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Analysis of Pattern of Congenital Anomalies Diagnosed in A Tertiary Care Hospital

Dr. Vanitha.V	MD, Senior Assistant Professor ., Institute of Obstetrics and Gynaecology., Madras Medical College
Dr. Shanthi.S	MD, DGO, Senior Assistant Professor ., Institute of Obstetrics and Gynaecology., Madras Medical College
Dr. S. Usha Rani	MD., DGO, Professor, Institute of Obstetrics and Gynaecology. Madras Medical College

ABSTRACT

This is a hospital based prospective analysis conducted in the department of obstetrics and Gynaecology, tertiary care government hospital, Chennai during the period Aug 2013 to April 2014.The aim of the study was to find the incidence, pattern of structural congenital malformations, the most common anomaly, its etiology and the role of prenatal USG in its early detection in our population. During the study period 9500 women were admitted for delivery. Since our hospital is a tertiary care centre, all anomalous fetuses referred had an USG done with varying levels of expertise from different centres. All these women were further evaluated and anomalies were confirmed by our in house radiologist. Genetic counselling and early termination of pregnancy was offered to all major lethal anomalies diagnosed. A thorough post-natal screening and examination was performed on all infants born and all anomalies were recorded. The pattern of congenital anomalies found included CNS(25%), CVS (21%), Musculoskeletal(15%), GIT(4%), Renal/GUT (8%). 40% of the major anomalies were detected prenatally. In 66% of those antenatally diagnosed with congenital anomaly, pregnancy was terminated. This study stresses the importance of a thorough clinical examination of all neonates and an echo when suspected as many women have missed a target scan at level 2 centres. CNS anomalies were the most common anomaly detected. Among CNS defects NTD was the common anomaly detected by USG. Serum folate level was deficient in 35% of the cases with neural tube defects. This study also stresses the need and significance of periconceptional folic acid supplementation, importance of target scan which aid in terminating an anomalous fetus at an earlier gestation thereby reducing perinatal mortality and parental anxiety.

KEYWORDS congenital anomalies,malformation,prenatal diagnosis,Neural tube defects

INTRODUCTION

Congenital anomalies can be defined as a structural or functional abnormalities present at birth. It occurs in 2 -3% of all births. These contribute to 8 -18% of perinatal mortality and morbidity [1] and 10 -15% of neonatal deaths. [2, 3, 4]

Survivors had severe mental and physical disability [5]. The goal of prenatal diagnosis and fetal medicine is to detect these 2 to 3 % of women with congenital anomaly and offer genetic counselling, further testing, early termination or delivery and to even plan perinatal surgeries. This study was intended to document the pattern of congenital anomalies detected prenatally and also the prevalent pattern of anomalies in the new born of this region. [6, 7]

Materials and methods:

This prospective study was done in a government tertiary care hospital, Chennai. 9500 women were admitted for delivery during this period of study from August 2013 to April 2014. Since our hospital being a tertiary care centre, all anomalous fetuses referred had an USG done with varying levels of expertise from different centres. All these cases were further evaluated and anomalies were confirmed by our in house radiologist. Genetic counselling and early termination of pregnancy was offered to all women diagnosed with major lethal anomalies. A thorough post-natal screening and examination was performed on all infants born and all major and minor anomalies were documented. The demographic details recorded were gestational age at diagnosis of fetal anomaly, type of anomaly, commonest anomaly prevalent, etiological, and risk factor if any, sex, mode of delivery/termination were recorded.

RESULTS:

Total number of deliveries during our study period was 9302. Congenital anomaly (major/minor) was found in 223 neonates detected either antenatally or postnatally. The incidence of congenital anomaly in our study is 2.4 %. The pattern of congenital anomalies found included CNS(25%), CVS (21%), Musculoskeletal(15%), GIT(4%), Renal/GUT (8%).(Fig-1) Antenatal USG detected 77 cases of which 60 were lethal anomalies and 17 were minor anomalies. Early termination of pregnancy was offered to these mothers (66%) (Table 1). This significantly contributed to the reduction in perinatal mortality and parental anxiety.

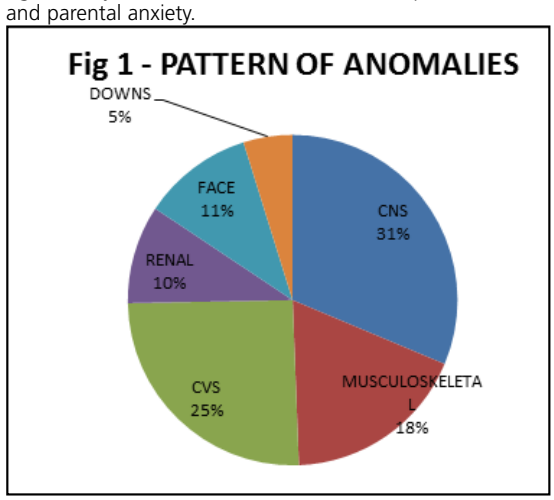


TABLE-1
Perinatal outcome of prenatally diagnosed congenital anomalies

PERINATAL OUTCOME	NO.OF CASES
Termination of pregnancy	43
IUD/still birth	7
LSCS	7
Labour natural	20

Postnatally 146 anomalies were detected of which 62 were major anomalies missed in antenatal USG.

TABLE- 2
Demographic distribution of anomalies

Maternal age<20 20-25 25-30 >30	29% (42) 40% (58) 22% (32) 9% (14)
Sex –female Male	51% (75) 49% (71)
Primi gravida Multi gravida	47% (69) 53% (77)
Consanguinity	5% (7)

S.No	TYPE OF ANAMOLY	N	%
1	Central nervous system Hydrocephalus,anencephaly,meningo-myelocele,holoprocencephaly,lissen-cephaly,schizencephaly,microcephaly,CP cyst,dandy walker variant	56	25%
2	CVS ASD, VSD,TOF,PDA,TGV,AVSD,hypoplas-tic left venytricle,isomerism	47	21%
3	Musculo skeletal CTEV,Polydactyly,OI,Achondroplasia,mi-cromelia,arthrogryposis,body stalk anomaly	34	15%
4	Genitourinary tract Hydronephrsis, pelvic kidney, renal agenesis, cystic dysplasia, LUTO, PUJ obstruction, ureterocele.	18	8%
5	Face Cleft lip/palate, facial dysmorphism	21	9%
6	Downs	8	4%
7	GIT Meconium ileus, duodenal atresia, malrotation of gut, anorectal anomalies, omphalocele.	9	4%
8	Multiple anomalies	11	5%
9	Respiratory system Brochopulmonary dysplasia, CDH and others	6	2%
10	Others Micropenis, hypospadias, single umbil-ical artery.	13	6%

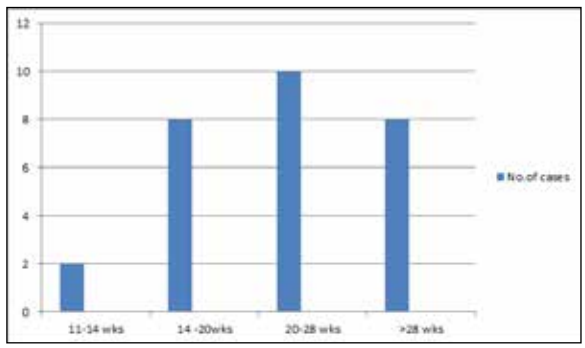
Table 3. Pattern of congenital anomalies

Antenatal USG diagnosed major anomalies in 60 of 119 total fetuses with major anomalies.75 % of major cardiac anomalies were detected antenatally. Subtle cardiac anomalies like ASD,VSD and PDA were missed in the antenatal USG.CNS defects were detected antenatally with highest accuracy in 65 % of cases.28 cases of NTD were reported of which 22 were identified prenatally in USG, making it the single most group of anomaly effectively diagnosed.The incidence of NTD was 2.6 % in our study.18 % of cases had history of consanguinous marriage and 60% occurred in primiparous women.32% of NTD occurred in parous women within 6 months of abortion.Serum folic acid levels were in the deficient range(<5.3 ng/ml) in 4 0f 11 (35%) patients diagnosed prenatally before 20 wks with NTD.71% of neural tube defect were in female fetuses.

Table 4.

TYPES OF NTD	NO.OF CASES
Anencephaly	6
Hydrocephalus with spinabifida	2
Acrania	1
Exencephaly	1
Encephalocele	5
Meningomyelocele	10
NTD with multiple defects	3

Types of NTD



Discussion

Congenital malformations account for 8-18% of perinatal mortality and the third most important factor in perinatal mortality next to birth asphyxia and prematurity. In the present study the incidence of congenital anomalies is2.4 %which is similar to the findings of ICMR, Annual report 1989(8,9).40 % of the total anomalies had been detected prenatally and termination was done early in 56 % of cases(Table.1).Analysis of overall distribution of malformations in the present study showed that CNS was the commonest system involved (25%) followed by cardiovascular(21%) and musculoskeletal(15%) (Table3).Similar observations of commonest CNS involvement(39%) was reported by S.Swain, A.AgarwalIndian paediatrics.net;oct 1994.Among CNS defects NTD were the commonest contributing to 50% of the total CNS Anomalies[10]. similar observations were reported in analysis of incidence of major fetal anomalies using USG in northern Kerala where USG done for 30030 cases,857 anomalies (25.9%)picked up and reported 39.20% CNS anomalies, of which 250 cases were NTDS by Balakumar K[10,11].Serum folic acid levels were in the deficient range in 4 0f 11(<5.3 ng/ml)35% in patients diagnosed prenatally before 20 weeks with NTD in our study. 71% of affected fetuses were female; such female sex bias in NTD is also reported from several studies. seller MJ Division of medical and molecular genetics, Guys hospital, London study 32% of NTD occurred in parous women within 6 months of abortion this stresses on the need for folic acid supplementation to this high risk group of women our finding is similar to Elahmmohasheri et al study from north Iran where folate deficiency was found in 35.4% of cases of NTD[12]. Primary prevention by periconceptional folic acid supplementation and maternal serum AFP and a target USG can prevent the occurrence and early detection of defects[13]. Our study emphasises the role of USG in early diagnosis of congenital anomalies and accurately identifying major anomalies and thereby reducing perinatal mortality.

Conclusion

This study helps to know the pattern of congenital anomalies prevalent in Chennai. Among the commonest CNS disorders the prevalence of NTD was high. Also our study detected folic acid deficiency in 30% of cases with neural tube defects (only 10 women were screened). Folic acid deficiency should be prevented by periconceptional folic acid supplementation. Screening strategies should be made accessible to all, since congenital malformation can occur even in a very low risk pa-

tients. With improved control of infection and nutritional deficiency, congenital malformations are becoming an important determinant in perinatal mortality. By early prenatal screening and diagnosis using USG and maternal serum markers lethal anomalies can be terminated and perinatal mortality can be decreased.

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