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A Newborn with Major Congenital Anomalies of Amniotic Band Syndrome Associated with Acyanotic Congenital Heart Disease VSD, Oral Facial Cleft and Hydrocephalus: A Case Report

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ABSTRACT

Background: congenital anomalies are structural or functional anomalies that occur during intrauterine life and might be identified before, at birth or later in life. An estimated 6% of babies worldwide are born with a congenital disorder, resulting in 17% to 42% deaths among infants and 20% to 30% of stillbirths. In Tanzania, its prevalence is estimated to be 60.5 per 1000 live births. Majority of these are due to multifactorial including chromosomal anomalies. Its impact in our African societies extends to social consequences including stigmatization, divorce and witchcraft suspicions.

Case Presentation: we present a case report of syndromic baby with major congenital malformation associated with Oral facial cleft, Congenital hydrocephalus, Acyanotic congenital heart disease VSD, Amniotic band syndrome on left lower limb and fingers of upper limbs. This is the first case to be reported in Tanzanian Context

Keywords: Congenital Anomalies; Amniotic band syndrome; Acyanotic congenital heart disease VSD; Oral facial cleft; Hydrocephalus

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BACKGROUND

Congenital anomalies, or birth defects, are structural or functional abnormalities that occur during intrauterine development and can be identified at birth or later in life. These anomalies are categorized into major malformations, which have medical or social consequences (e.g., cleft palate, tetralogy of Fallot), and minor malformations, which do not directly affect health (e.g., transverse palmar crease, broad forehead). Some congenital anomalies are associated with multiple malformations affecting various body systems, including the vertebral, cardiac, and renal systems (Cloherty et al., 2021; CDC, 2021).

The pathophysiology occurrence of Congenital anomalies can be due to disruptions, which are due to extrinsic events that alter structures already developed normally as a result of compromised circulation, for examples, amputation of digits by amniotic band due to physical compression. Deformations due to physical forces action on previously formed structures including uterine crowding or oligohydramnios that results in clubfeet (Glynn & Drake, 2022). Dysplasia which is an abnormal cellular organization or function within a specific tissue type throughout the body resulting in abnormality such as skeletal and ectodermal dysplasia (Cloherty et al., 2021; CDC, 2021; Cortez-Ortega et al., 2025; Mi et al., 2024; Phan et al., 2023).

The causes of congenital anomalies are often multifactorial, with factors ranging from genetic mutations to environmental exposures, including maternal conditions like diabetes, smoking, and advanced maternal age. Despite significant global research, approximately 50% of congenital anomalies have unknown causes. In Tanzania, the prevalence of congenital anomalies is notably high at 60.5 per 1,000 live births, with profound clinical and social implications (Falsaperla et al., 2022; Ilham & Febriani, 2026). Beyond the medical challenges, affected families often face social stigma, divorce, and even witchcraft accusations, highlighting the need for greater awareness and healthcare interventions (CDC, 2021; Howley et al., 2023; Gosavi et al., 2025).

This case report is particularly significant in the Tanzanian context, as it presents a rare and severe congenital anomaly from a zonal referral hospital in the Lake Zone. It sheds light on the challenges of diagnosing, managing, and socially coping with such cases in Tanzanian healthcare settings, where data on congenital anomalies is limited (Falsaperla et al., 2022; Ilham & Febriani, 2026). This case aims to raise awareness and encourage more research into the burden and trends of congenital anomalies in Tanzania.

CASE PRESENTATION

A 30 years old, gravida 2, para 1+2, living 1, women presented at emergency maternity unity with chief complain of prevaginal bleeding for past 6 hours. She has history of two previous abortion, both at 12 weeks. Her pregnancy had 39 completed weeks of gestational age. She attended antenatal clinic five times and all her laboratory

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work-up including blood count indices, blood grouping, urinalysis, stool examination and fasting blood sugar were normal. She was HIV negative and blood film for malaria parasites were negative. Record of the prenatal ultrasound scan taken at 27 weeks GA reported a normal single viable fetus with no anomaly. She also received iron and folic acid supplements, Tetanus Toxoid vaccine twice, sulfoxide-pyrimethamine (SP) tables twice and Mebendazole. Furthermore, she had no history of either long medication uses or chronic illness. Socially, the mother had no history of illicit drug use, smoking, alcohol consumption, chemical/ radiation exposure or phenotypic genetic abnormalities in her families.

On assessment, she was normal tensive, fetus in longitudinal cephalic presentation, with fundal height measuring 43cm. The Fetal heart rate not heard. Per vaginally, she had active vagina bleeding, a bed side ultrasound revealed placenta Previa grade three with no fetal cardiac activities. She then, planned for emergency caesarean section.

OUTCOME

Male baby weighing 5 Kilograms with Apgar score of 2 at first minute then 0 at 5 minutes. He was a syndromic baby with hydrocephalus (occipital frontal circumference measuring 46cm), low set ears, exophthalmia eyes, oral facial cleft and complete absent of nasal septum. The baby had normal chest cage, abdominal wall with male genitalia, while, he had missing of fingers on right and left upper limb with amniotic band syndrome of left limb. On systemic assessment, he has bulging of both frontal and posterior fontanelles with poor/reduced primitive reflexes. While on cardiovascular findings, he has cold extremities. On the other hand, advanced diagnostic tools such as cardiac echocardiography were not performed, which could have provided more detailed and informative insights into the condition.

The final diagnosis was Major congenital malformation with Oral facial cleft, Congenital hydrocephalus, Acyanotic congenital heart disease VSD, Amniotic band syndrome on left lower limb and fingers of upper limbs.

DISCUSSION

This case report illustrates the heterogenicity and profound clinical implications of complex congenital malformations across multiple organ systems (Cloherty et al., 2021). Amniotic band syndrome (ABS) has wide spectrum of clinical presentation of unknown etiology. Its incidence ranges from 1:1200 to 1:15000 live births. In this case, the ABS have associated with life threatening craniofacial deformity, Congenital hydrocephalus and Acyanotic congenital heart disease VSD. More less similar case was reported by Allen et., al 2020 in Tanzanian lake zone, where, a Down syndromic newborn baby had hydrocephalus, amniotic band, ambiguate genitalia. Similarly, the study done

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in Benin found congenital heart disease being associated condition (Eshete et al., 2021), while other study done in Ethiopia, found the condition to be severely associated with cleft lip and palate (Chaulo et al., 2024). On top of that, one of the analytical cross-sectional studies among survived young infants, reported, maternal factors including lack of taking folic acid were associated with anomalies involving CNS (Howley et al., 2023; Gosavi et al., 2025; Rasul et al., 2025; Karacan et al., 2022; Mi et al., 2024; Hamed, 2024).

The exact etiology of the disease is still uncertain, however, there are theories describing the condition. Theory on extrinsic factors insists that, disruption of amniotic membrane as early event, which might be due to abnormal germinal disc development or traumatic disruption of the membrane. The disruption sought leading to extradition of fetal part out of the cavity along with certain fetal parts which gets entrapped and subjected to vascular compression, whereby, the constriction band created. While, the other theory, believes such anomalies are due to genetic basis. Finding of this case support genetic basis as probable cause of the disease, as ABS occurred concomitantly with craniofacial anomalies and hydrocephalus, to which was our case (Phan et al., 2023; Kalk et al., 2024; Wibowo, 2022). However, study done in Northern found that, the anomalies are 83% likely to occurs among mothers who did not took folic acid (Chaulo et al., 2024).

Usually, the newborn baby with such complex congenital anomalies, bears multiple medical, surgical, ethical, psychosocial and physical issues for themselves and family in general. Management of such cases needs supportive care with multidisciplinary team of neonatologist, pediatric surgeons, geneticist, cardiologist, physiotherapists and psychologists, which literally, become so much difficult to our low- and middle-income countries. Hence the survival of such cases becomes almost negligible to our settings (Doi et al., 2011; Mi et al., 2024; Kalk et al., 2024; Abuhamda, 2022; Sylejmani et al., 2025).

The World Health Organization argues its member states to capitalize on the introduction of surveillance systems program in order to establish trends and prevalences of the cases. Furthermore, public awareness on minimization of environmental factors including exposures to hazardous chemicals and timely taking of folic acid supplements are another crucial angle of intervention. Though it has cost implication, countries are encouraged to roll out preconception medical genetic screening and counselling among higher risk groups (Cloherty et al., 2021; Glynn & Drake, 2022).

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Figure 1. Front View of a Newborn Baby with MCAs

CONCLUSION

This case underscores the urgent need for improving prenatal screening policies and services in Tanzania. Early and precise prenatal diagnosis is crucial, not only for better clinical management but also for providing psychological support and preparing families for potential challenges. Enhanced prenatal screening would allow for early detection of congenital anomalies, enabling timely interventions, reducing the social stigma surrounding birth defects, and improving overall maternal and child health outcomes. Public health policies must prioritize the integration of genetic counseling and prenatal diagnostic services to better support families, reduce the burden of congenital anomalies, and improve health outcomes across Tanzania (Wibowo, 2022).

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