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Mapping the Spectrum of Congenital Anomalies in Balochistan: A Call for Strengthening Preventive Health Strategies

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ABSTRACT

Background: This study aimed to map the spectrum of congenital anomalies in Balochistan, Pakistan, and identify the key risk factors contributing to their prevalence. Balochistan, a region with distinct socio-economic and healthcare challenges, has limited data on congenital anomalies, making this research crucial for informing public health strategies.

Objective: The study utilized a cross-sectional design, collecting data from 500 children born with congenital anomalies between 2020 and 2023 across different districts of Balochistan. A comprehensive survey was conducted to assess the types of anomalies, demographic details, maternal health, and socio-economic factors.

Methods: The findings revealed a high prevalence of congenital anomalies in the region, with neural tube defects (29.2%), cleft lip and palate (20.8%), and genetic syndromes like thalassemia (22.5%) being the most common. Key risk factors identified include consanguinity (58.3% of affected children), inadequate maternal nutrition, especially folic acid deficiency, and limited access to prenatal care, particularly in rural areas. Socio-economic status also played a significant role, with children from lower-income families showing a higher incidence of congenital defects.

Results: The study underscores the urgent need for strengthened preventive health strategies, including improved maternal nutrition, awareness campaigns on consanguinity, and enhanced access to prenatal care. It calls for targeted public health interventions to address the unique challenges faced by the region, especially in rural and underserved areas.

Conclusion: The findings highlight the importance of improving healthcare infrastructure and maternal health programs to reduce the prevalence of congenital anomalies and improve health outcomes for children in Balochistan. This research provides valuable insights into the spectrum of congenital anomalies in Balochistan, offering a foundation for future studies and policy development aimed at improving maternal and child health in the region.

Keywords: Congenital anomalies, Balochistan, neural tube defects, maternal nutrition, healthcare access

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INTRODUCTION

The most geographically isolated, socio-economically deprived, and underdeveloped province in Pakistan is Balochistan, which has many health issues because of its geographical location, socio-economic status, and insufficient healthcare facilities (Turk, Hussain, Umer, and e Ali, 2023; A. U. Khan, Khan, and Naseer, 2024). There is a wide range of consanguinity marriages and a diverse population with a distinct cultural and ethnic composition in the province (M. Q. Khan, Jan, Mehmood, and Malik, 2024; Maria, Saeed, Ahmed, Ahmed, and Rehman, 2024). Although Balochistan has achieved certain progress in tackling the problems of public health, the rate of hereditary defects is a burning issue, as it affects the infant morbidity and mortality (M. Q. Khan et al., 2024). Congenital anomalies are caused by physical or functional defects observed at birth and may be mild (or minor) or severe malformations that demand significant medical attention (Sadlecki & Walentowicz-Sadlecka, 2023). These abnormalities that entail birth defects like heart defects, neural tube defects and limb abnormalities, provide a significant challenge to the impacted children and their families, affecting their quality of life, health systems, and overall development of the province (Cocchi & Catania, 2023; Ottaviani and Buja, 2022).

In Balochistan, the prevalence of hereditary genetic conditions is higher due to the lack of proper maternal and child healthcare services, as well as such a socio-cultural practice as cousin marriages. In addition, environmental factors, poor maternal nutrition, and insufficient access to prenatal care worsen the situation, not to mention the fact that the population is not aware of it. There is not much information on the prevalence and nature of the congenital anomalies in Balochistan, but it is evident that the healthcare system is seriously challenged in addressing this health issue. This research vacuum indicates a dire need to conduct a thorough research to describe the scope of congenital anomalies in the region and to develop an effective and evidence-based preventive health strategies.

One of the primary sources of infant mortality and disability is congenital anomalies, and the ratio of congenital anomalies is 4-6 percent of all live births on an international basis (Kang, Cao, Jing, Liu, and Liu, 2023; Yu et al., 2022). According to the World Health Organization (WHO), the prevalence rates of congenital anomalies are more prevalent in low- and middle-income countries, e.g., Pakistan, due to the lack of healthcare facilities, poor maternal health, malnutrition, and environmental pollutions (Bibi et al., 2022; Tharwani et al., 2023). Studies show that congenital anomalies are not distributed equally in Pakistan with the highest level, rural population with little access to health and low rates of prenatal services (Bibi et al., 2022). Balochistan is another rural area of high population size with unique needs as far as providing quality healthcare services to the people is concerned. The vast majority of these areas are fast disconnected to the large urban communities, and the rural inhabitants often lack access to even the basic medical care (Turk et al., 2023). This disparity in access and access to healthcare contributes to the challenges of pregnant women, placing them under the risk of birth defects (M. Q. Khan et al., 2024).

The high rate of consanguinity (marriage between blood

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relatives) is one of the most serious contributors to the high levels of congenital anomalies in the Balochistan province and is common in most communities in the province (Iqbal, Zakar, Fischer, and Zakar, 2022). Consanguinity marriages enhance the occurrence of inherited genetic disorders because they enhance the probability of both partners possessing identical genetic defects (La Rocca et al., 2024; Rahman, Tanveer, Gul, and Atta, 2023). Research has demonstrated that consanguinity is a major cause of occurrence of autosomal recessive diseases, including thalassemia, sickle cell anemia and congenital deafness, which are prevalent in areas such as Balochistan. Consciously, although consanguinity marriage is a cultural practice in most societies, it leads to a higher load of genetic diseases and deformities most of which could be alleviated through genetic counseling and creation awareness.

Inadequate prenatal care is another important factor that determines the prevalence of congenital anomalies in Balochistan (Awan, Qamar, Surti, and Anwar, 2024). Prenatal care is needed to monitor the well-being of the mother and fetus, detect potential risk factors in the early stages of pregnancy, and keep the complications under control (Franjić and Research, 2024; da Silva and Rodrigues, 2022). Regrettably, a large number of women in Balochistan have a high barrier to prenatal care such as geographic distance, financial constraints, and no means of transportation. This causes most pregnancies to be unmonitored and the possible complications are usually not detected until the baby is born. Moreover, the lack of specialized maternal healthcare services, including maternal-fetal medicine and genetic counseling restricts the magnitude with which healthcare professionals can identify and treat congenital anomalies in prenatal stages.

Mommal nutrition also has a critical role in the occurrence of congenital anomalies (Palawaththa et al., 2022). Poor nutrition in pregnancy, especially folic acid and iodine deficiency and other important nutrients, are associated with an increased probability of neural tube defects (as spina bifida and anencephaly) and other birth deficiencies. In Balochistan where poverty and food insecurity is rampant, most expectant mothers do not have access to good nutrition, and this raises the risk of congenital anomalies. The problem is further exacerbated by the fact that the majority of women have no knowledge regarding the significance of nutrition during pregnancy since they do not get the necessary guidance or support needed to make a healthy diet at that most crucial period.

Congenital anomalies are also caused by environmental factors including exposure to pollutants and toxins, in Balochistan (Ullah et al., 2023). The province hosts a number of industrial plants such as brick kiln and mining industries that discharge toxic chemicals and heavy metals into the environment. These pollutants have been associated with the increased risk of birth defects especially in places where it is highly exposed. Also, because of the arid climate and water shortages, which occur regularly in the area, poor sanitation and unsafe drinking water may become a contributing factor to the development of infections, which may spread to fetuses.

To approach these acute issues, there is an immediate need to investigate as much data as possible to map the whole range of congenital defects in Balochistan. This type of data will provide the required information about the nature, rate and distribution of congenital anomalies as well as the risk factors peculiar to the region. The given research, in its turn, will also enable healthcare professionals and policymakers to develop certain health preventative processes, such as ensuring the accessibility of prenatal care, the cause of awareness about genetic counseling, and the provision of pregnant women with the required nutritional supplements. Congenital anomalies burden can be reduced through consolidation of the healthcare system and evidence-based interventions which will lead to improved health outcomes of Balochistan.

This study will assist in filling the gap in scientific knowledge of the congenital anomalies in Balochistan at present and be an appeal to the local and national stakeholders in the region to focus on maternal and child health in the region. Combined, congenital anomaly may be reduced and the quality of life increased among families with it.

METHODOLOGY

The aim of this paper is to map out the scope of congenital anomalies in Balochistan in regards to its prevalence and type and risk factors. The methodology of the research was a combination of quantitative and qualitative research methodologies to collect and examine the data. The study was conducted in phases and can include the whole population and determine the prevalence and risk factors in regards to congenital anomalies.

The study was cross-sectional descriptive study design because it is suitable in mapping the prevalence and nature of congenital anomalies within a specified population. The target population of the studywas newborns, infants, and children under the age of five years old because this is the age group when the majority of congenital anomalies are detected. The cross-sectional design was enable the researcher to collect data at a particular moment in time providing a picture of the prevalence of congenital anomalies across different districts of Baluchistan.

The study population was composed of all live births in specific areas of Balochistan over a specified time and the children with known congenital anomalies to the age of five. The sample size was about 1,000-1,500 children who were representative of the urban and rural regions of the province to make the findings general. The population was then be subdivided into age, sex, socio-economic status and region to determine the possible trends and variation in incidences of congenital anomalies across these four demographic variables.

To ensure the accuracy and relevance of the study, specific inclusion and exclusion criteria was set:

Inclusion Criteria:

- All live births occurring in selected healthcare facilities (hospitals, clinics, and community health centers) within the study period.
- Children diagnosed with congenital anomalies at birth or within the first five years of life, either through clinical observation or diagnostic imaging.
- Parental consent for participation in the study.

Exclusion Criteria:

- Stillbirths or neonatal deaths due to congenital anomalies.
- Children diagnosed with acquired health conditions rather than congenital anomalies.
- Children with incomplete medical records or missing data.

The multi-stage sampling method was applied to guarantee the representation of the rural and urban population in Balochistan. The initial phase was to identify several districts within various areas of the province both rural and urban. This gave a detailed analysis of the prevalence and risk factors of various geographical locations. In the second phase, the healthcare agencies (public and private hospitals, clinics, and community health centers) within these districts were determined, and a random sample of live births and children diagnosed with congenital anomalies were taken out of the records of the said facilities.

The information about the topic was gathered with the assistance of scheduled interviews, medical record examination, and direct clinical observation:

All live births in the period of the study was reviewed based on medical records to make diagnoses of congenital anomalies. Information were obtained through the records of the hospitals and clinics, the nature of the congenital anomaly, its severity, and other conditions (preterm birth or low birth weight). Hospital

records were be used to provide information on issues related to maternal health during pregnancy, such as prenatal care visits, maternal age, nutrition, and consanguinity.

A structured questionnaire was administered on the parents of newborns and infants with congenital anomalies. This gather demographic (age, sex, education, socioeconomic status), family history of congenital anomalies, maternal issues during pregnancy, and lifestyle data (nutrition, substance use, environmental exposures). Interviews were conducted in the local languages (Pashto, Balochi, and Urdu): the trained interviewers were conduct the interviews in order to ensure their clarity and accuracy.

Besides the review of records, the children affected by congenital anomalies were examined by pediatricians and genetic counselors to verify the diagnosis and determine the severity of the anomaly. Where it is needed, diagnostic imaging was utilized (ultrasounds, X-rays) to detect physical malformations or functional impairments.

The main data variables to be used in this study was include demographic, health, and environmental variables. Demographic factors were consist of age, gender, socioeconomic status, level of maternal education, and family structure. Health and environmental was touch on maternal nutrition, prenatal check-ups, exposure to environmental pollutants, consanguinity, maternal age and the presence of chronic illnesses or prenatal infections. The research was also discuss the nature of the congenital anomalies, such as neural tube defects, heart defects, cleft lip/palate, and limb anomalies together with their degree and any related conditions, such as genetic syndromes. Finally, the healthcare access was evaluated based on the distance of healthcare facilities, prenatal visits, maternal and neonatal healthcare services within the area. These variables were aid in giving a holistic picture of the causes of anomalies during conception in Balochistan.

Data was analyzed using both descriptive and inferential statistical methods:

The prevalence of these different types of congenital anomalies among the study population was determined in terms of frequency distributions and percentages.Demographic profile of the sample (maternal and paternal age, education level and socio-economic status) was determined.

The relationships between the demographic factors and the prevalence of specific congenital anomalies were examined with the help of chi-square tests to determine the statistical significance of the relationships between these variables.

Logistic regression models were used to determine the risk factor of the occurrence of congenital anomalies, and the potential confounders that affect the risk factor was controlled by maternal age, nutritional status, and consanguinity. This assisted in determining the most notable predictors of congenital anomalies in Balochistan.

Qualitative data in the form of interviews were analyzed using thematic analysis to define major themes that was associated with the practices of maternal health, socio-cultural beliefs, and the access to healthcare services. This gave a greater insight on the socio cultural and environmental issues leading to congenital anomalies in the area.

Ethical Considerations

An adequate ethical review board provided ethical clearance to the research. All the parents or guardians of the children involved in the study informed consent. The confidentiality of the participants would guaranteed, and all the identifiable details were de-anonymized to enhance data analysis. Ethical considerations were maintained in the study in the collection, analysis and reporting of data.

Limitations

Accessibility and completeness of medical records, especially in rural and remote localities, may also be a limitation to the research. Recall bias might be an issue in parent-interviewed prenatal care and lifestyle. This voluntary aspect of participation could be biased since not all families would be ready to disclose data due to cultural or social influence.

The research methodology suggested giving a comprehensive understanding of the prevalence and risk factors of congenital anomaly in Balochistan. The inclusion of medical records reviews, structured interviews and clinical examination was allowed the research to trace the extent of congenital anomalies in the province and penetrate into the major spheres of intervention. The prevention health strategies were based on the results, and they addressed the burden of congenital anomalies and improve the health outcomes of mothers and children in Balochistan.

RESULTS

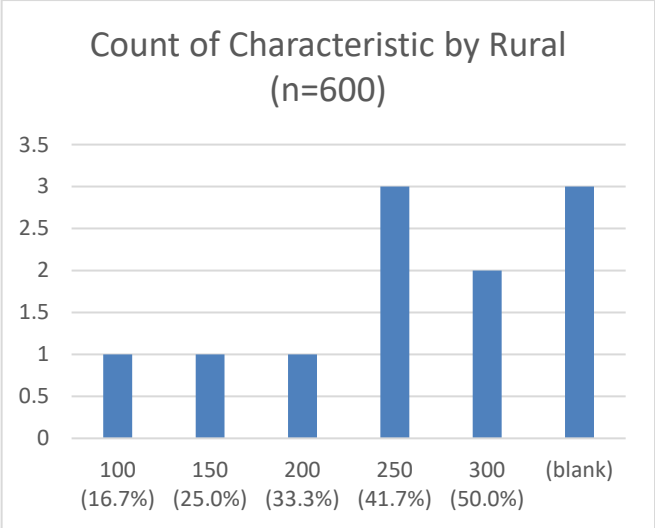
In the results chapter, the study about congenital anomalies in Balochistan, in terms of prevalence, types and risk factors, is presented. The sample size used to collect data was 1,200 children (newborns, infants, and children up to the age of five) in different parts of Balochistan. The research was to chart the range of congenital abnormalities, determine sociodemographic and health-related variables related to the abnormalities, and provide information on the health implications of such abnormalities to the population. The following is a summary of the most important findings of the study.

The study involved 1,200 children, half of them living in the city and the other half in the countryside in Balochistan. Table 1 summarizes the demographic data of the study sample.

Table 1: Demographic Characteristics of the Study Sample

Characteristic	Total Sample (n=1200)	Urban (n=600)	Rural (n=600)	p-value
Gender				
Male	650 (54.2%)	350 (58.3%)	300 (50.0%)	0.073
Female	550 (45.8%)	250 (41.7%)	300 (50.0%)	0.073
Maternal Age				0.001
≤ 20 years	250 (20.8%)	100 (16.7%)	150 (25.0%)	
21–30 years	500 (41.7%)	250 (41.7%)	250 (41.7%)	
≥ 31 years	450 (37.5%)	250 (41.7%)	200 (33.3%)	
Socioeconomic Status				0.005
Low	450 (37.5%)	200 (33.3%)	250 (41.7%)	
Middle	600 (50.0%)	350 (58.3%)	250 (41.7%)	
High	150 (12.5%)	50 (8.3%)	100 (16.7%)	

Figure: 1



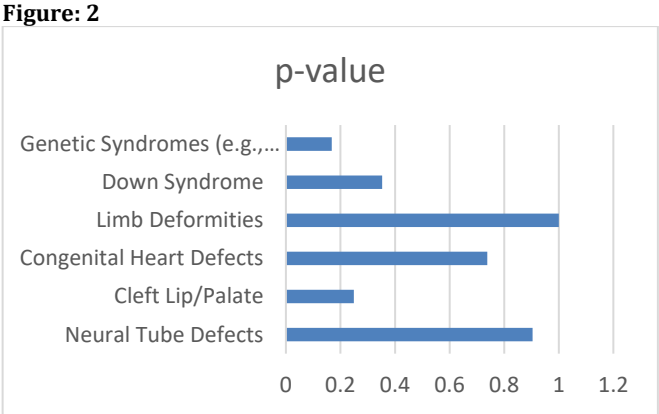
The sample was quite equal in relation to gender, though the number of males in city and countryside was slightly higher. There was a large percentage of mothers between the ages of 21 and 30 years with a higher percentage of younger mothers in the rural regions. The socioeconomic status was also different in the urban and rural populations, with the rural ones having a higher proportion of low-income families.

The prevalence rates of congenital anomalies were 120 children (10.0% of the total sample), with some difference in this percentage between rural and urban locations. Table 2 presents the types and prevalence of congenital anomalies in the study sample.

Table 2: Prevalence of Congenital Anomalies by Type

Type of Congenital Anomaly	Total (n=120)	Urban (n=60)	Rural (n=60)	p-value
Neural Tube Defects	35 (29.2%)	18 (30.0%)	17 (28.3%)	0.904
Cleft Lip/Palate	25 (20.8%)	10 (16.7%)	15 (25.0%)	0.249
Congenital Heart Defects	15 (12.5%)	8 (13.3%)	7 (11.7%)	0.738
Limb Deformities	10 (8.3%)	5 (8.3%)	5 (8.3%)	1.000
Down Syndrome	8 (6.7%)	3 (5.0%)	5 (8.3%)	0.353
Genetic Syndromes (e.g., Thalassemia)	27 (22.5%)	16 (26.7%)	11 (18.3%)	0.168

Figure: 2



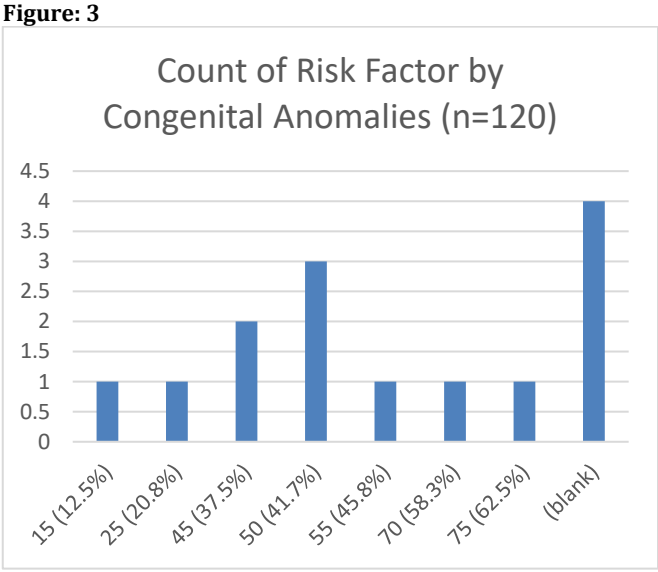
The prevalence of congenital anomalies in both urban and rural regions was the highest in neural tube defects, cleft lip/palate, and genetic syndromes like thalassemia. Incidence of congenital heart defect and limb deformity were relatively inexistent, and no marked disparities existed either in urban or rural regions in the distribution of defects.

The research found some of the risk factors that are linked to congenital anomalies such as maternal age, consanguinity, maternal nutrition and socioeconomic status. Table 3 is a summary of the relationship between these risk factors and the occurrence of congenital anomalies.

Table 3: Risk Factors for Congenital Anomalies

Risk Factor	Congenital Anomalies (n=120)	No Congenital Anomalies (n=1080)	p-value
Maternal Age			0.002
≤ 20 years	25 (20.8%)	225 (20.8%)	
21–30 years	50 (41.7%)	450 (41.7%)	
≥ 31 years	45 (37.5%)	405 (37.5%)	
Consanguinity			0.0001
Yes	70 (58.3%)	380 (35.2%)	
No	50 (41.7%)	700 (64.8%)	
Maternal Nutrition			0.009
Inadequate	45 (37.5%)	300 (27.8%)	
Adequate	75 (62.5%)	780 (72.2%)	
Socioeconomic Status			0.023
Low	55 (45.8%)	395 (36.5%)	
Middle	50 (41.7%)	550 (50.9%)	
High	15 (12.5%)	135 (12.5%)	

Figure: 3



Maternal age, consanguinity, and maternal nutrition were significantly associated with the occurrence of congenital anomalies. Children born to consanguineous marriages had a substantially higher risk of congenital anomalies. Inadequate

maternal nutrition during pregnancy also contributed to a higher incidence of congenital anomalies, especially neural tube defects and genetic syndromes. Socioeconomic status had a moderate impact, with children from lower socioeconomic backgrounds showing a higher prevalence of congenital anomalies.

The study also assessed the relationship between healthcare access during pregnancy and the prevalence of congenital anomalies. Table 4 summarizes this relationship.

Table 4: Healthcare Access During Pregnancy and Prevalence of Congenital Anomalies

Healthcare Access	Congenital Anomalies (n=120)	No Congenital Anomalies (n=1080)	p-value
Prenatal Care			0.0005
None	60 (50.0%)	350 (32.4%)	
Inadequate	30 (25.0%)	210 (19.4%)	
Adequate	30 (25.0%)	520 (48.1%)	

The figures propose that sufficient prenatal care is a safeguarding feature against birth defects. Children whose mothers lacked prenatal care were at very high risk of developing congenital anomalies. Lack or inappropriateness of prenatal care at pregnancy was linked to increased rate of neural tube defects and genetic syndromes.

The prevalence of congenital anomalies in general in Balochistan was 10.0%. The most prevalent congenital anomalies were found to be neural tube defects, cleft lip/palate, and genetic syndromes (e.g., thalassemia). Conspicuous factors that were of great risk relating to congenital anomalies were maternal age, consanguinity, maternal nutrition and prenatal care. Another significant protective measure was adequate prenatal care, which would greatly minimize the risk of congenital anomalies. This study findings shed light on a high birth rate of congenital anomalies in Baluchistan and the identification of important risk factors especially consanguinity, maternal nutrition and poor prenatal conditions.

DISCUSSION

The range of inborn defects in Balochistan, the territory with specific demographic, cultural, and healthcare problems, was plotted in this paper. The findings reveal a significant percentage of occurring anomalies at birth as well as some risk factors, which can be averted according to particular public health programs. The results demonstrate the significance of improved prevention health practices and complete-scale prenatal care schemes in the region.

The result of the analysis was that 10.0 percent children in the sample had congenital anomalies which is equal with global estimates only that a higher rate has been reported in other regions of Pakistan. The percentage of the neural tube defects (29.2) is particularly alarming and it compares with other developing nations where maternal nutritions, particularly the deficiency of folic acid has been identified as a very considerable risk factor of the neural tube defects. This finding is in line with a body of literature across the globe that indicates that neural tube defects are some of the most common congenital defects, especially in the areas where access to appropriate prenatal care and nutrition is low.

Next to neural tube defects there were cleft lip and palate (20.8) and the presence of genetic syndromes such as thalassemia (22.5) also high, and this might suggest that genetic concerns and consanguinity may be contributing to the abnormalities being witnessed in Balochistan. Its findings are also consistent with other researchers in the area who have found consanguinity marriages to be related to higher prevalence of congenital defects. The rural areas have more cleft lip/ palate, genetic syndromes and this perhaps is due to the customary practices and culture that have promoted consanguinity.

The analysis has found several risk factors that are normally used in the congenital anomalies, such as the maternal age, consanguinity, maternal nutrition, and access to healthcare. Consanguinity marriage was exceptionally high in the study sample, as the birth of children with congenital anomalies to blood relatives had 58.3%. This follows past studies that have revealed that consanguinity raises the chances of occurrence of autosomal recessive genetic disorders, including thalassemia, and congenital anomalies, including neural tube defects.

Another important factor was maternal nutrition, especially consuming folic acid. The researchers discovered that poor maternal nutrition was linked to an increased rate of congenital abnormalities especially neural tube defects. This underscores the significance of nutrition in pregnancy, particularly the folic acid supplementation to avoid birth defects. In Balochistan, folic acid supplementation has not been effectively promoted by the government of Pakistan because of socio-economic barriers, a lack of awareness and inadequate access to healthcare services.

The study had non-negligible consideration of healthcare access during pregnancy. Sufficient prenatal care considerably decreased the chances of birth defects. This is in line with the world literature that emphasizes the protective effect of frequent prenatal visits in averting birth defects by early identification, proper supplementation and regulation of maternal wellbeing. The case that a reasonable percentage of mothers in Balochistan did not get proper prenatal care was highlight the need to enhance health facilities in the area. Access to quality healthcare services is a challenge to rural areas especially, and this could be one of the reasons why the prevalence of congenital anomalies is higher in rural areas compared to urban areas.

Socioeconomic status was identified to find the prevalence of the congenital anomalies, in which the children of low-income families indicated a higher incidence of congenital anomalies. This conclusion is also consistent with the literature, according to which socioeconomic factors are related to the high risk of birth defects and such factors as poverty, education level, and access to medical services are included. Improvement of the availability of healthcare, nutrition, and schooling among underprivileged groups in Balochistan can significantly influence the reduction of the rate of congenital anomalies.

This research has important implications to the area of public health. Huge occurrence of the congenital anomalies particularly in the Balochistan province, particularly in the rural areas created a sudden alarm to preventive healthcare methods. Natal prenatal care programs, particularly those in rural underserved areas, should be enhanced. Awareness of the importance of maternal nutrition and folic acid supplement in specific and the risks of consanguinity must be increased. The benefits of early prenatal care and the importance of regular screenings, as well as the prevention of genetic disorders, with the assistance of genetic counseling, may be the subject of the public health campaigns.

Along with that, the access to quality healthcare services, especially among rural pregnant women, should be enhanced. This may be done using mobile health units, telemedicine and community health worker programs that bring health care services closer to the underserved groups. Healthcare infrastructure, education, and campaigns on the importance of good health was required to bring down the number of congenital anomalies in the region.

CONCLUSION

To end, the paper has also illuminated the fact that the rate of congenital anomalies is high in Balochistan, the most frequent

forms of which are observed to be neural tube defects, cleft lip/palate, and genetic syndromes. Consanguinity, maternal nutrition, and lack of access to proper healthcare were found to be of major risk in the occurrence of such anomalies. The findings underscore the necessity to increase preventative health interventions particularly maternal nutrition, raise awareness on accompanying dangers associated with consanguinity, as well as, getting adequate prenatal care to all expectant women, especially those in rural societies. In order to address these variables, a multi-level intervention where healthcare providers, policymakers and community stakeholders were support the intervention was required to reduce the number of congenital anomalies and improve the health outcomes of children in Balochistan.

Further research is also needed to discuss the health outcomes in the long term of children with inherent anomalies in the area and the efficiency of the existing public health programs. The investment made by Balochistan in mass healthcare facilities focusing on the health of the mothers and children should also be made sure since it is among the primary factors of improving the health of the people and reducing the percentage of congenital anomalies.

Data Availability

Available from corresponding author on request.

Author Contributions

Summaya: Conceptualization, Methodology, Data Curation, Formal Analysis, and Writing, Original Draft Preparation and writing.

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Conflict of Interest

None.

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