



STUDY OF CONGENITAL ANOMALIES AT TERTIARY CARE CENTRE

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ABSTRACT

Background: Congenital anomalies are the important causes for perinatal mortality and morbidity. Therefore early diagnosis of congenital anomaly is helpful for patients. **Method:** It is a prospective study carried out during period from December 2021 to November 2022 in Obstetrics and Gynecology Department of B. J. Medical College and Hospital, Ahmedabad. 50 cases of congenital anomalies out of 7320 delivered cases were studied in this study. **Results:** The present study was conducted in the hospital for evaluation of associated risk factors and antenatal diagnosis and foetal outcome. Among the 7320 deliveries, 6813 were live births and 330 still births. Out of the 50 cases studied, 35 were major anomalies and 15 were minor anomalies. Among those new born who survived; 90% were diagnosed antenatally, 10% were missed. The most common anomaly detected is cardiovascular system (24%) followed by craniospinal system and musculoskeletal system. Study found that congenital anomalies are associated with lack of appropriate antenatal care and fetal anomaly scanning. **Conclusion:** Prevalence of congenital anomalies remain high due to lack of Pre conceptional counselling, default in folic acid intake, lack of appropriate antenatal care, nutritional disorder, infectious diseases, life style changes. Need for focussed screening in the high-risk categories, pre scan counselling with karyotyping, Nuchal translucency test, triple screen and relevant serology should be done. A level 2 targeted scan should be undertaken at 18-20 weeks and again at 24 weeks to rule out anomalies.

KEYWORDS : Congenital anomalies, anomaly scan

INTRODUCTION:

Congenital anomalies are abnormality of structure, function or body metabolism that is present at birth which results in physical or mental disability or perinatal mortality.² Birth defect can be isolated abnormality or a part of syndrome and continue to be an important cause of perinatal mortality and morbidity. There are more than 4000 known birth defects. Congenital anomalies contribute to long term disability, which may have significant impacts on individuals, families, health care system and societies.⁸ As estimated 2-3% babies worldwide are born with congenital anomaly.⁽⁸⁻⁹⁾ In India incidence is around 2.5%¹⁰ and congenital anomalies account for 8-15% of perinatal mortality and 13-16% of neonatal deaths in India.⁽⁶⁻⁷⁾

The most common anomalies are heart defects, neural tube defect and chromosomal abnormality. Some congenital abnormalities are curable if they are detected early in the antenatal period (e.g. Cardiac anomalies). The advanced diagnostic modalities used for prenatal diagnosis with fetal anomaly scanning will help to reduce the birth prevalence with serious anomaly and increasing the chance of survival for those who are born.

Findings of correctable abnormality can be indication for delivery to take place at a tertiary center with facility for pediatric surgery or uncorrectable anomaly to lead to termination of pregnancy. This study was conducted to predict the variables which contribute in the incidence of congenital anomaly and timely diagnosis of anomaly so that we can reduce the related perinatal morbidity and mortality.

AIMS AND OBJECTIVES:

- To determine the frequency and type of different structural congenital anomalies in our hospital.
- To identify the most common congenital anomaly.
- To identify possible risk factors responsible for these anomalies.
- To evaluate the fetal outcome.

MATERIALS AND METHODS:

- Total 50 cases out of 7320 deliveries were prospectively evaluated for congenital anomalies and associated risk factors during period from December 2021 to November 2022 in Obstetrics and Gynecology Department of B. J. Medical College and Hospital, Ahmedabad.
- Antenatal women diagnosed with fetal congenital anomaly by imaging were included in this study.
- Variables like maternal age, parity, consanguinity, abortions, intrauterine deaths, nutrition, family history, maternal infections and diseases, usage of drugs, occupation.

RESULTS:

Table 1: Maternal Characteristics

	Number	Percentage
Maternal age		
<20 years	05	10%
20-25 years	37	74%
>35 years	08	16%
Consanguinity		
Consanguineous	28	56%
Non-consanguineous	22	44%
Gestational age		
Term	22	44%
Preterm	28	56%
Parity		
Primi	24	48%
Multi	26	52%
Birth weight		
<2.5 kg	28	56%
>2.5 kg	22	44%
Mode of delivery		
Vaginal delivery	30	60%
Caesarean section	08	16%
Abortion	12	24%
Booked status		

Booked	20	40%
Un-booked	30	60%

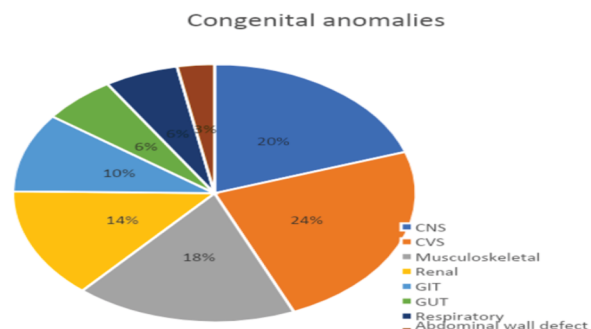
Most congenital anomalies are seen with age of 20-25 years and with multiple pregnancy.

Lack of appropriate antenatal care leads to nutritional deficiency and missed congenital anomaly in early pregnancy.

Table 2: Association of risk factors

Risk factors	Number	Percentage
Age >35 years	08	16%
Infections, Fever	06	12%
Nutrition	10	20%
Treatment taken for infertility	05	10%
Maternal diabetes	04	08%
Hypothyroidism	02	04%
Twins	03	06%
Oligohydramnios	08	16%
Polyhydramnios	06	12%

In present study, it was found that congenital anomaly was found in the mothers who had risk factors like multiparity, extreme ages, previous history of abortions/congenital defect, lack of appropriate antenatal care and nutrition and maternal disease



We found that major defect seen in cardiovascular system and least in abdominal wall defect.

Fetal echocardiography is directed towards selected pregnancies carrying a higher than normal risk of fetal cardiac anomalies.

Table 3: Patterns of anomalies observed

Anomalies	Number	Percentage
Cranio-spinal system:	10	20%
Hydrocephalus	02	4%
Anencephaly	03	6%
Meningocele	02	4%
Meningomyelocele	01	2%
Spinal bifida	01	2%
Encephalocele	01	2%
Musculoskeletal system:	09	18%
Cleft lip, cleft palate	03	6%
C'TEV	03	6%
Polydactyly	01	2%
Skeletal Dysplasia	01	2%
Congenital vertical talus	01	2%
Cardiovascular system:	12	24%
ASD	08	16%
Tricuspid atresia with hypoplastic right ventricles	02	4%
Hypoplastic aortic isthmus	01	2%
Tetralogy of Fallot	01	2%
Renal	07	14%
Pelvi-ureteric junction obstruction	02	4%
Bladder outlet obstruction	03	6%

Polycystic kidney	01	2%
Renal agenesis	01	2%
Gastrointestinal system:	05	10%
Ileal atresia	01	2%
Duodenal atresia	02	4%
Trachea-esophageal fistula	02	4%
Abdominal wall defects:	01	2%
Gastroschisis	01	2%
Genitourinary:	03	6%
Hypospadias with hydrocele	01	2%
Ambiguous genitalia	02	4%
Respiratory system:	03	6%
Congenital diaphragmatic hernia	03	6%

Table 4: Fetal outcome

Outcome	Number	Percentage
Abortions	12	24%
Preterm vaginal delivery	16	32%
Full term vaginal delivery	14	28%
LSCS	08	20%

Table 5: Postnatal Management required

Postnatal management required	Number	Percentage
Active management	08	21%
Conservative management	16	42%
No active management	10	26%
No management	04	10%

Out of 38 cases, 08 anomalies required active management in form of surgery along with medical management, 16 cases were managed conservatively 10 cases were observed and followed up subsequently and did not require active intervention.

DISCUSSION:

The study is focused on the structural congenital anomalies at tertiary care center. Congenital anomalies are important causes of still births and infant mortality, and are contributors to childhood morbidity.

Among the 7320 deliveries, 6813 were live births and 330 were still births. Out of the 50 cases, 35 were major anomalies and 15 were minor anomalies. 60% cases in the present study were not registered cases and had not taken antenatal care, while 16% cases were those of lethal anomalies diagnosed with level 2 targeted scan and timely terminated. The survival rate of major anomalies was 64% and minor anomalies was 98%. Among the new born babies that survived; anomalies were diagnosed antenatally in 90% cases and were missed in 10% cases.

Increased risk of congenital malformation seen in consanguineous marriages .⁴ As consanguineous marriages are still common in present day society, proper premarital counselling and carrier detection for common inherited disorders should be undertaken

Majority of mothers of babies with congenital anomaly belonged to age group 20-25 years. This similar observation was made by Kamble V et al.⁵The study by Kokate et al where maternal age >30 was the most important risk factor.¹² Most commonly implicated high-risk factor was increased maternal age.

In the present study, 52% cases of congenital anomalies seen in multi parity. Sarkar et al Study also found the incidence of congenital anomalies more common in multipara as compared to primipara.⁴

In the present study, most common system involved in congenital anomaly was Cardio-vascular system (24%) followed by cranio-spinal system(20%).In the study by kokate

et al cranio-spinal anomalies(44%) was commonest followed by musculoskeletal system(30%).¹²

Maternal anaemia and malnutrition pose a serious threat to both mother and foetus, hence fortification of food with iron, folic acid, iodine should be carried out. To reduce the incidence of neural tube defects, periconceptual use of folate should be encouraged.

A level 2 targeted done at 18-20 weeks and again at 24 weeks to rule out anomalies and those who takes decision for termination of pregnancy, it is easier for obstetrician and patient to offer termination. Sensitivity of USG was 74.03% and specificity was 98.86%.¹¹

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CONCLUSION:

The prevalence of congenital malformation in India is around 182 cases per 10,000 live births¹³. 2-3 per 100 children are born with birth defects around the world.¹

Antenatal care (ANC) can reduce the health risks for mother and their babies by screening and monitoring pregnancies and its complications.³ Hence, there is need for focussed screening in these high-risk categories, pre scan counselling with karyotyping, triple screen and relevant serology should be done. The mid trimester scan at 18-20 weeks clearly represents the best time to accomplish the most.

Once anomaly detected, various management options are to be discussed with the patient in consultation with neonatologist, pediatric surgeon and neurosurgeon if necessary. Lethal anomalies are terminated immediately irrespective of gestational age.

Hence enforcing the importance of mid trimester USG scan in identifying the cases, Pre conceptual counselling, folic acid intake, vaccination can help in reducing the incidence of congenital anomalies.

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