

A Study on System Wise Involvement of Congenital Anomalies in Foetuses and Their Association with Maternal Age and Parity

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Abstract

Background: Analyse the congenital malformations, involving various systems of the body systemwise & their association with maternal age and parity. **Methods:** This was a hospital-based cross-sectional study conducted at government maternity hospitals, Hyderabad over a period of one year months from October 2011 2015 to September 2012. All women attending Antenatal Clinic were screened by ultrasound examination. Foetuses of pregnant women were screened by clinical and ultrasonographic evaluation (12wks to 28wks). **Results:** The central nervous system and the musculoskeletal system topped the list with 16 cases (26.66%) out of total 60 cases. 11 cases of central nervous system belongs to 21-25 years age. **Conclusion:** Anomalies were most likely to be in the central nervous system. Maternal history of previous congenital anomalies, parental consanguinity, and history of medical disorders were associated with an increased risk of congenital anomalies. Early diagnosis will prevent mothers from psychological trauma of bearing and rearing an anomalous child.

Keywords: Congenital anomalies, system wise involvement, CNS, Musculoskeletal systems, maternal age, parity.

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INTRODUCTION

In India after the control of infectious diseases and reducing global malnutrition, the congenital malformations have become the major cause of perinatal mortality and morbidity.

Congenital anomalies by a broad definition are structural or functional abnormalities including metabolic disorders which are present at birth [1, 2]. As every year an estimated 7.9 million children are born with a serious birth defect, 3.3 million children under five years of age die from birth defects and 3.2 million who survive may be disabled for life [3]. As per the WHO fact-sheet of October 2012, congenital anomalies can be defined as structural or functional anomalies including metabolic disorders, which are present at the time of birth [4]. It accounts for 8-15% of perinatal deaths and 13-16% of neonatal deaths in India [5, 6]. So, it is difficult to prevent congenital malformations but the mortality and morbidity caused by them can be prevented by early detection and proper preventive and curative measures. Survivors of congenital anomalies might have lifelong physical, mental, visual, and auditory disabilities if not managed appropriately [7],

which can negatively affect the human and economic life of the person concerned, as well as their families and communities.

The knowledge of incidence, prevalence, etiological factors and different types of malformations and their severity can help the medical and paramedical personnel to identify at-risk individuals early and plan appropriate preventive measures and provide effective treatment of the condition.

In view of the importance of the congenital malformations in the contribution of perinatal mortality and morbidity, the present study was undertaken and aimed at presenting the spectrum of various congenital anomalies, epidemiological features of pregnant women with anomalous fetus.

METHOD

This is a hospital based study for one year from October 2011 2015 to September 2012. Relevant information regarding maternal age, parity, gestational age, birth weight, sex, and consanguinity was documented. Data includes demographic information,

maternal history, pregnancy and neonatal outcomes. All women attending the Antenatal Clinic were screened by ultrasound examination by radiologist for foetal well-being at around 18 weeks of gestation were recorded. All the aborted fetus and newborns were examined for congenital malformations soon after delivery. Echocardiography, x-ray, and cranial and abdominal ultrasonography were performed.

Using the International Statistical Classification of Diseases and Related Health Problems [8], we classified the patterns of congenital anomalies into:

- Congenital malformation of the nervous system;
- Congenital malformation of the musculoskeletal system;
- Congenital malformation of the digestive system;
- Congenital malformation of the circulatory system; and
- Congenital malformation of the eye, ear, face, and neck.

RESULTS

This study aimed to determine the pattern of congenital anomalies in the government maternity hospital in Hyderabad and assess the association between congenital anomalies and maternal characteristics.

Table-1: Systemwise Distribution Of Abnormalities

SL. NO	Abnormalities	Total no	%
1	CNS	16	26.66
2	Musculoskeletal	16	26.66
3	Genito urinary	11	18.33
4	GIT	4	6.66
5	Respiratory	2	3.33
6	CVS	2	3.33
7	Multiple	9	15
	Total	60	100

SYSTEM WISE DISTRIBUTION OF CASES

The central nervous system and the musculoskeletal system topped the list with 16 cases (26.66%) each, followed by genitourinary system 11 cases (18.33%), gastro intestinal system 4 cases (6.66%), and 2 cases each of respiratory (3.33%) and cardiovascular systems (3.33%). Multiple system involvement was seen in 9 cases (15%). The system wise distribution of cases is shown in Table-1.

Table-2: System wise distribution of cases based on Maternal Age

System	Mothers age			
	15-20	21-25	26-30	30+
CNS(16)	2	6	5	3
MS(16)	3	11	2	0
GUS(11)	1	4	4	2
GIT(4)	1	0	3	0
Resp sys(2)	1	0	1	0
CVS(2)	0	1	1	0
Multiple sys(9)	0	5	3	1
Total(60)	8	27	19	6

Table-2 shows the distribution of cases of congenital malformations in different maternal age groups. Out of 16 cases of central nervous system, 6 cases were seen in mothers of 21-25 years age group, 5 cases in mothers of 26 to 30 years age group and out of 16 cases of musculoskeletal system, 11 belonged to 21-25 years, 3 cases belonged to 15-20 years of maternal age group.

Table-3: System wise Distribution Of cases based on Birth Order

System	Birth order				
	I	II	III	IV	V
CNS(16)	5	3	5	1	2
MS(16)	10	6	0	0	0
GUS(11)	2	4	3	1	1
GIT(4)	2	1	1	0	0
Resp sys(2)	1	1	0	0	0
CVS(2)	1	0	1	0	0
Multiple sys(9)	4	0	2	2	1
Total(60)	25	15	12	4	4

Table-3 shows the distribution of cases of congenital malformations with respect to birth order. Out of 16 cases of central nervous system a maximum of 5 cases were seen in first and third birth order and 3 in second birth order, Out of 16 cases of musculoskeletal system a maximum of 10 cases belonged to first birth order.

DISCUSSION

The present topic of congenital malformations is gaining much importance now a days in view of increased awareness of various anomalies in the general population, improvement in diagnostic modalities, advancement of knowledge in pathophysiological aspects of the defects, identification of the teratogenic agents involved in causing such defects and increased rate of infertility followed by the treatment outcome contributing to various anomalies. The pattern and prevalence of congenital anomalies may vary over time or with geographical location reflecting a complex interaction of known and unknown genetic and environmental factors including socio-cultural, racial and ethnic variables [9].

Assessment of the incidence of congenital defects is very difficult due to early abortions,

stillbirths, prematurity, neonatal deaths, and late manifestation of the defects. Therefore the values depend on the selection criteria of the study group. Even the diagnostic tools used in the study forms an important criteria. Therefore the case selection criteria should be stringently defined and followed. In the

present study, fetuses of pregnant women attending two government hospitals in Hyderabad during one year period from October 2011 to September 2012, were screened through ultrasonographic evaluation in second trimester (12wks to 28wks) for congenital anomalies.

Table-4: Comparative Study Showing System Wise Distribution of Cases [10-16]

AUTHORS	TOT (n)	CNS %	MS %	GUS %	GIT %	RS %	CVS %	MULTIPLE %
Swain <i>et al.</i> , 1994	48	39.5	14.5	10.4	10.4	6.2	8.3	4.1
Kook Lee <i>et al.</i> , 1998	200	17	10	16.5	17.5	6.2	10.5	25
Neelu desai <i>et al.</i> , 2006	79	13.9	18.9	11.3	16.4	1.2	16.4	6.3
Nafees <i>et al.</i> , 2006	134	58.9	6.7	13.4	8.9	-	0.7	-
Somsri <i>et al.</i> , 2009	316	17.7	9.4	8.5	7.2	1.2	17.1	2.5
Sumit Gupta <i>et al.</i> , 2010	62	41.9	9.6	6.4	11.2	-	14.5	12.9
Fatema <i>et al.</i> , 2011	60	46.6	-	23.3	6.6	-	-	11.6
Present study, 2012	60	26.66	26.66	18.33	6.66	3.33	3.33	15

During the first two weeks of gestation, teratogenic agents usually kill the embryo rather than cause congenital malformations. Major malformations are more common in early embryos than in newborns; however, most severely affected embryos are spontaneously aborted during the first six to eight weeks of gestation. During organogenesis between days 15 to 60, teratogenic agents are more likely to cause major congenital malformations [17].

Each organ of an embryo has a critical period during which its development may be disrupted. The type of congenital malformation produced by an exposure depends upon which organ is most susceptible at the time of the teratogenic exposure. For instance, high levels of radiation produce abnormalities of the central nervous system and eyes specifically at 8 to 16 weeks after fertilization [18].

Maternal medical conditions can also produce teratogenic risks. Infants of diabetic mothers have an increased incidence of congenital heart disease, renal, gastrointestinal, and central nervous system malformations such as neural tube defects. Tight glycemic control during the third to sixth week post-conception is critical [18].

In our study, the malformations of central nervous system & musculoskeletal system with 16 cases each (26.66%) were sharing the top rank position followed by genitourinary system 11 cases (18.33%), gastro intestinal system 4 cases (6.66%), and 2 cases each of respiratory (3.33%) and cardiovascular systems (3.33%). Multiple system involvement is seen in 9 cases (15%). Central Nervous System anomalies were the commonest anomalies in our study, which is in accordance with some studies [19, 20], while some studies had different results. Some of the fetuses had open neural tube defect. This is comparatively higher than the previous study [21] and probably because of a higher incidence of Folic Acid deficiency in the study group. In current study, it was found that pregnancies

complicated by congenital anomaly in the foetus are at an increased risk of stillbirth. Our finding that there is an association between foetal abnormality and stillbirth is consistent with prior studies [21-23].

Congenital malformations involving central nervous system were reported to be the commonest in the studies done by Fatemaq *et al.*, Swain *et al.*, followed by musculoskeletal, gastro intestinal and genito urinary systems in some [10, 16]. Neelu Desai, found musculoskeletal system as ranking first and Samina Shamim *et al.*, have shown Gastrointestinal anomalies topping the list [12, 24].

The findings in our study were consistent with those in Munim *et al.*, (21.1%) and much less than in Nafees *et al.*, (58.9%), Fatema *et al.*, (46.6%) [13, 16, 25].

Advanced maternal age, defined as age 35 years and older at estimated date of delivery, has become increasingly common. Effective birth control, advances in assisted reproductive technology (ART), delayed marriage, increasing rates of divorce followed by remarriage, and women's pursuit of higher education and career advancement all contribute to this trend.

The explanation for the association of maternal age and chromosomal abnormalities is usually ascribed to biological aging of ova.

Maternal Age and Parity

Anencephalus, hydrocephalus, congenital cardiac disease, mongolism, hare lip and cleft palate are conditions which are more frequent in advanced maternal age groups. The percentage of incidence of anencephaly is ten times greater in the maternal age group 46–50 than in the age group 16–20 years. Higher frequency of anencephaly, hydrocephalus were born to multi para where as one third of Mongols were born to primi para along with spina bifida [26].

A study at Queen Charlotte's Hospital in London has shown the strong association of increased incidence of congenital malformations, with advanced maternal age, and mongolism association was most striking.

Obstetric care providers should be very conscious about counselling the patients diagnosed to be carrying anomalous child on Ultrasound, regarding subsequent prognosis including high risk of stillbirth.

CONCLUSION

The present study gave us an idea regarding the frequency of distribution of congenital anomalies and also its relation with associated maternal and various other risk factors. Awareness in the public regarding congenital anomalies in the fetus and the possible etiologies including environmental and genetic factors should be there. Educating adolescents and mothers is the best strategy.

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