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A study on fetal congenital anomalies and methods of termnation

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Abstract

Background: Congenital malformations are on rise in the current transition of the world from communicable to noncommunicable , chronic , genetic diseases. Congenital malformations form the fifth major cause of neonatal deaths in India . Most of the data is underestimated as the reports are mostly from hospital based studies only. With emerging and widespread use of prenatal USG as a diagnostic tool in Obstetrics,detection of fetal anomalies has become easy. Various methods of termination of pregnancy have been described for fetal anomalies in the 1st and 2nd trimesters.**Aims & Objectives** : The aim of the present study was to estimate the prevalence ,patterns , risk factors , distribution of various congenital malformations and to compare various methods of pregnancy termination.**Materials &Methods :**This was a cross-sectional study conducted in a tertiary care hospital over a period of 5 years i.e., from January 2016 till October 2020.**Results:** Out of 6400 deliveries, 100 cases of major congenital anomalies were noted,making a prevalence of 1.56%. CNS anomalies were most commonly reported. Most of the cases were reported in 2nd trimester. Most common method of termination was medical, using Mifepristone and Misoprostol.**Conclusion:** To conclude, the prevalence congenital anomlies is 1.56% . Of the congenital anomalies noted, CNS is the most effected system of which anencephaly is most frequently reported. Termination of pregnancy with T.Mifepristone & T.Misoprostol appears to give best results .The incidence may be reduced by increased awareness & proper counselling and surveillance.
Keywords: Congenital Anomolies, Termination Of Pregnancy.

Introduction

Congenital anomalies are also known as birth defects or congenital malformations.[1] According to WHO, congenital anomalies include structural and functional anomalies that occur during intrauterine life. They contribute to long term disability which has impact on families & societies. In global burden of disease study 2013 congenital anomalies are included in top 10 causes for perinatal mortality.[2] The incidence worldwide and in India is around 2-3 % . Most of the congenital anomalies are not compatible with life. Congenital anomalies involving the Central nervous system (CNS) seeking termination of pregnancy are the largest group at 10 per 1000 live births, followed by Cardiovascular system (CVS) 8 per 1000, Renal 4 per 1000 live births& limbs 1 per 1000 live births[3].The etiology of malformations is not known in 60% of cases. 20- 25 % occurs due to variety of causes such as genetic, infections, metabolic diseases, drugs, environmental factors.[4] Genetic causes include numerical, structural defects and mosaicism. Structural chromosomal defects include deletions, translocations & inversions.Sex linked chromosomal abnormalities are Turner's syndrome, Klinefelter's syndrome. Infections which could cause congenital anomalies are Rubella, Herpes, Cytomegalovirus, Toxoplasmosis, Syphilis. Increased use of irradiation, alkylating agents, antimetabolites,

smoking, alcohol & exposure to pesticides are known to cause congenital anomalies.[5] Medical disorders known to cause congenital anomalies are uncontrolled diabetes, hypothyroidism. Exposure to drugs such as anti-epileptics, warfarin, thalidomide, retinoic acid might increase the risk. The incidence of congenital anomalies can be reduced by preventive strategies like avoidance of exposure to radiation, antimetabolites, smoking, alcohol, Pre-conception& antenatal folic acid supplementation.Perinatal mortality and morbidity decrease to a marked extent by termination of pregnancy diagnosed to have congenital anomalies.[6]Advances in the prenatal diagnostic testing helps in detection of anomalies earlier in pregnancy leading to early intervention.

Materials and methods

This study was conducted in department of Obstetrics and Gynaecology, NRI medical college, Chinakakani, a tertiary care hospital in Andhra Pradesh, from January 2016 till October 2020.

Inclusion criteria

 Antenatal women with congenital anomalies to the foetus registered or referred to department of OBG at NRIGH were included. The data was collected on variables such as age, parity, gestational age, consanguinity, recurrent abortions,maternal infections, folic acid intake, obesity,history of previous anomalies, family history of congenital anomalies, any medical disorders such as diabetes, thyroid disease.

Results

100 cases of major congenital anomalies were noted out of 6400 deliveries. The prevalence of 1 .56% was noted.

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Table 1 : Age Wise Distribution

Maternal age in years	No. of cases
< 20 years	27
21- 25 years	41
26 – 35 years	25
36-40 years	7

Table 2: Distribution According To Parity of The Women

Parity Of Women	No . Of Cases
Primigravida	46
Multigravida	54

Table 3:Distribution according to gestational age

Gestational Age	No of cases
1st Trimester	7
2nd Trimester	86
3rd Trimester	7

Table 4:Consanguinity history distribution

Consangunity	No. Of Cases
Consanguinous	32
Non- Consanguinous	68

Table 5: Sex of The Fetus

Sex Of The Fetus	No. Of Cases
Male	36
Female	45
Ambiguous	19

Table 6: Weight of The Fetus

Weight of the fetus	NO. OF CASES
< 500 GMS	58
500 GMS-1KG	40
>1KG	2

Table 7: Risk Factors

RISK FACTORS	NO. OF CASES
BMI>25	88
Overt Diabetes	3
Gestational Diabetes Mellitus	8
Hypothyroidism	15
Hyperthyroidism	5
Family H/O Congenital Anomalies	12
Previous H/O Congenital Anomalies	13
Drug Usage (Anti Epileptics)	8
H/O Maternal Infection	8

Table 8: Method of Termination

Method Of Termination	NO. OF CASES
Medical (Mifepristone + Misoprostol)	65
Mifepristone Alone	3
Misoprostal Alone	2
Mechanical Dilation With Foley's Catheter	5
Extra-Amniotic Ethacridine Lactate	15
Dinoprostone Gel	5
Extra Amniotic Saline Infusion	3
Hysterotomy	1

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Table 9: Various Anomalies Detected During Study

Spectrum Of Anomalies	No.Of Cases
Central Nervous System	51
Cardiovascular System	26
Genito-Urinary System	11
Musculoskeletal System	13
Facial Anomalies	6
Abdominal Wall Defects	5
Gastro-Intestinal System	4
Respiratory System	2
Down`S Syndrome	4
Hydrops Fetalis	5
Beta-Thalassemia	3
Amniotic Fluid Abnormalities	8

Table 10: CNS Anomalies

CNS Anomalies	NO.OF CASES
Anencephaly	14
Spina Bifida	3
Ventriculomegaly	3
Dandy Walker Malformation	3
Microcephaly	2
Cystic Hygroma	4
Arnold Chiari Malformation	10
Hydrocephalus	2
Thanatotropic Dysplasia	2
Choroid Plexus Cyst	2
Spinal Dysraphism	4
Encephalocele+ Meningocele	2

Table 11: CVS Anomalies

CVS Anomaly	NO.OF CASES
Ventricular Hypoplasia	10
VSD (Ventricular Septal Defect)	8
ASD (Atrial Septal Defect)	1
TGA (Transposition Of Great Vessels)	3
TOF (Tetralogy Of Fallot)	1
Echogenic focus of ventricles	3
PDA (Patent Ductus Arteriosus)	0

Table 12: Renal anomalies

Renal Anomaly	NO OF CASES
Renal Agenesis	8
Polycystic Kidney Disease	3

Table 13: Musculoskeletal   Anomalies

Musculoskeletal Anomaly	NO OF CASES
Club Foot/Foot Deformity	3
Kyphoscoliosis	5
Sacrococygeal Teratoma	2
Muscular Dystrophy	3

Table 14: Facial Deformities

Deformity	NO OF CASES
Cleft Lip Alone	3
Cleft Palate Alone	0
Both Cleft Lip And Palate	3

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Table 15: Abdominal Wall Defects

Abdominal Wall Defects	No Of Cases
Gastroschisis	2
Omphalocele	3

Table 16: Digestive System

Digestive System	No Of Cases
Duodenal Atresia	3
Oesophageal Atresia	1

Table 17: Other Anomalies

Anomaly	No of cases
Diaphramatic Hernia	2
Down's Syndrome	4
Hydrops Fetalis	5
Beta Thalassemia	3
Anhydramnios	8

Discussion

Congenital anomalies are known to be important cause of still births, perinatal mortality and childhood morbidity. The prevalence of congenital anomalies in our study was 1.56% which is on par with the worldwide prevalence of 2-3 %. The occurrence of the congenital anomalies cannot be completely prevented, but due to the advancing technology and development of high-resolution ultrasound and increased expertise the detection rate is increased.[7]This has been beneficial to detect the anomalies at the gestational age where intervention can be done. Increased risk is noted in women with advanced maternal age.[8]Sugunabai et al [9]in their study quoted that maternal age over 35 years has got higher incidence of congenital malformations, but in our study pregnant women who were above 35 years were very few to come to a conclusion.Most of the patients registered belong to the age group of 21-25 years and more anomalies were noted in that age group. Higher incidence of congenital anomalies were noted in multigravida than that of primigravida which is comparable to the study by Mohanty C et al. [10]

The most common congenital anomalies noted in termination of pregnancy group belong to the CNS anomalies i.e, 51 % , followed by cardiovascular system 26% , musculoskeletal system 13% , Renal 11% , Facial 6 % , Gastrointestinal tract 4% , Respiratory system 2% , Hydrops 5 % . The 45 th annual report of Indian council of medical research, 2002-03 quotes that the most common cause of congenital anomalies are cardiovascular which is contrast to our study as the inclusion group includes patients registered for termination of pregnancy.The most common CNS anomaly detected in our study was anencephaly 14% which is comparable to study by Mohanty C et al.[10] The 2nd most common CNS anomaly was Arnold Chiari malformation, i.e., 10 %.In the CVS group, Ventricular hypoplasia was most commonly noted, we got a prevalence of 26 % which correlates with study conducted by Comstock CH [11]. They also quoted that sensitivity of ultrasound screening plays a role in detection of the cardiovascular anomalies. The most difficult defect to identify were isolated septal defects. The prevalence of Renal agenesis in genitourinary system was 11 %. The musculoskeletal anomalies detected were kyphosis, clubfoot, sacrococcygeal teratoma, muscular dystrophy. Facial anomalies detected were cleft lip , micrognathia; both cleft lip and palate together. Position of the fetus during USG examination, obesity, oligohydramnios may interfere with proper visualisation. W. Berge ST el al, in his study quoted that 3D USG is effective in the diagnosis of cleft lip and cleft palate. [12]The prevalence of congenital anomalies was more in female fetuses compared to male fetuses in our study,which was in contrary to the study conducted by Lavanya S et al[1].In our study the incidence of congenital anomalies is more in women with BMI>25 which is comparable to study conducted by J Rankin et al. [13]In our study, positive association was seen in congenital anomalies and maternal thyroid abnormalities.This was comparable to study conducted by Khoury MJ et al.[14]Most commonly used method of termination was combination of Mifepristone & Misoprostol. It was used in 39 women and is found to show less number of post abortal complications, reduced hospital stay which is comparable to study conducted by J E Dickinson et al.[15] Medical termination was complete in most of them with only one patient requiring hysterotomy. The different medical methods used were combination of Mifepristone + Misoprostol , Mifepristone alone , Misoprostol alone, Dinoprostone gel , Extra-amniotic Ethacridine lactate solution topped with Mifepristone, Extra-amniotic saline infusion, Mechanical dilatation using Foley's catheter.

Conclusion

Morbidity due to congenital anomalies can be minimised but not totally preventable by early diagnosis on USG and by proper intervention. The incidence may be reduced by pre-conception counselling, avoidance of exposure to risk factors, folic acid supplementation, prompt treatment of medical disorders like Diabetes mellitus. Early detection of anomalies helps in planning of termination of pregnancy earlier with less morbidity. Termination of pregnancy by combination of Mifepristone and Misoprostol shows best results.

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