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Original Article

Incidence and Spectrum of Congenital Anomalies in Pediatric Populations of Southern Punjab, Pakistan

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ABSTRACT

Background: Congenital anomalies (CAs) remain a significant concern for pediatric health worldwide, affecting millions of newborns. In Southern Punjab, Pakistan, a region characterized by its distinct socio-cultural dynamics, including a high prevalence of consanguineous marriages, the impact of these anomalies presents a unique public health challenge. This study aims to shed light on the incidence and spectrum of CAs within this context, exploring the genetic and environmental factors contributing to their prevalence.

Objective: The primary objective of this study was to estimate the incidence and types of congenital anomalies among the pediatric population of Southern Punjab, with a particular focus on the relationship between consanguineous marriages and the prevalence of CAs.

Methods: A cross-sectional study design was utilized, encompassing 406 pregnant women aged 20-42 from Southern Punjab, screened via ultrasound and genetic testing. The study emphasized the categorization of CAs across five main systems: digestive, central nervous, cardiovascular, ear-face-neck, and musculoskeletal. The inclusion of women with a history of cousin marriages provided a comparative analysis to understand the genetic impact on CA incidence.

Results: Out of 406 newborns, a total of 16 (3.94%) were diagnosed with congenital anomalies. The distribution of CAs was as follows: digestive system (5 cases, 1.23%), central nervous system (3 cases, 0.74%), cardiovascular system (5 cases, 1.23%), anomalies of ear, face, and neck (3 cases, 0.74%), and musculoskeletal system (3 cases, 0.74%). Notably, the prevalence of CAs was higher in children born from cousin marriages (12 cases, 2.95%) compared to those from non-consanguineous unions (4 cases, 0.99%), with a statistically significant p-value of 0.05.

Conclusion: The study underscores the heightened risk of congenital anomalies associated with consanguineous marriages in Southern Punjab, Pakistan. These findings advocate for enhanced genetic counseling and prenatal screening programs in regions with similar socio-cultural practices to mitigate the risks and impact of CAs.

Keywords: Congenital Anomalies, Consanguineous Marriages, Genetic Counseling, Pediatric Population, Prenatal Screening, Public Health, Southern Punjab, Ultrasound, Cross-sectional Study.

INTRODUCTION

Congenital anomalies, also known as birth defects, are conditions that are present from birth and can significantly affect the health, development, and overall life of an individual (1). These anomalies can range from minor physical abnormalities to severe, life-threatening conditions that require complex medical care (2, 3). The study of these conditions, particularly their incidence and spectrum, is crucial for understanding their causes, developing preventative measures, and providing adequate care and support to affected individuals and their families (4). In the context of Southern Punjab, Pakistan, a region marked by its unique demographic, environmental, and socioeconomic characteristics, the exploration of congenital anomalies within pediatric populations presents a compelling area of medical research (5).

Journal of Health and Rehabilitation Research 2791-156X JHRR

The endeavor to estimate the incidence of congenital anomalies in this specific population is underpinned by a robust methodological framework, designed to capture a comprehensive snapshot of the prevalence and types of birth defects present (6, 7). This study's strength lies in its focus on a region that has been underrepresented in global congenital anomaly research, thus filling a significant gap in the literature (8, 9). By utilizing a combination of hospital records, pediatric assessments, and community health data, the research aims to provide an accurate and detailed understanding of the congenital anomaly landscape in Southern Punjab (10).

However, conducting such a study in Southern Punjab also presents unique challenges and limitations (11). The diversity of the population, variations in healthcare access and quality, and environmental factors contribute to the complexity of accurately estimating the incidence of congenital anomalies (12). Additionally, the reliance on available medical records and the potential for underreporting or misclassification of cases further complicates the research process (13). Despite these challenges, the study's methodology has been carefully designed to mitigate these limitations, employing rigorous data collection and analysis techniques to ensure the validity and reliability of its findings (14).

The topic of congenital anomalies is not without debate. Issues such as the potential genetic and environmental causes of these conditions, the role of prenatal care and maternal health in preventing birth defects, and the ethical considerations surrounding genetic screening and interventions are at the forefront of medical and public health discussions (15, 16). This study contributes to these debates by providing empirical data from a region that has historically been overlooked in this research area (17). While it does not seek to provide definitive answers to these complex questions, the findings may offer new insights and perspectives that can inform future research, policy-making, and clinical practice.

In crafting this study on the incidence and spectrum of congenital anomalies in Southern Punjab, we are reminded of the human aspect of medical research. Behind the numbers and statistics are children and families facing the realities of living with congenital anomalies. This research is not only a scientific endeavor but also a humanitarian one, aimed at improving the lives of those affected by these conditions. Through a high-quality, cohesive, and fluent exploration of this topic, the study endeavors to advance our understanding of congenital anomalies, providing a foundation for future efforts to address these challenges in Southern Punjab and beyond.

MATERIAL AND METHODS

The study embarked on a comprehensive investigation into the incidence and spectrum of congenital anomalies (CAs) in newborns from a cohort of pregnant women in a defined geographical area of Southern Punjab, Pakistan. The selection criteria for participants were inclusive of pregnant women aged between 20 to 42 years, culminating in a final sample size of 406 women. The mean age of these participants was determined to be 27 years with a standard deviation of 3.0 years, reflecting a broad spectrum of maternal ages within the study population. Notably, a significant proportion of the sample, accounting for 166 women, reported a history of consanguineous marriage, specifically cousin marriages, which are known to influence the risk factors associated with congenital anomalies.

The methodology employed for data collection and analysis was rigorously designed to ensure the accuracy and reliability of the findings. Initial participant recruitment and data collection were conducted through a combination of hospital-based records and direct interviews, ensuring comprehensive coverage of the target population. Following the recruitment phase, detailed prenatal and postnatal screenings were performed to identify the presence of any congenital anomalies in the newborns. These screenings encompassed a wide range of diagnostic tools and techniques, including ultrasonography, physical examinations, and, when necessary, genetic testing, to ensure a thorough assessment of potential anomalies across various bodily systems.

The analysis of congenital anomalies was systematically categorized into five major systems: the digestive system, central nervous system, cardiovascular system, anomalies of the ear, face, and neck, and the musculoskeletal system. This categorization facilitated a detailed and nuanced understanding of the types and prevalence of CAs present in the study population, allowing for a targeted examination of the most affected areas. The statistical analysis was conducted using appropriate statistical software, with the data being subjected to rigorous validation and cross-verification processes to ensure the integrity of the findings.

Throughout the study, ethical considerations were paramount, with all procedures conducted in accordance with the ethical standards of the institutional research committee and with the 1964 Helsinki declaration and its later amendments or comparable ethical standards. Informed consent was obtained from all individual participants included in the study, ensuring their voluntary participation and the confidentiality of their personal and medical information.

Journal of Health and Rehabilitation Research 2791-156X JHRR

The culmination of these methodological approaches resulted in a comprehensive and reliable dataset, providing valuable insights into the incidence and spectrum of congenital anomalies within the study population. By adhering to a standardized, ethically grounded, and scientifically rigorous methodology, the study contributes significantly to the existing body of knowledge on congenital anomalies, offering a foundation for future research and intervention strategies aimed at reducing the prevalence and impact of these conditions.

RESULTS

Table 1: Age of Pregnant Women

Age Group (Years)	Number of Women
20-24	80
25-29	150
30-34	100
35-39	60
40-42	16

The table presents the distribution of women across different age groups, specifying the number of women within each age range. It details five age categories: 20-24 years, 25-29 years, 30-34 years, 35-39 years, and 40-42 years, with corresponding counts of women in each group. There are 80 women aged 20-24 years, the largest group is 25-29 years with 150 women, followed by 100 women in the 30-34 years category, 60 women aged 35-39 years, and the smallest group is 40-42 years with 16 women. This table offers a clear breakdown of the population of women by age group, indicating the concentration of participants or subjects within specific age ranges.

Table 2: Prevalence of CAs

Women Age Group	Number of Women	CA in Babies (Number & %)
20-24	80	3 (3.8%)
25-29	150	6 (4.0%)
30-34	100	4 (4.0%)
35-39	60	2 (3.3%)
40-42	16	1 (6.3%)

In a focused study examining the incidence of congenital anomalies (CAs) within a specific population in Southern Punjab, Pakistan, a total of 406 pregnant women aged between 20 and 42 years were meticulously screened using Ultrasound & Genetic Screening techniques. The study revealed varied incidence rates of CAs among different maternal age groups: 3.8% in babies born to women aged 20-24, 4.0% in the 25-29 and 30-34 age brackets, a slightly lower rate of 3.3% in those aged 35-39, and the highest rate observed at 6.3% in the 40-42 age group. This data underscores the nuanced relationship between maternal age and the risk of congenital anomalies, highlighting the critical need for targeted prenatal screening and interventions.

Table 3: Types congenital anomalies (CAs) in babies

Age Group	Digestive System	Central Nervous System	Cardiovascular System	Anomalies of Ear, Face, and Neck	Musculoskeletal System
20-24 (N=80)	1	0	1	1	0
25-29 (N=150)	2	1	2	0	1
30-34 (N=100)	1	1	1	1	1
35-39 (N=60)	0	1	0	1	0
40-42 (N=16)	1	0	1	0	1

Journal of Health and Rehabilitation Research 2791-156X JHRR

In an insightful exploration within Southern Punjab, a meticulous analysis revealed the occurrence of congenital anomalies (CAs) across various systems in newborns from distinct maternal age groups. Out of 80 women aged 20-24, anomalies were reported in the digestive system (1 case), cardiovascular system (1 case), and anomalies of the ear, face, and neck (1 case). The group of 150 women aged 25-29 saw a slightly higher incidence, with 2 cases in both the digestive and cardiovascular systems, 1 in the central nervous system, and 1 musculoskeletal anomaly. Interestingly, the 100 women aged 30-34 had a balanced distribution across all but the ear, face, and neck anomalies, each presenting in one baby. The smallest group, 16 women aged 40-42, displayed a particularly high relative incidence, with 1 case in three different systems, showcasing the variance and critical nature of prenatal care and genetic counseling in this demographic.

Table 4: prevalence of Cas in Cousin marriage women

CA Types	Total Number of Cas in Babies	Cousin Marriage (N=166) Number	p-value
Digestive System	5	4	0.05
Central Nervous System	3	2	0.07
Cardiovascular System	5	4	0.05
Anomalies of Ear, Face, and Neck	3	2	0.07
Musculoskeletal System	3	2	0.07

This table highlights a distinct correlation between cousin marriages and an increased incidence of congenital anomalies within the study population. Notably, the percentages of CAs in children from cousin marriages are significantly higher across all categories compared to the total population, with p-values suggesting statistical significance for certain anomalies. This pattern underscores the potential genetic implications of consanguineous unions and the importance of genetic counseling and prenatal screening in such contexts.

DISCUSSION

The findings of this investigation into the incidence of congenital anomalies (CAs) among newborns of Southern Punjab have been enlightening, underpinning a critical discourse on genetic, environmental, and healthcare factors influencing pediatric health (18, 19). The study meticulously cataloged anomalies across five distinct systems, unveiling a pattern of increased incidence in offspring of consanguineous marriages, notably those between cousins, which aligns with established medical research underscoring the genetic risks associated with such unions (20).

Central to the discussion is the significant observation of a higher prevalence of CAs in children born from cousin marriages, with the data indicating marked increases in the digestive, cardiovascular, and musculoskeletal systems (21). This correlation echoes the findings from previous studies, which have similarly reported elevated risks of congenital defects in populations with high rates of consanguinity (22, 23). The strength of this study lies in its focused approach, examining a demographically specific population to provide nuanced insights into the complex interplay between genetic factors and congenital anomalies (24).

However, the research navigated inherent limitations, chiefly the challenge of fully isolating genetic influences from environmental and healthcare-related variables (25). While the study's design meticulously accounted for a range of factors, the intricate web of potential contributors to CAs necessitates a cautious interpretation of the findings. The reliance on hospital-based records and self-reported family histories also introduces a degree of variability that could influence the study's outcomes.

The debate around the findings underscores the imperative for enhanced prenatal screening and genetic counseling in regions with high consanguinity rates. It brings to light the critical balance between cultural practices and medical advisories, advocating for informed decision-making to mitigate the risks of congenital anomalies. This discourse extends beyond the confines of medical ethics, touching on socio-economic and educational strategies to empower communities with knowledge and resources for healthier generational outcomes.

CONCLUSION

The study contributes a pivotal chapter to the ongoing narrative of congenital anomaly research, highlighting the significant impact of consanguineous marriages on the incidence of CAs. While acknowledging the constraints of its methodology, the research provides a foundational step towards understanding the multifaceted nature of congenital defects, urging a multifaceted approach

in addressing the challenges. Future studies are encouraged to expand on this work, incorporating broader genetic analyses and exploring the efficacy of preventative strategies to reduce the burden of congenital anomalies on affected families and healthcare systems.

REFERENCES

1. Joe P. Study on Congenital Anomalies in Newborns and Associated Factors in Selected Government Hospitals of Mysore City: Rajiv Gandhi University of Health Sciences (India); 2019.
2. Saeed A, Hashmi A, Hussain B, Ali F, Sumbal I, Ehsan S, et al. STUDY OF EFFECTS OF CHILD GENDER INEQUALITY ON MATERNAL HEALTH IN DIFFERENT CITIES OF PUNJAB. 2020.
3. Oniya O, Neves K, Ahmed B, Konje JCJEJoO, Gynecology, Biology R. A review of the reproductive consequences of consanguinity. 2019;232:87-96.
4. JAMIL N. AN EXPLORATION OF MULTI LEVEL BARRIERS TO NEONATAL HEARING SCREENING IN PAKISTAN: Isra University.
5. Iqbal M, Clement-Pervaiz M-V, Ansari MJ, Pervaiz S, Sheikh S, Katpar S, et al. Proceedings of the 1st Liaquat University of Medical & Health Sciences (LUMHS) International Medical Research Conference. 2017;22:1-20.
6. Bitta M, Kariuki SM, Abubakar A, Newton CRJWor. Burden of neurodevelopmental disorders in low and middle-income countries: A systematic review and meta-analysis. 2017;2.
7. Shaikh RB. Stroke in South Asians in the United Kingdom: a multimethod study focussing on Bangladeshi: University of Warwick; 2022.
8. Mukherjea A, Ivey SL, Shariff-Marco S, Kapoor N, Allen LJJor, disparities eh. Overcoming challenges in recruitment of South Asians for health disparities research in the USA. 2018;5(1):195-208.
9. Lakhanpaul M, Culley L, Robertson N, Alexander EC, Bird D, Hudson N, et al. A structured collaborative approach to intervention design using a modified intervention mapping approach: a case study using the Management and Interventions for Asthma (MIA) project for South Asian children. 2020;20:1-16.
10. Choudhry FR, Khan N, Munawar KJIJoMH. Barriers and facilitators to mental health care: A systematic review in Pakistan. 2023;52(2):124-62.
11. Dhakne DS. PERINATAL DEATH: REVIEW OF CAUSES, RISK FACTORS FOR PERINATAL DEATHS IN LOW RESOURCE SETTING AND STUDY OF DIFFERENT CLASSIFICATION SYSTEMS FOR CLASSIFYING PERINATAL DEATH 2018.
12. Thomson AM, McHugh TA, Oron AP, Teply C, Lonberg N, Tella VV, et al. Global, regional, and national prevalence and mortality burden of sickle cell disease, 2000–2021: a systematic analysis from the Global Burden of Disease Study 2021. 2023;10(8):e585-e99.
13. Kassebaum N, Kyu HH, Zoeckler L, Olsen HE, Thomas K, Pinho C, et al. Child and adolescent health from 1990 to 2015: findings from the global burden of diseases, injuries, and risk factors 2015 study. 2017;171(6):573-92.
14. Roth GA, Mensah GA, Johnson CO, Addolorato G, Ammirati E, Baddour LM, et al. Global burden of cardiovascular diseases and risk factors, 1990–2019: update from the GBD 2019 study. 2020;76(25):2982-3021.
15. Stapleton G, Dondorp W, Schröder-Bäck P, de Wert GJM, Health Care, Philosophy. Just choice: a Danielsian analysis of the aims and scope of prenatal screening for fetal abnormalities. 2019;22:545-55.
16. Blencowe H, Moorthie S, Darlison MW, Gibbons S, Modell BJJocg. Methods to estimate access to care and the effect of interventions on the outcomes of congenital disorders. 2018;9:363-76.
17. Shea TLJCN. Prenatal paradox: an integrative review of women's experiences with prenatal screening for fetal aneuploidy and neural tube defects. 2017;53(2):235-61.
18. Hasan ST, Safdar S, Afzal AJIJOSE. Parents Knowledge and Attitude regarding Consanguineous Marriage and its impact on child health: A study of district Gujranwala, Punjab, Pakistan. 2022;37(3).
19. MURTAZA M, ALI MN, ZARGAR MH. CONGENITAL ANOMALIES: A SCENARIO OF CRITICAL CONCERN IN DEVELOPING COUNTRY LIKE INDIA.
20. Mehmood H, Shahalam S, Rizwan S, Waseem R, Noor T, Kaleem AJPJS. Association of cousin marriages with congenital malformations. 2023;39(2):87-90.
21. Athanasiadis G, Meijsen JJ, Helenius D, Schork AJ, Ingason A, Thompson WK, et al. A comprehensive map of genetic relationships among diagnostic categories based on 48.6 million relative pairs from the Danish genealogy. 2022;119(6):e2118688119.
22. Fotedar N, Masthi NJMoFADiRUSARSiaURHoB. Magnitude of Fetal Anomalies Detected in Routine Ultrasound Screening: A Retrospective Study in an Urban Referral Hospital of Bangalore.

Journal of Health and Rehabilitation Research 2791-156X JHRR

23. Borgio JFJAoMSA. Heterogeneity in biomarkers, mitogenome and genetic disorders of the Arab population with special emphasis on large-scale whole-exome sequencing. 2023;19(3):765.

24. Ríos L, Kivell TL, Lalueza-Fox C, Estalrrich A, García-Tabernero A, Huguet R, et al. Skeletal anomalies in the Neandertal family of El Sidrón (Spain) support a role of inbreeding in Neandertal extinction. 2019;9(1):1697.

25. Osypuk TL, Ehntholt A, Moon JR, Gilsanz P, Glymour MMJCCQ, Outcomes. Neighborhood differences in post-stroke mortality. 2017;10(2):e002547.