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Prevalence of Congenital Anomalies at a Tertiary Care Centre in Tamil Nadu, South India

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Abstract: ***According to WHO congenital Anomalies are defined as structural (or) functional anomalies including metabolic disorders which are present at the time of birth. This study included the patients admitted with detected anomalous fetus in Government Rajaji Hospital, Madurai. Over all prevalence of congenital anomalies in GRH is 6% Central Nervous system is the most commonly affected system. Congenital diaphragmatic hernia was the most common lethal congenital anomaly. Male fetus had a higher Incidence of anomalies.***

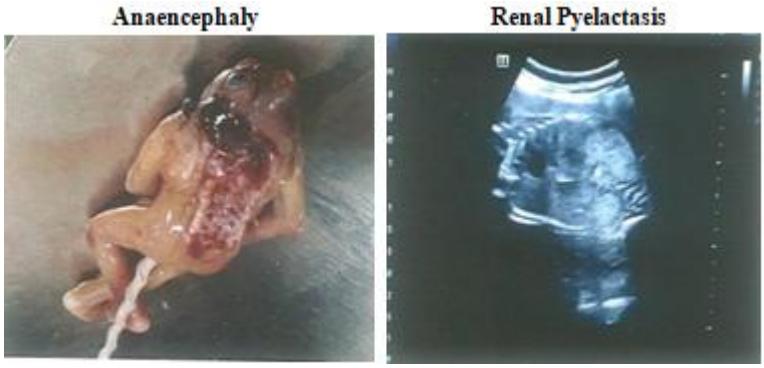
Keywords: Congenital anomaly, consanguinity, prevalence, lethal , USG, Diagnosis

1. Aim

A study to found the prevalence of congenital fetal anomalies in AN mothers admitted in Government Rajaji Hospital, Madurai.

2. Materials and Methods

It is a cross sectional, Retrospective, analytical study, was conducted from December 2018 to April 2019. Relevant Information regarding maternal Age, parity, Consanguinity, History of periconceptional Folic Acid intake, previous history of affected sibling was documented. Through Medical History regarding Diabetes mellitus complicating pregnancy was a evaluated. All neonates identified with congenital anomalies were further investigated. Lethal anomalies were terminated. For non lethal anomalies, fetal condition were discussed with paediatric surgeon and followed up till post partum in AN OPD. After taking written informed consent of parents of affected fetus. The work has been carried out in accordance to the code of Ethics. 100 AN mothers were followed up in this study.



3. Results

Total No. of Mothers with anomalous Fetus - 100

Age Distribution	<20	21-25	26-30	>30
	21	40	24	15

Consanguinity	NCM	10CM	20CM	30CM
	68	0	11	21

Lethal	Nonlethal
46	54

SEX	MCH	FCH
	47	16

ORDER OF BIRTH			
PRIMI	2nd Gravida	3rd Gravida	4th Gravida & above
46	31	12	11

MORE OF TERMINATION		
MIFEPRISTONE+ MISOPROSTOL	LABOUR NATURALIS	LSCS
25	25	4

Maternal age places an important role in birth of a child with congenital anomalies. Anomalies were noted commonly in 21 - 25 years (40%) of age group than elderly age group. In view of consanguinity 68% of cases had a history of Non-consanguinuos marriage followed by 30 degree consanguinuos marriage, in our study consanguinity was not significantly associated. Lethal anomalies were terminated at

1st and 2nd trimester and Non-lethal anomalies wee followed up till post partum. In association with other studies male fetus 74% has highest prevalence compared to female fetus. More number of incidence noted in primigravida (46%) compared to multigravida.

System involvement of conenital anomalies

cardiovascular system

S.No	Anomaly	Number
1	TOF	4
2	Hypoplastic (L) Heart Syndrome	1
3	Echogenic foci in (L) ventricle	1
4	Fetal VSD	3
5	Persistent (L) SVC	3
6	Ebstien Anomaly	1
7	Fetal Dextrocardia	3
8	Fetal Situs Inverse totalis	1
9	Fetal TGA	1

Renal System

1	Fetal Gross Hydronephrosis	3
2	B/L Renal Pyelactasis	3
3	Fetal (L) PCS Dilatation	3
4	Fetal Small Kidney / Bladder	1

Respiratory System

1	Congenital Diaphragmatic Hernia	10

Central Nervous System

1	Fetal choroid plexuses cyst	2
2	Down's syndrome	3
3	Corpus callosal agenesis	2
4	Arnold Chiari malformation	2
5	Anancephaly	7
6	Meningocele	5
7	Persistent cisterna magna	2
8	B/L ventriculomegaly	3
9	Hydrencephaly	1
10	Cystic Hygroma	3
11	Hydrocephalus	3
12	Exencephaly	1

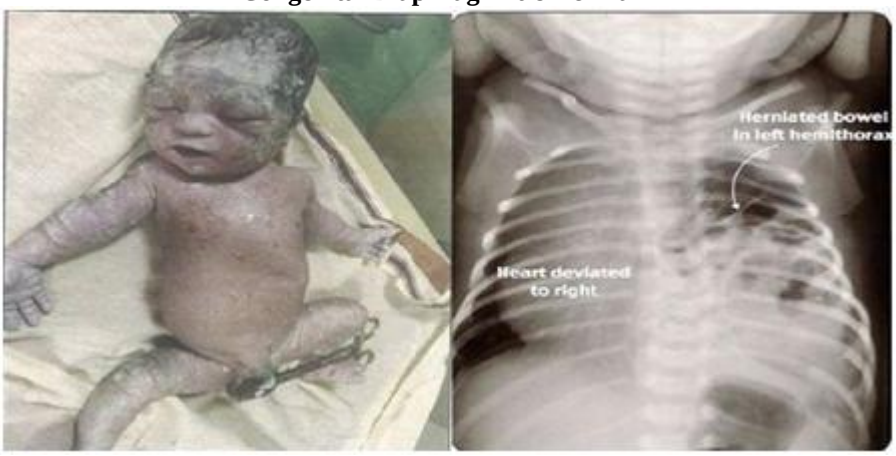
Musculoskeletal System

1	CTEV	3
2	Skeletal dysplasia	1
3	Osteogenesis Imperferta	1
4	Limb Anomaly	1

GIT

1	Omphalocele	3
2	Esophageal Atresia	1
3	Gastrochisis	1
4	Cleft Lip	1
5	Adrenal calcification	1
6	Anorectal Malformation	1

Congenital Diaphraghmtic Hernia



4. Discussion

Congenital anomalies are one of the major causes of still births and neonatal morbidity and mortality in India. Early detection of anomaly reduces significant fetal morbidity with the advancement of prenatal diagnostic techniques. Peri conceptional folic acid supplementation reduces the prevalence of fetal cranial anomalies. The other factors like socio economic status, environmental factors, Dietary habits and genetic component determine the occurrence of an anomaly. In past infectious diseases and nutritional problems were the major contributor in Infant mortality. But in present congenital malformations were one of the most common one. In our Study overall incidence of congenital anomalies is 6% compared with study of Bhat (1998) 3.7%, Moharty et al (1.6%) and Anne george chesian (12.5% per thousand and Prasanna Jeeth Kotake (0.9%) and Tenali ASL et al (March 2018) (6.3%) and Sumathy et al (1.9%). In this study the rate of Incidence of congenital malformation is higher in males (73%) compared to females (27%). But in a study of Tenali ASL et al reported a higher incidence of congenital anomalies in females (57%) compared to males. But in majority of studies like Amar taksunde (2010) reported higher Incidence among male babies compared to female babies. In our study we found that central nervous system were highest (33%) Followed by cardiovascular system[18%] and respiratory system[10%].

S. S. Abouel. Ella (2018), Baruah (2015) and Tenali ASL et al (2018) found higher Incidence of Musculoskeletal system malformation. Maternal age plays an important role in incidence of congenital malformation. In our study higher prevalence of congenital anomaly was found among women between 21-25 years of age (40%) it is similar with the study of S.S.Abouel - Ella et al and sumathi etal (2016) In our study consanguinity is controversial in Association with Egyptian study and Tenali ASL et al (2018) study. Lethal anomalies were terminated with mifepristone and

misoprostal (25%), labour naturalis 25%, and LSCS (4%) for obstetric indication. The causes of congenital anomalies can be classified into structural and functional anomalies. Genetic factors plays an important role in congenital anomalies. Single genedefects, chromosomal aberrations, multifactorial disorders, teratogenic factors incidence has increase now. Early and precise diagnosis of a fetus with multiple congenital anomalies is important for management, genetic counseling concerning etiology, recurrence risk, prenatal diagnosis, screening and recommendation for evaluation of other family members. Diagnosis of a child who presents with multiple congenital anomalies is still a complex issue. Congenital malformations account for about 20% of perinatal deaths and a substantial proportion of spontaneous abortions in the first trimester of pregnancy. Defects in the central nervous system (CNS) account for 50% of all serious malformations.

Congenital Diaphragmatic hernia is due to herniation of abdominal contents into the thoracic cavity through a diaphragmatic defect usually on the left side (75%). Prevalence is 1 in 3- 4 thousand births. The condition may be diagnosed prenatally with ultrasound. Pulmonary hypoplasia and pulmonary hypertension are the causes of mortality.

Prognosis depends upon the time of occurrence. If CDH occurs early in intrauterine life, it carries poor prognosis as the lungs would not have developed well. Anencephaly is a condition, in which there is a rudimentary brain with absent cerebral hemispheres and an absence of cranium and posterior skull bones. It is the only anomaly diagnosed in 1st trimester by absence of BPD. There is increased maternal serum alpha fetoprotein (MSAFP) in 2nd trimester. Anencephalous infants do not survive,so women are offered the option of MTP, if diagnosed before 20 weeks.

Conjoint Twins | Skeletal Dysplasia | Anophthalmia

5. Congenital Diaphragmatic Hernia

Prenatal Assessment of CDH

Although the fetal diaphragm can be visualized with high resolution equipment even in the first trimester, the diagnosis of CDH is typically suggested when abdominal organs are visualized in the chest or cardiac deviation is noticed on a four chamber cardiac view. Left sided CDH typically manifest with a rightward shift of the heart and mediastinum and coincident presence of echolucent stomach and intestines. The existence of abdominal viscera in the thorax may be identified by the presence of peristalsis. The liver may also be herniated in left-sided CDH, but it is more difficult to differentiate liver from lung sonographically.

Right sided CDH cases are more difficult to diagnose because liver has similar appearance to mid -trimester fetal lung. However, the right lobe of the liver typically shifts the heart and mediastinum to the left, and this abnormal arrangement provides the major clue to the presence of a diaphragmatic defect. Doppler interrogation of the umbilical vein and hepatic vessels or the location of the gallbladder may be used as additional landmarks to define the position of the liver. Cardiac compression and polyhydramnios are indirect signs of right-sided CDH.

The main differential diagnoses are other pulmonary pathologies, such as cystic masses (Cystic ademnomatoid malformation' bronchogenic, enteric, and neuroenteric cysts; mediastinal teratoma; and thymic cysts), bronchopulmonary sequestration, or bronchial atresia. In these conditions, intraabdominal organs are not displaced.

After the diagnosis of CDH, the first goal is to exclude additional abnormalities. Karyotyping is an essential step, but the finding of normal chromosomes does not exclude other genetic conditions and syndromes. Modern genetic techniques are therefore increasingly used and have been reviewed elsewhere. A customized array targeting genes previously linked to CDH has been developed and applied to fetuses with apparently isolated CDH.

Prenatal Intervention for CDH

In recent years, different strategies have been used to identify optimal candidates for fetal therapy for CDH; these were reviewed by Deprest and associates. Today, the clinical procedure consists of percutaneous fetoscopic endoluminal tracheal occlusion (FETO). The procedure is believed to work because it prevents egress of lung fluid, thereby increasing airway pressure, which promotes pulmonary tissue proliferation, increases alveolar airspace, and encourages maturation of pulmonary vasculature. In experimental conditions, sustained tracheal occlusion was shown to reduce the number of type II pneumocytes and hence surfactant expression, which can be improved by in utero release (Plug - unplug sequence).

Fetal Echocardiography in CHD

Detailed fetal echocardiography in the pregnancy at high risk for CHD provides an in-depth and comprehensive evaluation of fetal cardiovascular structure and function. In experienced hands, and particularly when performed beyond 18 weeks' gestation, fetal echocardiography has been shown to have high sensitivity for almost all forms of fetal heart disease.

First-trimester fetal echocardiography, performed between 10 and 14 weeks' gestation either transvaginally or transabdominally, does not have the same degree of sensitivity for CHD as second-trimester imaging, in part because of resolution considerations when imaging the smaller first-trimester heart, and in part because many forms of CHD (e.g. aortic and pulmonary valvar stenosis, ventricular hypoplasia, valvar regurgitation, arrhythmias, cardiac tumors, restriction of the ductus arteriosus or foramen ovale) evolve significantly between the late first trimester and term. Nevertheless, in experienced hands, the clinical role of first-trimester fetal echocardiography has increased along with first-trimester nuchal translucency screening and improvements in image resolution.

Pre-Implantation & Prenatal Diagnostic Procedures

Diagnostic procedures used in prenatal diagnosis include amniocentesis, chorionic villus sampling (CVS), and rarely fetal blood sampling. These enable characterization of an increasingly large number of genetic abnormalities before birth. Karyotype analysis has a diagnostic accuracy of more than 99 percent for aneuploidy and chromosomal abnormalities larger than 5 to 10 megabases. In the setting of a fetal structural abnormality, chromosomal microarray analysis (CMA) is recommended as the first-line genetic test performed, as it may detect clinically significant chromosomal abnormalities in approximately 6 percent of fetuses with normal standard karyotype. An exception would be if the structural abnormality strongly suggests a particular karyotype-such as endocardial cushion defect with trisomy 21 or holoprosencephaly with trisomy 13. In such cases, karyotyping with or without fluorescence in-situ hybridization (FISH) may be offered as the initial test (American College of Obstetricians and Gynecologists, 2016b). Among those without evidence of a fetal structural abnormality and with a normal karyotype, CMA has detected additional chromosomal abnormalities (pathogenic copy number variants) in approximately 1 percent.

Prevention of Congenital Anomalies

Genetic Counselling

It is a problem solving approach or communication process in relation to genetic disorders or congenital anomalies in the family. It is non-directive information to the individual or family who discuss the importance to their own situations. It is of two types. They are Prospective genetic counseling and Retrospective genetic counseling.

Prospective Genetic Counseling

It is for true prevention of disease. It aims at preventing or reducing heterozygous marriage by screening procedures and explaining the risk of affected children.

Retrospective Genetic Counseling

It is done after a hereditary disorder has already occurred.

6. Methods

- Contraception
- MTP
- Sterilization

Discourage consanguineous marriages. Avoid late marriage and pregnancy after 35 years. Promotion of health of girl child and pre pregnant health status of the females by prevention of malnutrition, anemia, folic acid deficiency, iodine deficiency etc., Encourage the immunization of all female child by MMR. Protection of individuals & whole communities against mutagens (X-ray, drugs, alcohol). Immunization by anti-D immunoglobin to the Rh-negative mothers after abortion. Elimination of active and passive smoking of tobacco by mothers. Avoidance of drug intake without consulting physician in the first trimester of pregnancy. Prevention of intrauterine infections and promotion of sexual hygiene. Efficient antenatal care. Promotion of therapeutic abortion after prenatal diagnosis. Discouraging reproduction after birth of a baby with congential anomalies. Increasing public awreness about the risk factors and etiological factors of congenital anomalies and their preventive measures.

7. Conclusion

During this study period, most of the women belonged to 21-25 years of age, congenital anomalies were seen more commonly in primi para (46%) compared to multipara, consanguinity is considered as controversial association with congenital anomalies. Congenital diaphragmatic hernia were the most commonest one. The predominant system involved was central nervous system (33%) followed by cardiovascular system. Non lethal anomalies (54%) was increased in Incidence compared to lethal anomalies. The rate of congential anomalies out numbered in males compared to Females. Anomaly rates can be reduced by preconceptional and antental folic acid supplementation and pre natal genetic testing in high risk cases can be offered. It is one of the most important cause of Neonatal morbidity and mortality.

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