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Spectrum of congenital anomalies detected through anatomy ultrasound at a referral hospital in Ghana

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Abstract

Background Africa has a high burden of congenital anomalies due in part to limited preconception care, infections, and environmental exposures. However, the true prevalence of congenital anomalies is unclear because of insufficient access to prenatal diagnostic services. We aimed to determine the rate of congenital anomalies, and characterize the anomalies detected prenatally at a referral hospital in Ghana.

Methods We performed a four-year retrospective review of all fetal anomaly ultrasounds performed and congenital anomalies detected from January 1st, 2020, to December 31st, 2023, at Korle Bu Teaching Hospital, Accra, Ghana. Data were extracted from the electronic database on maternal age, gestational age at time of ultrasound, and occupation. Detected congenital anomalies were identified, and each anomaly was categorized by ICD-10 code and EUROCAT classification. Descriptive statistics were performed.

Results The mean maternal age and median gestational age at the time of ultrasound were 31.1 (SD 6.3) years and 26.9 (IQR 22.5–31.0) weeks, respectively. 3,981 anatomy ultrasounds were performed during the study period, and 7.0% (280/3,981) of fetuses had anomalies. Most (70.7%, 198/280) had anomalies detected in an isolated organ system. Anomalies were most identified in the central nervous system (CNS) (45.0%, 126/280), genitourinary (GU) (28.6%, 80/280), and gastrointestinal (GI) systems (21.8%, 61/280). The most common CNS anomaly identified was ventriculomegaly (70.6%, 89/126), out of which 26.2% (33/126) had severe ventriculomegaly, with an overall detection rate of 0.8% (33/3,981). The most common GU anomalies were congenital hydronephrosis (70.0%, 56/80), and congenital posterior urethral valves (28.8%, 23/80). The most common GI anomalies were exomphalos (49.2%, 30/61), and duodenal atresia (23.0%, 14/61). Unrelated to a specific organ system, 3.2% (9/280) of cases had hydrops and 6.1% (17/280) had an associated soft marker of aneuploidy.

Conclusions Our study highlights the substantial burden of congenital anomalies detected through prenatal ultrasound at a tertiary referral center in Ghana, with a notably high detection rate of severe ventriculomegaly. This work underscores the feasibility and importance of performing detailed anatomy ultrasounds in Africa. Beyond the

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clinical benefit, these data lay the groundwork for studies to identify the underlying causes of high rates of anomalies to inform preventive policy and clinical interventions in low-resource settings.

Keywords Ultrasound, Anomalies, Defects, Congenital, Low-middle income country, Low-resource settings, Ventriculomegaly

Background

Globally, congenital anomalies affect approximately 1 in 33 infants, resulting in 3.2 million birth defect-related disabilities annually [1, 2]. In Africa, the burden of congenital anomalies is particularly pronounced due to a lack of preconception care, limited access to prenatal care, high rates of infectious diseases, nutritional deficiencies, and environmental exposures [3]. These anomalies can lead to fetal demise, neonatal mortality, long-term disabilities, and significant emotional and financial strain on families. They also impose substantial costs on healthcare systems, especially in resource-limited settings where long-term care for affected children is challenging.

The capacity to detect congenital anomalies during pregnancy varies widely across the continent. In many African countries, access to quality prenatal ultrasound services is restricted by shortages of trained sonographers, inadequate healthcare infrastructure, and the high cost of these diagnostic equipment [4–6]. Data on congenital anomalies in low- and middle-income countries (LMICs) are scarce, and most of the available information comes from postnatal studies in a limited number of countries. As a result, the reported prevalence rates of congenital anomalies in Africa are likely underestimates of the true burden. This may also be in part due to the under or missed reporting of congenital anomaly cases resulting in in-utero or perinatal demise.

Accurate antenatal detection of certain congenital anomalies is known to improve pregnancy outcomes by enabling the implementation of appropriate in-utero and perinatal multidisciplinary monitoring and management strategies. However, there are significant disparities in the access to these high-quality services across different geographic locations around the world. A systematic review reported significant variability in the frequency of antenatal ultrasound examinations, with a median of 50.0% in Africa compared to 90.7% in Asia [7]. These disparities highlight the limited availability of detailed anatomy scans in many LMICs, which likely contributes to differences in the reported prevalence of congenital anomalies. Despite advancements in prenatal care, significant challenges persist in resource-limited settings, including inadequate training, limited access to high-quality equipment, and inconsistent antenatal care practices [2, 7].

There is a pressing need to systematically analyze the spectrum of congenital anomalies detected through prenatal ultrasound scans to inform targeted interventions, enhance counseling for expectant families, and develop effective public health strategies [2, 8]. Risk factors for congenital anomalies in LMICs may differ from those in high-income countries (HICs) due to variations in genetic, environmental, nutritional, and infectious exposures. Consequently, the prevalence and types of anomalies seen in LMICs may also differ.

Methods

Study site and patient population

This is a four-year facility-based retrospective review of fetal anatomy ultrasound reports conducted between January 1, 2020, and December 31, 2023, at Korle Bu Teaching Hospital (KBTH), a leading tertiary referral center in Accra, Ghana. KBTH has an average annual delivery rate of 8,000 births and a broad catchment area that includes the Greater Accra Region, Central Region, Eastern Region, and other parts of Ghana. At KBTH, it is recommended that all pregnant women undergo a detailed anatomy ultrasound, usually scheduled between 18 and 22 weeks of gestation, or beyond this period, at the time of referral, to optimize prenatal diagnosis and care.

The study population comprised all pregnant women who underwent fetal anatomy ultrasound during the study period. This included women attending routine anatomy ultrasounds as well as those referred to KBTH from other facilities due to suspected anomalies or abnormal growth findings.

Study procedures

All ultrasounds were performed by maternal-fetal medicine (MFM) consultants or fellows in training who were certified by the International Society of Ultrasound in Obstetrics and Gynecology (ISUOG). All practitioners had completed the ISUOG 20 + 2 guidelines training for detailed anatomy evaluation and the Fetal Medicine Foundation course on fetal abnormalities [9]. Most of the scans were done using a GE LOGIQ 9 ultrasound machine.

Routine mid-trimester fetal anatomy scans followed the ISUOG 20 + 2 protocol, which includes a systematic assessment of 20 key anatomical structures, supplemented by two additional views under specific clinical indications. Central nervous system (CNS) areas assessed included the brain, ventricles, midline falx, choroid plexus, and posterior fossa. Cardiac evaluation involved the four-chamber view, outflow tracts, and the three-vessel and trachea view. Abdominal organs examined

included the stomach, kidneys, and bladder, along with a detailed assessment of the face profile, orbits, palate, nose, lips, diaphragm, spine, and extremities.

For suspected anomalies referred from other centers, targeted detailed morphology scans were performed. These extended beyond the standard protocol to include additional imaging planes, focused biometry, and Doppler interrogation where relevant. Extended neurosonography or comprehensive cardiac assessments were performed when indicated. All findings were systematically recorded using standardized templates and subsequently reviewed by a senior fetal medicine consultant for confirmation and quality assurance.

Detailed anatomy ultrasound reports were archived in a customized Microsoft SQL Server database (2017 version), which was established in 2019 as part of the Hospital's Maternal-Fetal Medicine Unit's initiative to enhance service delivery and training.

The research team comprehensively reviewed each ultrasound report, including the corresponding ultrasound images and videos, to verify the primary diagnosis and cross-referenced it with the documented ultrasound findings ensuring accuracy and consistency. Once the main diagnosis/anomaly was confirmed, it was classified using the EUROCAT classification system, and a corresponding ICD-10 code was assigned. Coding was performed independently by two trained research assistants to maintain reliability. Any discrepancies were resolved through discussion and consensus, while particularly complex cases were reviewed by the MFM team at KBTH to determine the final coding. Regular review meetings were held by the research team to address complex or ambiguous cases and ensure standardized coding practices. Severe ventriculomegaly cases were categorized based on a reported lateral ventricular dilation of ventricle atrial diameter of ≥ 15 mm and/or any comment in the ultrasound report indicating hydrocephalus was present. All coded data were securely stored in a password-protected Excel database, with patient identifiers removed or anonymized for confidentiality. Ethical approval was obtained from the Institutional Review Board at KBTH (KBTH-STC 000140/2024).

Maternal demographic data including age, occupation, residence, and date of delivery was obtained through chart review. Determination of formal versus informal employment was adjudicated by study staff. Estimated gestational age (EGA) at the time of the ultrasound was calculated from the estimated due date (EDD) reported on the booking ultrasound scan. For cases with missing data, efforts were made to contact patients directly through phone calls using the contact information available in their medical records. When direct outreach was unsuccessful, data were marked as missing after multiple attempts, ensuring transparency and consistency in data reporting.

Statistical methods

The primary outcome was the rate of congenital anomalies, defined as the proportion of fetuses with one or more anomalies (excluding isolated soft markers and isolated hydrops), out of the total number of fetuses who underwent detailed anatomy ultrasound. Multifetal pregnancies were included, with each fetus assessed individually. No multifetal pregnancy had more than one fetus affected by an anomaly. Isolated cases of hydrops were excluded unless a structural anomaly was present, as hydrops can result from non-structural causes such as immune conditions, anemia, or infection, and our study focused on structural anomalies. Secondary outcomes included maternal demographics, the frequency of anomalies by organ system, the frequency of anomalies categorized according to ICD-10 code per organ system, and the frequency of secondary CNS anomalies associated with ventriculomegaly. Descriptive statistics were performed for maternal demographics including percentages, medians and interquartile ranges. The frequency of anomalies by organ system was calculated from the number of fetuses with an anomaly in each organ system out of the total number of fetuses with an anomaly. The frequency of specific anomalies by ICD-10 code was calculated from the number of fetuses with a given ICD-10 code divided by the total number of fetuses with an anomaly in the corresponding organ system. The frequency of secondary CNS anomalies associated with ventriculomegaly was calculated by first dividing them into mild to moderate (10–15 mm) versus severe ventriculomegaly (≥ 15 mm lateral ventricle atrial diameter). Within these subgroups we then reported the frequency of fetuses with additional CNS anomaly by their ICD-10 codes. Statistics and graphs were generated using Microsoft Excel (2024).

Results

After excluding repeat ultrasounds ($n = 29$), 3,981 detailed anatomy ultrasounds were performed at KBTH from January 1st, 2020, to December 31st, 2023. Anomalies were detected in 280 (7.0%, 280/3,981) fetuses (Fig. 1).

Most cases (70.7%, 198/280) had anomalies detected in a single organ system, with 131 (46.8%, 131/280) cases having only an isolated anomaly detected. The mean maternal age and median estimated gestational age at the time of ultrasound were 31.1 (SD 6.3) years and 26.9 (IQR 22.5–31.0) weeks, respectively. Most women were multiparous (83.9%, 167/199) and reported informal employment status, which refers to work that is not regulated or protected by formal labor laws—such as petty trading,

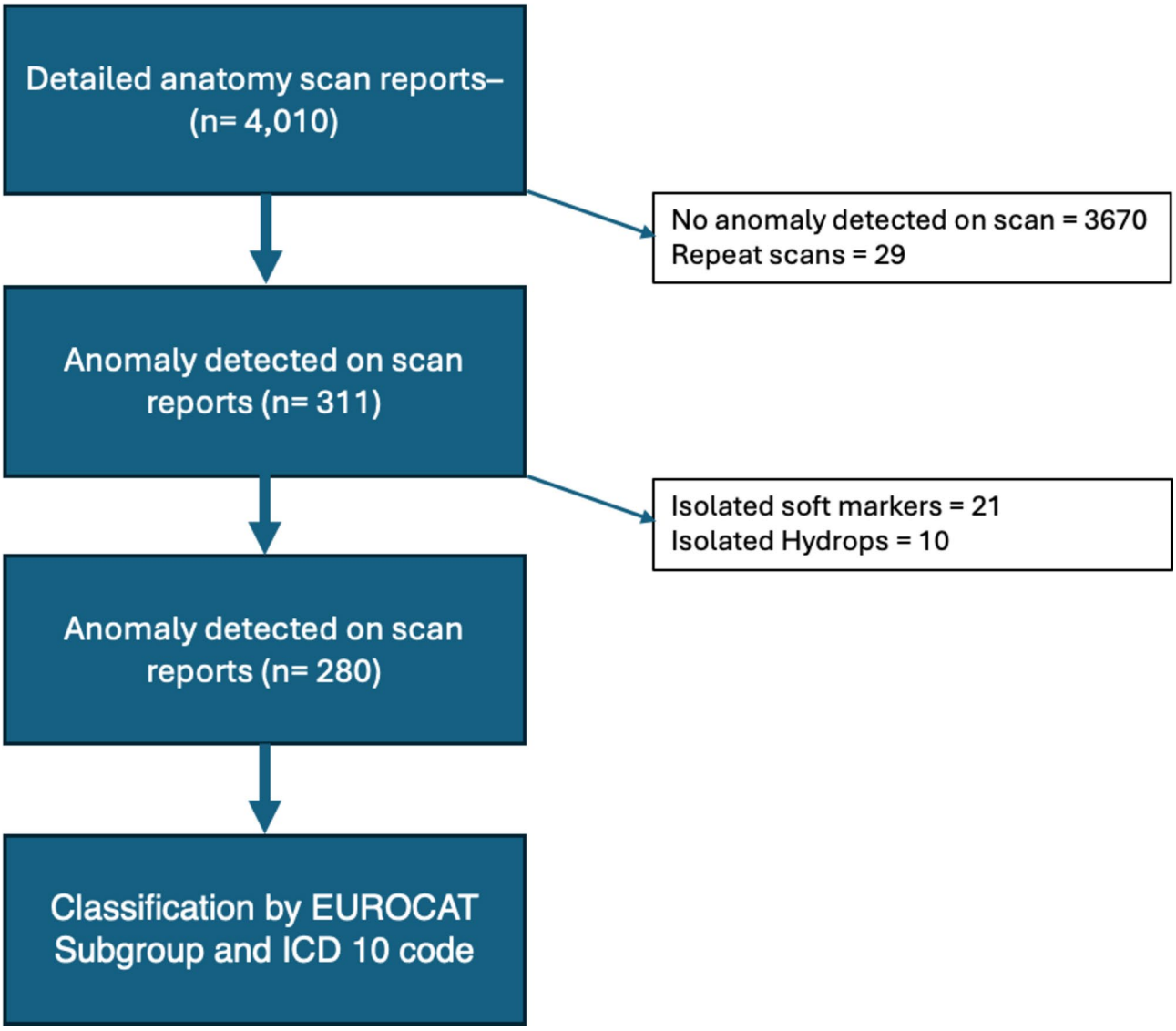

Fig. 1 Flow diagram of congenital anomaly report selection process. All detailed anatomy ultrasound reports performed in the four-year study period at KBTH were isolated, and any report without an anomaly identified or repeat scan for an identical patient was removed. Additionally, isolated soft marker and hydrops cases were removed to obtain the final selection of congenital anomaly reports for analysis

subsistence farming, domestic work, or self-employment without official registration (68.2%, 148/217) (Table 1).

The primary organ systems affected were the central nervous system (CNS) (45.0%, 126/280), genitourinary (GU) (28.6%, 80/280), and gastrointestinal (GI) systems (21.8%, 61/280) (Fig. 2).

The most common CNS anomaly ICD-10 code was "Other specified congenital malformations of brain" (70.6% of CNS cases, 89/126), which entirely comprised ventriculomegaly cases. In addition to ventriculomegaly, we identified 9 cases of anencephaly and 6 cases of spina bifida, which were recorded separately (Appendix 1). Among the ventriculomegaly cases, 33 (37.1%, 33/89) had severe ventriculomegaly (defined as a lateral ventricle atrial diameter ≥ 15 mm). Of these cases of severe ventriculomegaly (SVM), 25 were isolated cases of SVM with no other CNS lesion identified, 5 were associated with spina bifida, 2 with Dandy-Walker Malformation, and 1 with holoprosencephaly (Table 2). Additionally, 8 (24.2%, 8/33) of the SVM cases had no other associated CNS anomalies. Representative images of SVM captured at KBTH are shown in Fig. 3.

Of the mild-to-moderate ventriculomegaly (VM) cases, 33 (58.9%, 33/56) were isolated with no other associated CNS malformation identified, 4 (7.1%, 4/56) were associated with spina bifida, 9 (16.1%, 9/56) with Dandy-Walker Malformation, and 6 (10.7%, 6/56) with holoprosencephaly (Table 2). Additionally, 22 (39.3%, 22/56) of the mild-to-moderate cases had associated non-CNS anomalies.

Table 1 Maternal demographic characteristics for fetuses with congenital anomalies

Demographic Characteristic		*n (%) or median (IQR) Totaln = 280*
Age (years) (n = 280)	**< 25**	43 (15.4%)
	25–30	91 (32.5%)
	31–35	68 (24.3%)
	> 35	78 (27.9%)
Parity[a] (n = 199)	**0**	32 (16.1%)
	1	58 (29.1%)
	2–3	81 (40.7%)
	≥ 4	28 (14.1%)
Estimated Gestational Age at Diagnosis (in weeks) (n = 280)	**< 14wks**	3 (1.1%)
	14–24wks	87 (31.1%)
	> 24wks	190 (67.9%)
Employment[b] (n = 217)	**Informal**	148 (68.2%)
	Formal	53 (24.4%)
	Unemployed	16 (7.3%)

a: Only 199 out of the total 280 cases had a documented parity

b: Only 217 out of the total 280 cases had documented employment status

Table 2 CNS congenital anomalies associated with cases of ventriculomegaly

	Associated CNS Anomaly/Lesion	Reported number of cases (%)
Severe Ventriculomegaly (> 15 mm) (*N* = 33)	No other CNS lesion	25 (75.8%)
	Spina bifida[a]	5 (15.2%)
	Dandy-Walker Malformation	2 (6.1%)
	Holoprosencephaly	1 (3.0%)
Mild to moderate Ventriculomegaly (*N* = 56)	No other CNS lesion	33 (58.9%)
	Spina bifida[a]	4 (7.1%)
	Congenital cerebral cysts	1 (1.8%)
	Holoprosencephaly	6 (10.7%)
	Congenital malformations of corpus callosum	4 (7.1%)
	Dandy Walker Malformation	9 (16.1%)
	Other congenital malformations of brain	1 (1.8%)

a: one of these cases was also associated with microcephaly

The most common GU anomaly ICD-10 codes were "Congenital hydronephrosis" (70.0% of GU cases, 56/80) and "Congenital posterior urethral valves" (28.8% of GU cases, 23/80). The most common GI anomaly ICD-10

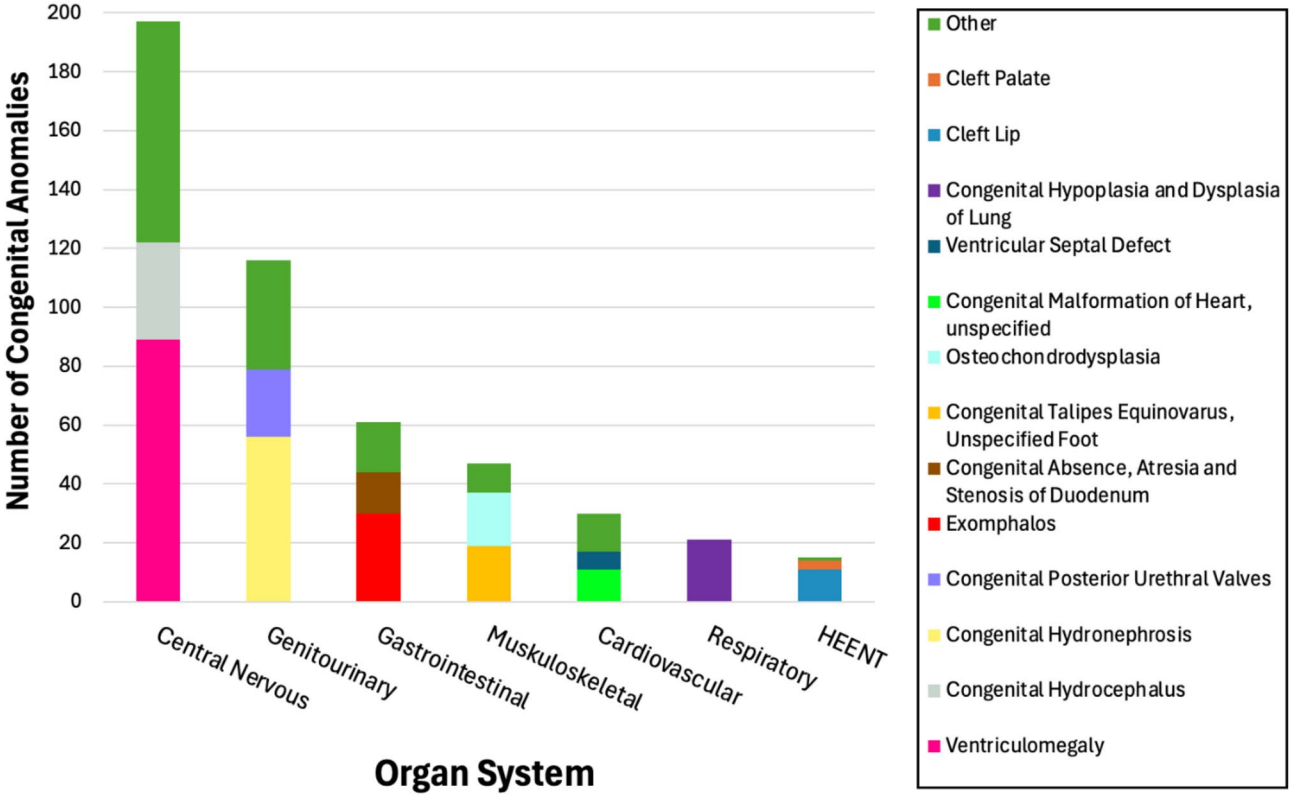

Fig. 2 Congenital Anomalies Identified in Detailed Anatomy Ultrasound by Organ System. Congenital anomalies as identified and reported by ICD-10 and EUROCAT classification were sorted by the organ system affected. The figure reports the absolute number of each reported anomaly, so a fetus with multiple anomalies may be accounted for multiple times. The top two most prevalent diagnoses were reported for each respective organ system. The "Other" category includes the combination of all other ICD-10 diagnoses that were not included in the top two most prevalent diagnoses for each organ system. HEENT: head, eyes, ears, nose, throat system

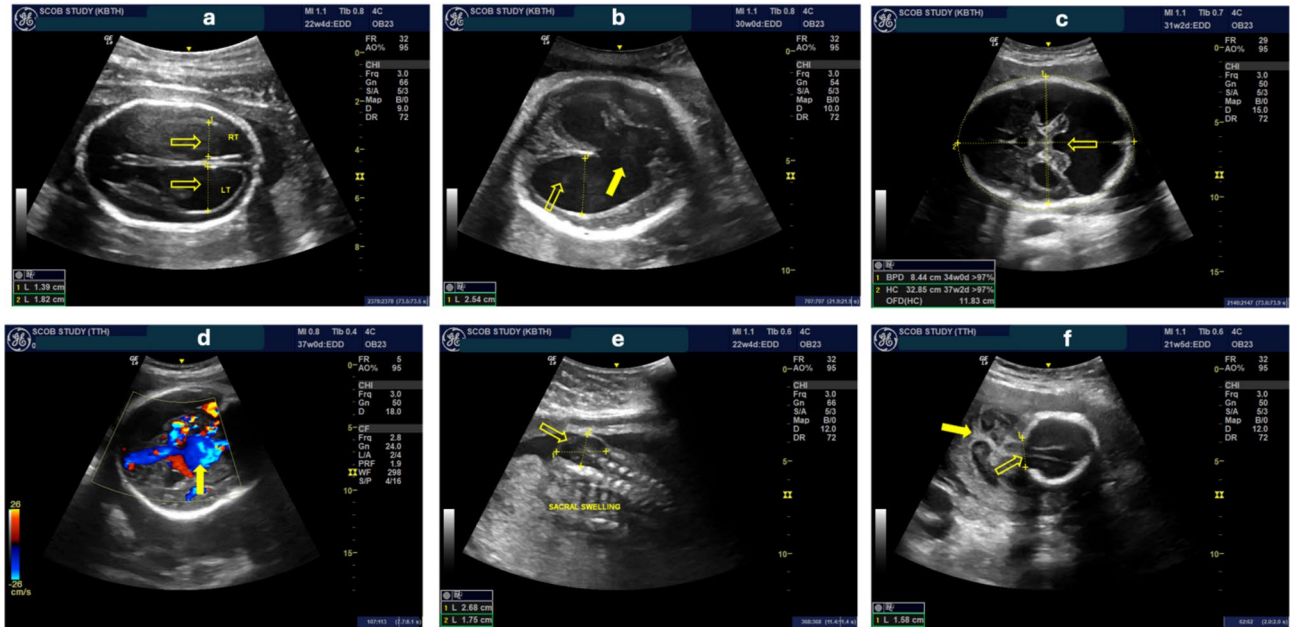

Fig. 3 Representative ultrasound images identified congenital central nervous system anomalies. **(a)** Severe ventriculomegaly with dilatation of the posterior horns of the lateral ventricles (hollow arrows); **(b)** Severe ventriculomegaly (hollow arrow) with fenestration of the falx cerebri (solid arrow); **(c)** Dandy-Walker malformation showing severe hypoplasia of the cerebellar vermis (hollow arrow) and an enlarged fourth ventricle; **(d)** Vein of Galen malformation presenting as a cystic lesion with bidirectional color flow on Doppler (hollow arrow); **(e)** Open spina bifida with associated myelomeningocele (hollow arrow); **(f)** Encephalocele with a large posterior cranial defect (solid arrow) and herniated brain tissue (hollow arrow)

codes were "Exomphalos" (49.2% of GI cases, 30/61) and "Congenital absence, atresia and stenosis of duodenum" (23.0% of GI cases, 14/61) (Fig. 2). Representative images of congenital hydronephrosis and gastroschisis captured at KBTH are shown in Fig. 4.

Musculoskeletal (MSK) and cardiovascular anomalies were also identified in 15.4% (43/280) and 10.0% of the anomaly cases (28/280), respectively. A comprehensive list of ICD-10 codes and associated EUROCAT classification systems for all the identified anomalies is available in Supplemental Table 1.

Discussion

This study provides the first review of antenatally-detected congenital anomalies in Ghana, demonstrating the utility of implementing routine anatomy ultrasounds in a resource-limited setting. Among 3,981 ultrasounds conducted at Korle-Bu Teaching Hospital over a four-year period, we identified a 7% rate of congenital anomalies, with most cases affecting an isolated system and CNS anomalies being the most common. Our findings highlight the potential impact of prenatal anomaly screening in improving the detection of congenital anomalies particularly in resource-limited settings.

The rate of congenital anomalies in our study (7%) is higher than reported rates in high-income regions such as the United States (3.0%) and Europe (2.4%) [10, 11]. Several factors may explain this high rate. First, our study was conducted in a tertiary referral hospital, where we receive a high proportion of complicated pregnancies, including referrals specifically for suspected anomalies. As such, our data includes both routine mid-trimester fetal morphology scans and detailed morphology scans for suspected anomalies, which likely enriched the study population for abnormalities. Second, like many studies from Africa, our findings are based on hospital-based populations, which tend to reflect a higher-risk cohort compared to the community- or population-based studies typically conducted in Western countries [12–15]. Additionally, the recent establishment of a maternal-fetal medicine unit at our center has expanded access to high-resolution anomaly scanning, contributing to improved detection rates.

In our study, congenital anomalies were detected in 7.0% of fetuses who underwent anatomy scans. This detection rate aligns with or exceeds rates reported from other hospital-based studies in African countries, including Egypt (7.4%), Ethiopia (18.5%), Kenya (19.4%) and Nigeria (11.1%) [12–15]. These figures are markedly higher than population-based prevalence estimates reported in countries such as India (1.8%), Iran (2.3%), and the United Kingdom (1.3%) [16–18], likely reflecting differences in study design, access to diagnostic services, and underlying risk profiles.

The elevated rates in African contexts may reflect a combination of environmental and socio-economic factors. Key risk factors include teratogenic exposures such as maternal infections (e.g., cytomegalovirus, rubella, and

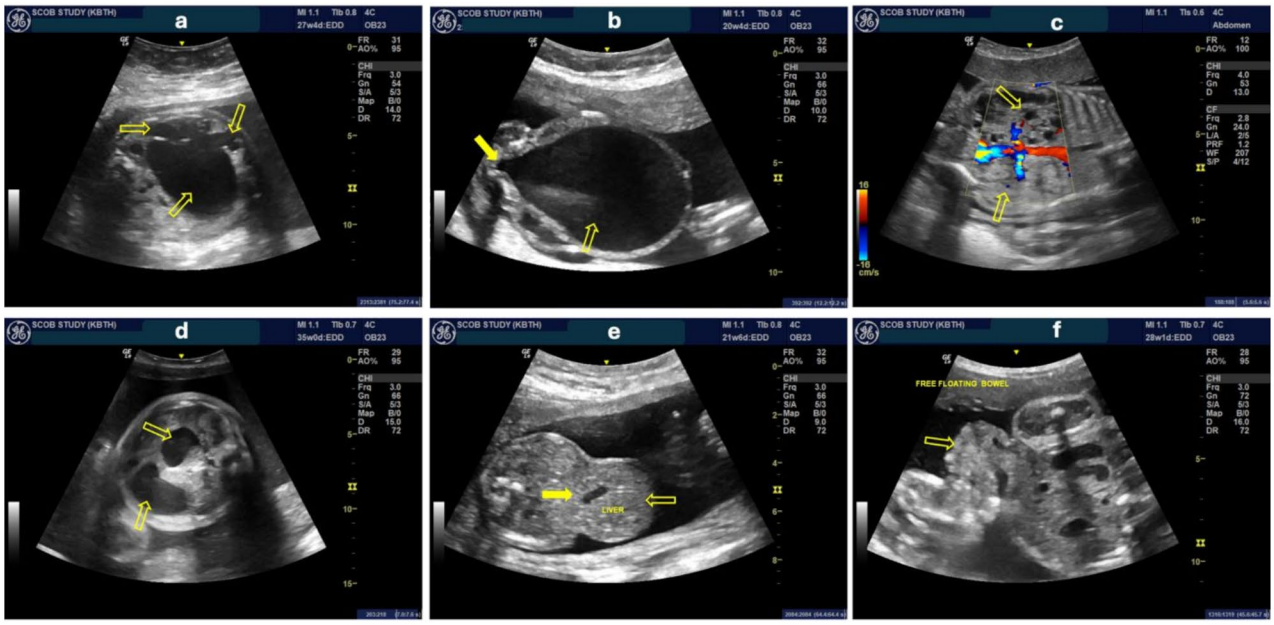

Fig. 4 Representative ultrasound images of identified congenital gastrointestinal and genitourinary anomalies. **(a)** Severe (Grade 3) hydronephrosis with associated ureteral dilation (hollow arrows); **(b)** Megacystitis (hollow arrow) with a "keyhole sign" indicating posterior urethral valves (solid arrow); **(c)** Multicystic dysplastic kidney showing an enlarged, hyperechoic kidney with multiple non-communicating cysts (hollow arrows); **(d)** "Double bubble" sign suggestive of duodenal atresia (hollow arrows); **(e)** Omphalocele with an abdominal wall defect (solid arrow) and herniated viscera enclosed in a membrane (hollow arrow); **(f)** Gastroschisis with free-floating bowel loops in the amniotic fluid (hollow arrows)

toxoplasmosis) and environmental pollutants [11]. Additionally, lack of folic acid supplementation, late initiation of prenatal care, and the presence of chronic diseases can further increase the risk of these anomalies. There may be genetic factors contributing to these elevated rates, such as higher rates of inherited abnormalities, founder effects, or consanguineous marriages. The use of certain drugs and uncontrolled herbal medications during pregnancy adds to these risks, highlighting the need for comprehensive prenatal care and public health interventions [19].

Central nervous system (CNS) anomalies were the most frequently detected anomalies in our study, comprising 45% of cases. Similar trends in CNS anomalies were observed in Ethiopia (66%) and Kenya (48%) [20, 21]. Our findings particularly highlighted the predominance of severe ventriculomegaly, where we found 33 cases /3981 total ultrasounds (829 per 100,0000). This elevated figure is even more pronounced compared to the reported values in the systematic review by Dewan et al., where they elucidated the highest incidence of congenital hydrocephalus in Africa (145 per 100,000 births) and Latin America (316 per 100,000 births) compared to HICs, such as the United States/Canada (68 per 100,000 births) [22]. It is unclear why rates of SVM are nearly 8-fold higher than what has been reported elsewhere in Africa. One key difference between this meta-analysis and our data is that the meta-analysis focused on cases of post-natally diagnosed hydrocephalus whereas this current study reviewed antenatally diagnosed cases following detailed ultrasounds. It is possible that fetal or neonatal demise may result in lower reported rates of infantile hydrocephalus. However, other factors may also play a role. Future research should explore the genetic, environmental, and socio-economic factors associated with SVM, and assess the impact of interventions such as folic acid supplementation and prevention of infections known to be associated with CNS anomalies, including cytomegalovirus, toxoplasmosis, and Zika virus.

Comparatively, genitourinary (GU) anomalies were the most frequently reported anomalies in studies from Nigeria and Saudi Arabia, while these ranked second in our findings (28.6%) [23, 24]. Variations in the distribution of anomalies across regions may stem from differences in risk exposures, such as nutritional deficiencies, infections, or environmental factors, as well as disparities in healthcare access and diagnostic capabilities. Genetic factors may also play a role, including undiagnosed inherited conditions, and possible consanguinity.

Limited access to routine antenatal care, diagnostic resources, and expertise could potentially lead to underdetection of certain anomalies, resulting in variations in reported prevalence across regions. Regions with better-equipped healthcare systems and trained personnel can identify a broader range of anomalies, while

resource-limited settings may only detect more apparent conditions.

Interestingly, the detection rate of cardiac anomalies in our study was relatively low (10.0% of total identified anomalies) compared to global trends, where cardiovascular defects are commonly reported as the most frequent severe congenital anomalies [7, 25]. This lower detection rate may reflect limitations in operator expertise and equipment, particularly since diagnosing cardiac anomalies often requires specialized skills and advanced imaging techniques like fetal echocardiography. In resource-limited settings, the lack of trained personnel and high-resolution equipment makes it challenging to identify subtle or complex heart defects. These disparities highlight the need for targeted training and technological advancements to improve diagnostic accuracy and enhance the detection of cardiac anomalies in the Ghanaian setting.

The median estimated gestational age at diagnosis in our study was 26.9 weeks, highlighting a delay compared to the recommended window of less than 24 weeks gestation for second-trimester anatomy ultrasounds [26]. This delay may be attributed to late initiation of antenatal care (ANC), as many pregnant women in low-resource settings face barriers such as significant travel distances to access ultrasound services or initiate care late in pregnancy [27, 28]. The timing of diagnosis plays a critical role in the subsequent management of fetal anomalies. Early ultrasound screening (< 14 weeks) can detect up to 33% of anomalies, with combined first- and second-trimester ultrasounds improving detection rates to 83% [26]. However, second-trimester ultrasounds remain crucial for identifying most structural anomalies, where major anomalies were primarily detected in the absence of first-trimester screening programs [26, 29, 30]. The late timing of detailed anatomy ultrasounds in our cohort limited the opportunities for early interventions, such as termination before viability or in-utero management, including procedures such as in-utero shunting for lower urinary tract obstruction, and fetal surgery for conditions like spina bifida. Moreover, delayed ultrasounds can compromise the accuracy of EDDs and EGAs, potentially impacting pregnancy outcomes by leading to incorrect assessments of fetal growth, mistimed interventions, or inappropriate decisions about delivery timing.

A critical challenge to address for improving antenatal care in Ghana is ensuring that the infrastructure and expertise demonstrated in this study can be scaled up and sustained across the country. While this study successfully performed detailed anomaly ultrasounds, this was made possible by the presence of maternal-fetal medicine specialists and access to appropriate ultrasound equipment at this tertiary referral center. While no universal benchmark exists, estimates from high-resource settings suggest one MFM specialist per 2,000–5,000 births and one sonographer per 750–1,000 scans per year [31]. Ghana, with fewer than 15 MFM specialists and limited numbers of trained sonographers for a population exceeding 30 million, falls significantly short of these estimates. Also, the limited availability of high-quality ultrasound equipment remains significant barriers to widespread implementation [32–34]. Addressing these challenges through standardized training programs and investments in diagnostic infrastructure is essential to replicate the quality of anomaly detection seen in this study across more facilities in Ghana and similar low-resource settings.

Strengths and limitations

There are multiple strengths to this study. First, it is the only study, to our knowledge, to report on the rate and spectrum of congenital anomalies identified antenatally in Ghana, providing essential baseline data for the region. This addresses a significant knowledge gap in the literature, and this can help inform clinical practice and health policy interventions. Second, the study employed a systematic approach to anomaly identification and classification, using ICD-10 and EUROCAT standards to ensure consistency and accuracy. Thirdly, the large and diverse sample size enhances the generalizability of the findings from this study. Finally, by focusing on antenatal detection, the study demonstrates the feasibility of performing detailed anatomy ultrasounds in a resource-limited setting and highlights the opportunities for pregnancy termination, preparation for immediate postnatal care, delivery planning, and patient counseling. This serves as a model for expanding antenatal care in similar low-resource regions.

There are also limitations to this study. Inherent to the retrospective study design, we were limited by missing data and the inability to confirm anomalies postnatally, which may have affected diagnostic accuracy. Additionally, data on the underlying causes of anomalies—such as genetic, environmental, or infectious etiologies—were not available for this retrospective analysis. Given the limited access to digital resources and unreliable internet access, demographic data and other data points such as gravidity, parity, and EGA were not reported for all patients. Additionally, most anomalies were detected at a late gestational age (median: 26.9 weeks). As the fetus matures, anomalies often become more apparent due to better anatomical differentiation. However, challenges such as fetal position and limited imaging windows, particularly in later gestation, can impact detection rates. Furthermore, as a referral center, anomalies are often identified at peripheral facilities and referred to the FMU for confirmation and management, contributing to the

later gestational age at detection. This pattern is consistent with referral practices worldwide.

Although KBTH records approximately 8,000 deliveries annually, only 3,981 detailed anatomy scans were included in this review. This discrepancy is partly due to financial barriers, as ultrasound is not covered by Ghana's National Health Insurance Scheme and must be paid for out-of-pocket. Additionally, some women presented as referrals in labor, and others with scans from external facilities, which were not captured in our database. As such, the anomaly rate reported reflects only those scanned at KBTH and does not represent the true prevalence in the broader population.

Despite these limitations, our findings have important clinical and research implications. On an individual level, the study provides essential insights into the spectrum and rate of congenital anomalies detected through antenatal ultrasounds, critical for clinicians counseling affected families and managing these complex pregnancies. At the hospital and national levels, the results inform public health policy for preventive interventions including public education, and resource allocation that prioritizes investments in training, diagnostic equipment, and maternal-fetal medicine services. On a global level, the study highlights differences in the rates and types of congenital anomalies between LMICs and HICs. These differences are crucial for shaping global health policies, understanding regional health disparities, and guiding research into the genetic, environmental, and socio-economic factors contributing to anomalies. This knowledge can help improve prenatal care worldwide and inform future research aimed at reducing the burden of congenital anomalies in LMICs.

The differences in the prevalence of congenital anomalies between LMICs and HICs highlight the need for region-specific approaches to patient counseling, resource allocation, and research prioritization. Future research should investigate the causes of congenital anomalies in Ghana, focusing on genetic, environmental, and infectious factors. Postnatal follow-up and long-term outcome studies are needed to understand the natural history and impact of these conditions. KBTH serves as a model for demonstrating the feasibility of conducting high-quality anomaly ultrasounds in LMICs, despite resource constraints. By categorizing anomalies by organ system and identifying trends, this research lays the groundwork for improving prenatal diagnostics, and corrective/therapeutic interventions in appropriately selected cases to optimize newborn outcomes.

Conclusions

We report a 7% rate of congenital anomalies at the largest referral hospital in Ghana. There was a variety of congenital abnormalities seen, with a significant proportion (70%) identified in the CNS, and a particularly high frequency of severe ventriculomegaly. This study demonstrates, that with access to the appropriate ultrasound technology, LMICs, such as Ghana, can identify these anomalies during the antenatal period. Timely antenatal diagnosis of such anomalies is critical to allow for patient decision-making, intervention, and planning. Given the significant number of patients currently receiving detailed anatomy ultrasounds late in pregnancy, after the period of feasible intervention, more concerted efforts are needed to encourage and complete such screening procedures earlier in pregnancy in Ghana. Additionally, given the high rate of CNS abnormalities among this patient population, further research is needed to investigate the inciting genetic, environmental, and infectious factors, as well as other factors contributing to such high rates of congenital anomalies.

Abbreviations

CNS	Central nervous system
GU	Genitourinary
GI	Gastrointestinal
LMICs	Low- and middle-income countries
HICs	High-income countries
KBTH	Korle Bu Teaching Hospital
MFM	Maternal-fetal medicine
ISUOG	International Society of Ultrasound in Obstetrics and Gynecology
EGA	Estimated gestational age
EDD	Estimated due date
HEENT	Head, eyes, ears, nose, throat system
VM	Ventriculomegaly
SVM	Severe ventriculomegaly

Supplementary Information

The online version contains supplementary material available at https://doi.org/10.1186/s12884-025-07640-x.

Supplementary Material 1

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Author contributions

ASD, PES, JC and SAP co-designed the study. ASD, MY, KDD, JOA, PS, AKB, TAM, PA, NEO participated in proposal writing and data collection. MY performed the data analysis. ASD & SB participated in data analysis and review. SB supervised proposal writing, data collection, and manuscript drafting. ASD, MY, and SB drafted the initial manuscript. KDD, JOA, PS, AKB, TAM, PA, NEO, PES, TAB, JC and SAO reviewed the manuscript. All authors participated in manuscript writing and review and have approved the final manuscript for submission.

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Data availability
The datasets generated and/or analyzed during the current study are not publicly available in order to maintain participant privacy but are available from the corresponding author on reasonable request.

Declarations

Ethics approval and consent to participate
This study was approved by the Korle-Bu Teaching Hospital Institutional Review Board (IRB) (KBTH-STC 000140/2024). As this was a retrospective study utilizing anonymized data from medical records, the requirement for individual informed consent was waived by the IRB in accordance with KBTH guidelines. We confirm that the study was performed in accordance with the ethical standards as laid down in the 1964 Declaration of Helsinki and its later amendments or comparable ethical standards.

Consent for publication
Not applicable.

Competing interests
The authors declare no competing interests.

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References

1. Organization WH. Congenital disorders 2023 [Available from: https://www.who.int/news-room/fact-sheets/detail/birth-defects
2. Zaki U, Qazi SH, Shamim U, Fatima S, Das JK, Bhutta ZA. Optimal Strategies for Screening Common Birth Defects in Children of Low- and Middle-Income Countries: A Systematic Review. Neonatology. 2024:1–21.
3. Moges N, Anley DT, Zemene MA, Adella GA, Solomon Y, Bantie B, et al. Congenital anomalies and risk factors in Africa: a systematic review and meta-analysis. BMJ Paediatrics Open. 2023;7(1):e002022.
4. Yetwale A, Kabeto T, Biyazin T, Fenta B. Prenatal ultrasound utilization and its associated factors among pregnant women in Jimma town public health institutions, Ethiopia. Health Serv Res Managerial Epidemiol. 2022;9:233339282210858.
5. Aliyu LD, Kurjak A, Wataganara T, De Sá RAM, Pooh R, Sen C et al. Ultrasound in Africa: what can really be done? J Perinat Med. 2016;44(2).
6. Luntsi G, Ugwu AC, Nkubli FB, Emmanuel R, Ochie K, Nwobi CI. Achieving universal access to obstetric ultrasound in resource constrained settings: A narrative review. Radiography (Lond). 2021;27(2):709–15.
7. Goley SM, Sakula-Barry S, Adofo-Ansong N, Isaaya Ntawunga L, Tekyiwa Botchway M, Kelly AH, et al. Investigating the use of ultrasonography for the antenatal diagnosis of structural congenital anomalies in low-income and middle-income countries: a systematic review. BMJ Paediatr Open. 2020;4(1):e000684.
8. Kamla I, Kamgaing N, Billong S, Tochie JN, Tolefac P, de Paul Djientcheu V. Antenatal and postnatal diagnoses of visible congenital malformations in a sub-Saharan African setting: a prospective multicenter cohort study. BMC Pediatr. 2019;19(1):457.
9. Salomon LJ, Alfirevic Z, Berghella V, Bilardo CM, Chalouhi GE, Da Silva Costa F, et al. ISUOG practice guidelines (updated): performance of the routine mid-trimester fetal ultrasound scan. Ultrasound Obstet Gynecol. 2022;59(6):840–56.
10. Parker SE, Mai CT, Canfield MA, Rickard R, Wang Y, Meyer RE, et al. Updated National birth prevalence estimates for selected birth defects in the united States, 2004–2006. Birth Defects Res Clin Mol Teratol. 2010;88(12):1008–16.
11. Dolk H, Loane M, Garne E. The prevalence of congenital anomalies in Europe. Adv Exp Med Biol. 2010;686:349–64.
12. ElAwady H, AlGameel A, Ragab T, Hassan N. Congenital anomalies in neonates in Fayoum Governorate, Egypt. East Mediterr Health J. 2021;27(8):790–7.
13. Birhanu K, Tesfaye W, Berhane M. Congenital anomalies in neonates admitted to a tertiary hospital in Southwest Ethiopia: A cross sectional study. Ethiop J Health Sci. 2021;31(6):1155–62.
14. Wagathu ROA. Describing congenital anomalies among newborns in Kenya: A hospital based study. Int J Health Sci Res. 2019;9(4).
15. Adeboye M, Abdulkadir M, Adegboye O, Saka A, Oladele P, Oladele D et al. A Prospective Study of Spectrum, Risk Factors and Immediate Outcome of Congenital Anomalies in Bida, North Central Nigeria. Annals of Medical and Health Sciences Research. 2016;6(6).
16. Rankin J, Pattenden S, Abramsky L, Boyd P, Jordan H, Stone D, et al. Prevalence of congenital anomalies in five British regions, 1991-99. Arch Dis Child Fetal Neonatal Ed. 2005;90(5):F374–9.
17. Bhide P, Kar A. A National estimate of the birth prevalence of congenital anomalies in India: systematic review and meta-analysis. BMC Pediatr. 2018;18(1):175.
18. Vatankhah S, Jalilvand M, Sarkhosh S, Azarmi M, Mohseni M. Prevalence of congenital anomalies in Iran: A review Article. Iran J Public Health. 2017;46(6):733–43.
19. Adane F, Afework M, Seyoum G, Gebrie A. Prevalence and associated factors of birth defects among newborns in sub-Saharan African countries: a systematic review and meta-analysis. Pan Afr Med J. 2020;36:19.
20. Mogess WN, Mihretie TB. Prevalence and associated factors of congenital anomalies in Ethiopia: A systematic review and meta-analysis. PLoS ONE. 2024;19(4):e0302393.
21. Onyambu CK, Tharamba NM, Onyambu CK, Tharamba NM. Screening for congenital fetal anomalies in low risk pregnancy: the Kenyatta National hospital experience. BMC Pregnancy Childbirth. 2018;18:1. 2018-05-23;18(1).
22. Dewan MC, Rattani A, Mekary R, Glancz LJ, Yunusa I, Baticulon RE, et al. Global hydrocephalus epidemiology and incidence: systematic review and meta-analysis. J Neurosurg. 2019;130(4):1065–79.
23. Akinmoladun JA, Ogbole GI, Lawal TA, Adesina OA. Routine prenatal ultrasound anomaly screening program in a Nigerian university hospital: redefining obstetrics practice in a developing African country. Niger Med J. 2015;56(4):263–7.
24. Eltyeb EE, Halawi MHA, Tashari TBM, Alharbi K, Alsayari OS, Albarrak DA, et al. Prevalence and pattern of birth defects in Saudi Arabia: A systematic review of observational studies. Pediatr Rep. 2023;15(3):431–41.
25. Boyd PA, Tonks AM, Rankin J, Rounding C, Wellesley D, Draper ES, et al. Monitoring the prenatal detection of structural fetal congenital anomalies in England and Wales: register-based study. J Med Screen. 2011;18(1):2–7.
26. Buijtendijk MF, Bet BB, Leeflang MM, Shah H, Reuvekamp T, Goring T, et al. Diagnostic accuracy of ultrasound screening for fetal structural abnormalities during the first and second trimester of pregnancy in low-risk and unselected populations. Cochrane Database Syst Rev. 2024;5(5):CD014715.
27. Amoah B, Anto EA, Osei PK, Pieterson K, Crimi A. Boosting antenatal care attendance and number of hospital deliveries among pregnant women in rural communities: a community initiative in Ghana based on mobile phones applications and portable ultrasound scans. BMC Pregnancy Childbirth. 2016;16(1):141.
28. Sendra-Balcells C, Campello VM, Torrents-Barrena J, Ahmed YA, Elattar M, Ohene-Botwe B, et al. Generalisability of fetal ultrasound deep learning models to low-resource imaging settings in five African countries. Sci Rep. 2023;13(1):2728.
29. Bardi F, Bergman JEH, Bouman K, Erwich JJ, Duin LK, Walle HEK, et al. Effect of prenatal screening on trends in perinatal mortality associated with congenital anomalies before and after the introduction of prenatal screening:

A population-based study in the Northern Netherlands. Paediatr Perinat Epidemiol. 2021;35(6):654–63.
30. Karim JN, Roberts NW, Salomon LJ, Papageorghiou AT. Systematic review of First-Trimester ultrasound screening for detection of fetal structural anomalies and factors that affect screening performance. Obstet Gynecol Surv. 2018;73(4):185–7.
31. Rayburn WF, Klagholz JC, Elwell EC, Strunk AL. Maternal-fetal medicine workforce in the united States. Am J Perinatol. 2012;29(9):741–6.
32. Sun H, Wu A, Lu M, Cao S. Liability, risks, and recommendations for ultrasound use in the diagnosis of obstetrics diseases. Heliyon. 2023;9(11):e21829.
33. Gorleku PN, Setorglo J, Ofori I, Edzie EKM, Dzefi-Tettey K, Piersson AD et al. Towards the scale and menace of unregulated sonography practice in Ghana. J Global Health Rep. 2020;4.
34. Gadeka DD, Esena RK. Barriers to quality care in medical imaging at a teaching hospital in Ghana: staff perspective. J Med Imaging Radiat Sci. 2020;51(3):425–35.

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