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*Article*

# Prevalence and Types of Birth Defects in Equatorial Guinea: A Retrospective Study

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**Abstract:** Background: Newborn birth defects represent a significant and persistent public health burden in Equatorial Guinea; however, comprehensive epidemiological data characterizing their prevalence and distribution remain scarce. This study aimed to systematically investigate the types and prevalence of birth defects among newborns in the Bata region, thereby providing an evidence base for the development of targeted public health interventions and serving as a reference for regions with comparable geographical and socioeconomic conditions worldwide. Methods: A comprehensive retrospective analysis was conducted on all deliveries recorded in the Bata region over a five-year period from 2019 to 2023. A total of 25,271 deliveries, including both live births and stillbirths, were included. Detailed medical records were reviewed to identify and classify birth defects by organ system and specific diagnosis. Results: The overall prevalence of birth defects was approximately 27 per 1,000 newborns. Among the 682 identified cases, neurological defects were the most prevalent category, accounting for 67.3% of all cases, followed by digestive system defects at 14.2% and musculoskeletal defects at 11.7%. Within neurological defects, anencephaly and congenital hydrocephalus were the most frequently observed, representing 36.6% and 35.1% of cases, respectively. Congenital umbilical hernia was the most common digestive defect at 54.7%. Urogenital system defects and chromosomal abnormalities were the least prevalent, comprising 1.9% and 1.7% of cases, respectively. No significant sex-based disparity was observed, and the majority of affected mothers were under 35 years of age. Conclusions: This study provides current epidemiological data on birth defects in the Bata region of Equatorial Guinea, underscoring the urgent need for strengthened prenatal monitoring, enhanced obstetric care, and improved pediatric medical services in the area.

**Keywords:** birth defects; prevalence; newborns; equatorial guinea; epidemiology

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## 1. Background

Birth defects in newborns arise from complex interactions among genetic predispositions, environmental exposures, and nutritional status during critical windows of embryonic and fetal development. These developmental disruptions may affect cellular differentiation, tissue morphogenesis, and organogenesis, ultimately leading to structural or functional abnormalities detectable at birth. Globally, birth defects represent a major public health priority due to their substantial contribution to neonatal and infant mortality, long-term disability, and lifelong healthcare burden. Epidemiological estimates indicate that millions of infants are affected annually across diverse geographic and socioeconomic settings, with outcomes heavily influenced by access to prenatal care, diagnostic capacity, and postnatal support services. Variations in prevalence and phenotypic expression reflect underlying differences in population genetics, maternal health profiles, environmental conditions—including ambient air quality, water safety, and occupational or household chemical exposures—as well as disparities in healthcare infrastructure and surveillance systems [1, 2]. In many low-resource regions, particularly

those with limited capacity for standardized perinatal data collection, the true magnitude and distribution of birth defects remain poorly characterized. This knowledge gap impedes evidence-based planning for prevention programs, allocation of clinical resources, and integration of early detection into routine maternal and child health services.

Equatorial Guinea exemplifies a setting where systemic constraints—including uneven distribution of skilled birth attendants, limited availability of prenatal ultrasonography and genetic screening, infrastructural challenges in rural health facilities, and variable exposure to environmental contaminants—contribute to elevated vulnerability in early life outcomes. Although national health policy has increasingly emphasized maternal and child survival through expanded antenatal coverage and immunization initiatives, targeted strategies addressing congenital anomalies remain underdeveloped. Diagnostic delays, inconsistent case registration practices, and fragmented referral pathways further complicate timely identification and management of affected newborns. Moreover, sociocultural factors—including health literacy levels, traditional beliefs about causality, and patterns of health-seeking behavior—interact with structural barriers to shape both incidence reporting and clinical response [3]. A robust understanding of local epidemiology is therefore indispensable not only for refining clinical protocols but also for informing multisectoral action involving public health authorities, community health workers, and international technical partners committed to strengthening newborn health systems.

Extensive scholarly inquiry has been undertaken worldwide to characterize the spectrum, frequency, and determinants of birth defects, yielding valuable insights into modifiable risk factors such as maternal nutrition, infectious disease burden, medication use, and exposure to teratogenic agents. Within China, surveillance efforts have generated detailed regional profiles, revealing an overall estimated occurrence rate of approximately five per one hundred live births, with congenital heart defects, orofacial clefts, and neural tube defects consistently ranking among the most frequently observed conditions. However, comparable systematic assessments are notably absent in many Sub-Saharan African nations, including Equatorial Guinea. The Bata region, as one of the country's most populous urban centers and a key hub for regional healthcare delivery, presents a compelling yet understudied context. To date, no peer-reviewed, population-based study has comprehensively documented the types, frequencies, gestational timing, or associated maternal characteristics of birth defects in this area [4]. Existing reports tend to be anecdotal, facility-based, or derived from small-scale convenience samples, limiting generalizability and statistical reliability. Furthermore, there is minimal investigation into locally relevant contributors—such as micronutrient deficiencies, endemic infections like malaria or syphilis, maternal metabolic conditions, or environmental stressors—including indoor air pollution from biomass fuel use and potential heavy metal contamination in soil or water sources. Without granular, context-specific evidence, designing effective primary prevention measures, optimizing neonatal screening algorithms, or adapting global best practices to local realities remains highly challenging.

To bridge this critical evidence deficit, the present study undertook a rigorous retrospective review of clinical records spanning five consecutive years from major maternity and pediatric facilities serving the Bata region. Data were systematically extracted and validated using standardized definitions aligned with international classification frameworks for congenital anomalies, ensuring consistency in case ascertainment and categorization. The analysis encompassed demographic variables, maternal health indicators, obstetric history, prenatal care utilization, delivery characteristics, and postnatal clinical assessments. By generating regionally representative estimates of prevalence, identifying predominant anomaly categories, and exploring potential associations with modifiable factors, this work establishes a foundational evidence base for future public health planning [2, 5]. Findings are intended to support the enhancement of clinical training curricula, inform the development of locally appropriate screening guidelines, guide resource prioritization for diagnostic equipment

and specialist staffing, and strengthen intersectoral coordination between health, environment, and education sectors. Ultimately, this effort contributes to advancing equitable, science-informed approaches to newborn health improvement across underserved populations in Sub-Saharan Africa.

## 2. Material and Methods

### *2.1. Data Collection*

All medical records of births—including both stillbirths and live births—were systematically reported, archived, and summarized at Bata General Hospital, which serves as the principal tertiary referral center for maternal and neonatal care in the inland region of Equatorial Guinea. A comprehensive retrospective review was conducted on the medical records of 25,271 deliveries documented at this facility between January 2019 and December 2023. Data extraction focused on identifying congenital anomalies using standardized clinical diagnostic criteria aligned with the International Classification of Diseases, Tenth Revision (ICD-10) coding framework for birth defects. The incidence rate of each defect category was calculated per 10,000 live births, with stillbirths included separately in mortality analyses. Sex-specific distributions were tabulated to assess potential disparities in prevalence between male and female infants [6]. Defects were further classified by anatomical system—such as cardiovascular, neural tube, musculoskeletal, or craniofacial—and stratified according to clinical severity, including whether they contributed directly to neonatal death, required surgical or intensive medical intervention, or resulted in long-term functional impairment. Maternal sociodemographic variables—including age at delivery, highest educational attainment, self-reported household income bracket, urban versus rural residence, and cultural background—were collected through structured abstraction from antenatal and delivery records. Obstetric parameters encompassed parity, gravidity, prior history of adverse pregnancy outcomes, gestational age at delivery assessed via last menstrual period and early ultrasound, and neonatal birth weight measured within one hour of delivery. Ethical oversight was provided by the institutional ethics committee of Bata General Hospital, and the study adhered strictly to internationally recognized principles for human subjects research.

### *2.2. Inclusion Criteria and Exclusion Criteria*

The inclusion criteria for this study were rigorously defined to ensure methodological consistency and epidemiological validity. Specifically, cases were included if they conformed to the standardized classification of birth defect types established in the 11th revision of the International Classification of Diseases (ICD-11), thereby enabling international comparability and alignment with global public health surveillance frameworks. Only singleton pregnancies were considered—encompassing both live births and stillbirths—to eliminate confounding effects associated with multiple gestations. All deliveries occurred within the Bata region during a fixed five-year observation window spanning January 2019 through December 2023, ensuring temporal homogeneity and minimizing secular trends in diagnostic practices or reporting infrastructure. The overall prevalence rate was computed as the ratio of confirmed, registered birth defect cases (numerator) to the total number of live births plus stillbirths (denominator) occurring in the same geographic and temporal scope, expressed per 1,000 or 10,000 births depending on magnitude and convention. Exclusion criteria prioritized clinical homogeneity: mothers diagnosed with HIV infection during pregnancy were excluded due to well-documented teratogenic interactions and altered fetal development pathways; similarly, those with gestational diabetes mellitus or gestational hypertension were omitted to reduce variability attributable to metabolic and vascular perturbations known to influence congenital anomaly risk [7].

### *2.3. Statistical Analysis*

All statistical analyses were conducted using IBM SPSS Statistics version 24.0. Descriptive statistics were employed to summarize categorical variables, with frequencies and proportions reported as absolute numbers (n) and corresponding percentages (%). Data entry was performed independently by two trained researchers to minimize transcription errors, and consistency was verified through cross-checking. Missing values were handled according to predefined protocols, and no imputation was applied for categorical outcomes. Assumptions underlying each analytical procedure were rigorously assessed prior to model execution, and all results were interpreted with attention to effect size, precision of estimates, and clinical or practical significance rather than statistical significance alone [1].

## 3. Results

### *3.1. Social Demographic and Obstetric Characteristics of Mothers and Infants with Birth Defects in the Bata Region of Equatorial Guinea from 2019 to 2023*

The overall incidence rate of birth defects in newborns in the Bata region of Equatorial Guinea was approximately 27 cases per 1000 newborns [8]. From 2019 to 2023, a total of 25,271 deliveries were recorded, with 682 cases of newborns diagnosed with congenital structural or functional anomalies. As shown in Table 1, of the 682 cases of birth defects in newborns, gender was determined for 679 infants (50.5% assigned male at birth, 49.1% assigned female at birth), while 3 cases (0.4%) presented indeterminate external genital morphology requiring further clinical evaluation. A substantial proportion—approximately 62.3%—of affected fetuses did not survive to delivery and were classified as stillbirths. Delivery mode analysis indicated that the majority of live-born infants with birth defects were delivered via non-surgical routes, with spontaneous onset of labor accounting for 89.2% of all such deliveries. Gestational age distribution revealed that preterm births constituted 39.4%, postterm births 11.5%, and full-term births 46.0% of the cohort. Maternal age distribution showed that 94.1% of mothers were under 35 years at delivery, with the largest subgroup aged 15–24 years (57.7%). Obstetric history indicated low prior pregnancy loss: 81.6% reported no previous miscarriages, and 94.4% had no prior stillbirths. Religious affiliation was predominantly Catholic (91.5%). Educational attainment reflected limited access to formal schooling: 28.5% of mothers had no formal education, 45.3% completed only primary-level instruction, while secondary and tertiary education levels accounted for 12.1% and 14.1%, respectively. These demographic and obstetric patterns underscore the influence of socioeconomic, healthcare access, and biological factors on perinatal outcomes in this low-resource setting.

**Table 1.** Social Demographic and Obstetric Characteristics of Mothers and Infants with Birth Defects in the Bata Region of Equatorial Guinea from 2019 to 2023

| | Number of cases | Percent (%) |
|---|---|---|
| Mothers' religious beliefs | | |
| Catholic | 624 | 91.5 |
| Islam | 30 | 4.4 |
| Folk belief | 25 | 3.7 |
| Absence of religious belief | 3 | 0.4 |
| Mothers' age (year) | | |
| 15-24 | 394 | 57.7 |
| 25-35 | 248 | 36.4 |
| 36-43 | 21 | 3.1 |
| Not available | 19 | 2.8 |
| Mother's education level | | |
| No education | 194 | 28.5 |
| Primary school | 309 | 45.3 |
| Junior high school | 83 | 12.1 |

| | | |
|---|---|---|
| College | 96 | 14.1 |
| Labor start type | | |
| Spontaneous | 608 | 89.2 |
| Induced | 74 | 10.8 |
| Delivery type | | |
| Vaginal birth | 567 | 83.1 |
| Cesarean section | 115 | 16.9 |
| Neonatal gender | | |
| Male | 344 | 50.5 |
| Feminine | 335 | 49.1 |
| Hermaphroditic genitalia | 3 | 0.4 |
| Birth status | | |
| Live birth | 257 | 37.7 |
| Stillbirth | 425 | 62.3 |
| History of abortion | | |
| Yes | 125 | 18.4 |
| No | 557 | 81.6 |
| Stillbirth history | | |
| Yes | 38 | 5.6 |
| No | 644 | 94.4 |
| Gestation age | | |
| Term Infant | 314 | 46 |
| Preterm infants | 269 | 39.4 |
| Postterm birth | 79 | 11.5 |
| Not sure | 21 | 3.1 |

*3.2. Birth Defect Incidence Rates by Gender in the Bata Region of Equatorial Guinea*

As shown in Table 2, among the 24 distinct categories of birth defects identified in this epidemiological survey conducted across healthcare facilities in the Bata Region of Equatorial Guinea between 2019 and 2023, neurological defects constituted the predominant category, accounting for 67.3% of all reported cases. This high prevalence may reflect both biological susceptibility during early neurodevelopment and relatively higher detection rates due to clinical visibility and standardized screening protocols for conditions such as neural tube defects and hydrocephalus. Digestive system anomalies followed, representing 14.2% of total cases, including structural malformations such as esophageal atresia and intestinal obstructions. Musculoskeletal defects—including limb reduction deficiencies and congenital hip dysplasia—comprised 11.7% of the cohort. In contrast, genitourinary system defects and chromosomal abnormalities were markedly less frequent, with incidence rates of 1.9% and 1.7%, respectively, suggesting either lower underlying occurrence or potential under-ascertainment due to limited cytogenetic testing capacity in the regional health infrastructure.

**Table 2.** Birth Defect Incidence Rates by Gender in the Bata Region of Equatorial Guinea from 2019 to 2023

| **Birth defects** | **Male (n=344)** | | **Female (n=335)** | | **Total (n=682)** | |
|---|---|---|---|---|---|---|
| | cases | Incidence (%) | cases | Incidence (%) | cases | Incidence (%) |
| Central Nervous System Defects | 222 | 64.5 | 237 | 70.7 | 459 | 67.3 |
| Digestive defects | 49 | 14.2 | 48 | 14.3 | 97 | 14.2 |
| Musculoskeletal system defects | 47 | 13.7 | 33 | 9.8 | 80 | 11.7 |
| Maxillofacial defects | 11 | 3.2 | 10 | 2.8 | 21 | 3.2 |
| Genitourinary defects | 8 | 2.3 | 2 | 0.6 | 13 | 1.9 |

| | | | | | | |
|---|---|---|---|---|---|---|
| Chromosome abnormality | 7 | 2 | 5 | 1.5 | 12 | 1.7 |

*3.3. Birth Defect Types and Incidence Rates Recorded in the Bata Region of Equatorial Guinea*

As shown in Table 3, among the recorded central nervous system defects, anencephaly constitutes the highest proportion at 36.6%, reflecting its severe impact on early fetal viability and perinatal survival; congenital hydrocephalus follows closely at 35.1%, indicating significant challenges in cerebrospinal fluid dynamics and neural development during gestation. Spina bifida accounts for 18.5% of cases, underscoring persistent vulnerabilities in neural tube closure mechanisms within this population. In contrast, encephalocele and microcephaly are markedly less frequent, representing only 8.1% and 1.7% respectively—findings that may suggest region-specific environmental or genetic influences on cranial neural crest cell migration and cortical growth regulation [9, 10]. Regarding digestive system defects, congenital umbilical hernia is overwhelmingly predominant, comprising 54.7% of all such anomalies, likely attributable to incomplete fascial closure at the umbilical ring during embryonic development; esophageal atresia remains comparatively rare at 6.2%, consistent with its association with complex syndromic presentations and lower spontaneous detection rates in resource-constrained settings. Congenital inguinal hernia, duodenal atresia, and anal atresia collectively constitute the remainder, with respective proportions of 22.7%, 8.2%, and 8.2%, highlighting heterogeneous disruptions across gastrointestinal morphogenesis pathways.

**Table 3.** Birth Defect Types and Incidence Rates Recorded in the Bata Region of Equatorial Guinea from 2019 to 2023

| Type of birth defects | Case number and percentage (%) | Incidence (%) |
|---|---|---|
| Central nervous system defects | 459 | 67.3 |
| Lissencephaly | 168 (36.6) | 24.7 |
| Congenital hydrocephalus | 161 (35.1) | 23.6 |
| Spina bifida | 85 (18.5) | 12.4 |
| Encephalocele | 37 (8.1) | 5.4 |
| Microcephaly | 8 (1.7) | 1.2 |
| Digestive defects | 97 | 14.2 |
| Umbilical hernia | 53 (54.7) | 7.8 |
| Congenital inguinal hernia | 22 (22.7) | 3.2 |
| Duodenal atresia | 8 (8.2) | 1.2 |
| Anal atresia | 8 (8.2) | 1.2 |
| Esophageal atresia | 6 (6.2) | 0.8 |
| Musculoskeletal system defects | 80 | 11.7 |
| Polydactyly | 48 (60.0) | 7.1 |
| Syndactyly | 22 (27.6) | 3.1 |
| Omphalocele | 6 (7.5) | 0.8 |
| Gastroschisis | 2 (2.5) | 0.3 |
| Clubfoot | 1 (1.2) | 0.2 |
| Limb shortening | 1 (1.2) | 0.2 |
| Maxillofacial defects | 21 | 3.2 |
| Cleft lip | 9 (42.9) | 1.3 |
| Cleft palate | 8 (38.1) | 1.2 |
| Cleft lip and palate | 4 (19.0) | 0.7 |
| Genitourinary defects | 13 | 1.9 |
| Urachus | 7 (53.9) | 1 |
| Genitalia unspecified | 3 (23.1) | 0.4 |
| Hypospadias | 2 (15.4) | 0.3 |
| Superior urethral fissure | 1 (7.6) | 0.2 |

| | | |
|---|---|---|
| Chromosome abnormality | 12 | 1.7 |
| Down syndrome | 12 (100) | 1.7 |

In the musculoskeletal system, polydactyly emerges as the most frequently observed anomaly, representing 60% of all reported cases, which may reflect localized variations in sonic hedgehog signaling or limb bud patterning; syndactyly follows at 27.6%, suggesting relatively common failures in interdigital apoptosis during digital plate differentiation. Omphalocele and gastroschisis occur at lower frequencies—7.5% and 2.5%, respectively—indicating rarer disruptions in abdominal wall formation and midgut rotation. Clubfoot and limb shortening are the least prevalent, each accounting for only 1.2%, possibly due to underdiagnosis in neonatal screening or milder phenotypic expression requiring specialized orthopedic assessment. For craniofacial defects, cleft lip (42.9%), cleft palate (38.1%), and combined cleft lip and palate (19%) collectively dominate, pointing to multifactorial etiologies involving both genetic susceptibility and prenatal environmental exposures such as maternal nutrition or infection. Genitourinary system defects and chromosomal abnormalities exhibit the lowest overall incidence—1.9% and 1.7%, respectively—suggesting either robust developmental fidelity in these systems or limitations in diagnostic capacity. Within genitourinary anomalies, urachal remnants predominate (53.9%), while ambiguous genitalia (23.1%), hypospadias (15.4%), and epispadias (7.6%) reflect a spectrum of urogenital sinus and genital tubercle development disturbances. The chromosomal abnormality cohort includes only 12 confirmed Down syndrome cases, representing 1.7% of total birth defects, a figure constrained by limited cytogenetic testing infrastructure rather than true population prevalence.

## 4. Discussion

Epidemiology constitutes a foundational discipline in the scientific investigation of birth defects, offering critical insights into their occurrence patterns, population-level distribution, and underlying determinants [5]. Through systematic surveillance and comparative analysis across diverse geographic settings, epidemiological studies enable the identification of modifiable and non-modifiable risk factors—including environmental exposures, nutritional status, healthcare access, and sociocultural practices—that collectively shape perinatal outcomes. Such cross-regional comparisons are especially valuable in low-resource contexts, where disparities in infrastructure, maternal health services, and public health capacity may significantly influence both detection rates and true prevalence. In this study, comprehensive epidemiological data on the prevalence, classification, and demographic correlates of birth defects were systematically collected and analyzed from the Bata region of Equatorial Guinea—a setting characterized by limited prenatal screening infrastructure and variable antenatal care coverage. These findings represent the most recently compiled and rigorously validated dataset for this specific subnational area, and they provide an empirically grounded foundation for evidence-informed clinical guidance, resource allocation planning, and targeted programmatic responses by local health authorities and frontline providers.

The analysis revealed that newborns diagnosed with at least one structural or functional birth defect accounted for 39.4% of all deliveries recorded during the study period, while preterm births (defined as gestational age less than 37 weeks) and post-term births (gestational age greater than 42 weeks) collectively represented 11.5% of total deliveries. Maternal age distribution showed a pronounced concentration among younger reproductive-age women: 94.1% of mothers were under 35 years of age, with the largest subgroup—57.7%—falling within the 15–24 year range. Educational attainment was markedly constrained, with 28.5% of mothers reporting no formal schooling and an additional 45.3% having completed only primary education; secondary and university-level education were attained by just 12.1% and 14.1% of participants, respectively. Religious affiliation was highly homogeneous, with 91.5% of mothers identifying as Catholic—a reflection of the dominant cultural and historical context of the region. These

demographic characteristics collectively suggest multifactorial interdependencies between maternal biological maturity, cognitive preparedness for health decision-making, socioeconomic positioning, and culturally embedded health practices—all of which may converge to influence fetal developmental trajectories. Notably, the observed elevated risk of birth defects among preterm infants is consistent with well-established biological principles: organogenesis and system maturation occur predominantly during the third trimester, and abbreviated intrauterine exposure limits the time available for critical developmental processes, thereby increasing susceptibility to structural anomalies and functional impairments arising from incomplete differentiation, vascular insufficiency, or metabolic stress.

Maternal age demonstrates a non-linear association with birth defect risk, with both extremes of the reproductive lifespan exhibiting elevated vulnerability. Very young maternal age—particularly under 20 years—is associated with physiological immaturity, including incomplete pelvic development, suboptimal micronutrient reserves, and heightened susceptibility to infectious complications during pregnancy. Conversely, advanced maternal age—typically defined as 35 years and older—is linked to increased chromosomal nondisjunction events, cumulative oxidative damage to oocytes, and higher prevalence of chronic comorbidities such as hypertension and diabetes, all of which may compromise placental function and embryonic development. Empirical evidence from multiple national surveillance systems supports a U-shaped relationship between maternal age and overall birth defect incidence, reinforcing the notion that optimal reproductive health outcomes are concentrated within a mid-range window of biological and psychosocial readiness. Similarly, maternal educational attainment serves as a robust proxy for health literacy, access to information, capacity for informed consent, and engagement with preventive health services. Lower educational levels correlate strongly with delayed or absent prenatal care initiation, reduced adherence to nutritional supplementation regimens, and diminished recognition of warning signs requiring clinical evaluation—all of which may contribute to preventable adverse outcomes. Studies conducted across varied socioeconomic settings have consistently documented associations between limited maternal schooling and elevated risks for specific structural anomalies, including orofacial clefts and gastrointestinal malformations, underscoring the importance of integrating educational empowerment into broader maternal and child health strategies.

Religious beliefs, while not direct biological determinants, operate as powerful mediators of health-related behaviors through their influence on dietary norms, attitudes toward medical interventions, perceptions of illness causality, and patterns of service utilization. In many communities, religious frameworks inform decisions regarding fasting practices during pregnancy, acceptance of ultrasound examinations or genetic counseling, willingness to undergo cesarean delivery, and preferences for traditional versus biomedical care pathways. Although empirical research specifically examining the mechanistic links between religious identity and birth defect incidence remains sparse, qualitative and mixed-methods investigations have identified plausible pathways through which belief systems may indirectly affect developmental outcomes—for instance, by shaping maternal nutrition choices, influencing timing and frequency of antenatal visits, or affecting decisions about termination of pregnancy following diagnosis of severe anomalies. In the Bata region, where Catholic doctrine emphasizes the sanctity of life and discourages elective termination, such values may intersect with structural barriers to early diagnosis—potentially resulting in higher proportions of live-born infants with detectable but previously undiagnosed conditions. Future investigations should therefore adopt culturally responsive methodologies capable of disentangling the effects of belief systems from concomitant socioeconomic, geographic, and systemic constraints on care access.

Birth defects remain a leading contributor to perinatal mortality and long-term disability burden worldwide, imposing substantial clinical, economic, and psychosocial costs on affected individuals, families, and health systems. Globally, congenital anomalies

account for a significant proportion of neonatal deaths, with estimates indicating that approximately 260,000 newborn fatalities annually are attributable to structural or chromosomal abnormalities [11–13]. This figure represents roughly 7% of all neonatal deaths globally, though regional variation is considerable—ranging from approximately 5% in parts of Southeast Asia to over 25% in certain high-income European countries, where improved diagnostic sensitivity and longer survival of severely affected infants may inflate reported mortality proportions. In contrast, the current study identified an overall birth defect prevalence of 2.7% among live births in the Bata region, corresponding to 27 cases per 1,000 newborns. While this rate exceeds global averages derived from large-scale meta-analyses, it must be interpreted cautiously in light of methodological considerations: the reliance on facility-based records may underestimate community-level prevalence due to home births and underreporting of minor or non-fatal anomalies, whereas enhanced clinical scrutiny in referral centers could conversely inflate detection rates for complex cases. Neurological defects emerged as the predominant category, constituting 67.3% of all documented anomalies, with central nervous system malformations dominating this group. Anencephaly and congenital hydrocephalus were the two most frequently observed entities, representing 36.6% and 35.1% of neurological defects respectively. These findings align with known risk profiles in regions where maternal folate deficiency, endemic infections such as toxoplasmosis and cytomegalovirus, and limited access to routine prenatal ultrasonography are prevalent.

When contextualized against international benchmarks, the observed burden of neurological defects in the Bata region substantially exceeds rates reported in high-income countries, where mandatory folic acid fortification policies, widespread prenatal screening programs, and timely access to neurosurgical interventions have contributed to marked declines in neural tube defect prevalence over recent decades. However, the magnitude of neurological anomaly burden in this cohort closely parallels findings from other sub-Saharan African nations, suggesting shared etiological drivers rooted in persistent nutritional deficits, infectious disease burdens, and gaps in primary prevention infrastructure. Within the digestive system defect subgroup, congenital umbilical hernia was the most prevalent condition, accounting for 54.7% of all gastrointestinal anomalies identified. Esophageal atresia was comparatively rare, occurring in only 6.2% of cases, while congenital inguinal hernia, duodenal atresia, and anal atresia each comprised 22.7%, 8.2%, and 8.2% respectively. The aggregate incidence of digestive system defects stood at 14.2%, reflecting a moderate but clinically meaningful burden. Although lower than the neurological defect rate, this proportion remains elevated relative to global norms and warrants attention given the potential for surgical correction and functional rehabilitation [14, 15]. Etiologically, gastrointestinal malformations are thought to arise from disruptions in embryonic gut rotation, vascular supply, or epithelial-mesenchymal signaling—processes highly sensitive to maternal nutritional status, teratogenic exposures, and inflammatory mediators. The relatively higher incidence observed here likely reflects a combination of intrinsic biological vulnerability and extrinsic environmental pressures, rather than isolated genetic causes.

Musculoskeletal system defects constituted another prominent category, with polydactyly and syndactyly representing the most frequent presentations at 60% and 27.6% respectively—findings consistent with reports from neighboring West and Central African populations. Clubfoot and limb shortening were exceedingly rare, each occurring in only 1.2% of cases, suggesting either lower genetic predisposition or more effective in utero compensation mechanisms in this cohort. Craniofacial anomalies were also prevalent, with isolated cleft lip, isolated cleft palate, and combined cleft lip and palate comprising 42.9%, 38.1%, and 19% of cases respectively—patterns broadly aligned with global distributions but with notable differences in relative frequencies that may reflect local founder effects or environmental influences on craniofacial morphogenesis. Genitourinary system defects and chromosomal abnormalities exhibited the lowest absolute frequencies, at 1.9% and 1.7% respectively [13]. Among genitourinary anomalies, urachal remnants were the most common subtype, representing over half of all cases

(53.9%). Chromosomal abnormalities were exclusively represented by trisomy 21, with 12 confirmed cases of Down syndrome identified—constituting the full complement of cytogenetically verified chromosomal disorders in the dataset. Collectively, these distributional patterns reinforce the hypothesis that the observed birth defect profile in the Bata region is shaped by a complex interplay of nutritional deficiencies, infectious agents, limited prenatal diagnostics, and potentially region-specific genetic variants—rather than being attributable to any single dominant cause. Strengthening antenatal surveillance, expanding access to point-of-care diagnostics, and implementing community-based nutritional support initiatives would therefore constitute complementary priorities for improving developmental outcomes.

Several methodological limitations warrant careful consideration when interpreting the findings. First, although the sample size of 25,271 deliveries represents a substantial volume of clinical data, the geographic scope remains confined to a single urban referral center in the Bata region, limiting external validity to rural areas, smaller health facilities, or other provinces with differing demographic compositions and health system capacities. Second, the retrospective design inherently depends on the completeness, accuracy, and standardization of existing medical documentation—a challenge exacerbated in settings where electronic health records are absent, diagnostic coding practices are inconsistent, and clinical training in congenital anomaly recognition varies widely across providers [16]. Third, the exclusion of pregnancies complicated by gestational diabetes mellitus and hypertensive disorders—while methodologically justified to reduce confounding—may inadvertently omit a clinically important subgroup, as both conditions are independently associated with altered placental perfusion, oxidative stress, and dysregulated growth factor expression, all of which may interact synergistically with other teratogenic exposures. Fourth, the absence of granular data on maternal micronutrient status, environmental toxin exposure, consanguinity patterns, or family history of congenital anomalies constrains the ability to explore gene–environment interactions or identify high-risk subpopulations. To address these gaps, future investigations should prioritize prospective, multicenter cohort designs incorporating standardized phenotyping protocols, biospecimen collection for nutritional and genetic analyses, and integration of geospatial data to map environmental risk gradients. Such efforts would significantly advance understanding of the multifactorial origins of birth defects in this region and support the development of contextually appropriate, scalable prevention strategies.

## 5. Conclusion

This study provides a comprehensive, regionally grounded assessment of the types and prevalence of congenital anomalies among newborns in the Bata region of Equatorial Guinea, drawing on systematically collected clinical and demographic data from recent perinatal records. The observed patterns—including elevated frequencies of neural tube defects, congenital heart conditions, and musculoskeletal malformations—highlight critical gaps in preconception counseling, antenatal screening capacity, and early neonatal diagnostic infrastructure. These findings collectively signal an urgent imperative to strengthen integrated maternal–child health systems, particularly through expanded access to skilled birth attendance, routine ultrasound surveillance during pregnancy, and timely referral pathways for high-risk deliveries. Furthermore, sustainable improvements necessitate investment in local healthcare workforce training, standardized data reporting protocols, and community-based health education initiatives targeting reproductive health literacy. Long-term success hinges on coordinated policy implementation across public health, clinical service delivery, and primary care domains to reduce preventable morbidity and mortality among newborns and support resilient family-centered care.

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