Original Article

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Frequency of Foetal Anomalies in a Tertiary Care Centre

Anatomy Section

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ABSTRACT

Objective: The present study was undertaken to explore the incidence of congenital foetal anomalies and the advantages of ultrasonography in detecting the foetal anomalies during the antenatal period.

Method: We focused our study on 1000 consecutive pregnancies that came for check up in the second and third trimesters, with major or minor clinically relevant malformations which were detectable by ultrasonography.

Results: The analysis revealed that they were 38 foetal anomalies in 37 foetuses. One had multiple anomalies, with the highest incidence of neural tube defects. There was also significant correlation with consanguinity.

Conclusion: The overall incidence of congenital foetal anomalies in the present study was 3.8%.This might be probably due to environmental pollution, radiation, exposure to different chemicals and teratogenic drugs.

Key words: Foetal anomalies, Ultrasonography, Premarital counseling

INTRODUCTION

Human evolution from a single cell, 'zygote' to a multi cellular organism, is an intricate and a complex process. Lucky are those foeti which travel through this wonderful journey without encountering any hindrance. The birth of a malformed baby is an unfortunate event for any family and equally for the society too! Influence of teratogens in the form of pathogens, extensive use of chemicals, causes of environmental pollution and use of drugs by the mothers indiscriminately in their day to day life, have resulted in an increased incidence of congenital abnormalities in the newly born children, [1]. Congenital anomalies are the vital causes for prenatal mortality and morbidity. Therefore, an antenatal diagnosis and foetal therapy have attained importance in the field of human embryology. According to Dolk, [2] "Environmental factors include any non – genetic factor that increases the risk of a birth defect for the exposed individual. Such factors are nutritional excesses or deficiencies (e.g. folic acid), maternal illnesses or infections (e.g. diabetes, rubella), drugs which are taken during pregnancy (e.g. thalidomide), chemical exposure in the workplace or home (e.g., to solvents or pesticides) and radiation (e.g., medical X–rays)." Scientific literature is interested in the association between congenital anomalies and the possible role of chemical contaminants [1], and foetuses are thought to be a further subgroup of the population who could be vulnerable to the effects of air pollutants [1,3].

Official records and studies on congenital malformations have confirmed the fact that, most of the common anomalies or birth defects occur in 2.5% of live births. Gaining hands on experiences show that birth defects pose multifarious social, economic and cultural problems, as well as mental trauma to the whole humanity. It has been widely noticed that many mothers are not aware of the impact of factors in causing congenital defects in their foetuses. For a better understanding of the aetiological factors of congenital anomalies, a knowledge on embryology, teratology, clinical genetics and diagnostic ultrasonography is very important [4,5]. Highly advanced imaging techniques such as 3-D and 4-D ultrasound have been helping largely in the diagnosis and treatment of birth defects in foetuses during the antenatal period. It has been strongly advised that antenatal ultrasonography has to be conducted compulsorily for a minimum of two times in all antenatal mothers. The first antenatal ultrasonography, preferably a transvaginal one, could be done between 14 and 16 weeks of gestation period. The second one, preferably a transabdominal one, could be done after the 26th week of pregnancy. This is because ultrasound imaging during antenatal period produces an anatomical record of embryological development of the human embryo. Further, an early detection of neural tube defects and an excellent medical management, which include termination of pregnancy and counseling the eligible couple, will result in betterment of the society by attaining eugenics.

An ultrasound examination will not, of course detect chromosomal anomalies, inherited disorders of metabolism or gene defects such as sickle cell disease. Detection of anomalies by ultrasound is based upon the direct visualisation of a structural defect (e.g. anencephaly), the demonstration of abnormal growth rates (e.g. short limbs in dwarfism), or the demonstration of a pathological process which is caused by the defect (e.g. a dilated stomach and duodenum in cases of duodenal atresia).

Some congenital abnormalities can be curable if they are detected early in the antenatal period (e.g. Cardiac anomalies). In–utero surgical interventions have been made possible by the advancement in the field of medicine. Pregnant women who carry anomalous foetuses can be counseled regarding the foetal anomalies and they can be sent to neonatal paediatricians for an early management or they can be advised to go for termination of pregnancy if the anomalies are of a incurable variety (e.g. anencephaly).

In view of the above, we were prompted to take up the present study "Frequency of foetal anomalies in a tertiary care centre". Apart from determining the incidence of congenital foetal abnormalities, such a study might bring to light certain factors which could possibly play an aetiological role in the production of foetal anomalies.

MATERIAL AND METHODS

The present study was based on the ultrasonographic diagnoses of 1000 consecutive pregnancies which were studied in the Department of Radiology, Government Maternity Hospital, which was attached to Osmania Medical College, Hyderabad, India, which is a tertiary care centre, in the years 2012-13 . The cases were selected from among women who were in the second and third trimesters, who were of the age group of 20-35 years, who had come for obstetric services.

The inclusion criteria were a history of suspected anomalies on clinical examination and women who were in their 2nd and 3rd trimesters, who have came for routine check ups. The women were selected, based on their ages as per criteria and parity and from all socio–economic classes. A detailed history of anomalies in other siblings, a history of consanguinity and age of the mother were also noted. A majority of the cases which were referred for scanning were referred for confirmation of the gestational ages and for exclusion of associated pathologies and anomalies. A more specific indication was the disproportionate uterine size as compared to the period of amenorrhoea.

RESULTS

The data was analysed by using SPSS, version 15.0 and the Microsoft Excel software. A p value of <0.05 was considered as significant. The results have been represented in the form of tables. Figures of cases which were followed up with terminations and autopsies which were done in Osmania Medical College were included.

Systems involved	No. of fetuses affected	Percentage (%)
CNS	17	45.94
URINARY SYSTEM	08	21.62
SKELETAL SYSTEM	03	8.10
GIT	06	16.22
CVS	03	8.10
TOTAL	37	3.8 % (overall)

[Table/Fig-1]: Showing the systemic distribution of anomalies

Foetal anomalies are significantly associated with consanguinity.

Total No cases	Without fetal anomalies	With fetal anomalies	p-value
Second trimester	11	10	<0.05
Third trimester	7	9	<0.05
Total	18	19	<0.05

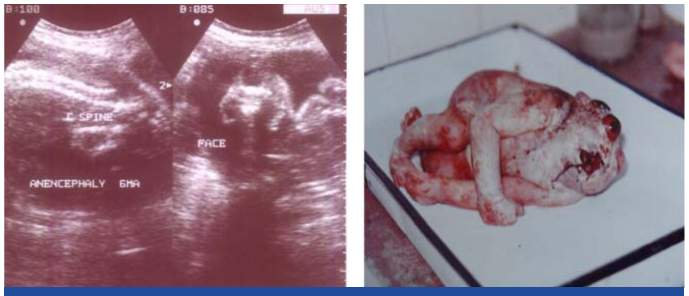
[Table/Fig-2]: Distribution of fetal anomalies in accordance with consanguinity

Overall, incidence of congenital anomalies was 3.8%. highest incidence of CNS anomalies observed.

CNS anomalies	No. of fetuses affected	Percentage (%)
Anencephaly	05	13.51
Meningo\ myelocele	02	5.41
Spina bifida	01	2.70
Encephalocele	01	2.70
Hydrocephalus	04	10.81
Holoprosencephaly	01	2.70
Dilatation of ventricles	03	8.10
Total	17	45.94 %

[Table/Fig-3]: The distribution of the CNS anomalies

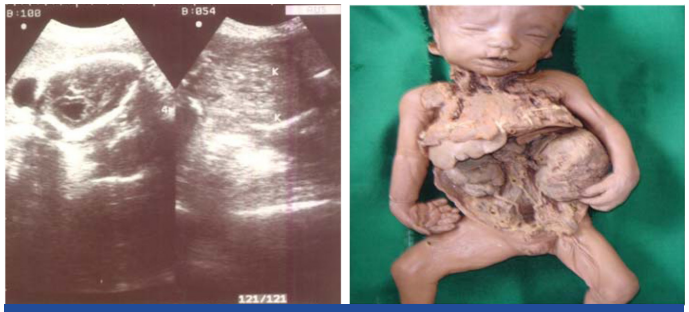
Total 5 anencephaly were noted in the observation.



[Table/Fig-4]: Showing anencephaly in USG and autopsy

Urinary system anomalies	No. of fetuses affected	Percentage (%)
Hydronephrosis	05	13.51
Cystic kidneys	02	5.41
Renal cyst	01	2.70
TOTAL	08	21.62 %

[Table/Fig-5]: Distribution of urinary system anomalies



[Table/Fig-6]: Picture showing polycystic kidney with hydronephrosis

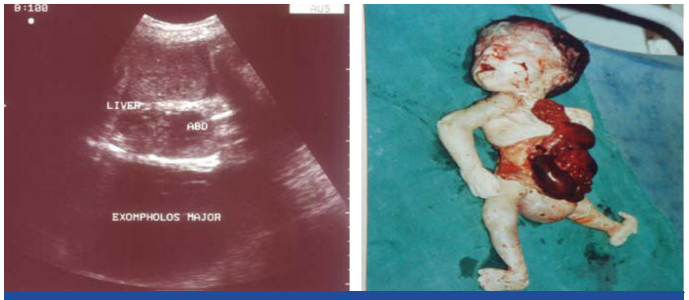
Hydronephrosis is common in urinary tract anomalies.

Skeletal Anomalies	No. of fetuses affected	Percentage (%)
Limb bone shortening	01	2.70
Achondroplasia	01	2.70
Club foot	01	2.70
TOTAL	03	8.10 %

[Table/Fig-7]: The distribution of skeletal system anomalies

GIT anomalies	No. of fetuses affected	Percentage (%)
Duodenal atresia	01	2.70
Diaphragmatic hernia	02	5.41
Cleft lip / palate	02	5.41
Omphalocele/Exomphalus	01	2.70
TOTAL	06	16.22 %

[Table/Fig-8]: The distribution of GIT anomalies



[Table/Fig-9]: Showing exompholos major in USG and autopsy

CVS anomalies	No. of fetuses affected	Percentage (%)
Echogenic left ventricle	01	2.70
Twin twin transfusion	01	2.70
Single umbilical artery	01	2.70
TOTAL	03	8.10 %

[Table/Fig-10]: The distribution of CVS anomalies

These are the overall findings which were observed in the present study.

DISCUSSION

[Table/Fig-1,2,3,4,5,6,7,8,9,10,11]

During the study period, 38 foetal malformations were identified in ultrasound, in the 1000 pregnancies, which corresponded to a prevalence of 3.8%. Among all anomalies, one foetus had multiple anomalies. The clinically relevant major and minor malformations [Table/Fig-1] were arranged system wise and tabulated. A detailed history, period of gestation and results of the present study were noted in a proforma.

It was seen that there was significant correlation ($p<0.05$) between the foetal anomalies and consanguinity [Table/Fig-2], which was inconsistent with the findings of Naeimeh Tayebi et al., [6].

1	Ogunyemi et al., [7]	2000	6877	3.1
2	Rashid SQ [8]	2002	5841	1.7
3	Tripale P et al., [9]	2003	20465	1.5
4	Nakling J et al., [10]	2005	18181	1.5
5	Salvador J et al., [11]	2005	99753	1.9
6	Sonka Ap et al., [12]	2006	1148	1.2
7	Becker R et al., [13]	2006	3094	2.8
8	Emilio et al., [14]	2012	8503	2.3
9	Present study	2013	1000	3.8

[Table/Fig-11]: The overall incidence of the fetal anamolies according

The findings of the present study correlated with those of Lee k et al., [15], Pagnotta G et al., [16] and Khronf N et al., [17]. Our values were higher as compared to those of the other studies. One of the possible reasons for this increased rate was that ours being a tertiary care centre, cases were referred here for further follow up and also radiation, environmental pollution and teratogens would have been other causes.

The most frequently observed abnormalities involved those of the central nervous system (17/38; 45%); 5/38 (13%) cases which were associated with anencephaly were detected in the prenatal period. A possible reason could be inheritance of abnormal genes from the parents, as well as new mutations in one of the germ cells that could have given rise to foetuses with such anomalies. This finding was in correlation with those of Vial Y et al., and Stefost et al., [18, 19].

Occurrence of other anomalies like Urinary anomalies [Table/Fig-5 and 6] and also skeletal, [Table/Fig-7] GIT [Table/Fig-8 and 9] and CVS anomalies [Table/Fig-10] have been represented in the descending order of their incidence in present study. Incidence of congenital anomalies in various areas is in consistent with other studies represented [Table/Fig-11].

Congenital foetal anomalies are a major cause of antenatal mortality and morbidity. The diagnosis of an anomalous foetus has been a challenging task over the years. Detection of foetal anomalies in the antenatal period is very important for the prognosis of foetuses, because they result in abortions, stillbirths and other foetal defects [Table/Fig-3 and 4]. An accurate and early diagnosis of congenital foetal anomalies will help in an early intervention of the foetus and this can prevent late irreparable damages. Prenatal ultrasonography is the best means for diagnosing malformed foetuses [20]. However, published results concerning the sensitivity of the screening vary greatly, depending on the population which was studied (high versus low risk), the quality of the equipment and in particular, the ultrasonographers' experience. Nowadays, high-resolution equipment allows the detection of minor malformations which are considered to be sonographic markers of specific conditions. These were not included in our study, to maintain the homogeneity of the population which was screened. We focused our attention on the severe abnormalities which could be detected by prenatal ultrasonography.

In conclusion, we share the view of Bucher and Schmidt [21], who in their meta–analysis insisted that, "a routine ultrasound screening in pregnancy is indicated only if it is explicitly performed to exclude congenital malformations". According to the results of the present study, we also recommend that all pregnancies, especially of consanguineous marriages, should be thoroughly examined and investigated for congenital anomalies. Premarital counseling, especially on the subject of parental consanguinity, is advised.

SUMMARY AND CONCLUSION

Congenital anomalies of foetuses are a great concern since time immemorial. Aetiological factors for these anomalies are plenty. There is a strong correlation between the congenital anomalies of the foetuses and chromosomal abnormalities, either structural or numerical. The overall incidence of congenital foetal anomalies in the present study was 3.8%. This correlated with the findings of some of the previous authors. But it was higher as compared to the findings of other authors. This might probably be due to environmental pollution, radiation and exposure to different chemicals and teratogenic drugs

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