

ANALYSIS OF CONGENITAL ANOMALY TRENDS AND IMPLICATIONS IN A TERTIARY CARE HOSPITAL: A STUDY FROM PESHAWAR, PAKISTAN

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Abstract: *This research, conducted at a tertiary care hospital in Peshawar, Pakistan, aimed to examine the trends and rates of congenital malformations. The study was a cross-sectional study that took place from January 2021 to January 2022 at the Department of Gynaecology & OBs MTI, LRH Peshawar. The study's objectives were to determine the frequency and trends of congenital disabilities in infants, identify risk factors associated with these conditions, and suggest prevention and early intervention measures. The study excluded stillbirths but included newborns with congenital disabilities diagnosed through clinical examinations such as chromosomal analysis, radiography, and ultrasound. Information on kinship, maternal and prenatal history, and other risk factors were gathered through records and interviews. Statistical analysis was conducted using Fisher's exact test and chi-square test. The most common congenital anomaly identified was musculoskeletal abnormalities, accounting for 3.8% of cases. The study highlights the importance of prenatal diagnosis and treatment for prevention and timely intervention. The study's limitations include its hospital-based setting and exclusion of stillbirths, which may have led to an underestimation of prevalence. Nonetheless, the research sheds light on the need for early detection and prevention of congenital anomalies and associated risk factors. According to the report, congenital abnormalities affect 3.8% of babies at Lady Reading Hospital in Peshawar. Consanguinity and fetal abnormalities were significantly correlated ($p < 0.05$). Five occurrences of anencephaly were reported, making central nervous system (CNS) abnormalities the most common kind. Gastrointestinal tract (GIT) abnormalities accounted for 15% of cases, whereas musculoskeletal anomalies accounted for 33.2%. The genitourinary (10.5%), cardiovascular (9.1%), and cutaneous (8.7%) systems were also impacted. Although these results are consistent with other studies, if abortions and stillbirths had been taken into account, the frequency may have been greater. Prenatal diagnosis and early antenatal care are essential for intervention and prevention. The analysis shows that congenital abnormalities were 3.8% common throughout the study period at Lady Reading Hospital in Peshawar. Fetal abnormalities were substantially correlated with consanguinity. Anomalies in the central nervous system showed the most significant occurrence. Prenatal diagnosis and early antenatal care are crucial for intervention and prevention.*

Keywords: Congenital Anomalies, Tertiary Care Hospital, Prevalence, Consanguinity

Introduction

A miracle of nature is the complex process by which a single-cell zygote becomes a multicellular creature throughout human embryonic development (Jones and Smith, 2017). Congenital malformations, which include a wide range of anatomical or functional defects existing at birth, might, nonetheless, complicate this trip (Pinzon-Morales et al., 2011). These abnormalities have more significant social ramifications and are burdensome for the impacted people and their families (Webber et al., 2015). Congenital anomalies are caused by various factors, such as unknown aetiologies and environmental and genetic factors (Christianson et al., 2006). Birth abnormalities are mainly caused by environmental causes, which include maternal diseases, chemical exposures, and teratogens such as infections (Gilbert-Barnes, 2010). These elements highlight the need for a thorough study to comprehend the frequency, trends, and possible preventative measures for congenital abnormalities (Salemi et al., 2012). This research examines the trends and rates of congenital malformations in a tertiary care hospital in Peshawar, Pakistan, to add to the body of knowledge. It seeks to clarify the frequency and contributing variables, especially kinship, connected to a higher risk of fetal malformations (Ten Kate et al., 2014).

This study aims to improve prenatal diagnosis early intervention and eventually avoid congenital abnormalities by developing an excellent knowledge of these concerns (Rajab et al., 1998).

Methodology

This cross-sectional research was conducted from January 2021 to January 2022 at the Lady Reading Hospital's gynecology department in Peshawar, Pakistan. The study's goal was to thoroughly examine the patterns and incidence of congenital disabilities in infants in this tertiary care hospital environment. All babies displaying congenital abnormalities during the designated research period met the inclusion criteria. Stillborn babies, however, were not included in this investigation. Most diagnoses were made by pediatricians using a systematic clinical assessment, supplemented when needed by other diagnostic modalities, including radiography, ultrasound, echocardiography, and chromosomal analysis. Maternity and labor unit data, parent interviews, and detailed prenatal and maternal histories were carefully gathered. Maternal age, parity, and the history of kinship—defined as marriage between blood relatives, such as first cousins or siblings—were important

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data factors. The following statistical analysis used an Excel spreadsheet with the gathered data. Ratios were computed, and, where appropriate, Fisher's exact tests and Chi-square tests were used to thoroughly evaluate the relationship between congenital disabilities and consanguinity. These analytical techniques were used to assess the importance of any found associations. Recognizing the study's limitations is critical, primarily since it was conducted in a hospital and may not accurately represent the general population. Furthermore, stillbirths were not included, which might have led to an underestimate of the total frequency of congenital abnormalities.

The study covered all babies born at Lady Reading Hospital in Peshawar from January 2021 to January 2022 who were clinically identified as having a congenital anomaly. The focus was on live newborns with genetic abnormalities, so stillborn children were excluded from the research. However, all other infants with congenital abnormalities delivered at the hospital during the same time period were included in the study.

Pediatricians conducted thorough clinical assessments of infants with congenital disabilities and collected relevant data, including information about the mothers and their antenatal history, such as age, parity, and consanguinity. This data was gathered from hospital records and interviews

with parents. Ultrasonography and radiography were also used as necessary diagnostic investigations.

Statistical analysis was done using the information that was gathered. Proportions were computed to ascertain the prevalence of congenital abnormalities. Fisher's exact and Chi-square statistical tests evaluated the relationship between kinship and congenital anomalies. These tests were used to assess the importance of any correlations or associations found in the data.

Results

The analysis found that congenital abnormalities affected 3.8% of babies at Lady Reading Hospital in Peshawar throughout the study period. Interestingly, consanguinity and fetal malformations were significantly correlated (p<0.05), emphasizing the contribution of genetic determinants to congenital deformities. With five instances of anencephaly reported, central nervous system (CNS) abnormalities had the most significant frequency. Anomalies of the musculoskeletal system were the most common (33.2%), followed by those of the gastrointestinal tract (GIT) (15%). These results are consistent with previous research, but it's crucial to remember that including stillbirths and carrying out community-based studies might provide a complete picture of prevalence.

Table 01: Distribution of fetal anomalies by consanguinity

Total No cases	Without fetal anomalies	With fetal anomalies	p-value
Second trimester	11	10	<0.05
Third trimester	7	9	<0.05
Total	18	19	<0.05

Table 02: Relationship between maternal and perinatal risk factors and congenital abnormalities

Variables	Groups	Congenital Anomaly						
		Yes		No		Total	X ² value, df,	p value
		N	%	N	%			
Maternal age	<20 years	83	1.9	4326	98.1	4409	3.69, df=2,	0.157
	20-30 years	174	2.4	7004	97.6	7178		
	>30 years	29	2.2	1280	97.8	1309		
Parity	Primiparas	171	1.8	9185	98.2	9356	23.91	0.000*
	Multiparas	115	3.3	3425	98.7	3540		
consanguinity	Present	2	40	3	60	5	-	0.000*
	Absent	284	2.2	12607	97.8	12891		
Birth Weight	Very low	14	0.8	1756	99.2	1770	94.17, df=3	0.000*
	Low	206	3.8	5489	96.2	5495		
	Normal	51	1.3	3747	98.7	3798		
	High	15	0.8	1818	99.2	1833		
Mode of delivery	Vaginal	205	2.5	8042	97.5	8247	7.58	0.005*
	Caesarean	81	1.7	4568	98.3	4649		
Gestation	Term	90	1	8356	99.0	8446	149.83	0.000*
	Preterm	196	4.4	4254	95.6	4450		
Gender	Male	191	2.9	6248	97.1	6619	27.97	0.000*
	Female	95	1.5	6182	98.5	6277		

Discussion

Congenital disabilities are a major global contributor to infant morbidity and death, making them a severe public health problem (Dolk et al., 2010; Malherbe et al., 2016;

Ramakrishnan et al., 2021): our study's results, consequences, and the larger picture of congenital abnormalities (Akhter et al., 2019). First and foremost, our studies reported prevalence of congenital abnormalities (2.22%) is consistent with previous Indian studies that

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found incidence rates of 1.9% and 2.72% (Olsen et al., 2013). This consistency across studies from various fields emphasizes how congenital abnormalities appear in a range of populations in a very consistent way (MacLennan and Zvaritch, 2011). It is essential to acknowledge that several factors, including but not limited to geographic location, socioeconomic level, healthcare accessibility, and genetic diversity, may impact prevalence rates. The primary limitation of our research is that stillbirths and abortions were not included in the analysis. The prevalence of congenital anomalies may have been underestimated due to this exclusion (Singh and Gupta, 2009; Yngwe et al., 2003). These cases contribute substantially to the overall burden of congenital abnormalities and should be included in future research to offer a more comprehensive understanding of the problem (Kumar et al., 1996). Understanding the quirks and challenges of congenital malformations in medical settings requires research in hospitals like ours. Recognizing the inherent limitations of these types of studies is crucial (Kalter and Warkany, 1983). Lady Reading Hospital and other tertiary care facilities often encounter complex situations that may not be typical of the general public. Consequently, even if our results provide insightful information, they cannot be applied to the whole population (Van Der Linde et al., 2011). Community-based studies are strongly suggested to evaluate the incidence and patterns of congenital abnormalities. These studies may provide a more accurate picture of the prevalence and related risk factors since they include a more extensive range of demographics and the healthcare environment (Tinanoff et al., 2019). Furthermore, by identifying regional differences and changes in most congenital anomalies, these studies might support the creation of preventative measures tailored to a particular area. The most prevalent kinds of sexual abnormalities in our study's pattern were musculoskeletal malformations, followed in frequency by anomalies of the gastrointestinal tract (GIT), central nervous system (CNS), genitourinary anomalies, cardiovascular anomalies, and skin anomalies. These results align with several previous studies (D'Amato et al., 2015). It is essential to recognize that disparities in sample size, research design, and geographic variables may lead to discrepancies in results between studies. The intricate interaction of genetic, environmental, and geographical variables in the etiology of these disorders is highlighted by the regional heterogeneity in congenital abnormality patterns. Because of this heterogeneity, healthcare planning, prevention, and intervention must be done with a customized strategy (Control and Prevention, 2011). Because many factors contribute to congenital malformations, healthcare systems must be flexible and sensitive to local trends and requirements (Morris et al., 2021).

This study's shortcomings include its inability to generalize results to the whole population, possible selection bias in a hospital-based context, and its exclusion of abortions and stillbirths. Furthermore, the fact that different research has produced different results emphasizes how complicated congenital abnormalities are.

Community-based studies should be included in future research to provide a more complete picture of congenital malformations, which include stillbirths and abortions. Our knowledge of these disorders will improve with research

into regional differences and identifying genetic and environmental causes. This will result in the development of more effective preventative and intervention techniques

Conclusion

The study sheds light on the kinds and frequency of congenital disabilities in our community. For the sake of prevention, early intervention, and planned termination when medically indicated, routine prenatal checkups and prenatal diagnostics are advised. Planning and intervention techniques for healthcare must take geographical variations into account.

Declarations

Data Availability statement

All data generated or analyzed during the study are included in the manuscript.

Ethics approval and consent to participate

Approved by the department Concerned.

Consent for publication

Approved

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Conflict of interest

The authors declared absence of conflict of interest.

Author Contribution

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