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A Study of Profile of Antenatal Mothers Detected To Have Congenital Anomalies in a Tertiary Care Centre, Thiruvananthapuram, Kerala

Dr. Simi Ayesha[1], Dr. Sreekumari Umadevi[2]
[1](Assistant professor, department of OandG, Govt. Medical College, Thiruvananthapuram)
[2](Assistant professor, department of OandG, Govt. Medical College, Thiruvananthapuram)

Abstract
BACKGROUND-The diagnosis of congenital malformation in the antenatal ultrasound puts the expecting motherand the rest of the family in distress and anxiety. High resolution ultrasound and fetal medicine has revolutionized ante natal diagnosis of congenital anomalies at earlier gestational ages. This allows the expectant mothers to identify the extend of anomalies and in consultation with pediatric surgeon and neonatologist frame appropriate mode of management and the option for termination of pregnancy in case of major anomalies. This study aimed at finding out the incidence of congenital anomalies among antenatal mothers registered in this institution for one year, any identifiable antenatal risk factors, classify them according to the system involved and compare with data from other studies
MATERIALS AND METHODS-*This is an institution based prospective observational study, conducted in the Department of obstetrics and Gynecology,SAT hospital, Government Medical College, Thiruvananthapuram,Kerala.The study was conducted for one year from July 2016 to June2017. Among those antenatal women registered, those detected to have congenital anomalies were studied in detail using structured proforma and analyzed.*
RESULTS-*Among the total 10331 antenatal admissions, 274 women were found to have congenital anomaly by ultrasound. Hence the incidence of congenital anomalies in this institution was found to be 2.6%. Urinary system anomalies(25.54%) were the majority, followed by circulatory system(21.89%) and nervous system(21.16%).*
CONCLUSION
Prenatal counselling in case of consanguinous marriages and periconceptional folic acid in cases of bad obstetric history, adequate blood sugar control in case of overt diabetes form methods of primary prevention First trimester aneuploidy screening with nuchal translucency and double markers should be made mandatory as it is evident from the study that congenital anomalies were seen in young, unsuspecting primi or multigravidas. In case of those identified of congenital anomalies, proper counselling in conjunction with fetal medicine consultant ,neonatologist, pediatric cardiology, paediatric surgeon and appropriate management options including MTP for lethal defects are the options available at present.
Keywords- *prenatal counseling ,aneuploidy screening*

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I. Introduction

Congenital anomalies can be defined as structural or functional anomalies that occur during intrauterine life. Also called birth defects, congenital disorders, or congenital malformations, these conditions develop prenatally and may be identified before or at birth, or later in life. An estimated 6% of babies worldwide are born with a congenital anomaly, resulting in hundreds of thousands of associated deaths. However, the true number of cases may be much higher because statistics do not often consider terminated pregnancies and stillbirths.

Some congenital anomalies can be treated with surgical and non-surgical options, such as cleft lip and palate, clubfoot, and hernias. Others, including heart defects, neural tube defects, and down syndrome, can cause lifelong impacts[1]. Thus the diagnosis of congenital malformation in the antenatal ultrasound puts the expecting mother and the rest of the family in distress and anxiety. Moreover congenital anomalies remain a significant but under recognized cause of mortality and disability among infants and children under five years of age. They can be life-threatening, result in long-term disability, and negatively affect individuals, families, health-care systems and societies[2].

How can we eradicate this social disaster? The report accompanying the resolution of the Sixty-third World Health Assembly (2010) on congenital anomalies describes the basic components for creating a national programme for the surveillance, prevention and care of congenital anomalies before and after birth[2]. Surveillance to detect the regional pattern of congenital anomalies,public health education in preventing the causative factors and early detection and correction of anomalies are the only way out. This study aims to analyse the profile of antenatal mothers detected to have malformed fetuses by USS getting admitted in a tertiary care center during the study period.

II. Materials And Methods

STUDY DESIGN- This is an institution based prospective observational study.

SETTINGS OF THE STUDY- The study is conducted in the Department of obstetrics and Gynecology, SAT hospital, Government Medical College, Thiruvananthapuram, Kerala.

STUDY DURATION- Duration of the study is for one year. (June 2016 - July 2017)

SAMPLE SIZE : 274 cases

STUDY SUBJECTS- All patients registered in SAT hospital with antenatally detected anomalies are taken as the study population and included in the study. Anomalies are defined as the gross structural defects as detected by antenatal ultrasound.

EXCLUSION CRITERIA.- Those mothers who do not give consent for the study are excluded.

PROCEDURE

Clearance from the institutional research committee and institutional ethics committee was obtained before starting the study. Cases were identified by visiting the antenatal wards and the identified cases were analysed in detail regarding pre existing medical conditions, previous obstetric history, history of any congenital anomalies in the family, folic acid prophylaxis,any drug intake, radiation exposure, history of fever with rash in the first trimester and substance abuse.

Their ultrasound findings regarding at what gestational age it was diagnosed and what type of anomaly was detected are noted. These cases are followed up till delivery and at birth the pediatrician' examination findings are noted. Any other associated anomalies detected, investigations done to confirm the malformations are noted.

ANALYSIS

The above data will be entered in a structured proforma and analyzed in excel sheet. Qualitative data will be expressed in proportions and quantitative data using mean and standard deviation.

III. Results

During the study duration of one year a total of 274 cases were detected to have one or more anomalies in their ultrasounds done from first trimester to third trimester. There were total of 10331 antenatal admissions. This comes to a 2.6% incidence of congenital anomalies.

Majority (n=199,72.6%) belonged to the age group of 21-30 years .fifty four(19.7%) belonged to 31-39 years age groups. And 21(7.66%) were less than 20 years of age.Regarding the residence majority (n=182,66.42%) were residing in rural areas and 92(33.57%) resided in urban areas. Regarding the socioeconomic status majority (n=227,82.8%) belonged to below poverty line and 47(17.2%) were above povertyline. Majority(n=186,67.88%) were booked that is had at least 3 antenatal visits in the study setting, 85(31.02%) were booked outside, three cases(1.09%) were unbooked or did not have any antenatal checkups.**(Table 1)**

Majority(n=130,47.4%) were primigravidas,90(32.8%)were second gravidas ,30(10.92%) were third gravidas ,22(8.02%) were fourthgravidas and there were one(0.36%) fifth and sixth gravida each.Going into further details of previous obstetric outcome 52(18.97%) had previous normal live born babies.58(21.16%) had at least one abortions in the past.4(1.45%) had medical termination of pregnancy previously and 11(4.01%) and 3(1.09%) had previous history of intra uterine deaths and neonatal deaths respectively. Only10(3.65%) had documented previous history of infertility treatment in the past. First ,second or third degree consanguinity were noted in 4 (1.5%) of mothers. First trimester risk factors like TORCH infections and no history of periconceptional folic acid intake were seen in 3(1.09%) and5(1.8%) respectively

Analysis of antenatal illnesses in mother's of anomalous fetus revealed that majority (n=115,41.97%)had no antenatal comorbidities. But diabetes complicating pregnancy was found to be a significant comorbidity in these mother's(n =58,21.16%) Of these 47(17.15%) had gestational diabetes and 11 (4.01%)had overt diabetes. Another 22(8.02%) had associated hypertensivedisorders. Other major comorbidities were hypothyroidism (n=36,13.13%) and anemia(n=22,8.02%). Another 28 mother's (10.2%) had infections like urinary infections and lower respiratory infections in the antenatal period.**(Table2)**

Due the anomaly in the fetus, associated fetal complications were looked for. Majority(n=145,52.9%) did not have any fetal complications detected antenatally but fetal growth restriction(n=45,16.4%),oligoamnios (n=42,15.3%), polyhydramnios(n=18,6.5%), and abnormal Doppler (n=10,3.6%) were found to be associated in these fetuses, which cannot be overlooked.**(Table 3)**

Majority of the anomalous fetuses (n=214,99.3%)were singleton pregnancies, only three were multiple pregnancies. Of these 203(47.08%) had vertex presentation s,12(7.29%) breech presentation and 1(0.36%) had transverse lie in the fetuses. Majority (n=124,45.2%) had vaginal delivery as the mode of termination of pregnancy. 93(33.9%) underwent lower segment cesarean section,3 patients (1.09%) had hysterotomy as the mode of termination as they had previous 2 caesarean sections in the past.54(19.7%) underwent medical termination of pregnancy as they had the anomaly detected before 20 weeks which is the legal government accepted cutoff for MTP in the study setting[3].

Caesarean section rate was high in the study. Majority (n=29,31.18%) underwent cesarean as they had previous history of one or more cesarean sections..FGR and abnormal Doppler became the indication in 5(1.82%) and 112(12.9%) of cases respectively .Failed induction, fetal distress and malpresentations were seen in 17(18.2%) ,12(12.9%) and 12(12.9%) respectively. At birth206(94.9%) were live babies 11(5.1%) were dead born and another11(5.1%) went for neonatal death.

The congenital malformations were classified as per ICD 10 chapter 17[4] classification. Majority (n=70,25.54%) had urinary system anomalies followed by circulatory system anomalies (n=60,21.89%) and nervous system anomalies (n=58,21.16%). Anomalies involving multiple systems were 19 in number(6.93%). Anomalies involving the respiratory system and musculoskeletal system were 17 (6.20%). Anomalies of eyes,ear, face ,neck were 10 in number(3.6%) followed by 8 (2.91%) cleft palate and cleft lip detected in the antenatal scans. There were only 4(1.45%) anomalies of the digestive system and 3(1.09%) involving genital organs. There was one(0.36%) case of aplasia cutis or malformation of the skin. Seven(2.55%) cases of chromosomal syndromes were detected in antenatal scans.**(Table 4)**.

Majority of anomalies were that of the urinary system (CAKUT). Of these renal pelvis dilatation as detected in antenatal scans at various gestational age was the commonest (n=37,52.85%) and hydroureteronephrosis was (n=15,21.43%) the next commonest, followed by multicystic kidneys (n=13,18.57%). There were three (4.28%) cases of pelviureteric junction obstruction and 2 cases (2.8%) of congenital absent kidneys **(Table 5)** .

Circulatory system anomalies were next in abundance 14 cases of Transposition of great arteries (23.34%), 6 cases (10%) each of Tetralogy of fallot, ventricular septal defect, echogenic foci in heart, pericardial effusion with or without cardiomegaly were detected. Next commonest cardiac lesion detected was the univentricularheart(n=7,11.6%). There were 4 cases (6.66%)of rhythm abnormalities. Three cases(5%) each of Atrial septal defect and Coarctation of aorta and two cases(3.3%) of rhabdomyoma were detected. One case each of double outlet right ventricle, Ebstein anomaly and dilated tricuspid valve was also detected.

Nervous system anomalies accounted for 21.16% of total. Of these neural tube defects(n=19,32.75%) were the commonest. Ventricular system anomalies (n=15,25,86%) were next followed by 6(10.34%) midline anomalies like holoprosencephaly,corpuscallosal agenesis and cavum septum pellucidum abnormalities. There were 5 (8.62%) of destructive lesions of the cortex like hydranencephaly,intraparenchymal dystrophy and intracranial lesion. There were 4(6.8%) cases of choroid plexus cyst and dolichocephaly each. There were 2(3.44%) cases each of microcephaly and posterior fossa abnormalities and one (1.7%) case of vein of galen aneurysm **(Table 5).**

Of the respiratory tract anomalies, majority were congenital diaphragmatic hernia(n=15,88.2%)and rest(n=2,11.7%) pulmonary airway malformation. Of the musculoskeletal abnormalities, limb abnormalities like CTEV(6) and one case of rhizomelia were detected (n=7,41.2%). Skeletal dysplasias also made up 41.7%(n=7) in this category. Anterior abdominal wall defects like omphalocele formed 17.6%(n=3) of this category. Multiple organ anomalies (n=19,6.93%) were seen. Chromosomal syndromes like Turner's Downs were detected in antenatal scans. Digestive system anomalies detected had 1 case each of duodenal atresia, segmental bowel dilatation, anorectal malformations and choledochal cyst.

IV. Discussion

The incidence of congenital anomalies in the study setting was determined to be 2.6%. This is in agreement to studies done in other parts of the country like Ahmedabad[5] and Lebanon[6]. Some other studies from India give a lower incidence of anomalies [7,8].

This wide variation was said to be due to various racial, socio-cultural and ethnic influences[9]. Moreover the study setting is one of the largest tertiary care centers in Kerala, India where antenatal mothers from all across the state get referred to in view of its proximity to premium institutes like SCTIMST where corrective surgeries for cardiac anomalies are being done.

Majority of mothers belonged to the age-group 20-30yrs. This observation is not in agreement to the proven association between increasing maternal age and increased incidence of anomalies. This association needs further ratification as there are studies from other parts of Kerala [7] and Assam[8] where the majority of mothers belonged to 20-30 years . The increased incidence of anomalies in lower socioeconomic class was reaffirmed in this study.

Majority of fetal anomalies were seen in primigravidas who were taken for surprise due the detection of anomaly in the ultrasound. But having a history of abortions , intrauterine deaths, neonatal deaths in the past was seen to be more common on these antenatal women as seen in studies from other parts of kerala[7] and Mumbai[10]. Thus antenatal mothers with bad obstetric history should be worked up for fetal anomalies.

Diabetes complicating pregnancy was found to be the most common associated maternal comorbidity as seen in studies from Egypt[11,12]. Other important associations were that of hypothyroidism, maternal anemia and maternal infections like urinary infections or lower respiratory tract infections. The incidence of major malformations in women with type 1 diabetes is at least doubled and approximates 11 percent [13].Gestational diabetes is defined as carbohydrate intolerance of variable severity with onset or first recognition during pregnancy[14]. This definition undoubtedly includes some women with previously unrecognized overt diabetes.

Hypothyroidism being an endemic disease in India the prevalence in the study setting is comparable to that in the general population of south India.[15] and is not a proven risk factor for congenital anomalies. Maternal infections like upper and lower respiratory tract infections caused by respiratory viruses are known to cause fetal malformations like anencephaly[16] moreover diarrheal illnesses caused by coxsackie viruses are also teratogenic[17]. This along with TORCH infections predispose to fetal malformations. Even though anemia is not an independent risk factor for fetal malformations anemia and poor nutritional status in the setting of immunocompromised status of pregnancy predisposing to infections could be increasing the risk for fetal anomalies. Prevention of anemia in adolescents and antenatal period by iron and folic acid supplementation remains very much relevant.

Fetal effects caused by anomalies varied from fetal growth retardation, oligoamnios and polyhydramnios. These strong associations are also seen in other studies done in Thiruvalla[7] and Odisha[18]. This finding can help us in counselling antenatal mothers detected to have FGR and amniotic fluid abnormalities without any known cause.

Most common congenital anomaly detected was of the urinary system, as seen in other studies done in kerala[7], and Belgium[19]. The most common anomaly that was detected was renal pelvis dilation or pyelectasis -52.8%.. The detection of fetal pyelectasis by ultrasound has gained importance for two main reasons: first, as a marker for aneuploidy and, second, as a precursor of postnatal urinary tract pathology like pelvi-ureteric junction obstruction (PUJO), vesico-ureteric reflux (VUR), early signs of outflow obstruction, duplex systems, multicystic dysplasia and upper tract dilatation in the absence of obstruction. The detection of prenatal pyelectasis should prompt a detailed anomaly scan looking for extra-renal anomalies and other markers of aneuploidy[20].

The second common malformation was involving the circulatory system. Commonest anomaly in this group being transposition of great arteries TGA-23.34%.TGA a mostly surgically correctable disorder was one of the reasons for reference to the study setting, not only from all over the state of Kerala but from neighbouring state of Tamilnadu also

The third most common anomaly was that of the nervous system commonest being neural tube defects- 32.5% . This was seen in many studies from Assam[8], Ahmedabad[21] in India and Iran[22] . Among the musculoskeletal system anomalies limb abnormalities like CTEV and skeletal dysplasias were found to be common.

V. Conclusion

Preventive measures against congenital anomalies should start right from adolescence, in the form of correcting anemia, having a balanced nutritious diet and regular exercise to prevent obesity. Government supported programmes like WIFS [23,24] for iron and folic acid supplementation should help in reducing the prevalence of anemia especially in rural areas. Prenatal counseling in overt diabetes and women taking medications for medical illnesses is very important. Such counseling and evaluation is also mandatory in

women with previous history of abortions, intrauterine death or neonatal death along with prenatal folic acid supplementation.

After conception, expectant mothers should avoid contact with viral fevers, have home prepared meals in a clean environment. Regular and timely antenatal visits with a proper scan for nuchal translucency and nasal bone is imperative. This along with double markers before 14 weeks can help detect congenital anomalies at the earliest and terminate the pregnancy if needed. Moreover early detection of anomalies in rural settings can allow the patients being referred to a higher center where fetal medicine experts are there or in utero procedures,or EXIT procedures can be done in consultation with a pediatric surgery team. First trimester aneuploidy screening with nuchal trancluscency and double markers should be made mandatory as it is evident from the study that congenital anomalies were seen in young, unsuspecting primi or multigravidas

Table 1:

Age (years)	No.	Percentage(%)
< 20	21	7.66
21 - 30	199	72.6
31 - 39	54	19.7
Residence		
Rural	182	66.42
Urban	92	33.57
Income		
APL	47	17.2
BPL	227	82.8
Total:	274	100

Table 2:

Maternal Diseases	Frequency	Percentage(%)
Nil	115	41.97
Diabetes GDM Overt	 47 11	 17.15 4.01
Hypothyroidism	36	13.13
Maternal Infections	28	10.2
PIH	22	8.02
Anemia	22	8.02
PIH & GDM	12	4.37
Fibroid	5	1.80
Bronchial Asthma	4	1.4
Heart Disease	3	1.09
Epilepsy	3	1.09
ITP	3	1.09
SLE	3	1.09
Drug Allergy	4	1.4

Table 3:

Foetal Complications	Frequency	Percentage(%)
Nil	145	52.9
FGR	45	16.4
Oligamanios	42	15.3
Polyhydramnios	18	6.5

Abnormal Doppler	10	3.6
Bradycardia	4	1.4
Macrosomia	3	1.09
IUFD	3	1.09
Antepartum Hemorrhage	2	0.7
PPROM	2	0.7

Table 4:

System involved	frequency	percentage(%)
Urinary system	70	25.54
Circulatory system	60	2189
Nervous system	60	21.16
musculoskeletal	17	6.2
respiratory	17	6.2
Cleft lip cleft palate	8	2.91
Chromosomal syndromes	7	2.55
digestive	4	1.45
genital	3	1.09
Eyes, ear,face and neck	10	3.6
skin	1	0.36
Multiple system involvement	19	6.93

Table 5

Nervous system		
• Neural tube defects	• 19	• 32.75
• Ventricular abnormalities(Hydrocephalus,Aqueductal stenosis)	• 15	• 25.86
• Microcephaly		
• Midline anomalies (like, holoprosencephaly, corpus callosal agenesis)	• 2	
• Posterior fossa abnormalities(like arnoldchiari and hypoplastic cerebellum)	• 6	• 3.4 • 10.34
• Vein of Galen aneurysm		
• Destructive lesions(like hydranencephaly, intraparenchymal dystrophy, etc)	• 2	• 3.4
	• 1	• 1.7
	• 5	• 8.62
Urinary system (CAKUT)*		
• Renal pelvis dilatation	• 37	• 52.85

• Hydronephrosis • Multicystic kidney • Renal agenesis • PUJ Obstruction	• 15 • 13 • 2 • 3	• 21.43 • 18.57 • 2.8 • 4.28
Circulatory system • TGA • Univentricular heart • TOF • Echogenic foci in heart • Pericardial effusion, cardiomegaly • VSD • Rhythm Abnormalities • ASD • Coarctation of Aorta • Rhabdomyoma • Dilated TV, DIlated atria • DORV • Ebstein	• 14 • 7 • 6 • 6 • 6 • 6 • 4 • 3 • 3 • 2 • 1 • 1 • 1	• 23.34 • 11.6 • 10 • 10 • 10 • 10 • 6.66 • 0.6 • 5 • 3.3 • 1.6 • 1.6 • 1.6
Respiratory system • CDH • Pulmonary airway malformation	• 15 • 2	• 88.2 • 11.7
Musculo-skeletal system • Limb abnormalities (like CTEV, Rhizomelia) • Skeletal dysplasia • Omphalocoele	• 7 • 7 • 3	• 41.2 • 41.2 • 17.6
Digestive system • Duodenal atresia • Segmental bowel dilatation • Ano-rectal malformation • Choledochal cyst	• 1 • 1 • 1 • 1	• 0.25 • 0.25 • 0.25 • 0.25

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