

Spectrum of Fetal Congenital Anomalies: A Hospital-Based Study from North India

Nishupriya, Pallavi Sharma, Anil Mehta

Abstract

Background: Congenital anomalies is a global health issue and plays a major role in perinatal morbidity and mortality. Birth defects have significant impact on individuals, families, healthcare system and societies. **Objective:** This study was done with the objective of estimating the incidence of congenital anomalies and presenting the spectrum of various congenital anomalies. **Material and Methods:** This cross-sectional study was conducted in the department of obstetrics and gynaecology Govt. Medical College Kathua, Jammu J&K w.e.f. 1st Oct 2019 to 31st March 21. Patients attending the antenatal OPD with ultrasound diagnosed congenital malformations and patients admitted in labour room who aborted on delivered congenital anomalous baby were included in the study. **Results:** During the study period 5663 births were recorded out of which 64 babies had congenital anomalies. Incidence of birth defects in present study was 1.1%. The predominant system involved was central nervous system 54.6% followed by cardiovascular system 12.5% musculoskeletal system 7.8%, Renal system 6.2% and Gastrointestinal system 4.6%. **Conclusion:** Congenital anomalies cannot be prevented totally but with awareness amongst masses and good antenatal care majority of the defects can be detected in early antenatal period when medical termination of pregnancy can be offered to women with lethal defects which will reduce maternal morbidity and mental agony of the parents.

Key Words

Congenital Anomalies, Birth Defects, Target Scan

Introduction

WHO defines congenital anomalies as structural or functional anomalies including metabolic disorders which are present at the time of birth. [1] According to a study done by March of Dimes (MOD) global report on birth defects worldwide 7.9 million births occur every year worldwide with serious congenital anomalies and 94% of these births occur in the middle and low income countries. [2] According to joint World Health Organisation

(WHO) and March of Dimes (MOD) meeting report birth defects account for 7% of all neonatal mortality and almost 3.3 million under five deaths. [3]

True magnitude of birth defects is not known though research on congenital malformation has reported incidence rate 0.8-3.8%. [4-12]

Rashtriya Bal Swasth Karyakaram (RBSK) is an initiative under the National Health Mission of Govt. of India setting

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District early intervention centre for prompt recognition of congenital anomalies and treatment at zero cost to the family. Every year 3rd March is observed as World birth defect day but still every limited data is available at local level on birth defects. Study of pattern of congenital anomalies at local level provides effective tool for necessary intervention & without comprehensive data it is difficult to evaluate possible teratogens and to implement effective preventive and care services. This hospital based study was done with the objective of estimating the incidence of congenital anomalies and presenting the spectrum of various congenital anomalies.

Material and Methods:

It is a hospital based cross-sectional study conducted in the department of obstetrics and gynaecology Govt. Medical College Kathua Jammu J&K w.e.f. 1st Oct. 2019 to 31st March 2021. All the booked and unbooked cases were taken into consideration. Patients attending the antenatal OPD with ultrasound diagnosed congenital malformation and patients admitted in labour room who aborted or delivered congenital anomalous baby were included in the study. A detailed history was taken regarding patients age and parity, gestational age, previous obstetric outcome, family history of birth defects and exposure to radiations, teratogens and intake of periconceptional folic acid. All the aborted fetuses and newborns were examined for congenital malformations soon after delivery. Systemwise distribution of the anomalies was performed.

Results

During the study period w.e.f 1st Oct 2019-31st March 2021. 5,663 births were recorded, out of which 64 had congenital anomalies. Incidence of births defects in present study was 1.1%.

Majority of our women were in the age group of 26-30 years and association between age and birth defects is not significant.

No relation was found between parity and congenital malformation.

6.25% of the malformed babies were born to mothers

with previous history of pregnancy wastage and one patient (1.56%) had malformed baby in previous pregnancy. No patient gave history of teratogens or radiation exposure. Preconceptional folic acid was not taken by single patient. Majority received folic acid only after registration.

45.31% of birth defects were detected after 20 weeks. In 7.8% patients birth defects were detected at birth out of which in 40% case USG was done in the periphery and findings were missed while 60% cases were totally uninvestigated unsupervised pregnancies who were admitted in labour room in active labour.

Discussion

Congenital anomalies is a global health issue and play a major role in perinatal morbidity and mortality. Birth defects have significant impact on individual, family, healthcare system and society.

During the study period, 5,663 births were recorded out of which 64 were diagnosed with congenital anomalies. In the present study, incidence of congenital malformation at birth was found to be 1.1% which is comparable with the earlier studies from India, Kokate P *et al* 0.9% (4), Datta *et al* 1.2% ^[5], Naik M 1.22% ^[6], Taksande 1.9% ^[7] In a hospital based study from Nepal by Khanal GP ^[8] 1.02% pregnant women were diagnosed as having congenital anomalies). Some studies observed higher incidence like Sharma Vishal M 2.4%. ^[9] Mithlesh et al 2.5% ^[10], Desai *et al* 3.6% ^[11] Hoangthi Nam Giang 3.8%. ^[12] This disparity may be because some of the studies were conducted in referral hospitals getting a lot of referred patients. Other reasons might be availability and accessibility of advanced services and technology, sociodemographic factors and pregnant mother's attitude towards screening in various parts of the world. Since ours is a hospital based study, the data cannot be projected to the general population. Some women might have aborted or delivered at home or in private hospital. Community based study should be ideal for true estimation of incidence of congenital anomalies in a population. Most anomalies were detected late beyond the legal gestational age of medical termination of pregnancy which

Table 1. Distribution of congenital malformations according to age.

Age	Number of deliveries	Number of Congenital anomalies	Percentage %	
< 20	132	3	2.27%	p = 0.34
21-25	2153	22	1.02%	
26-30	3186	34	1.06%	
> 30	192	5	2.6%	

Table 2. Distribution of congenital Malformation by parity

Gravidity	Number of deliveries	Number of Congenital anomalies	Percentage %	
Primigravida	3246	35	1.07%	p=0.1
Multigravida	2417	29	1.19%	

Table 3. Gestational age at which congenital anomalies detected

Gestational age (Weeks)	Number of Patients	Percentage%
< 12	3	4.6%
13-20	32	50%
20-28	16	25%
29-40	8	12.5%
At birth	5	7.8%
Total	64	100%

Table 4. Spectrum of congenital anomalies according to body system

Congenital Anomalies	No. of Cases	Congenital Anomalies	No. of Cases
Central Nervous System	35	Musculoskeletal system	5
Anencephaly	9	CTEV	2
Spina Bifida	5	Sacrococcygeal teratoma	1
Hydrocephalus	3	Short Limbs	1
Anencephaly	2	Polydactyly	1
Choroid Plexus Cyst	2	Renal System	4
Ventriculomegaly	2	Hydronephrosis	2
Alobar holoprosencephaly	2	Multicystic Kidneys	1
Arnold Chiari	2	Posterior Urethral Valve	1
Acrania	1	Gastrointestinal	3
Spinal agenesis	1	Duodenal Atresia	1
Cerebellar hypoplasia	1	Omphaloule	1
Exencephaly	1	Diaphragmatic Hernia	1
Agenesis of septum pellucidum	1	Respiratory System	3
Enlarged Cisterna Magna	1	CCam	2
Exencephaly	1	Hypoplastic Lung	1
Hypoplastic Vermis	1	Genitourinary	2
Cardiovascular System	8	Undescended testis and abdominal distension	1
Ventricular Septal Defect	3	Imperforate anus	1
Cardiomegaly	2	Mixed	4
Hypoplastic left Ventricle	1	Ventriculomegaly and omphalocele	1
Transposition of great arteries	1	Anecephaly and clift lip and palate	1
VSD with overriding of Aorta	1	Cleft Palate with Pierre robin syndrome	1
		Cystic hygroma, Irregular spine, shortlimbs, short nasal bone	1

is similar to other studies in India, Vishal *et al*^[9] Lavanya *et al*.^[13] In the present study 45.31% birth defects were detected after 20 weeks. Only 4.6% were detected in first trimester. The psychological trauma to the women can be well imagined who knows that the baby she is carrying is anomalous and is left with no choice but to continue pregnancy till term.

Knowledge of birth defects seems to be influenced by traditional beliefs. It is important to educate the community, health professionals and public about the need for preconceptional screening for high risk factors and various preventive measures available. Because of lack of education and awareness about the safety of USG during pregnancy and its importance in detecting birth defects. Many women still present late for antenatal checkups when they are advised for USG. In some cases, USG was done in 1st trimester and then repeat USG was done in III trimester. Anomaly or target scan was missed. It is, therefore, very important that whichever centre women comes for first registration, she should be advised to get target USG at 18-20 weeks to rule out birth defects so that this crucial time is not missed and if lethal anomalies detected, MTP can be offered to her. 3% cases were missed in USG which may be because of lack of expertise and availability of good resolution machines in peripheral centres,

In the present study, the most common system involved was central nervous system (54.68%) followed by cardiovascular system (12.5%), Musculoskeletal system (7.8%), Renal system and Gastrointestinal system (4.6%). Anencephaly was the most reported CNS defects followed by Spina bifida. Similar observations were made by Naik M *et al*^[6] and Sharma *et al*.^[9] Maximum number of congenital malformations were of nervous system in their studies. Lavanya *et al* in their study found that the predominant system^[13] involved is central nervous system followed by musculoskeletal system. Taye M *et al*^[14] and Silesh *et al*^[15] in their studies also found central nervous system as the most commonly affected body system. CNS malformations are better detected in the antenatal ultrasound than any other system

and also CNS defects are obvious to the birth attendants requiring no additional confirmation. In our study, not even a single patient took periconceptional folate. Folic acid was taken only after registration. This gap can be filled by ASHA workers who have direct contact with the community. Folic acid distribution to the eligible couples and periconceptional counselling can be done by them. The annual report of Indian Council of medical research says that the commonest congenital malformations are cardiac in nature.^[16] Mithlesh *et al* in their study also found that cardiovascular system malformations were the commonest in live births^[10] Kumar *et al* reported that cardiovascular system anomalies were the commonest followed by musculoskeletal, gastrointestinal and central nervous system.^[17] Anomalies of the cardiovascular system were the commonest in the cohort studied by Ajao followed by anomalies of digestive system.^[18] Detection of cardiovascular system defects require expertise and high resolution imaging and also minor cardiac defects do not manifest at birth and are diagnosed in early or late neonatal period and in our study the anomalies detected only till birth were included. However, Anne George *et al* in their study observed that musculoskeletal deformities were the commonest out of which CTEV was seen in 73.6% of patients.^[19] Sarkar *et al* found that predominant system involved was musculoskeletal followed by gastrointestinal and central nervous system.^[20] Musculoskeletal abnormalities were the most frequently reported (33%) congenital anomalies in the study by Forci *et al* out of which 17.4% had club foot.^[21] Genitourinary system was the most common system involved followed by musculoskeletal system and cardiovascular system in the study done by Shrestha S *et al*.^[22]

Conclusion

Central nervous system was predominantly affected system. Public healthcare awareness campaigns should be launched involving media, social media, Asha Workers to make public aware of birth defects, their prevention and management. Birth defect registry should be maintained.

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Conflicts of Interest

There are no conflicts of interest.

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