

A Study on Congenital Malformations in a Tertiary Care Medical College

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Abstract

Aim: This study was conducted to analyze the congenital malformations- their incidence, types, maternal socio-demographic profile and probable risk- factors. *Results:* our study showed that unwanted pregnancies, lack of antenatal checkups, no periconceptional folic acid intake, maternal diabetes mellitus, hypertensive disorders including Pre Eclampsia & Eclampsia, fever, maternal infections and drug intake were important factors associated with congenital malformations in the baby. *Conclusion:* Regular and proper antenatal checkups and care, periconceptional folic acid supplementation can significantly reduce and prevent the incidence of congenital malformations and there by decrease the mortality and morbidity associated with birth defects.

Keywords: Congenital Malformations; Birth Defects; Live Births; Morbidity; Mortality.

Abbreviations:

CMF- Congenital malformations
ANC- Ante-natal care
NICU- Neonatal intensive care unit
NTD- Neural tube defects

Introduction

Pregnancy and child birth is one of the most pleasant and anticipated

journey for an expected couple more so for the would-be mother but when a baby is born with congenital malformations, the parents are devastated and feels helpless, hopeless and betrayed. One cannot imagine the physiological, physical, social, mental and financial burden for the parents, family and also the child. A congenital malformation is defined as any structural or functional abnormality, including metabolic disorders which are present from birth [1].

World statistics reveal that 7-8% of all births globally result in the birth of congenitally malformed babies annually which end up as dead or a lifelong disability in them [2]. The total burden of such births comes to a whopping 9 million a year [2]. Among these, neural tube defects (NTDs) constitutes the commonest and most serious set of anomalies affecting approximately 3 lakh new born every year [1]. Though the exact etiology of congenital malformations are not always known nevertheless, many factors namely genetic, infectious, environmental factors have been implicated in there pathogenesis. Chromosomal abnormalities, single gene defects & multifactorial disorders are some of the genetic causes thought to be causing congenital malformations. Teratogens e.g. .drugs, substance abuse, toxins, alcohol and other constraints are the main non-genetic etiological factors associated with birth defects [1, 3].

Teratogenic factors are those fetal environmental factors which cause damage in the embryonic phase of the fetal development (4-10 weeks of intra-uterine life).many such factors are known e.g. Maternal infections (Toxoplasmosis, Rubella or German measles, CMV (cytomegalovirus) herpes simplex virus HSV) , and HIV/AIDS, (TORCH) maternal conditions e.g. Diabetes mellitus , epilepsy,

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maternal exposure to high -dose radiations, drugs and substance abuse, alcohol, tobacco, cocaine, retinoic acid, some antibiotics e.g. tetracycline, streptomycin, anticancer drugs e.g. methotrexate, thalidomide etc, anticoagulants e.g. warfarin, some antiepilepticse.g. sodium valproate, phenytoin, mood stabilizers' e.g. lithium, environmental teratogenic pollutants e.g. mercury are some of the known teratogenic agents, Many other risk factors associated with congenital malformations like maternal medical complication & smoking. This study was undertaken for a better understanding of the several possible-known and hethertho unknown- maternal factors which predispose to the occurrence of congenital malformations and this aid in formulating preventive measures thereof.

Material & Methods

- Study type: Hospital based , observational study.
- Study duration: Two years (Jan 2014- Jun 2016).
- This study was conducted in a tertiary care medical college in North India.

Inclusion Criteria

- All live-born infants

- Fetal deaths(>20 weeks)
- Pregnancy termination at any gestational age with birth defects

Exclusion Criteria

All live births, fetal deaths or abortions without any congenital malformation.

- All the patients, after fulfilling the above criteria – were subjected to informed, written consent-were included in the study. Cases were then identified so as to having isolated, multiple or complex congenital malformations. A pre-designed, standardized questionnaire was filled up following a thorough & detailed history taking from each patient so as to identify the factors possibly related to the congenital malformation in question.

All the relevant data thus collected were sorted, tabulated and statistically analyzed.

Results & Observations

In our study, a total of 75 congenital malformations were noted during the study period. The results and observations thereof are represented in a tabulated form as follows:

Table 1: Socio-demographic profile of the mothers

Factor		Number of Patients	Percentage%
Parity	Nulliparous	50	66.66
	multiparous	25	33.34
Age	<18	23	30.66
	18-34	46	61.33
	>35	16	21.31
ANC care	Unbooked	56	74.66
	Booked	19	25.34
Sex	Male	33	44.00
	Female	39	52.00
	Ambiguous	03	4.00
Birth weight	<2500	41	54.66
	>2500	34	45.34

75 (3 fetuses had ambiguous genitalia)

Table 2: Types of congenital anomalies

Congenital Malformation	Number of cases	Percentage (%)
Neural tube defects (NTD)	28	37.33
Anencephaly	8	
Spina bifida	2	
Encephalocele	2	
Meningocele	3	
Meningomyelocele	4	
hydrocephalus	6	
Holoprosencephaly	3	
Orofacial defects	8	20.05

Cleft lip + palate	3	
Cleft palate	2	
Cleft lip	3	
Skeletal defects	13	17.33
TelipesEquinoVarus	5	
Polydactyly	6	
Other limb defects	2	
Congenital heart anomalies	7	9.33
Congenital lung anomalies	5	6.66
Diaphragmatic hernia	4	5.33
Abdominal wall defects	4	5.33
Omphalocele /	2	
Gastrorchi-sis		
Genito-urinary anomalies	6	8.00
Renal agenesis	3	
Renal duplication	1	
Congenital PCKD	1	
Renal cysts (single)	1	

Table 3: Maternal socio-demographic factors

Maternal factors	Present		Absent	
	Number of patients	(%)	Number of patients	(%)
Booked	17	22.66	58	74.34
AN checkup	30	40.00	45	60.00
Unwanted pregnancies	15	20.00	60	80.00
DM	34	45.33	41	54.67
Hypertensive Disorders of Pregnancy (PE+E)	16	21.33	59	78.64
H/O febrile illness in pregnancy	28	37.33	47	62.67
Folic acid intake	20	26.66	55	73.34
H/o tobacco,smoking,alcohol	10	13.33	65	86.67
Family history of CMF	13	17.33	62	82.67
Consanguous marriage	30	40.00	45	60.00

Discussion

In our study period, the total number of deliveries was 7182. There were 75 babies with congenital malformations. Incidence was 1.04% Prevalence of birth defects was – 104.82/10,000 live births. Neural Tube Defects (NTDs) were the commonest type of congenital malformations – 28 (37.33%) followed by orofacial – 8 (20.05%) and skeletal defects 13 (17.33). These results were quite comparable to those stated by BDRI (Birth defects Registry of India – 2019 [4] and that stated by a study done by Bhattacharjee et al [1].

In the present study – 50 (66.6%) patients were nullipara and 25 (33.4%) were multipara, 30.66% were < 18 years and 21.31 % were above 35 years. 74.66% were unbooked with no AN checkup or care, 44.00 % were male babies, 52.00% were female babies & 3 babies (4.00%) had ambiguous genitalia, 54.66% babies weighed < 2500gm at birth while 45.34 % weighed >2500 gm. Our results were similar to those stated by Bhattacharjee et al [1], less than 18 years age , 16.1% above 35 years [1]. There lies a significant association between maternal age and congenital malformations as has been suggested by various studies worldwide. Gill S. K et al [5] reported

increased chances of congenital malformations in ages below 20 years and above 40 years. They also stated increased risks of association with total anomalous pulmonary venous return, amniotic band sequence & gastrorchi-sis when maternal age was below 20 years and various cardiac defects age was below 20 years and various cardiac defects, oesophageal atresia, hypospadias and craniosynostosis when maternal age was more than 40 years [5].

We observed that orofacial defects e.g. cleft lips and palate were more common in female babies whereas skeletal and genitourinary defects were more common in male babies. Our findings were consistent with the studies by Bhattacharjee et al [1] and Azeez et al (Africa) [6].

We also observed that these babies were mostly of low birth weight (<2500gms) Preterm labour and congenital malformations have other common risk factors eg history of febrile illness in the mother, hypertensive disorders of pregnancy including PE-E, and various maternal infections.

Similar observations were stated by Khawry et al [7] where they found a significantly high number of preterm births in babies born with congenital malformations.

Another interesting observation from our study was that approximately 20% of these babies were unplanned, unwanted pregnancies and they were history of intake of abortifacient drugs. Similar observations were made by Bhattacharjee et al [1]. Only 40.00% had AN checkup and care or undergo any ultrasonography at any time during their pregnancy. 33% had hypertension, 45.33% had diabetes in our study. It is a known fact that women who had worst blood sugar control at periconceptional period had very high risk of having babies with congenital malformations [8]. Alissa R et al showed a very strong association between maternal hypertension and birth defects [9].

73.34% women had no folic acid supplementation during this pregnancy thus confounding the fact that folic acid deficiency had very high incidence of NTD [10, 11, 12]. 17.33% had family h/o CMF thus holding relevance considering the genetic basis of CMF [5]. 40.00% had consanguineous marriages which is significant.

Conclusion

From our study we found that Congenital Malformations are quite common in our society with a prevalence of 104.82/10,000 live births and incidence of 1.04% with Neural Tube Defects being the most common type followed by skeletal, genitourinary & orofacial defects. NTD are easily preventable by folic acid supplementation and thus it is very unfortunate that we still have to encounter such a large number of babies with NTD because of lack of Antenatal care and awareness among the general population. Other factors like unwanted pregnancies, unplanned pregnancies with history of abortifacient intake and attempted abortions, maternal diabetes, hypertension, febrile illness etc also let to increased chance of babies being born with congenital malformations. The most important factor being lack of folic acid supplementation and also consanguineous marriages up to a large extent. Thus it is observed that there are several maternal risk factors associated with congenital malformations. Improvement in general health awareness, regular and proper antenatal visits and checkups, proper and timely diagnosis and management of maternal disease e.g. diabetes, hypertension, febrile illness and maternal

infections are the tools which when implemented will go a long way in reducing the incidence of babies born with congenital malformations and the associated social psychological, emotional and economical burdens thereof.

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