

Study of Gross Congenital Anomalies in the Tertiary Care Hospital

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Abstract

Introduction: Congenital anomaly is a structural or functional defect that occurs during intrauterine life and can be identified prenatally, at birth or sometime later in infancy. Congenital anomalies may be caused by genetic or environmental factors. Most congenital anomalies, however, show the familial patterns expected of multi-factorial inheritance. **Methods:** Cross sectional observational hospital based study conducted during the period of August 2020 to August 2021. All new-borns delivered in the hospital with congenital anomalies were included in study. Relevant information regarding maternal age, parity, gestational age, sex and the outcome were documented. **Results:** Incidence of congenital anomalies are more associated with increasing maternal and paternal age. Higher incidence was found in higher order pregnancy. Maximum cases of congenital anomalies affected musculoskeletal system followed by gastrointestinal system and genitourinary system. **Conclusion:** With the help of proper antenatal screening, diagnostic modalities and better health care facilities, congenital anomalies can be diagnosed earlier and interventions planned accordingly. More than one risk factors can be linked with congenital anomalies. Earlier Central nervous system anomaly was commonly involved but increase folate supplementation in target population reducing the incidence.

Keywords: Antenatal scan, Gross congenital malformation, New born

Introduction

Congenital anomalies can be defined as structural or functional anomalies of any type that occur during intrauterine life and can be identified prenatally, at birth, or sometimes may be detected later in infancy¹. Indian people are at high risk of factors for birth defects, e.g. universality of marriage, high fertility rate, large number of unplanned pregnancies, and poor coverage of antenatal care, poor maternal nutritional status and high rate of consanguineous marriages^{2,3}. It accounts for 8-18% infant mortality rate and 10-15% of neonatal death in India. Congenital anomalies are responsible for increasing preterm birth, childhood & adult morbidity. Non-lethal anomalies also accountable for psychological and financial burden on family as well as society. India having a very high population rate, birth defects can be matter of concern. By strategical approach we can prevent birth of anomalous child. Population oriented strategies like iodization, flour fortification with multivitamins, folic acid supplementation⁴. Preconceptional care and counselling, carrier screening is crucial for control of birth defect⁵.

Aim of our study was to determine the distribution of various congenital anomalies in fetus and new born delivered at our teaching hospital, as well as their relation to various fetomaternal risk factors. Our aim is also to diagnose few anomalies by amniocentesis and to terminate pregnancy if it's lethal.

Materials and Methods

The cross sectional observational study conducted in Sardar Vallabhbhai Patel Institute of Medical Science and Research (SVPIMSR), tertiary care hospital, Ahmedabad during the period of August 2020 to August 2021. All new-borns delivered in the hospital with congenital anomalies were included in study. Relevant information regarding maternal age, parity, consanguinity, gestational age, history of drug intake, associated maternal condition, history of previous affected children, nutritional status of mother, complications of labor and antenatal 3D-4D USG findings were noted. In few indicated patients who gave consent for triple marker test, NIPT, amniocentesis or fetal echo done according to indication and findings were documented. In the new born babies with congenital anomalies ultrasound, echocardiography or karyotyping and X-ray were done to confirm the suspected anomalies and rule out internal anomalies and Review and comparison of antenatal USG was also done. Diagnosis of congenital anomalies was done based on clinical evaluation of a new born by a neonatologist. Soon after birth each patient received protocol-based management and advice. Data analysis was done in MS EXCEL software.

Results

During the study period of August 2020 to August 2021 at our tertiary care hospital 10,642 babies were born, of which 268 had congenital anomalies making the incidence 2.51%. The incidence of congenital anomalies are more associated with increasing maternal and paternal age.

Table 1: Maternal Age

Characteristics	Total no of deliveries (n=10642)		No. of anomalous baby (n=268)		Incidence (in %)	
	Maternal Age(M)	Paternal Age(P)	Maternal Age(M)	Paternal Age(P)	Maternal Age(M)	Paternal Age(P)
Up to 20 years	993	1211	21	20	2.11	1.65
21-25 years	4212	3088	112	52	2.65	1.68
26-30 years	3584	3996	88	94	2.45	2.35
31-35 years	1636	1635	39	68	2.38	3.18
> 35 years	217	812	8	34	3.68	4.18

Table 2: Parity

Characteristics Order of Baby (Parity)	Total number of deliveries(n=10642)	No. of anomalous baby(n=268)	Incidence (%)
Primipara	4632	106	2.28
2 nd	2948	74	2.51
More than 2 nd	3062	116	3.78

Congenital anomalies are more seen with increase in parity of patients.

Table 3: Gestational Age

Characteristics Maturity	Total number of deliveries(n=10642)	No. of anomalous baby(n=268)	Incidence (%)
Pre-term (<37 weeks)	1732	158	9.12
Full term (>37 weeks)	8910	110	1.23

Congenital anomalies are more associated with pre-term births.

Table 4: System-Wise Incidence Of Congenital Anomaly

System	No. of anomalous baby(n=268)	Incidence (%)
Musculoskeletal	76	28.35
Gastrointestinal	58	21.64
Genitourinary	36	13.43
Nervous system (CNS+Neural Tube defects)	29	10.82
Cardiopulmonary	24	8.95
Others	45	16.79

In our study musculoskeletal system is most common affected system among all anomalous babies

Table 5: Risk Factors Associatedwith Congenital Anomalies

Risk factors	No. of Cases (%)
Consanguineous marriage	26
Fever during first trimester (>100° C for 48 hours)	9
Anaemia (Uncorrected moderate anaemia)	8
Pre-eclampsia (BP >140/90 mmHg with albuminuria)	11
Diabetes Mellitus	21
Polyhydramnios (AFI > 25 cm)	23
Oligohydramnios (AFI < 5 cm)	15
History of Recurrent Pregnancy Loss (>3 Spontaneous abortions)	7
History of anomalous child in previous pregnancy	11
Epilepsy	4
Malformation of Uterus	8
Conceived after treatment for Infertility	3
Unknown	56

More than one risk factors can be associated with congenital anomalies

Discussion

Congenital anomalies are one of the leading cause associated with new born deaths within first few weeks of birth and can result in long term disability with a significant impact on individual, families, societies and health care system

During the study period, 10642 babies were born out of them 268 were having congenital anomalies. The incidence is 2.51% in our study. Amar⁶ et al had done study of Congenital Anomalies at birth in Central India, a rural medical college and hospital based data which shows incidence of 1.91%. The incidence in our study is higher because of new diagnostic modalities & regular ANC visits. Swain⁹ et al also found the incidence of congenital anomalies to be 1.2% in study. This rate also co-relates with countries having similar sociodemographic profile.⁷

Congenital anomalies are more common with increasing maternal and paternal age. Conception and delivery at later age has more probability of occurrence of genetic defect due to chromosomal anomaly. In our studies, 2.11% of anomalies are seen below 20 years of age, 5.1% of anomalies are seen between 21-30 years of age, 3.68% of anomalies are seen when maternal age is more than 35 years of age. Even in Sarkar¹² et al study, prevalence of congenitally anomalous babies was 1.8% in mother <20 years of age, 2.4% in mothers 20-30 years of age and 2.2% in mothers of >35 years of age.

In our study 2.28% of anomalies were present in primi parous patients, 2.51% of anomalies were present in 2nd para patients, 3.78% of anomalies are seen when parity is more than 2. The probable explanation of this phenomenon can be the fact that higher order pregnancies are usually seen at higher maternal age.

According to Sarkar¹² et al, incidence of congenital anomaly is 1.8% in primi para patients and 3.3% in multi para patients which.

Among preterm babies 9.12% had congenital anomalies, while only 1.23% of full term babies had congenital anomaly. This can be explained by the phenomenon of natural selection. It is consistent with earlier reports of Sarkar¹² where occurrence of congenital anomalies were 4.5 times higher in preterm deliveries than full term deliveries and El Koumi⁸ where occurrence of congenital anomalies 3.03% in preterm deliveries and 2.01% in term deliveries. 28.35% of anomalies in this study were seen in the musculoskeletal system, followed by the gastrointestinal system(21.64%) and genitourinary system(13.43%). The incidence of CNS malformation(10.82%) has reduced than observed in previous studies which may be linked to awareness about antenatal folic acid supplementation. This result can also co-relate with studies of Gandhi PR¹³ where 46% anomalies were seen in musculoskeletal system followed by genitourinary system(21.35%) and gastrointestinal tract(14.02%) and Vinodh SL¹⁴ where anomalies were of musculoskeletal system (24%), central nervous system(21.4%), genitourinary system(16.4%) and gastrointestinal tract(14.2%).

Congenital anomalies are commonly seen in consanguineous marriages. In our study 26% of anomalies are seen in patients with consanguineous marriage. This finding corresponds with Mosayebi¹⁰ et al study where incidence of congenital anomalies was 7% in consanguineous marriages compared to 2% of non-consanguineous marriages. Any anomaly identified but the risk factors can be linked with the causation of malformation. CAs are multifactorial in origin and exact etiological association can't be made out. This data co-relates with studies of Aiyar RR¹¹ and Anand JS.¹⁵

Anomaly scan is advised to all antenatal patients at about 20-22 weeks of gestation. If any anomaly is detected by USG, then patient is advised to do further evaluation.

There are few limitations of this study as this study was conducted in our tertiary care hospital having a greater number of referred patients, hence the incidence of our study cannot be applied to total population. For that the community-based studies are ideal.

Conclusion

The present study concluded that incidence of congenital anomaly is 2.51%. Though with the help of proper antenatal screening, different diagnostic modalities, food fortification, and better healthcare facilities congenital anomalies are diagnosed earlier and interventions planned accordingly. Earlier Central nervous system anomaly was commonly involved but increase folate supplementation in target population reducing the incidence of neural tube defect. Increasing awareness of maternal care, early diagnosis, antenatal 3DUSG, NIPT, amniocentesis, proper counselling for this pregnancy and subsequent pregnancy can reduce this dreaded complication of pregnancy. Early detection and termination of fetus with congenital anomalies can ease the economic burden, psychological trauma to patient and family. Team work of obstetrician, physician, geneticist and sonologist is required for management of viable congenital anomalies.

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