CASE REPORT

Spontaneous Fetal Decapitation: A Rare Lethal Developmental Anomaly

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Abstract

Background: Spontaneous fetal decapitation or acephaly, is an extremely rare and universally fatal condition resulting from disruption of craniovertebral development during early embryogenesis. The absence of fetal head is usually found in twin reverse arterial perfusion (TRAP) syndrome in twin gestation. It is unusual in singleton pregnancy; only nine cases of acephaly have been reported to date. Amniotic band syndrome has been considered as a possible etiology for spontaneous decapitation in singleton gestation.

We present a case of a 32-year-old gravida 3 para 2 woman who was diagnosed with an acephalic fetus at 16 weeks of gestation. Ultrasonography (USG) revealed a fetus with a complete absence of cranial vault, brain tissue, and facial structures, along with abrupt widening of the cervical spine—findings consistent with spontaneous fetal decapitation. Medical termination with mifepristone and misoprostol resulted in the expulsion of an acephalic fetus.

Keywords: Acephaly, Case report, Craniovertebral anomaly, Lethal malformation, Prenatal ultrasound, Spontaneous fetal decapitation.

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Introduction

Spontaneous fetal decapitation, also known as acephaly, is among the rarest congenital anomalies documented in fetal medicine, with fewer than 8 to 9 cases reported worldwide.[1,2] It is characterized by the complete absence of the fetal head or cessation of development at the craniovertebral junction, resulting in the absence of the cranial vault, brain tissue, and facial structures.[3,4]

The anomaly originates during the fourth week of embryogenesis, when the rostral neural tube closes, and occipital somites fuse with cervical sclerotomes to form the craniovertebral junction.[5] Disruption in this highly sensitive developmental window—whether due to failed neuraltube closure, vascular insufficiency, or intrinsic mesenchymal weakness—can lead to the complete absence of skull-base formation.[6–8]

Several mechanisms have been proposed:

- Neuraltube closure defects: Acephaly is considered the most severe form of rostral neuraltube failure.[8–10]
- Vascular disruption: Early interruption of cephalic blood flow may result in ischemic necrosis of forming cranial structures.[11]
- Intrinsic craniovertebral developmental arrest: Mazzitelli et al. found the absence of amniotic bands in most cases, strongly supporting intrinsic developmental failure.[1]
- Amniotic-band sequence (ABS) – rare: Vitale et al. described an ABS-related decapitation, but most acephaly cases—including this one—lack limb defects or constriction rings typical of ABS.[3]
- Mechanical instability of early fetal mesenchyme: Proposed in older literature, suggesting malformed craniovertebral tissues may not withstand fetal movements.[7,11]
- Genetic contribution: Consanguinity and prior neuraltube defects have been noted in isolated cases.[12]

Across reported cases (Mazzitelli et al., Proffitt et al., Belhaouz et al.) diagnosis occurs between 11 and 18 weeks using ultrasonography (USG), which reliably demonstrates absent cranial structures and abrupt cervical termination.[1,2,12]

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Maternal histories generally show no teratogenic drug intake, no radiation exposure, and no systemic illness, supporting a sporadic developmental origin.[2,4] Given the uniformly fatal nature of acephaly, early antenatal care and first-trimester anomaly scans are essential for timely diagnosis and counseling.

Case Presentation

A 32-year-old G3P2L2 woman presented to the gynecology outpatient department (OPD) with a complaint of on-and-off bleeding for 1 week following a 2-month amenorrhea. Her last menstrual period (LMP) was 23 March 2025; she has a consanguineous marriage with a second-degree cousin. Her previous pregnancies were uneventful, resulting in healthy term infants.

She had no history of teratogenic drug exposure, including antiepileptic medications, retinoids, methotrexate, misoprostol, or cytotoxic agents. She denied any radiation exposure, fever, rash, illness, or symptoms suggestive of TORCH infections during early

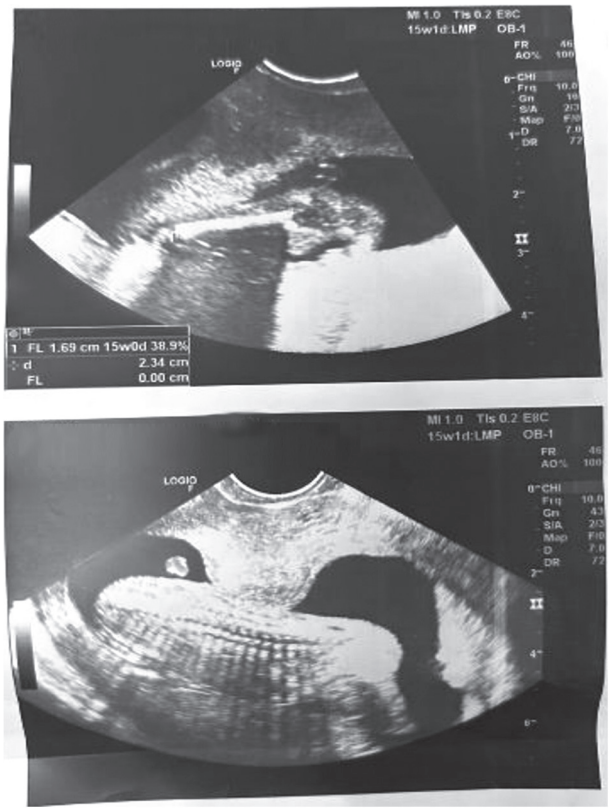


Fig. 1: Obstetric USG Showing complete absence of the fetal cranial vault , brain tissue and facial structures

pregnancy. No chronic medical conditions such as hypertension, thyroid disease, diabetes, or epilepsy were present. There was no family history of congenital anomalies. Routine antenatal investigations, folic acid supplementation, and progesterone were advised and were called for routine check-ups after 1 month.

After that, the patient was lost to follow-up and again presented to OPD at 16 weeks for an antenatal check-up. She was advised by USG obstetrics; her report shows a single live fetus of 15 weeks. Fetal cranial vault, brain tissue, and face are not visualized, with dilation of the cervical spine suggesting spontaneous decapitation (lethal anomaly) (Fig. 1).

After detailed counseling regarding prognosis and management options, medical termination was initiated with 200 mg mifepristone, followed by 800 µg vaginal misoprostol after 24 hours. Spontaneous expulsion of an acephalic fetus with placenta and membranes occurred after 48 hours (Fig. 2). Immediate recovery was uneventful, and she was discharged on the third day after counseling, with emphasis on slightly increased risk due to consanguinity, preconception folic acid supplementation and early first-trimester anomaly scans in future pregnancy.

Discussion

Spontaneous fetal decapitation is a profoundly rare congenital anomaly, with few cases described globally. The condition arises during early embryogenesis and results from severe disruption of the craniovertebral junction, where the skull base fuses with the upper cervical spine.[8]

Proffitt et al.[2] reported a case of intrauterine fetal decapitation secondary to amniotic band sequence diagnosed by prenatal ultrasonography. Vitale et al.[3] described a case associated with an ABS, but this involved limb defects, unlike most intrinsic cases. Mazzitelli et al.[1] reviewed earlier cases and concluded that most acephaly cases are intrinsic developmental disruptions, not ABS-related.

This case closely aligns with the intrinsic developmental category, owing to:

- Symmetrical absence of cranial structures.
- Normal limbs.
- No amniotic bands.
- No trauma.
- No teratogenic exposure.
- Presence of consanguinity, which may increase the risk of recessive anomalies.[12]

Pathogenetic Considerations

Neuraltube closure failure, vascular insufficiency, and intrinsic tissue instability are the most widely accepted mechanisms.[8,10,11] Vascular disruptions in early gestation have been proposed in earlier literature as a cause of sudden cranial resorption.[11]

USG Diagnosis

Ultrasound is diagnostic when showing:

- Absent cranial vault.
- Abrupt cervical termination.
- Intact thoracoabdominal anatomy.
- Absence of amniotic bands or trauma markers.

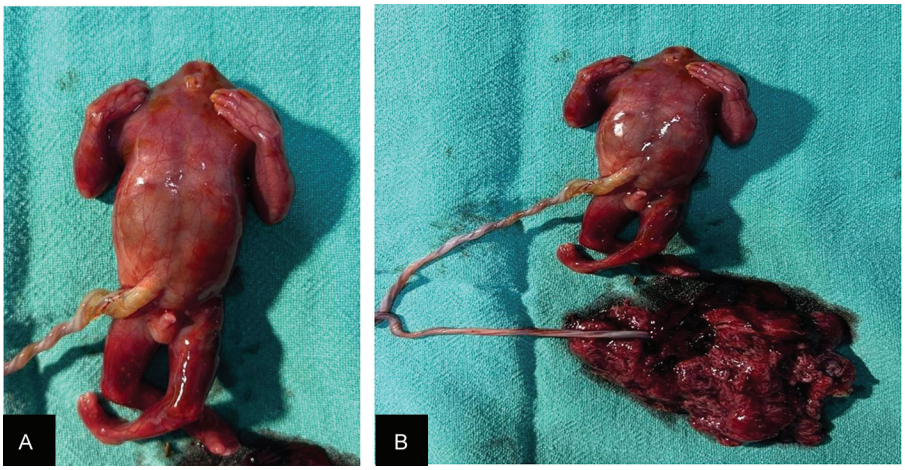
These criteria are consistent across previous reports.[2–4] Although amniotic bands were not visualized in our patient, the literature shows that they are frequently difficult to identify on ultrasound. Belhaouz et al. reported an acephaly case where the fetal head could not be identified even after delivery, noting that amputated fetal parts in amniotic band syndrome may undergo rapid autolysis or remain adherent to membranes. This supports the theoretical possibility—though unlikely in our case—that retained products noted 2 weeks later could represent degraded cranial remnants. Because the condition is uniformly fatal, termination is the recommended management once confirmed.[6]

Conclusion

Spontaneous fetal decapitation is an exceptionally rare and lethal congenital anomaly resulting from profound embryologic disruption of the craniovertebral junction. This case demonstrates classical sonographic findings, absence of external disruptive forces (such as amniotic bands), and relevance of consanguinity as a possible contributory factor. This case reinforces key clinical principles:

- Early antenatal registration is crucial.
- First-trimester anomaly scans enable early detection of lethal defects.
- A thorough maternal history, including teratogenic and radiation exposure, is vital.
- Genetic counseling is essential, especially in consanguineous marriages.
- Documentation of rare anomalies contributes to global understanding and improved diagnostic accuracy.

Early detection facilitates informed decision-making and reduces maternal physical and psychological morbidity. In addition,



Figs 2A and B: Gross specimen of an Acephalic fetus following medical termination of pregnancy

literature indicates that a detached fetal head may not always be identifiable at delivery due to autolysis or adherence to membranes, an important consideration when evaluating retained intrauterine tissue after expulsion of an acephalic fetus.

Declarations

Artificial Intelligence (AI) Disclosure

The authors declare that no AI tools or AI-assisted technologies were used in the preparation of this manuscript.

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Nil.

Authors' Contributions (CRediT Statement)

Data Collection: Surbhi Chauhan; Writing—Original Draft Preparation: Nidhi Gupta; Writing—Review and Editing: Shivani Singh. All authors have read and approved the final version of the manuscript.

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