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A Study On Distribution Of Pattern Of Congenital Anomalies In Fetus-Tertiary Care Centre, South India.

Sreelakshmi.U[1], K. Saritha[2], B. Indira[3]

Corresponding author K. Saritha

1. *Professor, Department Of Obstetrics And Gynaecology, Mallareddy Medical College For Women, Hyderabad, Telangana.*
2. *Professor, Department Of Obstetrics And Gynaecology, Mallareddy Medical College For Women, Hyderabad, Telangana.*
3. *Professor, Department Of Obstetrics And Gynaecology, Mallareddy Medical College For Women, Hyderabad, Telangana.*

Abstract:
Background*: Congenital malformations represent defects in morphogenesis during early fetal life. These anomalies are permanent changes produced by an intrinsic abnormality of development in body structure during fetal life. Aim of our study is to study frequency and patterns of congenital anomalies in the fetuses and associated risk factors.*
Methods: *The present study is a Prospective Observational Study carried out during the period of January 2021-March 2024 in the department of Obstetrics and Gynaecology, Mallareddy Narayana hospital, Hyderabad, Telangana. All the pregnant women diagnosed with congenital anomalies attending Antenatal outpatient department were recruited for this study. A detailed history was recorded in a proforma All pregnant participants were subjected to a first trimester ultrasound scan, NT scan, and TIFFA scan at 18-20 weeks and fetal 2D -Echo at 24 weeks. Anomalous fetuseswere confirmed by ultrasonography recorded by 2 different sonologists. Study participants were followed up until delivery and termination of pregnancy***.**
Results: *During the study period total of 41 fetuses with congenital anomalies (CAs) were observed. Majority of the congenital anomalies were in maternal ages between 20 years to 30 years 31(75.6%). The predominantly affected system was central nervous system. Medical termination of pregnancy was done before 20 weeks of gestation with major and multiple anomalies.*
Conclusion:*In this present study Central nervous system anomalies were most common followed by Musculoskeletal system and Renal system. Folic acid supplementation, regular ante natal visits, prenatal diagnosis and early detection of Congenital anomalies were recommended.*
Keywords*: Congenital anomalies, Maternal age, Prenatal diagnosis, Ultrasound scan, central nervous system,*

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I. Introduction:

Congenital malformations represent defects in morphogenesis during early fetal life. Congenital malformations account for 8-15% of perinatal deaths and 13-16% neonatal deaths in India. [1, 2, 3]These anomalies are permanent changes produced by intrinsic abnormality of development in body structure during fetal life. The causes of congenital anomalies are divided into four broad categories genetics, environmental, multifactorial and unknown. Among the multifactorial causes can be factors like pollution, radiation, maternal factors like Diabetes mellitus, late age at pregnancy, increasing birth order, consanguineous marriages, uncertain medications, nutritional deficiencies, neonatal infections, obesity etc.

Prenatal diagnosishas greatly benefited from advances in ultrasound technology and in our ability to detect microscopic and submicroscopic chromosome abnormalities as well as single gene disorders, leading to substantive improvements in detection of such congenital anomalies. At present, invasive prenatal diagnosis continues to be the gold standard for pregnancies at increased risk for chromosomal anomaly or other genetic disease, with chorionic villus sampling being the procedure of choice for the first trimester. Whereas mid-trimester amniocentesis continues to be the most common form of invasive procedure for prenatal diagnosis [4]. In the low-risk population prenatal diagnosis generally consists of screening procedures by means of ultrasound and maternal serum biochemistry.Ultrasound screening for fetal structural abnormalities is generally

recommended at 19-21 weeks of gestational age.A major impact of antenatal diagnosis of malformations is related to the severity of the malformations detected. Most severe defects are reportedly detected earlier than minor ones, which is especially relevant in many countries where only before viability is termination of pregnancy authorized by law [5]. Aim of our study is to study frequency and patterns of congenital anomalies in the fetuses and associated risk factors.

II. Methods:

The present study is a prospective observational study carried out during the period of January 2021-March 2024 in the department of Obstetrics and Gynaecology,Mallareddy Narayana hospital,Hyderabad,Telangana.All the pregnant women diagnosed with congenital anomalies attending to Antenatal Outpatient Department were recruited for this study, before conducting the study approval was obtained by institutional ethical committee.A written informed consent was obtained from all study participants prior to enrolment.

A detailed history was recorded in a proformawhich included maternal age, paternal age, gravida status, history of previous abortion, history of prior congenitally malformed babies, education status, occupation of both parents, history of consanguinity socio-economic status, residence-rural, urban, history of chronic diseases like Thyroid disorders, Diabetes, epilepsy, hypertension etc. History of smoking-passive and active, alcohol, any drug intake during organogenesis period.History of folic acid intake during preconceptional period taken or not.

All pregnant participants were subjected to a first trimester ultrasound scan, NT scan,and TIFFA scan at 18-20 weeks and fetal 2D -Echo at 24 weeks. Anomalous fetuses with confirmed ultrasonography recorded by 2 different sonologists. Pregnant women with congenital anomalies in fetus willing to participate in this study included, those who were not willing excluded.Study participants were followed up until delivery and termination of pregnancy.

III. Results:

During study period total of 41 fetuses with congenital anomalies were observed. All were singleton pregnancies. **Table-1** showing Demographic profile of study participants.Majority of the congenital anomalies were in maternal age between 20 years to 30 years31(75.6%). Most of the affected cases were primigravida19(46.3%). Partner age of majority affected cases was 31-40 years of age23(56%),remaining were less than 30 years of age. Previous history of abortions wasidentified in 9 (21.9%)cases. Majority were home maker 13 (31.7%). Most of the partners Occupation was as Driver16 (38.9%). Majority resided in Urban area 23(56%). Predominantly affected babies were male 22(53.6%).History of passive smoking was there in 21 (51.2%) cases. History of Alcohol intake was there in 11 cases. History of Consanguinity was present in 5cases. Co-morbid conditions like Hypothyroidism in 5 cases, Diabetes mellitus in 2 cases, Bronchial asthma in one case, epilepsy in one case, Polyhydramnios in one case were identified. History of regular Folic acid intake was there in 9 (21.9%)cases and infrequent intake of folic acid intake was identified in 12 (29.2%) cases because of vomiting and hyperemesis. Antenatal checkups were not there in 11 (26.8%) cases.

Table-1: Demographic profile of the study participants:

Characteristics	**No of cases[n=41]**	Percentage(%)
Maternal AGE(YEARS)		
<20	4	9.7
20-30	31	75.6
>30	6	14.6
GRAVIDA status		
G1	19	46.3
G2	11	26.8
G3	9	21.9
G4	1	2.4
G5	1	2.4
Occupation(maternal)		
Home maker	13	31.7
Laborer	12	29.2
Agriculture	4	9.7
Professional	12	29.2
H/O previous abortion	9	21.9
Partner occupation		
Driver	16	38.9
Labor	13	31.7
Agriculture	4	9.7
Professional	4	9.7
Traders	3	7.2

Education		
Primary school	25	60.9
Secondary school	10	24.3
Intermediate	5	12.1
Degree	1	2.4
Residence		
Rural	18	44
Urban	23	56
Religion		
Hindu	33	80.4
Muslims	5	12.1
Christian	3	7.2
Socio-economic status		
Lower class	32	78
Middle class	6	14.5
Higher class	3	7.2

Table -2 showing system wise type of congenital anomalies. The predominantly affected system was central nervous system followed by musculoskeletal system and Renal system. With increasing use of ultrasound examination in first and second trimester of pregnancy and use of double marker, triple marker and quadruple markers for high-risk pregnancies diagnosis of congenital anomalies was become much easier. The pregnant women diagnosed with major congenital anomalies by doing the above tests and TIFFA scan sought termination of pregnancy if the fetuses were not compatible with life.

Table-2: showing System wise distribution of type of structural Anomaly.

System involvement	Type of structural anomaly -number of cases (%)
CNS: (n=27) 65.8%	Anencephaly-11 (26.8%) Spina Bifida -5 (12.1%) Meningomyelocele -2 (4.8%) Acrania-Anencephaly sequence -1 (2.4%) Anencephaly with Meningomyelocele-1 (2.4%) Exencephaly with meningomyelocele-1 (2.4%) Occipitoencephalocele -2 (4.8%) Choroid plexus cyst-3 (7.2%) Arnold Chiari malformations -1 (2.4%)
Musculoskeletal: (n=11)26.8%	Omphalocele -2 (4.8%) Kyphoscoliosis -2 (4.8%) Gastroschisis -3 (7.2%) Congenital diaphragmatic Hernia-1 (2.4%) Congenital talipesequinovarus -1 (2.4%) Club foot-1 (2.4%) Short long bones -1 (2.4%)
Renal: (n=4)9.7%	Anhydromnios Renal hypoplasia -1 (2.4%) Anhydromnios with multicystic kidneys -1 (2.4%) multicystic kidneys -1 (2.4%) Hydroureter with PUJ obstruction-1 (2.4%)
CVS: (n=3)7.2%	Hypoplastic left heart -2 (4.8%) Atrial septal defect-1 (2.4%)
GIT and RS: (n=2)4.8%	Imperforate anus and plural effusion -1 (2.4%) Tracheo - esophageal fistula-1 (2.4%)
Chromosomal: (n=1)2.4%	Down syndrome -1 (2.4%)

CNS: Central nervous system, CVS: Cardio vascular system, GIT: Gastrointestinal system
RS: Respiratory system (Multiple Anomalies were seen in 5 cases)

Table-3showinggestational age at which delivery or termination of pregnancy occurred. Medical termination of pregnancy was done before 20 weeks of gestation in 19(46.3%) cases(Anencephaly and multiple anomalies-Anencephaly with meningomyelocele and kyphoscoliosis, Acrania-anencephaly sequence,Exencephaly with meningomyelocele andGastroschisis, Spina bifida with kyphoscoliosis and multicystickidnyes, Short long bones andanhydromnios with renal hypoplasia, anhydromnios with multicystic kidneys, hypoplastic left heart, Down syndrome). Spontaneous abortion was seen in 6(14.6%) cases.Preterm deliveries were seen in 11(26.8%) cases, Term deliveries were 5(12.1%). Immediately after delivery all babies were transferred to paediatric surgeon for further management.Upto 14weeks anomalies like Anencephaly, meningomyeloceleand multiple anomalies were terminated medically by Mifepristone and PGE1 tablets.14-20weeks foetuses which had multiple anomalies and not compatible with life were terminated by Mifepristone followed 12hours later by PGE2 gel smeared on a bougie introduced intracervically. Withanhydromnios fetuses

were terminated after taking consent from parents with Mifepristone followed by PGE2 gel smeared on a bougie.

Table-3: showing Gestational age at which Delivery and termination of pregnancy occurred:

Gestational age at which delivery	No of cases (n=41)	Percentage (%)
Term delivery (37-40 weeks)	5	12.1
Preterm delivery (20-37 weeks)	11	26.8
Spontaneous Abortion (<20 weeks)	6	14.5
MTP (<20 weeks) (Medical termination of pregnancy)	19	46.4
Gender of the baby		
Male	22	53.6
Female	19	46.4

IV. Discussion:

In early pregnancy, major fetal congenital malformations like anencephaly, Myelomeningocele which can be reliably diagnosed at 10-14 weeks of pregnancy. Most of the structural anomalies are increasingly detected with advancing gestation [6,7]. The fetuses with lethal CNS anomalies often die in-utero or in the immediate perinatal period.

In our study majority of thefetal congenital malformations were found maternal age between 20-30 years 31 (75.6%) similar to Bhalerao A, Bhalerao K study[8], this increased risk among

younger parents could be due to genetic or environmental factors. Majority of the congenital anomalies were observed in primigravidas in our study 19(46.3%) similar to Sunitha et al study[9]. In our study majority were from Urban area 23(56%) similar to Silesh M et al [10]study 73%. In this study predominantly affected fetuses were male 22(53.6%) similar to Zahirulalam Md et al[11] study(57.5%). In this present study, the highest proportion of congenitalanomalies were Central nervous system anomalies 27(65.8%) like anencephaly11 (26.8%), followed by spinal bifida 5(12.1%), and the least common was chromosomal type of CAs.Studies conducted bySheeba et al.[12] and Akruti et al.[13]from India and SileshMet al[10]alsoreported CNS anomalies to be the most frequent anomaliesin their studies.Incontrast OkonEkwere E et al study[14]showed Gastrointestinal system anomalies were most commonandinBhalerao A, Bhalerao K study [8]Musculoskeletal system anomalies were most common.Table -4: showing distribution of pattern congenital anomalies in different studies. Pattern of congenital malformations in various geographic regions may also change with ethnicity, socioeconomic level, diet, environmental variables, maternal age, and lifestyle[17].

Table 4: showing frequency of distribution of patterns of congenital anomalies in different studies:

Studies	CNS	Musculoskeletal system	GIT	Renal	CVS
1.Bhalerao A, Bhalerao K[8]	25%	36.9%	16.6%	10.7%	3.57%
2. ZahirulAlam Md et al[11]	30%	24%	9%	-	13%
3.Ekwochi U et al [15]	10.5%	>50%	16.5%	-	-
4 Chimah OU et al[16].	33%	-	30%	-	57%
5.Our study	65.8%	26.8%	4.8%	9.7%	7.2%

CNS: Central nervous system, CVS: Cardio vascular system, GIT: Gastrointestinal system

In this study lack of folic acid supplementation observed in 20 cases (48.7%) with congenital anomalies similar to Tanzania study[18]. In our study those who have migrated for labour work from different parts of India not followed regular antenatal checkups, not taken folic acid tablets during periconceptional period were identified and those women were counselled them about importance of folic acid supplementation during periconceptional period and to seek medical advice in next pregnancy.

V. Conclusion:

In our study congenital anomalies were more likely to be associated with maternal age between 20-30 years of age,primigravida,urban residence. The congenital anomalies affected significantly higher proportion of male babies than female. In this present study Central nervous system anomalies were most common followed

by Musculoskeletal system and Cardiovascular system.Folic acid supplementation pre-pregnancy and ante natal period, regular ante natal visits as per WHO guidelines,prenatal diagnosis and early detection of Congenital anomalies were recommended.

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