ORIGIONAL ARTICLE

PREVALENCE, OUTCOME AND RISK FACTORS ASSOCIATED WITH CONGENITAL MALFORMATIONS IN NEONATES AT A TERTIARY CARE HOSPITAL -A RETROSPECTIVE STUDY.

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ABSTRACT

INTRODUCTION: There is a high burden of congenital anomalies worldwide and more so in the developing countries. These birth defects contribute significantly to the neonatal mortality rate in our country, which is one of the highest globally. Addressing and identifying the risk factors for these conditions is imperative for their prevention .**OBJECTIVE:** this study was done to determine the prevalence of congenital malformations and the risk factors associated with them. **METHODOLOGY:** this cross-sectional retrospective study was carried out at the Pediatric unit of Ziauddin Hospital from 1st January 2020 till 1st January 2023.Data was extracted from the electronic health records and entered on a pre-designed proforma. Information regarding maternal risk factors , antenatal visits ,neonatal characteristics and type of congenital anomalies was noted .Data was analyzed using SPSS version 20 and expressed as frequencies and percentages and chi square test was applied with P-value of <0.05 considered statistically significant. **RESULTS :**The prevalence of congenital malformations was 4.5%.Anomalies related to the central nervous system were the highest ,followed by cardiac birth defects .A smaller percentage undertook regular antenatal visits and folic acid supplementation. Family history of birth defects is a significant risk factor. **CONCLUSION:** Congenital malformations are a frequent occurrence in our set up .Effective antenatal care and screening are the cornerstone of preventing these disorders.

Key words: Congenital Anomalies; Birth Defects; Risk Factors; Prevalence; Malformations; Neonates; Mortality.

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INTRODUCTION

Birth defects are a prime reason of illness and death particularly in decreased income communities. Research information in this respect is nonetheless not gathered properly in such areas. Though perinatal impairment in oxygenation, decreased gestational age and septicemia mainly cause of adverse consequences, birth deformities are responsible for major ill health and expiries in newborns.[1] The neonatal mortality world-wide substantially multiplied as a result of these birth defects. Sadly about 90% of birth defects arise in developing countries with low-resource settings.[2, 3] Major congenital anomalies are described as anomalies with an immense impact on existence, life span and that they arise in 2–3% of still- births and in 20–30% of live babies'. However, their occurrence changes periodically and regionally showing a complicated interplay among inborn and ecological factors.[4] Understanding the underlying causes of congenital anomalies is essential in avoiding them and in guidance related to inherited disorders which could assist decreasing their prevalence. Predisposing elements can be genetic (10–30%), environmental (5–10%) or these malformations could be as a result of an inheritance that involves multiple (20–35%) factors but in 30–45% the cause is undetermined. [5, 6] generally, the etiology of congenital malformations is multifactorial. In developing countries, infectious diseases seem to be responsible for a huge proportion of these cases. Implicated maternal elements consist of age, lifestyle, and ailments for the duration of pregnancy, improper antenatal care, side-effects of drugs used during pregnancy and not using folic acid prior to conceiving. [7] Parental consanguinity, preceding miscarriages and stillbirths, and inheritable congenital ailment are different essential eti ological elements.[8] Despite the massive burden of congenital anomalies in the developing part of the world, there is nonetheless a scarcity of research data on

the prevalence and etiology of birth defects as well as a lack of congenital malformation registries.[1] There is massive under-estimation of these conditions in developing countries because of not seeking medical care at health facilities, and inadequate ability to identify these disorders. Research studies on the prevalence of birth defects are conducted to set up baseline rates, exhibit adjustments that arise over the years and deliver clues to etiology. Furthermore, there is considerable variance in the kinds and occurrence of birth defects across geographical areas due to dissimilarities in the way of living, ethic and social elements. In addition, there is a dearth of research data with regards to the prevalence of congenital malformations in Pakistan as there is no national registry and surveillance in this regard. [9] This research study though hospital-based, was conducted at one of the biggest private sector hospitals of the country .It aimed to assess perinatal and maternal elements that can be related to the prevalence and outcome of these congenital anomalies in the local context. The observations made by this study will thus, help to identify makers for monitoring and surveillance of these congenital defects as we lack a national surveillance system in this regard. We therefore, intended to evaluate the frequency, predisposing elements and result of congenital anomalies amongst neonates delivered in a tertiary care facility as a number of risk factors if addressed could lead to a decrease in the frequency of these conditions in the developing countries like Pakistan. [10]

Operational definition

Congenital malformation is defined as anatomical or functional abnormality detected in the organ-system externally or internally was defined as a CM based on WHO definition.[11]

METHODOLOGY

This retrospective study was conducted at Unit of Ziauddin University hospitals Karachi .All birth records of neonates delivered with one or more congenital malformation during the last three years i.e. from 1st January 2020 till 1st January 2023 were reviewed. The data was collected from electronic medical record and entered on a structured pre-designed proforma adapted from previous literature. Data was recorded the proforma by two postgraduate residents. The inclusion criteria comprised of records of all live born neonates diagnosed with at least one birth defect an admitted in the neonatal intensive care unit of Ziauddin hospital. All records with incomplete information (those record that miss more than two variables) and the records of still births were excluded from the study. Measures were taken to ensure the quality and confidentiality of collected data by using a coding system. Variables such as number of birth defects ,types of congenital anomaly, parental consanguinity ,mode of delivery , whether a still birth or live birth , family history and factors associated with congenital anomaly (i.e. maternal factors and infant factors) were recorded .The proforma was pretested before actual study and necessary changes were made to maximize the reliability of data. Finally, the data was examined for completeness by the

principal investigator. Data was analyzed using SPSS version 20. Qualitative data was expressed as mean and standard deviation, quantitative data as frequencies and percentages and chi-square was applied for categorical variables. To adjust for a large number of outcome variables p-value <0.05.was taken as significant.

RESULTS

There were 2520 deliveries during the study period, of these 94 live births had congenital anomalies. Out of these 54 were males and 38 females while in 2 the gender was ambiguous. Majority of these neonates i.e.43 (45.7%) were low birth weight while 31(32.9%) were very low birth weight i.e. <1500 grams. Most of the neonates 65 (69.1%) were delivered via Lower segment caesarian section (LSCS) and 29 (30.9%) by spontaneous vaginal delivery (SVD). Table 1 shows characteristics of neonates with congenital anomalies .A large proportion of the neonates 68(72.3%) with malformations were preterm.

Table 1. **Characteristics of neonates with congenital anomalies**

(Total N = 94)

		(N %)
Gender	male	54 (57.4)
	female	38 (40.5)
	ambiguous	2 (2.1)
Birth weight (grams)	<1000	5 (5.3)
	1100-1500	26 (27.7)
	1600-2000	21 (22.3)
	2100-2500	23 (24.5)
	>2500	19 (20.2)
Gestational Age (weeks)	<28	6 (6.4)
	29-32	28 (29.8)
	33-36	34 (36.2)
	39-40	18 (19.1)
	>40	8 (8.5)
Outcome	expired	19 (20.2)
	discharged	69 (73.4)
	Transferred to other facility	6 (6.4)

Distribution of anomalies with regards to body system involved is shown in table 2 .

Malformations related to the nervous system had the highest prevalence 26 (27.6%) ,followed by cardiac 24 (25.5%) and gastrointestinal 18(19.1%) .

Table 2.   Distribution of congenital anomalies according to body system

SYSTEM	ANOMALIES	N (%)
Central Nervous System	Meningomyelocele	7 (7.4)
	Hydrocephalus	6 (6.4)
	Encephalocele	2 (2.1)
	Anencephaly	4 (4.3)
	Microcephaly	7 (7.4)
Cardiovascular system	Complex CHD	2 (2.1)
	Cyanotic heart disease	3 (3.2)
	PDA	8 (8.5)
	VSD	6 (6.4)
	Hypo plastic left heart	2 (2.1)
	ASD	1 (1.1)
	Others	2 (2.1)
Gastrointestinal system	Cleft lip	2 (2.1)
	Cleft Palate	3 (3.2)
	ankyloglossia	2 (2.1)
	omphalocele	2 (2.1)
	duodenal atresia	3 (3.2)
	imperforate anus	2 (2.1)
	TEF	4 (4.3)
Genitourinary system	polycystic kidney	1 (1.1)
	Ambiguous genitalia	2 (2.1)
	Hydronephrosis	5 (5.3)
	Hypospadias	2 (2.1)
	Renal agenesis	2 (2.1)
Respiratory system	Renal Hypoplasia	3 (3.2)
	Diaphragmatic Hernia	4 (4.3)
	Choanal atresia	2 (2.1)
	Lung hypoplasia	3 (3.1)
Musculoskeletal system	Polydactyly	3 (3.1)
	Vertebral anomalies	5 (5.3)
	talipes equinovarus	6 (6.4)
Skin	Hemangioma	4 (4.3)
	Cutis Aplasia	2 (2.1)
	Ichthyosis	3 (3.2)
	cystic hygroma	2 (2.1)
	Others	2 (2.1)
Cardiovascular system	Down Syndrome	5 (5.3)
	Trisomy 18	2 (2.1)
	Turner Syndrome	1 (1.1)
	TAR Syndrome	1 (1.1)

CHD= Congenital Heart disease; PDA= patent ductus arteriosus;

VSD= ventricular septal defect; ASD= Atrial Septal defect;

TEF= Tracheoesophageal fistula.   TAR syndrome =Thrombocytopenia Absent Radius syndrome

Table 3 shows maternal characteristics of neonates with congenital anomalies. Maternal age of greater than 40 years was present in 29(30.9%) and multiparty was a risk factor present in majority 62(66%) of these births. History of consanguinity was found in a large number 61(64.9%). Majority of the mothers 87(92.6%) were educated but only 57(60.6%) underwent regular antenatal care .Folic acid supplementation was present in just 38(40.4%). In 18 (19.1%) there was a previous child with congenital malformation and in 7 (7.4%) there were two or more previous children with birth defects .Gestational Diabetes and maternal hypertension as risk factors were present in 29(30.9%)and 23(24.5%) respectively .

Table. 3. Distribution of Maternal risk factors among study population (n = 94)

		N (%)
Maternal Age (Years)	<20	4 (4.3)
	20-25	12 (12.7)
	26-30	21 (22.3)
	31-35	28 (29.8)
	>40	29 (30.9)
Parity	Primigravida	32 (34)
	Multiparous	62 (66)
Maternal Education	No Education	2 (2.1)
	Undergraduate	5 (5.3)
	Graduate	65 (69.2)
	Post Graduate	22 (23.4)
Working Mother	Yes	38 (40.4)
	No	56 (59.6)
Consanguinity	Yes	61 (64.9)
	No	33 (35.1)
Folic Acid Intake	Yes	38 (40.4)
	No	56 (59.6)
IVF Conception	Yes	19 (20.2)
	No	75 (79.8)
Regular Antenatal Visits	Yes	57 (60.6)
	No	37 (39.4)
	Diabetes	29 (30.9)

Maternal Co-morbid	Hypertension	23 (24.5)
	Hypothyroidism	4 (4.3)
	TORCH Positive	6 (6.4)
	Anemia	17 (18.1)
Birth Defect in previous pregnancies	One	18 (19.1)
	Two Or More	7 (7.4)
Mode of Delivery	LSCS	65 (69.1)
	SVD	29 (30.9)

LSCS =lower segment caesarian section; SVD=spontaneous vaginal delivery;

TORCH=Toxoplasmosis others Rubella, Cytomegalovirus, Herpes; IVF =in vitro fertilization

DISCUSSION

Congenital anomalies have a high global occurrence, affecting 3% to 5% of live births. They contribute 5.5% to mortality in under 5-year-old Pakistani pediatric population. [12] The prevalence of birth defects in our population has been reported to be as high as 7 % by some local researchers .[13] We however, observed a prevalence rate of 4.5 % which is similar to the results reported by Gillani et al .[8] In a study from Peshawar Khan et al observed a prevalence rate of 2.9% which is lower than observed by us. [14] Silesh et al however , have reported a prevalence rate of 5.95% in Ethiopia [15] whereas in Iran[16] it has been reported as 2.3%.Research data from India has reported the prevalence of congenital anomalies in neonates as 1.9%. The variation in prevalence rates may be due to different ethnicities, genetic, socio-demographic and regional factors and is due to a complex interplay of these elements .[17] In our study, anomalies related to the nervous system had the highest prevalence (27.6%) followed by cardiac malformations (25.5%). Agadoorappa et al [18] in their study

found a 2.3% prevalence of congenital defects in babies of Pakistani ethnicity and out of these 30% were heart defects .Similar to our findings , Shabbir et al have reported neurological birth defects to be the commonest in their study [19] while Rafi et al in their study reported gastrointestinal anomalies to have the highest occurrence[20] similar to the findings of Shamim et al in their study at a tertiary care facility of Karachi .[9] An epidemiological study on birth defects done in Sialkot, Pakistan, reported that limb defects were the commonest malformation with an occurrence of 47%. [21]A study by Sheikha et al from Bahrain have reported musculoskeletal defects to be the most prevalent anomalies observed in neonates .[22] Perveen et al observed that congenital defects were more common among primiparous mothers at a tertiary care center in Karachi.[10] They also reported a history of consanguinity to be the commonest risk factor for malformations being present in 44.74% of their study population .Butt et al have made similar observations and reported high prevalence of congenital anomalies in babies of primiparous .[23] We however, noted birth

defects to be more prevalent in multiparous mothers. Gestational Diabetes was the most prevalent comorbid maternal risk factor in our study, being present in 30.9% of the mothers. These findings are in contrast to the study observations of Qadir et al who observed only 2% of mothers with gestational diabetes to have delivered babies with birth defects .[24]This may be explained by the fact that their study population might have achieved a good glycemic control hence, minimal prevalence of congenital malformations . We observed that in a significant percentage (59.6%) of our study population intake of folic acid in preconception period was not present despite the fact that 92% of the mothers in our study population were well educated. A study from Khyber Pakhtunkhwa observed that regular antenatal care had been provided to a mere 32.3% mothers who delivered babies with birth defects received antenatal care.[25] In a number of studies[8, 9, 26] family history of birth defects was noted to be a significant risk factor for congenital defects, our study findings are in concert with their observations, as we found that in 26.5% of our study population a positive family history of birth anomalies was present. This finding highlights the need for early detection of birth defects through prenatal screening in all cases where the especially the family history is significant .The main emphasis should be on early recognition , fetal echocardiography in cases of positive family history ,antenatal counseling and public awareness campaigns focusing on the importance of preconception folic acid supplementation and regular antenatal check-ups .

CONCLUSION

There is a high prevalence of congenital anomalies in Pakistan. Neurological and cardiac malformations are the commonest among all birth defects .Thus, the need for improving antenatal care, preconception folic acid use and early antenatal screening are imperative.

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