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Original Research Article

Study of congenital malformation in tertiary care centre, Mumbai, Maharashtra, India

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ABSTRACT

Background: Birth defects are important cause of neonatal morbidity and mortality. Congenital anamolies are defined as structural and functional abnormalities including metabolic disorders present at birth. There are several known factors that are associated such as maternal infection like TORCH, genetic factors, drugs, maternal age, Consanguinity. Screening in late first and second trimester is important tool to reduce the prevalence.
Methods: A retrospective study was done. Data was collected and analyzed. Fetal outcome was assessed. Variables like maternal age, parity, consanguinity, abortions, sibling with malformation, nutrition, smoking ,alcoholism, family history of congenital anomalies, conceived after infertility treatment, maternal diabetes, infections, fever, drugs, history of intrauterine deaths were critically evaluated.
Results: Out of total 5020 deliveries, 50 babies with congenital anomalies identified. Incidence being 0.9%, commonest congenital anomalies involving craniospinal system (44%). Second most common is musculoskeletal system (30%). Consanguinity is single most important factor which was found to increase the risk of congenital anomalies in our study. In 40% of the cases consanguinity was noted. Most common perinatal risk factors are preterm labor (22%), polyhydramnios (8%) and breech (16%). The fetal outcome was 80% of the babies were compatible with life and 20% were non compatible.
Conclusions: In the present study, most of the mothers who had anomalous fetuses had risk factors like consanguinity and previous history of abortions. Hence the need for focused screening in this high risk category. A level II targeted scan is done at 18-20 weeks and again at 24 weeks to exclude anomalies and reduce the prevalence. Once an anomaly is detected, various management options are to be discussed with the patients in consultation with neonatologist, pediatric surgeon and neurosurgeon when necessary. If parents are willing to continue the pregnancy with compatible congenital anamolies in baby then pregnancy may be continued. But if the congenital anamoly is imcompatible with life then pregnancy should be terminated. This study was conducted to study the incidence of various congenital anamolies in babies and their possible etiological factors in the population visiting to tertiary care hospital at Mumbai.

Keywords: Anamolies, Consanguinity, Targeted scan

INTRODUCTION

Birth defects are important cause of neonatal morbidity and mortality. These 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.[1] Congenital anamolies are defined as structural and functional abnormalities including metabolic disorders present at birth. There are several known factors that are associated

such as maternal infection like TORCH, genetic factors, drugs, maternal age, and consanguinity. 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. This study was conducted to evaluate the incidence of structural congenital anomalies and to predict the variables which contribute in the incidence of congenital anomalies so that we can reduce the related perinatal morbidity and mortality.[2,3]

METHODS

A retrospective study was done. Data was collected and analysed.

Fetal outcome was assessed. Variables like maternal age, parity, consanguinity, abortions, sibling with malformation, nutrition, smoking ,alcoholism, family history of congenital anomalies, conceived after infertility treatment, maternal diabetes, infections, fever, drugs, history of intrauterine deaths were critically evaluated.

RESULTS

Table 1: Age.

Age	Number	Percentage
<20	-	0
20-30	38	76
>30	12	24

Table 2: Parity.

Parity	Number	Percentage
Primigravida	18	36
2nd Gravida	17	34
3rd Gravida	12	24
4th or More	03	6

Table 3: Gestational age.

Gestational age	Number	Percentage
<28 wks	10	20
28-37 wks	13	26
> 37 wks	24	48
After birth	03	06

Out of total 5020 deliveries, 50 babies with congenital anomalies identified. Incidence being 0.9%, commonest congenital anomalies involving craniospinal system (44%) (Table 8). Second most common is musculoskeletal system (30%) (Table 8). 88% of cases were registered at our hospital. 76% cases were in the age group of 20- 30 yrs and 24% were in the age group of >30 yrs (Table 1). 36% cases were primigravidae (Table 2). In 10% of cases history of abortions was present (Table 5). About 20% congenital anomalies were detected before 28 wks (Table 3). 26% of the cases were diagnosed between 28-37 wks; most of them have no previous antenatal scans due to infrequent antenatal visits (Table 3). Most common perinatal risk factors are preterm labor (22%), polyhydramnios (8%) and breech (16%) (Table 6). Congenital malformations contribute to 20 % of perinatal mortality. Even though congenital anomalies of minor degree, prematurity along with associated maternal contributing factors are responsible for the perinatal mortality.

Table 4: Blood group.

Blood group	Number	Percentage
O Positive	14	28
A Positive	14	28
B Positive	14	28
AB Positive	05	10
A Negative	03	6

Table 5: Risk factors.

Risk factor	Number	Percentage
Consanguinity	20	40
Abortions	5	10
History of intrauterine fetal death	4	8
Maternal diabetes	3	6
Age > 30 yrs	12	24
Infections, fever	4	8
Sibling with malformation	2	4

Table 6: Associated risk factors.

Risk factor	Number	Percentage
Preterm	11	22
Polyhydromnios	4	8
Breech	8	16
IUGR	3	6
Oligohydramnios	2	4
Risk factor	Number	Percentage

DISCUSSION

We found the incidence of congenital anomalies in our hospital was 0.9% in our study which is equal to the general incidence in developing countries.[2-5]

In our study 44% (Table 8) of cases involved craniospinal system. Meningomylocele amounting to 12% cases of NTDs and most common factor contributing to perinatal mortality.

Table 7A: Distribution of anomalies: Craniospinal - 22 (44%).

Hydrocephalous	5	10
Meninomylocele	6	12
Encephalocele	1	2
Spina bifida	1	2
Holoprosencephaly	1	2
Dolicocephaly	1	2
Acrania	1	2
Anencephaly	5	10
Lisinocephaly	1	2

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Lisinocephaly	1	2

Table 7B: Distribution of anomalies: Abdominal wall defects - 2(4%).

Imperforated anus	1	2
Gastroschisis	1	2

Table 7C: Distribution of anomalies: Cardiovascular – 3 (6%).

VSD	1	2
PDA	1	2
Complex cardiac anamoly	1	2

Table 7D: Distribution of anomalies: Renal – 2 (4%).

Bilateral hydronephrosis	1	2
Renal agenesis	1	2

Table 7E: Distribution of anomalies: Musculoskeletal- 15 (30%).

Cleft lip	1	2
Cleft Palate	3	6
Cleft lip and Palate	7	14
Limb defects	3	6
Polydactyly	1	2

Second most common congenital anomalies involved facial and neck structures but most of them are non-fatal but contributing to perinatal morbidity (Table 8).

Though most of the anomalies are compatible with life, the increase in perinatal mortality was mainly due to associated preterm labor, prematurity, polyhydramnios, maternal diabetes and IUGR (Table 6).

Table 8: Gross distribution of anamolies.

Craniospinal	22	44
Cardiovascular	2	4
Renal	3	6
Abdominal	2	4
Musculoskeletal	15	30
Multiple congenital anamolies	6	12

Table 9: Fetal outcome.

Abortions	1	2
Preterm vaginal delivery	11	22
Term vaginal delivery	14	28
LSCS	15	30
MTP	09	18

Consanguinity is single most important factor which was found to increase the risk of congenital anomalies in our study.[22] In 40% of the cases consanguinity was noted (Table 5).

Maternal Age >30 is also the most important risk factor found to increase the risk of congenital anamoly in our study.

The fetal outcome was 80% of the babies were compatible with life and 20% were non compatible.

Social awareness about the consanguinity and if unavoidable, genetic counselling are important measures that can be done to reduce the consanguinity.

Preconceptional counselling and Supplementation of folic acid can be done to reduce the incidence of NTD.

CONCLUSION

In the present study, most of the mothers who had anomalous fetuses had risk factors like consanguinity and previous history of abortions. Hence the need for focused screening in this high risk category. Pre scan counselling with karyotyping, triple screen and relevant serology has to be done.

A level II targeted scan is done at 18-20 weeks and again at 24 weeks to exclude anomalies. A single ultrasound examination is allowed per pregnancy, the mid trimester scan at 18- 20 weeks clearly represents the best time to accomplish the most. Once an anomaly is detected, various management options are to be discussed with the patients in consultation with neonatologist, pediatric surgeon and neurosurgeon when necessary.

If parents are willing to continue the pregnancy with compatible congenital anamolies in baby then pregnancy may be continued. But if the congenital anamoly is imcompatible with life then pregnancy should be terminated. This study was conducted to study the incidence of various congenital anamolies in babies and their possible etiological factors in the population visiting to tertiary care hospital at Mumbai.

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