis of anorectal malformation. In view of the imperforate anus and to prevent complications related to bowel obstruction and sepsis, a staged surgical management plan was formulated.

The presence of multiple melanocytic naevi, along with anorectal malformation and antenatal SUA, raised concern for possible syndromic or developmental associations. The parents were counseled regarding the diagnosis, need for staged surgical correction, possible associated anomalies, and the importance of long-term follow-up. The neonate remained clinically stable during the hospital stay and continued to receive multidisciplinary care involving neonatology, pediatric surgery, and dermatology services. Table 3 shows the diagnostic evaluation and management summary.

Table 3: Diagnostic Evaluation and Management Summary

Domain	Findings / Plan
Primary diagnosis	Anorectal malformation (anal atresia)
Associated antenatal anomaly	Single umbilical artery
Associated postnatal finding	Multiple cutaneous melanocytic naevi
Initial laboratory investigations	Within normal neonatal limits
Abdominal ultrasonography	No gross renal anomalies detected
Systemic screening	Planned (genitourinary, spine, cardiovascular)
Pediatric surgery consultation	Obtained
Initial feeding strategy	Nil per oral
Supportive management	Intravenous fluids
Surgical approach	Planned staged surgical management
Multidisciplinary involvement	Neonatology, Pediatric Surgery, Dermatology
Parental counseling	Provided
Clinical status during hospital stay	Stable

Fig 1, 2, 3: Cutaneous examination revealed multiple melanocytic naevi

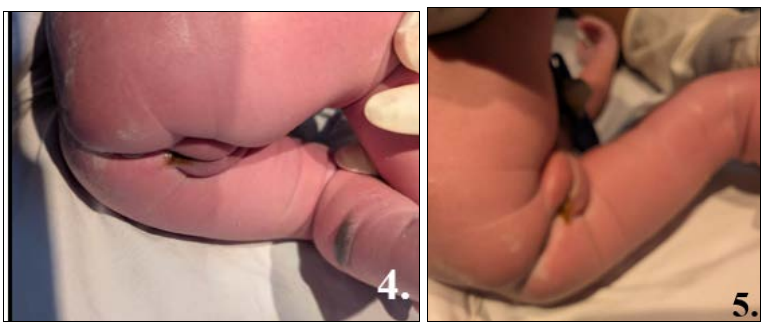

Figure 4, 5: Perineal examination confirmed anal atresia

Discussion

Anorectal malformations (ARMs) constitute a diverse group of congenital anomalies arising from abnormal development of the distal hindgut and cloacal structures during early embryogenesis. Their incidence has been reported to range between 1 in 4, 000 and 1 in 5, 000 live births, with significant variability in anatomical complexity and associated anomalies [1, 2]. The prognosis and long-term functional outcome in ARM are strongly influenced by the presence of associated congenital anomalies rather than the anorectal defect alone.

In the present case, anal atresia was associated with an antenatally detected single umbilical artery (SUA) and multiple congenital melanocytic naevi, an unusual and rarely documented combination. SUA is one of the most common umbilical cord abnormalities, occurring in approximately 0.5-1% of singleton pregnancies [3]. Although SUA may be isolated, several studies have demonstrated a significantly increased risk of associated congenital anomalies, particularly involving the cardiovascular, renal, gastrointestinal, and musculoskeletal systems [4]. Gastrointestinal anomalies, including anorectal malformations, have been reported with increased frequency in neonates with SUA, underscoring its value as an important antenatal marker [8].

The antenatal detection of SUA in this case prompted detailed fetal surveillance. Prenatal diagnosis of anorectal malformations remains challenging, as direct visualization of the anal opening is often not feasible, especially in the third trimester. However, indirect sonographic markers such as non-visualization of the perianal muscular complex, distal bowel dilatation, or associated anomalies may raise suspicion [5, 6]. In the present case, inability to satisfactorily demonstrate the perianal muscular complex on antenatal ultrasound allowed early anticipation of anal atresia and facilitated timely postnatal evaluation and surgical planning.

A distinctive and rare feature of this case was the presence of multiple congenital melanocytic naevi. Congenital melanocytic naevi result from abnormal migration or proliferation of neural crest-derived melanocytes and are observed in approximately 1% of neonates, although extensive or multiple lesions are uncommon [7]. While melanocytic naevi are primarily cutaneous findings, their coexistence with structural congenital anomalies raises the possibility of a shared embryological basis. Neural crest cells contribute not only to melanocyte development but also to elements of the enteric nervous system, suggesting a potential mechanistic link between pigmentary abnormalities and gastrointestinal malformations [9].

Classically, anorectal malformations are associated with the VACTERL spectrum—vertebral, anorectal, cardiac, tracheoesophageal, renal, and limb anomalies [8]. Cutaneous manifestations are not considered defining features of this association. However, sporadic reports have described pigmentary or neurocutaneous abnormalities coexisting with complex congenital malformations, indicating that broader developmental field defects may occasionally be involved [10]. The association of anal atresia, SUA, and multiple melanocytic naevi in the present case supports the need for a comprehensive and multidisciplinary approach to evaluation.

From a clinical standpoint, all neonates with anorectal malformations should undergo systematic screening for associated anomalies, including evaluation of the genitourinary system, spine, and cardiovascular system, even in the absence of overt clinical signs [2, 9]. Early identification of associated conditions allows optimization of surgical timing, anticipatory guidance, and long-term follow-up. The staged surgical approach planned in this case aligns with standard management principles aimed at reducing morbidity while optimizing functional outcomes.

Reporting rare associations such as the present case expands the existing literature on anorectal malformations and highlights the diagnostic importance of antenatal markers like single umbilical artery. Additionally, documentation of unusual cutaneous associations may stimulate further research into shared embryological pathways and improve awareness among clinicians managing neonates with complex congenital anomalies.

Conclusion

This case highlights a rare and unusual association of anal atresia with multiple cutaneous melanocytic naevi and antenatally detected single umbilical artery. While anorectal malformations are commonly associated with multisystem anomalies, the coexistence of significant cutaneous pigmentary abnormalities and single umbilical artery is infrequently reported. The antenatal identification of single umbilical artery and indirect sonographic markers facilitated early suspicion of anorectal malformation and enabled prompt postnatal evaluation and multidisciplinary management. This case underscores the importance of meticulous antenatal surveillance, thorough postnatal systemic evaluation, and a multidisciplinary approach in neonates with anorectal malformations. Reporting such rare associations contributes to the expanding phenotypic spectrum of anorectal malformations and may offer insights into shared embryological mechanisms. Long-term follow-up is essential to monitor surgical outcomes, associated anomalies, and potential neurocutaneous implications.

Declarations

Ethical Approval

Ethical approval was not required for this case report, as it describes a single patient and does not involve any experimental intervention. The case was managed as per standard institutional clinical protocols.

Consent for Publication

Written informed consent was obtained from the parents/legal guardians of the neonate for publication of the clinical details and accompanying images. All identifying information has been anonymized to ensure patient confidentiality.

Availability of Data and Materials

All data supporting the findings of this case report are included within the manuscript.

Competing Interests

The authors declare that they have no competing interests.

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Authors ’Contributions

All authors contributed to the clinical management of the patient, data collection, literature review, and preparation of

the manuscript. All authors read and approved the final manuscript.

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