Original Research Article

Pattern of Congenital Malformations and Associated Risk Factors During Pregnancy at A Tertiary Care Centre

Krishna Patel[1], Shabdika Kulshreshtha[2], Anupama Hada[3], Triguna Rupalal[4]

[1]Third Year Resident, Department of Gynaecology and Obstetrics, Geetanjali Medical College and Hospital, Udaipur, Rajasthan

[2]Associate Professor, Department of Gynaecology and Obstetrics, Geetanjali Medical College and Hospital, Udaipur, Rajasthan

[3]Assistant professor, Geetanjali Medical College and Hospital, Department of Gynaecology and Obstetrics, Udaipur, Rajasthan

[4]Third year Resident, Department of Gynaecology and Obstetrics, Geetanjali Medical College and Hospital, Udaipur, Rajasthan

Abstract

Background: Congenital malformations include structural or functional anomalies that occur during intrauterine development and represent a major cause of perinatal morbidity and mortality.

Aim: To identify and study the maternal risk factors, the various patterns of congenital anomalies in the fetus of $\geq$ 16 weeks of gestation by USG and in neonates.

Methods: A prospective follow-up study was conducted at Geetanjali Medical College and Hospital, Udaipur from Jan 2020 to June 2021. 53 pregnant patients after 16 weeks of gestation with anomalous foetus and malformed neonate detected at the time of birth were included in the study. They all underwent a fetal anomaly scan at 18-20 weeks of gestation.

Results: 43.3% patients were above 30 years of age. Maternal exposure to radiation, smoking, drugs and alcohol was seen in 32.3 % patients giving birth to CM neonate. CNS was the most common system involved in 34% CMs. More than half of the patients (56.6%) delivered at less than 28 weeks of gestation. Majority of the neonates (67.6%) expired due to fatal congenital malformations of CNS/ CVS. All the live neonates were admitted in NICU.

Conclusion: This study explores the possible causes and effects of congenital malformations. The result of this study supports that history of consanguinity is the most important risk factor seen in more than half of the patients. The booked antenatal cases have lesser chances of carrying a CM fetus till the term.

Keywords: congenital malformations, associated risk factors, pregnancy, tertiary care centres.

Introduction

According to the World Health Organization (WHO), congenital anomalies, congenital disorders or congenital malformations (CMs) can be defined as structural or functional anomalies (e.g. metabolic disorders) that occur during intrauterine life which are identified prenatally, at birth or postnatally. CM may be minor or major. The minor malformation is a structural abnormality present at birth with minimal clinical effect but may have a cosmetic effect, for example preauricular tag. Major malformations have a significant effect on function or on social acceptability, for example, ventricular septal defect and cleft lip[1]. About 20–30% of infant deaths and 30–50% post-neonatal deaths are attributed to CMs. The first trimester, especially between the 3rd and 8th weeks of gestation, is the crucial period for morphogenesis of organs. Any insult during this period such as teratogenic drugs, alcohol abuse, radiation exposure, certain infections, micro-nutrient insufficiency can cause congenital abnormality. Ultrasound is the best possible non invasive technique available to detect any CM in pregnant women which will help to identify to pregnancy termination or provide an opportunity for fetal therapy or better neonatal care. This is the period where preventive intervention strategy will reduce the incidence of developing CMs[2].

Causes and risk factors include:

- Genetic factors
- Socioeconomic and demographic factors
- Environmental factors
- Infections
- Maternal nutritional status

According to the March of Dimes global reports on birth defects, worldwide at least 7.9 million people are born with a birth defect every year, and of those, at least 3.3 million babies die every year before the age of 5 years[3]. CMs continue to haunt every obstetrician. Therefore, this study explores the possible causes and effects of congenital malformations. Being a developing country, analysing the demographic factors, socio-economic factors, genetic factors and feto maternal outcome in congenitally malformed neonates should be conducted to reduce the neonatal morbidity and mortality. Some congenital anomalies are preventable through vaccination, adequate intake of folic acid and fortification of staple foods by iodine, food supplementation, and adequate antenatal care[4].

Material and Method

After obtaining approval from Institutional Research Ethical board and patient's written informed consent, this study was conducted at Geetanjali Medical College and Hospital Udaipur from Jan 2020 to June 2021. 53 pregnant patients after 16 weeks of gestation with anomalous foetus and malformed neonate detected at the time of birth were included in the study. They all underwent a fetal anomaly scan at 18-20 weeks of gestation. All the live, still birth, intra uterine dead foetus were included in this study. Fetal outcome was assessed by obstetrician and paediatrician for details of malformations. A detailed, structured performa including following details like maternal age, parity, consanguinity, abortions, siblings with malformations, nutrition, smoking, alcoholism, family history of congenital malformations, conception after artificial reproductive techniques, maternal diabetes, infections, fever, history of teratogenic drugs intake were recorded and analysed. All the data collected was statistically analysed and distribution percentage was calculated.

Results

Out of 53 patients 46 (85%) were above 25 years of age. More than half (approximately 31) i.e. 58%, had history of consanguineous

marriage. 11 (20.8%) had personal history of drug use and 3 (5.7%) each, had personal history of smoking and alcohol. 7 (13.2%) had previous history of congenital malformations, most of them 45 (84%) consumed folic acid tablets only.Among them 7(13%) had history of infections & history of fever in firsts trimester. 18% of them received infertility treatment, 8 (15%) had uterine anomaly & 10 (18%) had a history of IUD/abortion. 18 (34%) had CNS involvement, 11 (20.8%) had CVS system involvement, 10 (18.9%) had GIT involvement, 5 (9.3%) had involvement of urogenital system & in 1 (1.9%) respiratory system was involved.

All the surviving neonates (17) required NICU admission, while 7 (41%) were operated and 36 (67.9%) expired. 34 (64.1%) were male and 19 (35.9) were female. 41 (77.4 %) neonates were low birth weight (with less than 2.5kg), while remaining 12 (22.6%) weighed more than 2.5 kg. Among them 30 neonates (56.60%) delivered at less than 28 weeks of gestation (WOG), 5 neonates (9.43%) delivered in between 28 to 32 weeks while 18 neonates (33.96%) delivered in between 32 to 37 weeks.

Table1: Personal history wise distribution of study participants.

Personal history	Number of patients	Percentage (%)
Radiation	0	0.0
Smoking	3	5.7
Alcohol	3	5.7
Drug	11	20.8
Total	17	32.2

Table 2: Distribution of study subjects according to obstetric ultrasound

Presentation		No. of patients	Percentage (%)
Breech		10	18.9
Cephalic		43	81.1
AFI (Amniotic Fluid Index)			
Normal		43	81.1
Oligohydramnios		4	7.5
Polyhydramnios		6	11.3
System involved	**Type Of Anomaly**		
CNS	**Acrania**	1	1.9
	Arnold Chiari Malformation	1	1.9
	Choroid Plexus Cyst	1	1.9
	Dandy Walker Malformation	1	1.9
	Dorsolumbar Meningoencephalocele	1	1.9
	Encephalocele	2	3.8
	Anencephaly	1	1.9
	Hydrocephalus	3	5.7

	Macrocephaly	1	1.9
	Meningocele	2	3.8
	Microcephaly	1	1.9
	Occipital Encephalocele	1	1.9
	Spina Bifida	2	3.8
CVS	CHD (congenital heart disease)	8	15.1
	VSD (Ventricular septal defect)	1	1.9
	ASD (Atrial septal defect)	1	1.9
	Fetal Cardiomegaly	1	1.9
GIT	Atresia	1	1.9
	Cleft lip	2	3.8
	Fetal Hydrops	1	1.9
	Mesenteric Cyst	1	1.9
	Diaphragmatic Hernia	3	5.7
	Omphalocele	2	3.8
Musculoskeletal system	Achondroplasia	2	3.8
	Club foot	2	3.8
	CTEV	1	1.9
	Radial Anomaly	1	1.9
	Rocker Bottom Feet	1	1.9
	Short Bone	1	1.9
GUT	Hypospasdias	1	1.9
	Bladder Absent	1	1.9
	Kidney absents	1	1.9
	Hydronephrosis	2	3.8
Respiratory System	CCAM (congenital cystic adenomatoid malformation)	1	1.9

Table 3: Distribution of study subjects according to the systemic involvement

Systemic involvement	No. Of patients	%
CVS	11	20.8
CNS	18	34.0
GIT	10	18.9
Respiratory	1	1.9
Urogenital	5	9.3
MSK	8	15.1

Table 4: Distribution of study subjects according to the neonatal outcome.

Neonatal Outcome	No. Of patients	%
Alive neonates	17	32.1
Neonates operated	7	41
IUD/Stillbirth	36	67.9

NICU admission	17	100

Discussion:

In present study, 35.8% study participants were between 30 -35 years of age while 7.5% of the participants were above 35 years. While the study conducted by Rabah M. Shawky et al[5] among CMs in Egyptian children found that 59.96% mother were above 35 years at conception. Hollier LM et al[7]. found in their study that maternal age is an important parameter in the birth of a congenitally malformed fetus.

In present study more than half i.e. 58.5% of the patients had the history of consanguineous marriage. While the study conducted by Rabah M. Shawky et al.[5] among Egyptian children found that Consanguineous marriage was present in 45.8% of parents of patients while study conducted by Rizk Francine[6] from Lebanon found that a significant positive association exists between the consanguinity and CA in the Lebanese population.

18% patients in the present study (18.9%) had a family history of congenital malformations while Awad Elkhadir et al[8] in their study found that family history of CMs was reported to occur in 12.8 % and it was considered statistically significant.

In the present study, 3 of the antenatal patients (5.7%) had a history of smoking and alcohol respectively. 11 patients (20.8%) gave a personal history of drug consumption. While in a study done by Rizk Francine et al[6] found that history of alcohol and smoking was present in 2.2% and 3.2% patients respectively. The history of drug intake was 6.9%. In the study by Rabah M Shawky et al[5] observed that history of active and passive smoking was 55.44% while drug consumption was seen in 36.32%. In a study by Ameen et al[9], history of smoking was found to be present in 12.3%, compared with 3.8% in the control group.

In our study we found that among 53 patients, 7 (13.2%) were having previous history of congenital malformations, most of them 45 (84.9%) were consuming folic acid tablets while Rabah M. Shawky et al[5] in their study found that 27.5% of mothers received folic acid or multivitamin which is significantly lower than that in the control group. In our study we found that only 7 (13%) were having infections & history of fever in 1st trimester. 18.9% of them were receiving infertility treatment. Only 8 (15%) were having uterine anomaly & 10 (18.9%) were having history of intra uterine death (IUD)/abortion. The study conducted by Mamatha Shivanagappa et al[10] noted history of previous abortions in 27.7%, consanguinity in 10.6%, Rh negative pregnancy in 6.4% and anomalous uterus in 6.4% of the mothers with CM neonates.

In the present study, 4 of the antenatal patients (7.5%) had history of oligohydramnios and 6 patients (11.3%) had history of polyhydramnios. In the study done by Rabah M Shawky et al[5] found that oligohydramnios (9.8%) and polyhydramnios (10.8%) were present in high frequency. Stoll et al found that 55% of cases with polyhydramnios had more than one malformation, 13.4% of them had chromosomal aberration and 32% had multiple malformation.

In the present study, among them 30 neonates (56.60%) delivered at less than 28 weeks of gestation, 5 neonates (9.43%) delivered in between 28 to 32 weeks while 18 neonates (33.96%) delivered in between 32 to 37 weeks. According to Marwa Shawky Mohammed abdou et al[11] 10% neonate delivered at preterm, 86% delivered at full term and 4% delivered at post term.

In the present study, 41 (77.4 %) neonates were low birth weight (with less than 2.5kg), while remaining 12 (22.6%) weighed more than 2.5 kg. A study conducted by Ashish Marwah et al[12] found a higher incidence of congenital malformations in low-birth-weight babies (2.19%) as compared to babies with birth weight more than 2.5 kg (1.57%). A study conducted by Patel ZM and Adhia et al[13] found that 59.8% of babies were low birth weight (<2.5kg) and 40.2% of babies have weight more than 2. 5kg.

In the present study, out of a total of 53 neonates, 34 (64.1%) were male and 19 (35.9%) were female. A study by Samira T. Abdulghani et al[14] observed that 58% of the neonates were male, 42% were female and 2% were born with ambiguous genitalia. Likewise, in a study by R Padmanabhaan et al[15] 54% were male babies and 45% were female babies. According to a study published by Ashish Marwah et al[12] males (2%) were affected more as compared to females (1.43%).

In the present study, according to which, all 17 surviving neonates required NICU admission, while 7 (41%) were operated and 36 (67.9%) expired. In the study by Samira T Abdulghani et al[14] 66.6% were malformed live birth, 24.2% were abortions and 9.2% were still birth.

Conclusion

The results of this study support that the history of consanguinity is the most important risk factor seen in more than half of the patients. The booked antenatal cases have lesser chances of carrying a CM fetus till the term. Maternal exposure to radiation, smoking, drugs and alcohol was seen in 32.3 %. In our study polyhydramnios and non-cephalic presentation was seen in 11.3% and 18.9% of the cases. All the live neonates were admitted in NICU as they required

intensive care support for managing their CMs in an optimal manner. Weakness of this study is that other common risk factors which are frequently seen (as described earlier) in congenitally malformed pregnancies are not found to be associated here. The sample size calculation and sampling population can be increased for this. But overall more studies and a larger amount of data is required to reach to a meaningful conclusion.

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