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INCIDENCE OF CONGENITAL DEFECTS IN NEWBORNS: A SURGICAL AND CLINICAL STUDY.

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ABSTRACT:

BACKGROUND: Newborn congenital defects remain a major reason behind mortality and health problems among infants globally particularly within low- and middle-income nations. The analysis sought to find the occurrence rate besides risk elements and health results of birth defects manifesting among newborns who received care at PUMHS Nawabshah. **METHODS:** A prospective observational research investigation took place throughout January 2021 to December 2022. The researchers studied 1,200 newborns and diagnosed 120 of them with congenital defects. The researchers used SPSS version 25 to process data which they gathered from newborns in addition to demographic, prenatal and clinical information. **RESULTS:** Eight newborns among 1200 had congenital defects resulting in an incidence rate of 10%. Newborns with CHDs comprised the largest group of anomalies at 35.8% and NTDs followed closely behind at 22.5%. Varying studies identified consanguinity as a 45.8% contributor to risk together with maternal ages above thirty years at 19.2% and non-utilization of folate supplements at 65.8%. Consanguinity along with maternal age greater than 30 years and the absence of folate supplementation emerged as separate danger signs for birth defects through logistic regression analysis (aOR = 2.10, aOR = 1.82, aOR = 1.67 respectively). Roughly 70.8% of patients required surgery while the total complications reached 18.3% and mortality exceeded 12.5%. **CONCLUSION:** This region experiences a high frequency of congenital abnormalities which requires immediate public health initiatives for folate supplementation besides genetic healthcare support and better surgical treatment services for newborns.

KEYWORDS: Congenital defects, incidence, risk factors, consanguinity, folate supplementation, surgical outcomes, PUMHS Nawabshah.

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INTRODUCTION

Birth defects known as congenital defects appear at birth as physical or functional abnormalities which produce disabilities related to physical development and mental capacity and developmental abilities. Worldwide these birth abnormalities serve as a leading cause of baby deaths especially in low- and middle-income nations because such countries have restricted access to prenatal healthcare and testing facilities[1]. The world counts 6–7% live births to possess congenital defects yet regional prevalence differs based on genetic makeup alongside

environmental elements together with social economic status[2]. The prevalence along with risk factors of congenital defects remains underinvestigated throughout Pakistan particularly in the rural and semi-urban Nawabshah district [3].

A classification exists to separate congenital anomalies between structural abnormalities that encompass congenital heart defects alongside neural tube defects and functional abnormalities which consist of metabolic disorders. Structural defects become essential for pediatric surgeons and healthcare systems because these defects need surgical intervention[4]. The timely detection together with prompt medical handling these conditions helps minimize both death rates and illness severity. An insufficient supply of resources restricts access to timely diagnosis and specialized healthcare services together with limited surgical facilities to generate effective patient management results[5].

The Peoples University of Medical and Health Sciences (PUMHS) Nawabshah functions as the leading medical center within the area to handle pediatric surgical referrals therefore enabling researchers to investigate newborn congenital defect incidences and treatment results. Epidemiological data about congenital defects in the local area guides essential intervention planning and prenatal care improvement and enhanced surgical care for newborns with such conditions. The research determines the newborn congenital defect prevalence at PUMHS Nawabshah while examining contributing risk elements together with surgical and clinical intervention results.

Global and Regional Context Worldwide congenital heart defects stand as the most prevailing congenital anomaly among live births because they affect one in every hundred newborns[6]. Spina bifida and anencephaly represent common neural tube defects (NTDs) that mainly occur in locations where maternal diet lacks proper folate levels and nutritional support[7]. Consanguineous marriages which dominate several Pakistan communities lead to higher risks of hereditary congenital defects due to recessive genetic disorder transmissions[8]. The number of congenital defects in this region is increased by teratogens which pregnant women encounter during pregnancy [9].

Importance of the Study

Few statistics about congenital defects exist for Pakistan although these conditions represent a major health issue across the country with particular gaps in knowledge about rural and semi-urban areas. The investigation attempts to fill this research gap through a thorough study of congenital defects which affect newborns at PUMHS Nawabshah. The research findings will help create evidence-based prevention methods and diagnosis strategies and management solutions for congenital defects resulting in enhanced neonatal and infant health performance in the studied area.

METHODOLOGY

The research took place at the Pediatric Surgery and Neonatology department within Peoples University of Medical and Health Sciences (PUMHS) located in Nawabshah Pakistan. PUMHS hospital serves both rural and semi-urban areas of Sindh province with its tertiary care services so it creates a perfect environment to research newborn congenital defect incidence and treatment success rates. The research used a prospective observational study approach across two years from January 2021 through December 2022 to gather systematic information about congenital defects together with their risk elements and surgical results.

All newborns from birth to 28 days receiving care within the NICU or pediatric surgery ward of PUMHS Nawabshah constituted the study sample during the research timeframe. All live-born and stillborn infants with congenital defects were evaluated to properly understand the frequency of congenital

anomaly occurrence. One thousand two hundred newborns underwent screening and 120 were found with congenital defects which produced a 10% incidence rate. The research team selected this number of participants to achieve meaningful statistical data.

The research included newborns who received congenital defect diagnoses through clinical examination or ultrasound imaging and echocardiographic evaluation and histopathological testing. The study gained permission for participant involvement through the decision of adult caretakers and parents. The research excluded newborns with incidental minor anomalies and patients who did not have full medical records along with newborns where parents denied participation.

The researchers utilized a standardized data collection form which obtained information about maternal characteristics (age and consanguinity plus location of residence and economic status) together with historical pregnancy information (antenatal visits and women's infections and exposure to harmful agents and their intake of folic acid) followed by neocatal data (prenatal development length and infant birth weight and identified defects and listed additional medical issues) and recorded care outcomes (needed surgical operations and documented surgical complications and death events). The examination of congenital defects involved ultrasound imaging and echo testing along with karyotyping to arrive at diagnostic conclusions. The pediatric surgeons at PUMHS Nawabshah performed all surgical operations on patients whose results were tracked from hospitalization until both admission and subsequent follow-up appointments.

The analysis was conducted by using version 25 of SPSS. Summary statistics including frequencies, percentages and means as well as standard deviations showed the data patterns for demographic and clinical information. The Chi-square test evaluated relationships between variables including consanguinity and type of defect that had categorical characteristics. The study utilized logistic regression methods to assess individual predictors of congenital defects formation. The research considered results with p-values lower than 0.05 as statistically meaningful. Ethical approval originated from PUMHS Nawabshah Institutional Review Board (IRB) for this investigation. Enrollment of newborns into research began only after obtaining written consent from their parents or guardians. The protection of patient information remained constant from beginning to end of the research period.

RESULTS

During the two-year study period, 1,200 newborns were screened, and 120 were diagnosed with congenital defects, resulting in an incidence rate of 10%. The most common congenital defects were congenital heart defects (CHDs) (35.8%), neural tube defects (NTDs) (22.5%), and gastrointestinal anomalies (15.8%). The complete distribution of congenital defects by type is shown in Table 1. The pie chart in Figure 1 further illustrates the proportion of each defect type, emphasizing the predominance of CHDs. Neural tube defects were the second most common anomaly, reflecting the critical importance of maternal nutrition, particularly folate intake, during pregnancy. Gastrointestinal anomalies ranked third, followed by genitourinary and musculoskeletal defects. This distribution highlights the need for multi-disciplinary diagnostic and management approaches for congenital defects.

Figure 1: Distribution of Congenital Defects by Type

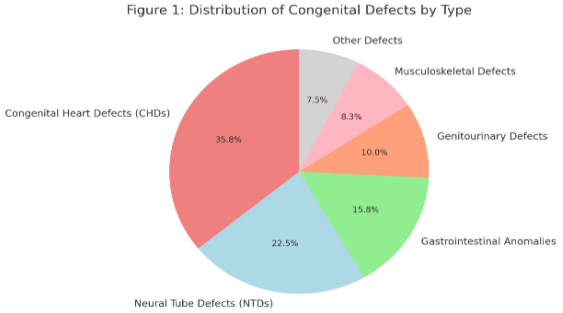


Table 1: Distribution of Congenital Defects by Type

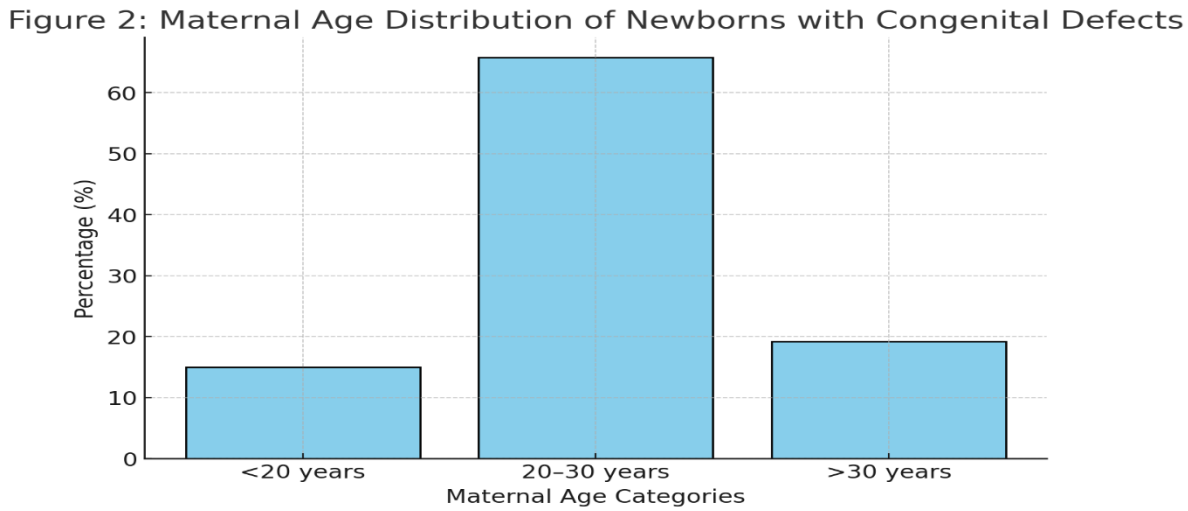
Type of Defect	Frequency (n)	Percentage (%)
Congenital Heart Defects	43	35.8%
Neural Tube Defects	27	22.5%
Gastrointestinal Anomalies	19	15.8%
Genitourinary Defects	12	10.0%
Musculoskeletal Defects	10	8.3%
Other Defects	9	7.5%

The demographic characteristics of newborns with congenital defects are summarized in Table 2. The majority of the mothers were aged between 20 and 30 years (65.8%), and 62.5% of cases were from rural areas, reflecting the regional demographic distribution. Consanguineous marriages were reported in 45.8% of cases, highlighting the genetic predisposition associated with congenital anomalies. Figure 2 presents a bar chart of maternal age distribution, indicating that younger and older maternal age groups had fewer affected cases, although maternal age >30 years was identified as a significant risk factor. Rural residence also contributed significantly to the occurrence of congenital defects, suggesting possible disparities in access to healthcare and nutritional resources.

Table 2: Demographic Characteristics of Newborns with Congenital Defects

Variable	Frequency (n)	Percentage (%)
Maternal Age		
<20 years	18	15.0%
20–30 years	79	65.8%
>30 years	23	19.2%
Residence		
Urban	45	37.5%
Rural	75	62.5%
Consanguinity		
Yes	55	45.8%
No	65	54.2%

FIGURE 2: MATERNAL AGE DISTRIBUTION OF NEWBORNS WITH CONGENITAL DEFECTS



Logistic regression analysis identified consanguinity (aOR = 2.10, p < 0.001), maternal age >30 years (aOR = 1.82, p = 0.001), and lack of folate supplementation during pregnancy (aOR = 1.67, p = 0.002) as significant independent risk factors for congenital defects. The results are shown in Table 3. Consanguinity doubled the risk of congenital defects, emphasizing the genetic component of these conditions.

Advanced maternal age increased the likelihood of chromosomal abnormalities, and the absence of folate supplementation was particularly associated with neural tube defects, highlighting the preventable nature of certain anomalies through maternal health interventions.

TABLE 3: INDEPENDENT RISK FACTORS FOR CONGENITAL DEFECTS (LOGISTIC REGRESSION)

Risk Factor	Adjusted Odds Ratio (aOR)	95% Confidence Interval (CI)	p-value
Consanguinity	2.10	1.50–2.94	<0.001
Maternal Age >30 years	1.82	1.35–2.45	0.001
Lack of Folate Supplementation	1.67	1.25–2.23	0.002
Rural Residence	1.45	1.12–1.88	0.004

Out of the 120 newborns with congenital defects, 85 (70.8%) required surgical intervention, with the most common procedures being the repair of congenital heart defects (35.8%), closure of neural tube defects (22.5%), and correction of gastrointestinal anomalies (15.8%). The overall complication rate was 18.3%, primarily due to wound infection (8.3%) and postoperative sepsis (6.7%). The mortality rate was 12.5%, with the highest mortality observed in cases of neural tube defects (18.5%) and complex congenital heart defects (16.3%). The high mortality and complication rates indicate the severity of certain congenital anomalies and underscore the importance of timely surgical intervention and post-operative care.

Histopathological examination of surgical specimens confirmed the diagnosis of congenital defects in all cases, with additional findings of ischemic changes in 15% of cases and infectious complications in 10% of cases. This confirmation highlights the importance of integrating histopathological evaluation into the diagnostic process to refine the clinical understanding of congenital anomalies and related complications. The key findings from this study reveal that congenital heart defects were the most prevalent congenital anomaly, and significant risk factors included consanguinity, maternal age >30 years, and lack of folate supplementation. Surgical intervention was required in most cases, with moderate complication and mortality rates, emphasizing the need for early diagnosis, preventive strategies, and comprehensive surgical and post-operative management.

DISCUSSION

The research results demonstrate that PUMHS Nawabshah newborns exhibit congenital birth defects at a rate reaching 10% which exceeds the WHO-reported global average of 6–7% [1]. The elevated incidence indicates the special difficulties faced by resource-limited environments because three converging factors of genetic elements and environmental elements together with socioeconomic elements boost the burden of congenital anomalies. These study findings contribute essential information regarding population trends and risk elements and consequences of defects in this regional birth population thereby establishing important operational and public health directions.[10-13]

The study results showed Congenital heart defects as the most prevalent anomaly with 35.8 percent incidence rate among cases. The prevalence of CHDs at 1% matches international observations which study congenital heart disease occurrence in

newborns[6]. CHDs encompass a wide range of structural abnormalities, from simple defects such as ventricular septal defects (VSDs) to complex anomalies like tetralogy of Fallot (TOF)[14]. The study indicates that CHDs represent a major cardiovascular concern as they require improved medical diagnostic technologies and better pediatric heart surgery services for resource-constrained areas.

Spina bifida and anencephaly made up the second most common group of defects seen in newborns resulting in 22.5% of total cases. Records show that the occurrence of birth defects at 3.3 per 1,000 live births is much higher than worldwide rates of 0.5–2 per 1,000 live births [7]. The excessive number of NTD cases seems connected to inadequate maternal nutrition combined with deficient folate consumption because these elements represent proven risk factors for NTD development [15]. Neural tube failure occurs when maternal diets lack sufficient folate levels during pregnancy thus creating these life-threatening anomalies [16]. The area needs immediate public health measures containing folate supplementation programs through food enhancement and pregnancy education to decrease the occurrence of NTDs.

Esophageal atresia together with intestinal malrotation constituted 15.8% of diagnosed birth defects in this study. These birth defects require urgent surgical handling to stop both life-threatening conditions including aspiration pneumonia and bowel obstruction [17]. The substantial number of gastrointestinal malformations documented in our research proves the need for early disease detection and sufficient pediatric operative services.

A study analysis showed that consanguineous marriages appeared in 45.8% of recorded cases which corresponds with different research from Pakistan and other regions known for consanguineous customs[8]. The strong association between consanguinity and congenital defects (aOR = 2.10) supports the role of recessive genetic disorders in the etiology of these anomalies. Genetic disorders along with metabolic diseases and structural abnormalities become more severe in people with consanguinity because their inheritance increases the risk of expressing deleterious alleles homozygously [18]. Community education initiatives need to work together with genetic counseling services to lower the occurrence of consanguineous marriages coupled with their linked dangers.

Age in excess of 30 years at childbirth functioned as an individual risk factor for developmental defects in newborns (aOR = 1.82) according to findings which match results from international research regarding Down syndrome risks in mature mothers [19]. A woman's advanced age presents two risk factors for multifactorial birth defects such as CHDs and NTDs because it impacts oocyte quality and creates more opportunities for environmental trigger exposure [20].

Insufficient prenatal folate intake during pregnancy directly correlated with higher rates of congenital defects (aOR = 1.67) because of its critical role in embryological development. During embryogenesis the reproduction and fix of DNA need folate to advance regular development but structural anomalies will emerge whenever this nutrient is deficient [21]. Low intake of folate supplements persists in numerous low- and middle-income countries because healthcare access for expectant women remains limited and public awareness regarding supplementation remains insufficient [22].

Medical staff conducted surgery on 70.8% of newborns requiring intervention mostly because of CHDs and NTDs. The treatment results reveal a complication occurrence of 18.3% and a mortality number of 12.5% whereas high-income nations achieve better results due to their advanced neonatal care along with surgical techniques 10. The high mortality numbers for NTD (18.5%) and complex CHD (16.3%) show the difficulties in managing

these conditions when healthcare facilities are limited in resource-poor areas [11].

Postoperative complications such as wound infection (8.3%) and sepsis (6.7%) proved to be primary causes of both mortality and morbidity within this research study. Without proper infection control and restricted antibiotic availability neonatal outcomes become worse due to the poor health condition of newborns [23.] NICUs require improvement alongside better infection control methods because these measures decrease complications during and after surgery.

The research findings from this study generate significant ramifications which affect both medical care delivery along with health policies for the public. Healthcare providers must raise both physician and public understanding about folate treatments for pregnant women especially in regions with high nutritional need. Rural communities should benefit from public health programs and community education along with food fortification policies because these initiatives enhance both folate consumption and the prevention of NTDs [24].

Secondarily genetic experts should perform testing on familial units who practice consanguineous relationships to decrease their risk of hereditary diseases. Providing community-based genetic counseling together with prenatal screening and diagnostic services allows medical staff to detect pregnancies that need early intervention in high-risk cases [25].

The improvement of access to prenatal diagnostic solutions and neonatal surgical treatments needs special focus on high-risk pregnancy cases and complex congenital malformations. Setting expert pediatric surgery and neonatal care centers throughout the region will improve delivery services for affected newborns and boost their treatment outcomes [26].

The study's defect incidence of 10% exceeds the worldwide average yet matches findings in other developing and middle-income countries according to research by [12]. Available research from India and Bangladesh demonstrates equivalent incidence statistics which demonstrates the mutual difficulties in managing congenital defects across limited resource areas [13]. Planned public health interventions should target the unusually high number of newborns with NTDs found in our research due to their worrisome frequency.

LIMITATIONS

This study has several limitations. These findings lose their ability to be applied generally across different areas because the study takes place exclusively at one center. Hospital-based data collection appears to show lower rates of congenital defects than the actual figures since there are some cases which remain unidentified in the community. This study lacked appropriate follow-up data which prevented researchers from studying the permanent surgical and clinical management effects.

CONCLUSION

This research demonstrates high congenital birth defect occurrence at PUMHS Nawabshah as investigators found consanguinity along with maternal ages above thirty years and non-supplementation of folate to be important risk factors. Research outcomes demonstrate that prenatal healthcare improvements combined with folate supplementation promotion and access enhancements to neonatal surgical treatment are necessary. More research needs to investigate congenital defects through prospective evaluation with extended postnatal monitoring to uncover additional risk elements affecting this population.

ETHICS APPROVAL: The ERC gave ethical review approval.

CONSENT TO PARTICIPATE: written and verbal consent was taken from subjects and next of kin.

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AUTHORS' CONTRIBUTIONS:

All persons who meet authorship criteria are listed as authors, and all authors certify that they have participated in the work to take public responsibility of this manuscript. All authors read and approved the final manuscript.

CONFLICT OF INTEREST: No competing interest declared

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