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Research Article

The incidence of congenital malformations at birth and mid-trimester abortions and possible etiological factors in Pune camp, Maharashtra, India

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ABSTRACT

Background: The leading cause of infant morbidity and mortality in poorer countries are malnutrition and whereas in developed countries they are accidents, cancer, and congenital malformations. The aim of the study was to find out the incidence of congenital malformations at birth and mid-trimester abortion. The objective of the study was to elucidate and evaluate the possible etiological factors in the studied group.
Methods: An Observational study was carried out at the department of obstetrics and gynaecology, Tertiary Care Hospital at Pune over a period of two years. Total of 98 mid-trimester abortions and 4450 live births were examined to look for visible congenital anomalies and associated etiological factors in studied group.
Results: The incidence of congenital malformation in mid-trimester abortions was 50% (49/98) and in live births was 1.32% (59/4450). Central nervous system (CNS) anomalies were most common in mid-trimester abortions and cardiovascular (CVS) anomalies were in live births. Possible etiological factors were the lack of folic acid supplementation and maternal diabetes.
Conclusions: CNS anomalies were most common in mid-trimester abortions and CVS anomalies were most common in live births. There is need for improve in diagnostics modalities to detect malformations at earliest and avoid morbidity and mortality burden.

Keywords: Congenital malformations, Anomalies

INTRODUCTION

Human reproduction is an inefficient enterprise. This inefficiency may relate to the complexity of the process of reproduction and manifests as pregnancy wastage. The biologist, the pediatrician, and the obstetrician are interested in studying different aspects of the multifarious problem related to human reproduction.

Major congenital malformations occur in generally around 3 to 4% of live birth, but minor anomalies are more frequent. Birth defects can be isolated or present in a characteristic combination or pattern.[1]

The leading cause of infant morbidity and mortality in poorer countries are malnutrition and whereas in developed countries they are accidents, cancer, and congenital malformations. Congenital malformations are

increasing as a result of the reduction of mortality due to other causes or due to improvement in a perinatal and neonatal period. In coming decades, this is going to be a leading cause of mortality in centers providing good neonatal care. Congenital anomalies may result in long-term disability which may have significant impacts on individuals, families, health care systems and societies

The most severe congenital anomalies are heart defects, neural tube defects, and chromosomal anomalies. Although congenital anomalies may be genetic, infectious or environmental in origin, most often it is difficult to identify the exact causes.

Keeping this in mind a study of congenital anomalies at birth occurring in the hospital over a period of two years was carried out. The primary aim of this study was to determine the incidence of congenital malformations and evaluate the possible etiological factors in malformed abortus and fetus. The aim of study is to find out incidence of congenital malformations at birth and midtrimester abortion. And objective is to elucidate and evaluate the possible etiological factors in the studied group.

METHODS

The present study was observational study carried out over a period of 2 years from 01 November 2012 to 30 September 2014 at public sector medical college in Pune. Congenital anomalies found in term births and midtrimester abortions were included in the study. Congenital anomalies in still birth were excluded from study. Total of 98 midtrimester abortions and 4450 live births were examined to look for congenital anomalies. Cases with congenital anomalies were studied in detail to find out possible etiological factors.

To cover all relevant findings, a proforma was designed. Complete maternal medical history, antenatal history, obstetric history, any exposure to drugs, radiation was revealed to find out possible etiological factors. In midtrimester abortions, congenital malformations were detected with the help of USG for anomaly scan. Examining all newborns identified congenital malformations in live births. Any admissions to NICU by neonatologist for further evaluations were followed up and cases with congenital malformations were included in my study.

All live births with malformations were evaluated as per advice given by neonatologist. Some of the cases were investigated with USG (KUB, Trans cranial, abdomen and pelvis), ECHO, Kidigram, TORCH for mother and baby, Karyotyping, Hormonal assay in case of suspected ambiguous genitalia, to find out anomalies.

RESULTS

The present study included 4548 patients those reported to the hospital over period of a period of 2 years from 01 November 2012 to 30 September 2014. The incidence of congenital malformations in midtrimester abortions was 50% (49/98) and in live births it was 1.32% (59/4450). Total malformations over a period of 2 years were 108 making incidence of 2.37%.

Table 1: Incidence of congenital malformations.

Number of abortions	Malformed abortus	Incidence
98	49	50%
Total no of live birth	**No of malformed foetus**	**Incidence**
4450	59	1.32%

Table 2 : Distribution of congenital anomalies according to age of the mother.

Age in years	Total no of abortions	Malformed abortus	Percentage of malformed abortus
<20	24	02	8.33%
20-30	50	35	70%
>30	20	12	60%
Age in years	**Total no of abortions**	**Malformed abortus**	**Percentage of malformed abortus**
<20	1060	05	0.47%
20-30	2900	40	1.37%
>30	490	14	2.85%

The distribution of congenital malformation in patients offered midtrimester abortion and those who had live birth are depicted in Table 2 below. The abortions were more common in reproductive age group in patients offered midtrimester abortion rather than extremes of age.

In our study we also studied the association of previous obstetric history and anomalous abortus and foetus. Highest number of congenital malformations were found in primigravida undergoing midtrimester abortion 19/30 (19.38%) while lowest number of congenital malformation were found in patients with previous live

birth 07/24 (7.14%). The patients who delivered a live baby had increased incidence in with at least one live birth 23/1000 (2.3%). System wise distribution of anomalies had shown that CNS was the most commonly (23.46%) affected system in midtrimester anomalies and CVS (0.44%) in live births anomalies.

Table 3: Association of previous obstetric history and anomalous abortus and foetus.

Previous obstetric history	Total no of abortions	Malformed abortuses	% normal abortuses	% abnormal abortuses
No previous pregnancy	30	19	30.61%	19.38%
Only live birth	23	13	23.46%	13.26%
Live birth and pregnancy loss	24	07	24.48%	7.14%
Only loss	21	10	21.42%	10.20%
Previous obstetric history	**Total no of Births**	**Malformed fetuses**	**% normal fetuses**	**% abnormal fetuses**
No previous pregnancy	1500	26	98.26%	1.73%
Only live birth	1000	23	97.7%	2.3%
Live birth and pregnancy loss	1200	08	99.33%	0.66%
Only loss	880	02	99.77%	0.22%

Table 4: System wise distribution of congenital anomalies.

System	Incidence in mid-trimester abortions (per 100 abortion)	Incidence in live births (per 100 abortion)
Cardiovascular	01(1.02%)	20(0.44%)
Central nervous system	23(23.46%)	06(0.134%)
Respiratory	02(2.04%)	04(0.089%)
Alimentary tract and body wall	02(2.04%)	12(0.269%)
Musculoskeletal system	05(5.10%)	04(0.089%)
Hematological	02(2.04%)	01(0.022%)
Urinary system	03(3.06%)	01(0.022%)
Genital system	-	05(0.112%)
Chromosomal	02(2.04%)	01(0.022%)
Multiple defects	09(9.18%)	03(0.067%)
EYE	-	02(0.044%)
Total	49(50%)	59(1.32%)

Table 5: Distribution of malformations in live births according to gender of fetus.

Sex of fetus	No in midtrimester abortions	No in live births
Male	25	39
Female	24	20
Total	49	59

Most common cardiac anomaly was VSD (0.134%).[2] cases of congenital heart block were found in mothers who were known case of SLE with Anti Ro and Anti La positive status among live births. Distributions of anomalies according to sex of fetus shown those male fetuses were most commonly affected than female fetus. There were many possible etiological factors, which were found in my study. In our study DM, Lack of FA intakes were the preventable factors. So many of factors are required to take into considerations to evaluate the possible etiological factors.

DISCUSSION

In this study, attempts were made to find out the incidence of congenital malformations in midtrimester abortions and live births. The overall incidence of congenital malformations in our study was 2.37%. Compares with other studies, Francine R et al showed 2.4%, Shah K et al found 2.38% Jehangir W et al observed the prevalence of 2.95% also Irvine et al.[2-5]

Maternal age is an important parameter in the birth of a congenitally malformed fetus. Age groups of 20-30 years were with 50 midtrimester abortions out of which 35 are malformed (70%). Kapoor K et al in a study with fetal autopsy found age group of 20-25 years were with

maximum numbers of anomalies.[6] Out of 4450 live births, 490 were with mother's age more than 30 years out of which 14 had congenital malformations (2.85%). Takasande A et al observed 3.60% of malformations in mother aged more than 30yrs and were comparable with the study.[7]

The study showed increased incidence malformations were noted in primigravida (19.38%) undergoing midtrimester abortion. and 7 malformations in 24 with live birth and pregnancy loss Group (7.14%). These observations of associations with first births were consistent with observations by McNeese ML et al and Csermely G et al.[8,9] Further research on specific parameters viz biological, behavioral, and environmental factors and their association with first births need further exploration.

Table 6: Possible etiological factors in malformations.

System	No of malformations in abortions and live births	Possible etiological factors
Cardiovascular	21	1) Type 2 DM 2) SLE (anti Ro and anti La antibodies) 3) Congenital heart diseases in mother 4) Advanced maternal age
Central nervous system	29	1) Lack of FA intake 2) Consanguinity 3) H/o febrile illness in 1st trimester 4) ART procedure(Post FET,) 5) Overt DM 6) Previous baby affected with anomaly
Respiratory	06	Nil
Alimentary tract and body wall	14	1) Advanced maternal age 2) Polyhydramnios 3) ART procedure(Post IVFET)
Musculoskeletal system	09	1) Consanguinity-Rhizomelia 2) DMD- a)mother carrier, b) previous baby affected 3) Oligohydramnios
Hematological	03	1) Husband and wife B thalassemia minor 2) Mother B thalassemia heterozygous
Urinary system	03	Nil
Chromosomal	03	Advanced maternal age
Genital system	05	1) ART procedure (Post IVF ET) 2) Warfarin intake by mother
Urinary system	01	Nil
Eye	02	Nil
Multiple defects	03	Advanced maternal age

In the present study in midtrimester abortions, the most common system with congenital malformations was CNS (23.46%) followed by multiple defects (9.18%) followed by the musculoskeletal system (5.10%) and other systems as mentioned above. The most common involved system in the live births was CVS (0.44%) followed by alimentary tract and body wall (0.269%), this was followed by CNS (0.134%) and other systems as mentioned above.

Congenital malformations were more common in the male. In midtrimester abortions; 25 male abortus versus 24 female abortus and in live births 39 male versus 20 females were noted. No abortus or fetus with ambiguous genitalia was seen in the present study. Shah K et al and Mohanty et al had also shown the similar findings in their research.[3,10]

In the present study 6/108 (5.55%) cases of congenital malformations were associated with single umbilical artery. A case of Rh isoimmunisation (A negative) was noted with a fetus affected with hypospadias.

History of consanguinity was observed in 5/108 (4.62%) mothers in our study, out of which four cases were in midtrimester abortions. Those were distributed like rhizomelia, B thalassemia major, anencephaly and one had multiple defects. One of the live births with NTD had a history of consanguinity.

History of a previous fetus affected with congenital malformations was seen in 7/108 (6.48%). Assisted reproductive techniques resulted in 4/108 (3.70%) cases in the present study with congenital malformations. CNS malformations were seen in 5/108 (4.62%) cases of congenital malformations in an absence of prophylactic folic acid supplementation. Febrile illness in the first trimester resulted in 3/108 (2.77%) cases of congenital malformations. In the present study 2/108 (1.85%) cases of congenital malformations were found in mothers with overt diabetes mellitus, which included one case of exencephaly in midtrimester abortions group and one case of PDA+VSD in the live birth group.

Two cases of renal agenesis (1.85%) were seen with oligohydramnios and pregnancy terminated in midtrimester abortion. One (0.92%) case of a mother with twin gestation with oligohydramnios had CTEV. 3/108 (2.77%) cases of congenital malformations like duodenal atresia, oesophageal atresia with or without tracheal stenosis detected to have polyhydramnios.

A case of hypospadias was seen in mother taking anticoagulation prophylaxis with warfarin for past history of dural venous thrombosis was found. A case of congenital ASD was seen in a baby whose mother also had congenital ASD. In the present study 2/108 (1.85%) cases of congenital malformations were found in the mothers with SLE. Both the fetus affected with congenital heart block and their mothers were positive for Anti RO and Anti LA antibodies. Amniocentesis helped to reveal two cases of B thalassemia major where both parents were B thalassemia minor with past history of the previous fetus affected with B thalassemia major.

CONCLUSION

The present study educated us regarding distribution and incidence of congenital anomalies. It also amplified its relation with associated maternal and fetal factors. The study showed that the incidence of congenital anomalies of central nervous system was highest compared to other types of congenital anomalies (of which neural tube defects being the commonest).

Various possible etiological factors in our study were consanguinity, the previous baby affected with congenital malformations, ART procedure, lack of prophylactic folic acid supplementation, febrile illness in the first trimester, overt diabetes mellitus, oligohydramnios, polyhydramnios, teratogenesis, congenital heart diseases in the mother, twin gestation, SLE and both parents B thalassemia minor.

More stress should be laid on prevention by regular antenatal care. During preconception and in pregnancy the probable teratogenic agents should be avoided. Antenatal diagnosis, genetic counseling, better diagnostic and management facilities should be provided to improve the outcome. There is a need for improving in diagnostics modalities to detect malformations at earliest and avoid morbidity and mortality burden of the state.

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