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Anatomy

INCIDENCE OF CONGENITAL ANOMALIES AND ITS CORRELATION WITH CONSANGUINITY – AN ULTRASOUND STUDY

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ABSTRACT

Objective - The present study was undertaken to explore the incidence of congenital fetal anomalies and its correlation with consanguineous marriages by ultrasonography during the antenatal period.

Method- We focused our study in 1470 consecutive pregnancies that came for check up in the second and third trimesters with major and minor clinically relevant malformations which were detectable by ultrasonography.

Result –The analysis revealed that they were 31 fetal anomalies in 29 fetuses. Two fetuses had multiple anomalies. There was significant correlation with consanguinity also.

Conclusion –The overall incidence of congenital fetal anomalies in the present study was 1.97% and there was a significant correlation between familial marriages and the prevalence of fetal anomaly.

KEYWORDS : Fetal Anomalies, Familial Marriages, Consanguinity And Ultrasonography.

INTRODUCTION

Human evolution from a single cell, 'zygote' to a multi cellular organism, is an intricate and a complex process. Lucky are those fetuses 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.[1]

While infections and malnutrition are the dominant causes of infant morbidity and mortality in the poorer countries, in the developed countries these causes are cancer, accidents and congenital malformations.

Congenital malformation (CM) began to emerge as one of the major childhood health problems and it refers to any abnormality, whether genetic or not, which is present at birth. Treatment and rehabilitation of children with CM is costly and complete recovery is usually impossible.

The etiology of CM is genetic (30-40%) and environmental (5 to 10%). Among the genetic etiology, chromosomal abnormality constitutes 6%, single gene disorders 25% and multifactorial 20-30%; however, for nearly 50% of CM, the cause is yet to be known.

Consanguineous marriages have been described as an important factor contributing to increased congenital malformations. A consanguineous marriage can be characterized by the degree of relatedness between the spouses: first cousins, double first cousins, half first cousins, first cousins once removed, second cousins, second cousins once removed and third cousins. Genetic effects of consanguinity can be traced to the fact that the inbred individual may carry two copies of a gene that was present in a single copy in the common ancestor of his/her consanguineous parents. A recessive gene may thus come to light for the first time in an inbred descendant after having remained hidden for generations. For this reason, consanguinity influences the incidence of some inherited diseases.[2]

In the past, congenital anomalies were detected after birth, that too, only the exterior malformations were detected earlier, while the anomalies inside the body cavities were detected much later. With the development of real time ultrasonography there was a revolution in antenatal diagnosis of congenital anomalies.[3]

Early antenatal diagnosis of congenital anomalies is crucial for early counselling, intervention and possible fetal therapy.[4]

RATIONALE OF THE STUDY:-

Congenital anomalies are the vital causes for prenatal mortality and morbidity.

Therefore, an antenatal diagnosis and fetal therapy have attained importance in the field of human embryology.

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 fetuses can be counseled regarding the fetal anomalies and they can be sent to the neonatal pediatrician for early management or they can be advised to go for termination of pregnancy if the anomalies are of an incurable variety (e.g. anencephaly).

In the view of the above, we were prompted to take up the present study "Incidence of congenital anomalies and its correlation with consanguineous marriages". Apart from determining the overall incidence of congenital abnormalities, such a study might bring to light certain factors which could possibly play an etiological role in the production of fetal anomalies.

MATERIALS & METHODS

The antenatal mothers referred for routine as well as target imaging to a specialist ultrasound diagnostic center in Hanamkonda, Warangal district from January 2017 to June 2017 contributed the population for this study. All the pregnancies of 9 to 41 weeks gestation were analyzed. Those with major anomalies were serially assessed and followed up for confirmation of the diagnosis.

INCLUSIVE CRITERIA:

1. Pregnant women those referred for routine as well as targeted imaging contributed the population for this study.
2. A history of consanguinity is noted.

EXCLUSIVE CRITERIA:

1. Those with vesicular mole and intrauterine demises were excluded.

The ultrasound machine used for the study is Logic 200 pro and probe used is convex probe of 3.5 - 5.5MHz.

PROCEDURE

A total of 1470 pregnant women of 9 to 41 weeks gestation were included in this study for a period of 6 months that is from January 2017 to June 2017.

Early pregnancy

For ultrasounds early in pregnancy the best views are obtained if the pregnant woman's bladder is full. The uterus is often hidden behind the bowel making it difficult to see. When the bladder is full the bowel is pushed out of the way.

Starting about an hour before the appointment time, the patient will need to drink about 500ml of clear fluid (water, juice, cordial or clear tea - not milky or fizzy drink) finishing half an hour before the scan. Do not empty the bladder before the ultrasound.

The scan can determine the viability of pregnancy, number of baby/babies, baby's age and to investigate possible reasons for vaginal bleeding and/or pain during pregnancy. In some cases, it is very difficult to assess baby through the abdomen due to bowel gas or the thickness of tissue under the skin. Increased fatty tissue commonly makes it harder to see the baby clearly. An internal scan can be the best option to get a better look at baby.

Trans-vaginal ultrasound – through the vagina

In this procedure, women are asked to remove any piece of clothing from their waist down including their undergarments. A firm wedge is then placed under the woman's bottom to raise the pelvis. The transducer is a long tubular structure with a handle. It is covered with a condom and sterile gel is put on the tip so that it can be placed into the vagina easily. If the patient consents to this, the transducer is inserted into the vagina, which gives a clearer view of baby.

In most cases, some pressure is applied on a patient's abdomen and transducer to move overlying bowel gas. It is important to let the sonographer know if you feel any pain. This procedure is uncomfortable but should not be painful.

Trans-vaginal scan are commonly done in early pregnancies for dating scans (early scans).

Morphology scan – a look at baby's body

This scan is best done from 19-20 weeks to fully assess baby. It is a very detailed examination which usually takes about 45 minutes to 1 hour depending on the baby's position.

The woman is asked to come on empty bladder for scanning. The woman is laid supine on the couch and abdomen is exposed up to pubic symphysis. The ultrasound gel is applied over whole abdomen. The convex probe is applied at right angles to abdomen. The probe is moved in all four quadrants of abdomen while observing the monitor.

During this scan, the sonographer looks at the baby's head, face, spine, heart, abdomen, kidneys, arms and legs to ensure that they can be seen and are normal. (Some problems may not be picked up by this scan).

OBERVATIONS AND RESULTS

The majority of cases referred for scanning were for confirmation of the gestational age and for exclusion of associated pathologies and anomalies. A more specific indication was the disproportionate uterine size compared to the period of amenorrhoea and history of consanguinity.

TABLE - 1 INCIDENCE OF FETAL ANOMALIES DETECTED BY ULTRASOUND

Total no. of cases	**1470**
No. of fetuses with anomalies	**29 (Overall 1.97%)**

During the study period, 31 fetal malformations were identified by ultrasound in the 1470 pregnancies, which correspond to a prevalence of 1.97%. Among all anomalies, two fetuses had multiple anomalies. The systemic distribution of fetal anomalies is represented in the table no.2. The smaller entities were grouped as "others". The central nervous system and genitourinary system were more commonly involved followed by gastrointestinal system, cardiovascular system and respiratory system.

TABLE-2 SYSTEMIC DISTRIBUTION OF FETAL ANOMALIES

Systems involved	No. of fetuses affected	Percentage(%)
CNS	09	29.03%
GUS	08	25.8%
GIT	05	16.13%
CVS	04	12.9%
RS	03	9.68%
SK	01	3.23%
OTHERS	01	3.23%
TOTAL	29	1.97%(overall)

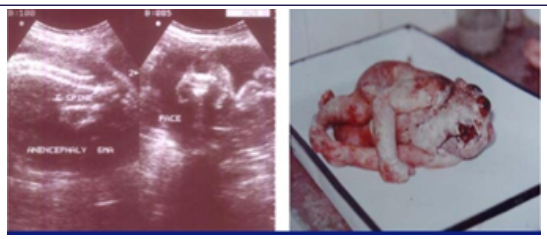

Figure showing Anencephaly in Ultrasound

CONSANGUINITY

From 1470 cases, 350 were consanguineous marriages and 1120 were non-consanguineous marriages. From 29 cases with anomalies, 13(3.7%) cases were from familial marriages, while 16(1.4%) cases were from non-familial marriages. There was a significant correlation between familial marriages and the prevalence of fetal anomaly.

The data was analyzed by using Graph pad instant 5.0 calculator and the Microsoft Excel software.

The results have been represented in the table no-3.

TABLE NO. 3 DISTRIBUTION OF FETAL ANOMALIES IN ACCORDANCE WITH CONSANGUINITY

GROUPS	NO. OF CASES WITH FETAL ANOMALIES	NO. OF CASES WITHOUT FETAL ANOMALIES	TOTAL NO. OF CASES
CONSANGUINITY	13(3.7%)	337(96.3%)	350
NON-CONSANGUINITY	16(1.4%)	1104(98.6%)	1120
TOTAL	29(1.97%)	1441(98.03%)	1470

p value of 0.0133 was considered as significant by Chi square method.

DISCUSSION

Most children who are born with major congenital anomalies and survive infancy are affected physically, mentally or socially and can be at increased risk of morbidity due to various health disorders.[2]

Early detection of birth defects could reduce their impact on the quality of life of disabled people, allowing the optimization of perinatal clinical management, the use of specialized neonatal and pediatric intensive care, and the timely application of surgical techniques.[5]

Antenatal ultrasound examination is the procedure of choice for the diagnosis of the malformed fetus. However, the results obtained vary greatly depending on the study population and, in particular, on the conditions in which the examinations are carried out.[6]

The overall incidence of the major congenital fetal anomalies is 1.97% in the present study. Different authors have reported an incidence ranging from 1.5% to 4%.

J.M. Carrera et al[6] in their study described the results of routine ultrasound examination over a period of 22 years and the prevalence of fetal anomalies was 3.03%.

Markov D et al[7] studied 1135 singleton pregnancies between 11±0 and 14±6 weeks gestations and noticed the overall prevalence of structural fetal anomalies was 4.6% (53/1135).

Balakumar.K[8] carried out a prospective study of 15 years and 7 months among the live 30,030 singleton pregnancies of 9 to 41 weeks gestation by subjecting them to ultrasound scanning.

The overall incidence of major anomalies was 2.59% in this region of Northern Kerala ($p<0.05$).

Sharada B. Menasinkai[9] has taken this study to know the incidence of congenital anomalies, in Cheluvamba Hospital attached to Mysore Medical College & Research Institute Mysore. Among 3000 births during May September 1999, there were 61 babies with congenital

anomalies, among these, 32 babies had neural tube defects. Overall incidence of congenital anomalies was 2.03%, incidence of CNS anomalies was 49.19% and incidence of Neural Tube Defects was 1.06%.

Nayab Alia et al[10] concluded that the prevalence of major congenital anomalies in their population appears to be similar to international figures as 2.97%. The study showed the preponderance of neural tube defects.

Dr. Rameswarapu Suman Babu et al[1] focused the study on 1000 consecutive pregnancies that came for check up in the second and third trimester, with major or minor clinically relevant malformations which were detectable by ultrasound. The analysis revealed that there were 38 fetal anomalies in 37 fetuses. One had multiple anomalies, with the highest incidence of neural tube defects.

They concluded that the overall incidence of congenital fetal anomalies in the present study was 3.8%. This might be probably due to environmental pollution, radiation, exposure to different chemicals and teratogenic drugs.

CONSANGUINITY

The prevalence of congenital anomalies was mostly observed in consanguineous marriages compared to non- consanguineous marriages. The comparative study of consanguinity with various studies is shown in the table below.

COMPARATIVE STUDY OF CONSANGUINITY WITH VARIOUS STUDIES

It was seen that there was significant correlation ($p<0.05$) between the fetal anomalies and consanguinity [Table no.-3], which were in consistent with the findings of Naeimeh Tayebi et al and R Suman Babu et al.

Study	Total no. of fetal anomalies	No.of anomalies associated with consanguinity	No.of anomalies not associated with consanguinity
NAEIMEH TAYEBI et al(2010)	45	34	11
R SUMAN BABU et al(2013)	37	19	18
PRESENT STUDY	29	13	16

Naeimeh Tayebi et al[2] concluded that the prevalence of congenital anomalies was mostly observed in consanguineous marriages compared to non consanguineous marriages. From 1195 neonates, 300 (25%) were from consanguineous marriages and 895 (75%) were from non-familial marriages. From 45 cases with anomalies, 34 (2.8%) cases were from familial marriages, while only 11 (0.9%) cases were from non-familial marriages. There was a significant correlation between parental marriages and the prevalence of anomaly ($p=0.018$).

SUMMARY &CONCLUSION

The present study was undertaken to find the overall incidence of congenital anomalies by ultrasonography and the prevalence of congenital anomalies in consanguineous marriages.

Congenital anomalies of fetuses 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 fetuses and chromosomal abnormalities, either structural or numerical.

The overall incidence of major anomalies among pregnancies of 9 - 41 weeks gestation was 1.97% in this specialist ultrasound center, Warangal.

The commonest system affected was the central nervous system, followed by the genitourinary system and gastro intestinal system.

This study strongly advises that, antenatal ultrasonography has to be conducted compulsorily for a minimum of two times in all antenatal mothers.

The first antenatal ultrasonography, preferably a trans-vaginal one, should be done between 9 and 12 weeks of gestation period. The second one, a trans-abdominal one, preferably morphology scan or targeted imaging for fetal anomalies (TIFFA) should be done after the 20th week of pregnancy.

This is because ultrasound imaging during antenatal period produces an anatomical record of embryological development of the human embryo. An early detection of incurable congenital defects and further management, which includes termination of pregnancy and counselling the eligible couple, will result in betterment of the society by attaining eugenics.

In conclusion, I would like to share the view of Bucher and Schmidt, 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". [11]

According to the results of the present study, I 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.

Abbreviations

CNS-central nervous system **GUS**-genitourinary system **GIT**-gastrointestinal system **CVS**-cardiovascular system **RS**-respiratory system **SK**-skeletal system

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