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Original Research Article

Incidence of congenital anomalies in Navodaya Medical College

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ABSTRACT

Background: Congenital anomalies are defined as structural or functional anomalies including metabolic disorders, that occur during intrauterine life and can be identified antenatally, at birth or later in life. It accounts for 11% of neonatal deaths globally and accounts for 8–18% of perinatal deaths and 10–15% of neonatal deaths in India. Aims and objectives of the research were to study incidence and risk factors associated with congenital anomalies in Navodaya Medical College.
Methods: The clinical study was done on 3008 patients over 1 year at Navodaya Medical College Hospital and Research Centre Thorough history, antenatal ultrasound, blood tests, new born babies were examined thoroughly by the paediatrician to detect the congenital malformation. If any internal congenital malformation were suspected further investigation like ultrasonography (USG), echocardiography (ECHO), X-ray, computed tomography (CT), and magnetic resonance imaging (MRI) were done.
Results: Out of 3008 cases, 40 babies had congenital anomalies, incidence is 1.3%, most commonly involved system is musculoskeletal system followed by cardiovascular system. Major risk factors associated are extremes of age, parity, lack of ante natal check-ups, no intake of folic acid, maternal diabetes mellitus.
Conclusions: Congenital malformations though cannot be prevented totally but can be minimised and if detected early which reduces mental agony in mother and family. Prenatal counselling, periconceptional folate, anomaly scan, prenatal diagnosis reduces the incidence of neonatal and infant morbidity and mortalities in India.

Keywords: Congenital anomalies, Anomaly scan, Preconceptional counselling, Infant mortality

INTRODUCTION

Congenital anomalies are defined as structural or functional anomalies including metabolic disorders, that occur during intrauterine life and can be identified antenatally, at birth or later in life.[1,2]

According to World Health Organization (WHO) congenital anomalies are also known as birth defects, congenital disorders or congenital malformations. It accounts for 11% of neonatal deaths globally and accounts for 8–18% of perinatal deaths and 10–15% of neonatal deaths in India.[3] The aetiology of congenital abnormality may be genetic (30–40%) or environmental (5–10%) and 50–60% are multifactorial.[4]

Defects are of prenatal origin resulting from defective embryogenesis or intrinsic abnormalities in the process of development. Birth defects can be isolated abnormalities or part of a syndrome and continue to be an important cause of neonatal and infant morbidity and mortality.[5]

Several known factors that are associated such as maternal infection like TORCH, genetic factors, drugs, maternal age, and consanguinity, increase in the use of irradiation, alkylating agents, antimetabolites, smoking and drinking

and environmental pesticides—all known to cause congenital anomalies.[6]

Congenital malformations remain one of the least focused areas of disease surveillance in India compared with communicable and some chronic diseases.[7] Unlike the situation in developed countries, where congenital malformations are the leading cause of infant mortality, in India low birth weight, prematurity, sepsis and infections are still the leading causes of neonatal and infant mortality.[8]

Royal College of Obstetricians and Gynaecologists (RCOG) classified malformations into lethal, severe and moderate. Lethal defects are anencephaly, bilateral renal agenesis, giant hygroma, osteochondrio dysplasia, icthyosis congenita. Severe defects include hydrocephalus, spina bifida, esophageal atresia, TOF, absent uterus, ectodermal dysplasia, posterior urethral valves (PUV), atrial septal defect (ASD), and patent ductus arteriosus (PDA). Moderate include imperforate hymen, septal deviation, choanal atresia, craniosynostosis, and eyelid defects.[9]

Screening in late first and second trimester is important tool to reduce the prevalence. Ultrasound is the best possible non-invasive technique available to detect any congenital anomalies in pregnant women which will help to identify the severity of the disease, its outcome leading to pregnancy termination or gives an opportunity for fetal therapy or better neonatal care.[10]

Congenital malformations though cannot be prevented totally but can be minimised and if detected early will reduce the mental agony in the mother and family. Prenatal counselling, periconceptional folate, anomaly scan, Prenatal diagnosis reduce the incidence of neonatal and infant morbidity and mortalities in India.[11]

METHODS

Methods of collection of data include outpatient department (OPD) and inpatient department (IPD) patients attending obstetrics and gynaecology department, Navodaya Medical College Hospital and Research Centre.

Study site

The study was conducted at Navodaya Medical College Hospital and Research Centre, Raichur.

Study design

The design of the study was observational.

Inclusion criteria

Women aged between 18–40 years, patients willing to give informed consent, and both booked and unbooked cases were included in the study.

Exclusion criteria

Patients not willing to give informed consent and patients who were not delivered in centre were excluded.

Methodology

After obtaining institutional ethical committee approval and written and informed consent from the patient, It was observational hospital based study conducted in Department of Obstetrics and Gynaecology in Navodaya Medical College. Those women who had diagnosed to have anomalous foetus either in antepartum period or after delivery, during neonatal period were included in study after informed consent. Detailed information regarding maternal age, booking status, order of pregnancy, gestational age, and consanguinity was documented. Antenatal history like maternal illness, ingestion of drugs, exposure to radiation and complications of labour was recorded. The screening for congenital malformation was done by antenatal ultrasound, and blood tests. All the new born babies were examined thoroughly by the paediatrician to detect the congenital malformation. If any internal congenital malformation were suspected further investigation like ultrasonography (USG), echocardiography (ECHO), and X-ray were done. Computed tomography (CT), magnetic resonance imaging (MRI), and cystourethrography were performed in selected cases only. All the foetuses and babies having congenital malformation were taken into account and were analysed for the study.

Data collection

All data of patients were collected in approved proformas. Later relevant data for analysis and comparison were recognized into Microsoft excel data sheet format.

Statistical analysis

Data was analysed using IBM statistical package for the social sciences (SPSS) 20 software. Chi squared test and independent t test were used.

RESULTS

In the present study, we studied the cases of congenital anomalies detected antenatally or after delivery in Navodaya Medical College, Raichur for a period of one year from 01 January 2020 to 01 January 2021. A total of 3008 cases, total numbers of malformed babies were 40, so total point incidence of congenital anomalies turned out to be 1.3%.

The results and observations thereof are represented in a tabulated form in Table 1.

Among the risk factors studied, multiparity was an important association. Regarding the maternal age, a higher proportion of anomalies were noted among teenage

mothers 25% of anomalous babies being born to mothers less than 18 years and another 35% to mothers between more than 35 years.

Table 1: Socio demographic profile of the mothers.

Type	No. of cases	Percentage (%)
Parity		
Nulliparous	12	30
Multiparous	28	70
Age (years)		
<18	10	25
18-34	16	40
>35	14	35
ANC care		
Booked	21	52.5
Unbooked	19	47.5
Sex		
Male	18	45
Female	19	47.5
Ambiguous	03	7.5

Table 2: Gestational age at which malformations were detected.

Gestational age	Number
1st trimester	02
2nd trimester	25
3rd trimester and after delivery	13

Table 3: Malformations detected by USG.

Malformations	No. of cases	Percentage (%)
Detected	23	57.5
Not detected	10	25
USG not done	7	17.5

Table 4: Risk factors.

Risk factor	No. of cases	Percentage (%)
Consanguity	13	32.5
Abortions	8	20
H/O of IUFD	4	10
Overt DM	10	2
PIH	6	15
Anemia complicating pregnancy	2	5
Varicella infection	1	2.5
UTI	1	2.5
Sibling with malformation	2	5

Among older mothers (>35 years) there was 14 have anomalous baby. Mothers between 18-34 had 16 anomalous babies (40%). With regards to maternal problems, overt diabetes mellitus was found to be associated with birth of an anomalous baby, with nearly 25% of anomalous neonates being born to diabetic mothers.

The regular antenatal scans done to detect anomalies missed 42.5% of cases. Majority of which were cardiac and neural tube defects which emphasizes the need for more targeted modalities like foetal echocardiogram and MRI to be a part of antenatal screening in the presence of maternal risk factors, though cost could be a limiting factor.

Table 5: Outcome in malformed fetuses.

Outcome	Cases
Live births	20
MTP	12
IUD	8

Table 6: Distribution of anomalies: system wise.

System	No. of cases	Percentage (%)
Musculoskeletal system	11	27.5
CVS	10	25
CNS	9	22.5
Genitourinary	5	12.5
GIT	2	5
Abdominal wall defects	2	5
Endocrine	1	2.

Table 7: Musculoskeletal system: distribution of anomalies.

Malformation	No. of cases
Cleft lip	2
Cleft palate	1
Cleft lio and cleft palate	2
Limb defects	1
Polydactyly	5

DISCUSSION

Total number of cases was 3008. There were 40 babies with congenital malformations. Incidence was 1.3%. Out of 40 patients primi patients were 12 and multiparous were 28, age group less than 18 years in 10 cases,18-34 years in 16 cases and greater than 35 years in 14 cases. Out of 40 cases 2 cases diagnosed in the first trimester, 25 in the second and 13 in third trimester and after delivery. Out of 40, 20 were detected in USG, 10 are not detected, and USG not done in 7 cases.

Among the risk factors studied, multiparity was an important association. Regarding the maternal age, a higher proportion of anomalies were noted among teenage mothers 25% of anomalous babies being born to mothers

less than 18 years and another 35% to mothers between more than 35 years.

Among older mothers (>35 years) there was 14 have anomalous baby. Mothers between 18-34 had 16 anomalous babies (40%). In the present study, 12 (30%) patients were nullipara and 28 (70%) were multipara, 25% were <18 years and 35% were above 35 years. 52.5% were booked and 47.5% unbooked with no AN check-up or care, 45% were male babies, 47.5% were female babies and 3 babies (7.5%) had ambiguous genitalia.

With regards to maternal problems, overt diabetes mellitus was found to be associated with birth of an anomalous baby, with nearly 25% of anomalous neonates being born to diabetic mothers.

The regular antenatal scans done to detect anomalies missed 42.5% of cases. Majority of which were cardiac and neural tube defects which emphasizes the need for more targeted modalities like foetal echocardiogram and MRI to be a part of antenatal screening in the presence of maternal risk factors, though cost could be a limiting factor.

Table 8: Comparison with other studies.

Study group	Incidence (%)
Present study	1.3
Swain et al	1.2
Taksande et al	1.91
Anand et al	
Karla et al	1.98
Desai et al	3.6
Datta et al	1.24
Saifullah et al	3.6

Other studies like Datta et al, Swain et al, Taksande et al, Anand et al and Karla et al showed incidence of congenital anomalies was 1.24%, 1.2%, 1.91%, 2% and 1.98% respectively.[12-15] Studies like Desai et al and Saifullah et al showed slightly higher incidence (3.6%) than our study. The true incidence of congenital malformations depends upon several factors and no two studies are strictly comparable. It depends upon ethnic background, population sample (hospital or community based, live birth or total birth), nature of study (prospective or retrospective), age at the time of diagnosis, duration of follow up, autopsy rate, diagnostic facility available and enthusiasm and acuteness of physician.

Musculoskeletal defects were the commonest type of congenital malformations – 28 (37.33%) followed by cardiovascular – 8 (20.05%) and CNS defects 13 (17.33). The present study results are comparable to studies done by Swain et al and Baht et al in South India (3.7%), Dolk et al in Europe (2.39%) and also with the western data from the EUROCAT surveillance.[16,17]

Gill et al reported there lies a significant association between maternal age and congenital malformations as has been suggested by various studies worldwide increased chances of congenital malformations in ages below 20 years and above 40 years.

In our study total 32.5% cases had consanguineous marriage. Our study also showed a higher incidence of congenital malformation in parents having consanguineous marriage.[18-20]

Other condition like diabetes, severe preeclampsia was also associated with increased incidence of congenital malformation. Ordonez et al showed positive association of diabetes mellitus, hypertension, and hypothyroidism with congenital malformation. Taksande also concluded similar pattern of association.[21]

CONCLUSION

Factors like unwanted pregnancies, unplanned pregnancies with history of abortificient intake and attempted abortions, maternal diabetes, hypertension, febrile illness etc also led to increased chance of babies being born with congenital malformations. The most important factor being lack of folic acid supplementation.

Musculoskeletal defects being the most common type followed by cardiovascular, CNS, genitourinary. Thus, it is observed that there are several maternal risk factors associated with congenital malformations. Improvement in general health awareness, preconceptional counselling, regular and proper antenatal visits and check-ups, proper and timely diagnosis and management of maternal disease e.g. diabetes, hypertension, febrile illness and maternal infections peri conceptional folic acid supplementation 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 economic burden.

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