

A Study of Lethal Congenital Anomalies Undergoing Termination of Pregnancy at a Tertiary Care Hospital

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Abstract

Background: India does not have national birth defects surveillance in spite of congenital anomalies causing significant neonatal and foetal mortality. There is paucity of data regarding lethal congenital anomalies which are mostly diagnosed late in pregnancy making their termination risky to pregnant women.

Aims and objectives: This study was conducted in pregnant women who underwent termination of pregnancy, for lethal congenital anomaly in foetus, to find their demographic and clinical profile, types of congenital malformations, risk factors and methods of termination.

Material and methods: This was a retrospective hospital based study of women undergoing MTP for lethal anomalies in foetus at Medciti institute of medical sciences from October 2020 to October 2021. Data about women's age, gravidity, occupation, consanguinity, periconceptional folic acid, previous obstetric outcome, gestational age at diagnosis, type of congenital anomaly in foetus, lactational status in pregnant women, risk factors in pregnancy, foetal sex, last child birth time period and MTP method was recorded. Data were analysed and frequencies and percentages presented in descriptive statistics.

Results: Majority (44%) of women were 21-25 years age. 62% were housewives 48% women were 2nd gravida. 28.56% had abortion in previous pregnancies. 64% anomalous foetuses were female. 86% anomalies were detected from 16-20 weeks' gestation. Consanguinity was noted in 20% women. Fever in first trimester in 80%, urinary tract infections in 1st trimester in 86%, hypertension in 8%, hypothyroidism in 8%, anaemia in 4% and gestational diabetes in 6%. 97.22% parous women continued breastfeeding in pregnancy and 66.66% had last child birth between 7-9 months. 96% did not take periconceptional folic acid. Majority (52%) were CNS anomalies. Anencephaly was commonest. All pregnancies were terminated medially.

Conclusion: There is need for better health facilities including periconceptional care, educating women on birth spacing, early pregnancy booking and ultrasound. Folic acid food fortification is good option.

Keywords: Lethal congenital anomaly, MTP, Risk factors.

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Introduction

Birth defects are also known as congenital abnormalities, congenital disorders or congenital malformations. They can be defined as structural or functional anomalies (for example, metabolic disorders) that occur during intrauterine life. An estimated 240 000 new-borns die worldwide within 28 days of birth every year due to birth defects. [1] According to Rashtriya Bal Swasthya Karyakram, India with a birth cohort of 26 million annually accounts for highest share of congenital birth defects globally. Here, congenital birth defect mortality (8.6%) ranked as the fourth leading cause of neonatal death. [2] Lethal congenital malformations are fatal birth defects that are an important cause of fetal/neonatal death. There is a lack of informative data about these malformations in India. Lethal congenital anomalies diagnosed in late fetal life still pose a management dilemma for healthcare professionals regarding the fetus as well as the mother. [3] These serious birth defects require termination of pregnancy. While pregnancy is considered a forward development for the mother and the family, diagnosis of foetal abnormalities is an unexpected incident during this period and is associated with severe emotional injuries for women. [4] Women undergoing second trimester abortion are at risk for significant morbidity which increase with gestational age at abortion. Although birth defects may be the result of one or more genetic, infectious, nutritional or environmental factors, it is often difficult to identify the exact causes.[1] The process of induced abortions due to fetal anomalies demands accurate and immediate diagnostic examinations. A structured investigation process is needed to allow the patient

decision time and to minimize terminations near the legal limit. [5]

Aims and Objectives:

The study is undertaken to know the demographic and clinical profile of women with lethal congenital malformation who underwent termination of pregnancy and secondary objective is to find out types of congenital malformations, risk factors associated and methods of termination.

Material and methods:

This was a retrospective study based on hospital records conducted at a rural tertiary care centre. The case records of women who underwent termination of pregnancy for lethal anomalies at Mediciti institute of medical sciences over a period of one year that is from October 2020 to October 2021 were included in the study. We considered those malformations as lethal congenital anomalies which had a high possibility of foetal or neonatal death. Necessary permissions for retrieval of records were obtained. We retrieved the data about various factors which included mother's age, mother's gravidity, mother's occupation, consanguineous marriage, periconceptional folic acid intake, previous obstetric outcome, gestational age at diagnosis, type of congenital anomaly, lactational status in pregnancy, risk factors in pregnancy, gender of the foetus, time interval from last child birth and method of termination.

Ultrasonography can identify at least 35 - 50% of major foetal malformations with a specificity of 90 to 100%. Though other methods of screening like biochemical markers and karyotyping are available, ultrasonography has the advantage of being

non-invasive, safe, fast, accurate and reproducible with real time display, causing no discomfort to the patient at any time of gestation [6]

Hence prenatal diagnosis in these cases was done by USG and all anomalies diagnosed in other centre were confirmed by obstetrician and or radiologist of our hospital. Termination was done by methods suitable for the respective gestational age.

Data were analysed and frequencies and percentages were presented in descriptive statistics.

Results:

The total no. of medical records, of cases which underwent termination for lethal congenital anomaly ,retrieved was 50.

Table 1: Distribution of congenital anomalies with age of mother

Age group in years	Number	Percentage (%)
18-20	7	14%
21-25	22	44%
26-30	13	26%
31-35	7	14%
36 and above	1	2%
TOTAL	50	100%

Table 1 shows the age of the women ranged from 18-37 yrs. Mean age was 25 years and majority(44%) were in 21-25 years age group.

Table 2: Distribution of congenital anomalies with patient's occupation

Occupation	Number	Percentage (%)
Housewife	31	62%
Farmer	9	18%
Laborer	10	20%
Total	50	100%

Table 2 shows that among the total women, 31(62%) were housewives, 10(20%)women were labourers and 9(18%) were farmers.

Table 3: Distribution of congenital anomalies with antenatal Care

Antenatal care	Number	Percentage (%)
Booked	11	22%
Unbooked	39	78%
Total	50	100%

Table 3 shows most of these women (78%) were unbooked .They were referred from other centers. Only 11(22%) women were booked.

Table 4: Distribution of congenital anomalies with obstetric score (gravidity)

Gravida	Number	Percentage (%)
Primi	8	16%
G2	24	48%
G3	14	28%
G4	3	6%
G5	1	2%
Total	50	100%

Table 4 shows 24 (48%) of the women were 2nd gravida followed by 14 (28%) women who were 3rd gravida. 8 (16%) women were primigravida, 3(6%) were 4th gravida and only 1 woman was 5th gravida.

Table 5: Previous Pregnancy Outcome

Outcome	Number	Percentage (%)
Previous normal babies	30	71.42%
Previous spontaneous abortion	6	14.28%
Previous missed abortion	6	14.28%
Previous pregnancy with anomaly	0	0%
Total	42	100%

Table 5 shows among the multigravida women, 30(71.42%) of women had normal babies in previous pregnancies. 12(28.56%) of women had abortion in previous pregnancies. None of the women had anomalous babies in the previous pregnancies.

Table 6: Distribution of anomalous foetuses with sex of fetus

Sex of fetus	Number	Percentage
Male	18	36%
Female	32	64%
Total	50	100%

Table 6 shows of the total 50 anomalous foetuses, 32(64%) were female and 18(36%) were male

Table 7: Gestational age at which anomaly was detected in the study subjects

Weeks of gestation	Number	Percentage
<16 weeks	7	14%
>16	43	86%
Total	50	100

Table 7 shows maximum no of anomalies 43 (86%) were detected late , that is after 16 weeks. Only 7(14%) were detected between 12 to 16 weeks.

Table 8: Risk factors among subjects

Risk factor	Number	Percentage (%)
Consanguineous marriage	10	20%
Fever in 1 st trimester	40	80%
UTI in 1 st trimester	43	86%
Hypertension	4	8%
Anemia	2	4%
Gestational diabetes	3	6%
Hypothyroidism	4	8%

Table 8 shows the risk factors which were documented.

History of consanguinity was noted in 20% of women. Fever in first trimester was present in 80% of women, Urinary tract

infections in 1st trimester was present in 86%. Other medical disorders like hypertension was found in 8% of women, hypothyroidism was noted in 8% of women, anaemia in 4% and gestational diabetes was found in 6%.

Table 9: Distribution of parous women among the subjects with last child birth

Last childbirth in months	Number	Percentage
7-9	24	66.66%
10-12	8	22.22%
13-14	3	8.33%
15-19	1	2.77%
20 and more	Nil	0%
Total	36	100%

Table 9 shows among the total subjects, 36 were parous women. All these parous women had last child birth less than 2 yrs. 24(66.66%) had last child birth between 7-9 months which shows a very short

interpregnancy interval (interval between delivery and conception of the subsequent pregnancy) in them. 8(22.22%) women had last child birth between 10-12 months

Table 10: Distribution of the subjects with lactational status

Breast feeding the previous child	Number	Percentage (%)
Yes	35	97.22%
No	1	2.78%
Total	36	100%

Table 10 shows that among the total subjects who were parous, 35(97.22%) were continuing breastfeeding their previous child in the present pregnancy.

Table 11: Distribution of women with folic acid prophylaxis

Folic acid prophylaxis	Number	Percentage
Taken	2	4%
Not taken	48	96%
Total	50	100%

Table 11 shows only 2(4%) women took periconceptual folic acid and 48(96%) of women did not take.

Table 12: System affected among the subjects

System affected	Number	Percentage
CNS	26	52%
CVS	5	10%
Genitourinary System	4	8%
GIT	1	2%
Multiple	14	28%
Total	50	100%

Table 12 shows majority 26(52%) of the anomalies were involving the central nervous system. In 5(10%) fetuses, the anomalies were involving cardiovascular system and in 4(8%) genitourinary system. Only 1 fetus had isolated gastrointestinal anomaly. In 14(28%) fetuses the anomalies were involving multiple systems

Table 13: Distribution of the subjects with the type of anomaly

Anomaly	Number of subjects (singly occurring)	Number of subjects (occurring along with other anomalies)
Anencephaly	8	3
Lissencephaly	4	1
Exencephaly	1	-
Semilobarholoprocencephaly	1	-
Arnold chairi malformation	4	1
Encephalocoele	1	-
Macrocephaly	-	1
Aqueductal stenosis	2	1
Spina bifida	3	4
AV Malformation	-	4
TAPVC	3	-
Absent left atrium	1	-
Absent right atrium	1	-
Aplastic right heart	-	1
AV septal defect	-	1
Omphalocoele	1	-
Duodenal atresia	-	2
Esophageal atresia	-	3
Skeletal dysplasia	-	3
Right radial ray aplasia	-	1
Right renal aplasia	-	1
Absent nasal bone	-	1
Dysplastic kidney	2	7
Polycystic kidney disease	2	1

Table 13 shows the different anomalies that were seen that were found as isolated anomalies or as a part of multisystem anomalies. Anencephaly was seen most commonly, in 8 cases as isolated anomaly and in 3 cases along with other anomalies. Lissencephaly, Spina bifida, Arnold Chairi malformation, AV malformation, aqueductal stenosis were other common CNS anomalies occurring singly or along with other system anomalies. Total Anomalous Pulmonary Venous Connection was commonest in CVS anomalies. Absent right heart, Absent left heart and Right heart aplasia and Atrioventricular septal defect were other CVS anomalies occurring singly

or with other anomalies. Omphalocoele was the only anomaly of gastrointestinal system which occurred alone. Duodenal atresia and Oesophageal atresia occurred along with other lethal anomalies (dysplastic kidney and skeletal dysplasia, polycystic kidney disease). Polycystic kidney disease and Dysplastic kidney were the anomalies observed in genitourinary system. The anomalies of musculoskeletal system seen were Skeletal dysplasia and Radial ray aplasia present along with lethal anomalies of other systems. 1 case of Absent nasal bone was seen along with Dysplastic kidney.

Table 14: Mode of delivery in previous pregnancies

Mode of delivery	Number	Percentage
No previous delivery (Primigravida and G2A1)	14	28%
All vaginal deliveries	24	48%
1 previous lscs	7	14%
2 previous lscs	5	10%
Total	50	100%

Among the women, previous mode of delivery was also documented to assess the risk involved in termination and to decide on the method of termination. Table 14 shows that of the total women, 24 (48%) had previous vaginal deliveries, 7 (14%) had 1 previous LSCS, 5(10%) had 2 previous LSCS and the rest of them ,14 (28%)had not undergone any deliveries before.

Table 15: Method of MTP used to terminate the anomaly

Mifepristone 200 mg 24 hr. apart	Misoprostol 50 micrograms 6 hrs. apart	Number of cases
1 dose	2 doses	22
1 dose	3 doses	9
1 dose	4 doses	7
2 doses	2 doses	9
0	4 doses	1
2 doses	0	1
1 dose	0	1

Table 15 shows that all the pregnancies could be terminated medially using mifepristone followed by misoprostol after 24 hrs. However the no. of doses required for each case varied. Risk consent was taken prior to the procedure. Maximum no. of cases could be terminated with 1 dose of Mifepristone followed after 24 hrs. with 2 doses of misoprostol 6 hrs. apart.

Discussion:

In this study majority (44%) of women were in younger ,21-25 years age group. In a similar study, Papa Dasari et. al., [7] reported that 41% of subjects were in the age group of 21-25 years. Regarding occupation of the subjects in our study, among the total women, 31(62%) were housewives.10(20%) women were labourers and 9(18%) were farmers. In an observational study of profile of neonates born with congenital birth defects in a tertiary care hospital Abhilash Sinha et. al., [2] reported 76.1% mothers were

housewives, 21.7% were doing skilled job and 2.2% were labourers. Patients attending to our hospital are mostly from rural background which probably explains the 18% of women being farmers. Majority 39(78%) of the women in this study were unbooked. In a study of congenital anomalies of fetus by Kanhere AV et. al., [8] 65% of women were unbooked.

In our study most women were multipara. Of that 24 (48%) of the women were 2nd gravida followed by 14 (28%) women who were 3rd gravida. A similar study by Tiwari et. al.,[3] showed 59.21% of the subjects as multigravida whereas 40.79% were primigravidas. In the study done by Kanhare AV et. al., [8] multigravidas were 62% whereas primigravidas were 38%. In a study done on major congenital anomalies, Pusayapaibul et. al., [9] also documented 60.1% women as multigravidas and 39.9% as primigravidas. Among the multigravida women in our study, 30(71.42%) of women had normal babies in previous pregnancies.

12(28.56%) of women had abortion in previous pregnancies. In the study done by Papa Dasari et. al.[7],20.5% of women had history of previous abortions. In the study done by Pusayapaibul et. al. [9] also previous history of abortions was seen in 26.3 %of women.

Maximum no of anomalies 43 (86%) in our study were detected late, that is after 16 weeks. Only 7(14%) were detected between 12 to 16 weeks. In the study done by Papa Dasari et. al.,[7] 78% of the major congenital anomalies which underwent termination of pregnancy were detected between 16 to 20 week. In a study conducted by Kapoor K et. al., [10] when the gestational age was taken into account, the presentation and subsequent foetal loss (either by spontaneous or induced abortions) occurred maximum (41%) in >15-20 wks.

Some of the risk factors were documented in our study. History of consanguinity was noted in 20% of women in this study. High statistical significant association between consanguinity in the study participants and presence of congenital fetal malformations in second trimester was shown by Ali El-Shabrawy Ali et. al. [6] Papa Dasari et. al. [7] documented 27.35% association of congenital anomalies with consanguinity in the women in their study

Fever in first trimester was present in 80% of women in our study. Waller et. al., [11] in their study found that maternal report of cold or flu with fever was significantly associated with 8 birth defects (anencephaly, spina bifida, encephalocele, cleft lip with or without cleft palate, colonic atresia/stenosis, bilateral renal agenesis/hypoplasia, limb reduction defects, and gastroschisis) with elevated adjusted odds ratios ranging from 1.2 to 3.7. Palmer et. al., [12] also documented in his prospective cross-sectional study on congenital anomalies that fever in the 1st trimester is the commonest among the etiological factor found. However, they

found etiological factor in only 12% of subjects in their study.

Urinary tract infections in 1st trimester was present in 86% in our study. The journal *Birth Defects Research (Part A)* has published a study looking at the relationship between reported antibiotic use among women with kidney, bladder, or just urinary tract infections (all referred to as UTIs) just before or during the first trimester of pregnancy and birth defects. The findings showed that women who used the antibiotics trimethoprim-sulfamethoxazole, nitrofurantoin, or cephalosporin were at higher risk for one or more major birth defects, as compared to women who only took penicillin [13].

Gestational hypertension was found in 8% of women. Hypothyroidism was noted in 8% of women, anaemia in 2 4%) and gestational diabetes was found in 6%. These risk factors were less commonly present when compared to other studies. [2,7]

Of the total 50 anomalous fetuses, 32(64%) were female and 18(36%) were male. Incidences of congenital malformation were slightly more in female with F: M ratio 1.6: 1 in Palmer et. al., [7] Many studies like Tiwari et. al., [3] and Kumbhar S. et. al., [14] have documented male preponderance amongst congenital malformed babies.

All these parous women in our study had last child birth less than 2 yrs. 24(66.66%)had last child birth between 7-9 months which shows a very short interpregnancy interval (interval between delivery and conception of the subsequent pregnancy) in them. There was no documentation of interpregnancy interval in most of studies on congenital anomalies. In one study by Peterson et al., [15] short interpregnancy intervals were associated with a trend of higher risks for several defects, notably in the absence of folic acid supplement use. _ORs for <6 compared with

18-23 mo. were >1.2 for 4/8 noncardiac and 3/6 cardiac malformations.

Among the total subjects who were parous, 35(97.22%) were continuing breastfeeding their previous child in the present pregnancy. This factor was also not looked into in many similar studies. Among few studies which studied effects of breastfeeding while being pregnant, Shahan et. al., [16] showed increased risk low birth weight, anaemia, IUGR, increased caesarean section and prolonged labour. Molitoris. [17] showed increased risk of miscarriage. However risk of birth defects was not studied.

Only 2(4%) women in our study took periconceptual folic acid and 48(96%) of women did not take. Many studies documented this as a risk factor for congenital anomalies in the fetus. In the study done by Dasari et. al., [7] 92.24%, in study by Tiwari et. al., [3] 66% women did not take periconceptual folic acid. 40% of NTDs can be prevented with preconception folic acid supplementation, the intervention would reduce about 30 000 affected births in India, considering complete compliance. [18] Since anencephaly and spina bifida together form a significant percentage of anomalies in the present study, the observation of lack of periconceptual folic acid in women is an important risk factor for the anomalies.

In the present study, the most common system with congenital malformations was CNS, 52% occurring alone and CNS was also commonly involved in multiple system anomalies. CNS was most commonly involved in majority of the studies like, Dasari et. al., [7] (55.5%), Bhide P et. al. [19], (75.85%), Palmer et. al., [12] (64.56%). Kapoor.K.et.al., [10] (32.6%). Anencephaly was most commonest anomaly in present study and this finding was consistent with most studies. The other anomalies which involved CNS alone were lissencephaly, spina bifida, Arnold Chiari malformation, aqueductal stenosis, exencephaly, encephalocoele and

semilobarholoprocencephaly.

Anencephaly, lissencephaly, Arnold Chiari malformation, macrocephaly, aqueductal stenosis, spina bifida and AV malformations were seen in the multisystem anomalies. Next common single system involved was cardiovascular system (10%). Total anomalous pulmonary venous connection was the commonest among these. Others were absent left ventricle, absent right ventricle, atrioventricular septal defect and right heart aplasia. Genitourinary system (8%) was the next common system which was involved comprising of polycystic kidney disease and dysplastic kidney and right renal aplasia. However these anomalies were a part of anomalies involving multiple systems more than occurring singly. Omphalocele was the only gastrointestinal system anomaly which occurred in single system anomaly. Esophageal atresia and duodenal atresia were a part of multiple system anomalies which were lethal like skeletal dysplasia, dysplastic kidney and polycystic kidney disease. Anomalies involving musculoskeletal system, skeletal dysplasia and radial ray aplasia were present only as a part of multisystem disorder and did not occur singly. One case of absent nasal septum was seen which was again along with lethal anomaly, dysplastic kidney. Cases involving more than one system had one or more lethal anomalies.

In the present study about 24% of the cases had a history of previous LSCS which actually increases the risk involved in the termination of a second trimester abortion especially in cases with 2 previous LSCS as shown by a study by Frick et. al., [20]

All the cases could be terminated by medical methods in the present study using Mifepristone and/or Misoprostol in 100%. In the study done by Dasari et. al., [7] also all the cases were terminated by medical methods using mostly misoprostol and/or mifepristone in 50% of cases whereas other medical methods were used for rest of cases.

As the studies in the literature evaluating factors particularly for lethal congenital anomalies are less, our results were compared in some instances with other studies that evaluated all the congenital malformations. This may be a limitation of this study.

Conclusion:

In this study of lethal anomalies in second trimester terminations of pregnancy, CNS was most commonly involved. The anomalies which could be prevented by folic acid, Anencephaly and spina bifida summed up to 22% of anomalies in women when single system was involved and as a part of multisystem anomalies in 14%. Most of women were unbooked, lacking good antenatal care due to which they could not get folic acid prophylaxis and also the advantage of an early USG. Most of the women in the present study had short interpregnancy interval. Most of the women conceived when they were breastfeeding the previous child. This must have caused the delayed diagnosis of pregnancy which was probably mistaken as ongoing lactational amenorrhoea by the women resulting in late booking for antenatal care. Further studies may be needed to look into other factors, such as unmet nutritional demands, as the cause of increased risk of congenital anomalies in breastfeeding pregnant women and women with short interpregnancy intervals. This study found that the major or lethal congenital anomalies which required termination of pregnancy were detected mostly in advanced weeks increasing the risk to mother due to the procedure. Consanguinity was documented as risk factor in 20% of women. Medical disorders, diabetes, hypertension and hypothyroidism risk factor in only few women. Fever in first trimester found in most of the women as risk factor needs further research with prospective studies to evaluate its association with congenital anomalies. Urinary tract infection in first trimester documented in majority of women calls for

the review of antibiotics used to treat this condition regarding their probable teratogenicity. There is a need for better health care facilities including proper preconceptional care and counselling, educating women regarding importance of birth spacing and early ultrasound. Folic acid food fortification as suggested by many studies should be considered by the health policy holders.

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