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Original Research Article

STRUCTURAL ANOMALIES DETECTED ON ANTENATAL ULTRASOUND AND ITS ASSOCIATION WITH CONSANGUINITY, PREVIOUS ANOMALIES, MATERNAL DIABETES AND HYPERTENSION IN PATIENTS ATTENDING THE TERTIARY CARE HOSPITAL, ADILABAD, TELANGANA

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Abstract

structural congenital anomalies have been known since time immemorial and are considered as abnormalities of structure present from birth. Serious birth defects are life-threatening or have the potential to result in disability. It is a global problem, but their impact is particularly severe in middle and low-income countries. This study is done to assess the magnitude of congenital anomalies, patterns of congenital anomalies and the association of congenital anomalies with consanguinity, previous anomalies, maternal diabetes and hypertension. This is a retrospective observational study done in the department of radiology, Rajiv Gandhi institute of Medical Sciences. A total of 38,303 antenatal ultrasound scan reports (from July 2017 to March 2022) were reviewed. The overall percentage of fetuses with visible congenital malformation in the present study is 1.45%, the most common organ system anomaly was central nervous system and involved in 149(26.8%) patients. Other systems involved are the following, genitourinary system141 (25.3%), multiple systems 90 (16.2%), cardio vascular system 58 (10.3%), musculo skeletal system 56(10%), facial 36 (6.5%), gastrointestinal system in 10 (1.7%), lymphatic system in 15 (2.6%), Respiratory system in 3 (0.5%). There is history of consanguinity was there among 27.5% of the antenatal mothers. Early identification of gross structural anomalies by second trimester ultrasound scans and counselling of antenatal mothers, genetic screening will help in reduction in the incidence of congenital anomalies at birth.

Key words: Structural anomalies, Antenatal ultrasound

Introduction

Congenital anomalies can be defined as structural or functional anomalies that occur during intrauterine life and can be identified prenatally at birth or sometimes may only be detected later in infancy, such as hearing defects. In simple terms, congenital refers to the existence at or before birth[1].

Congenital anomalies are one of the main causes of the global burden of disease. An estimated 6% of babies worldwide are born with a congenital anomaly, resulting in hundreds of thousands of associated deaths[1]. The impact of congenital anomalies is severe in middle and low-income countries, it is estimated that approximately 95% of the children who die from birth defects are from those countries[2]. In India congenital anomalies constituted the fifth largest cause, being responsible for an estimated 9% of neonatal deaths in the year 2010[3].

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About 60% of the causes of congenital anomalies in humans are still unknown[3]. However, in about 25% of congenital anomalies, the causes seem to be "multifactorial", indicating a complex interaction between genetic and environmental risk factors[4].

Genes play an important role in many congenital anomalies. This might be through inherited genes or through mutations. Consanguinity also increases the prevalence of rare genetic congenital anomalies and nearly doubles the risk[1].

Maternal age is also a risk factor for abnormal intrauterine fetal development. Advanced maternal age increases the risk of chromosomal abnormalities, including Down syndrome. Maternal exposure to certain pesticides and other chemicals, as well as certain medications, alcohol, tobacco and radiation during pregnancy, may increase the risk of having congenital anomalies. Maternal infections such as syphilis and rubella are a significant cause of congenital anomalies in low and middle-income countries. Maternal folate insufficiency increases the risk of having a baby with a neural tube defect while excessive vitamin A intake may affect the normal development of an embryo or fetus[1].

The pattern of congenital anomalies varies from region to region and also over time[5]. Generally, congenital anomalies that involve the CNS and the cardiovascular and musculoskeletal systems have been reported to be the most common [6, 7, 8].

Understanding the patterns and risk factors associated with congenital anomalies and early prenatal detection of congenital anomalies is very essential in tackling the problem effectively. Ultrasound can be used to screen for Down syndrome and major structural abnormalities during the first trimester and for severe fetal anomalies during the second trimester. Ultrasound is a highly specific and sensitive imaging modality for detection of congenital anomalies in fetuses during sonographic evaluation of antenatal mothers [9, 10].

The true magnitude of the number of births affected by congenital anomalies in this region is unknown due to lack of established birth defects surveillance.

By this study we will be able to assess the magnitude of congenital anomalies, patterns of congenital anomalies and the association of congenital anomalies with consanguinity, previous anomalies, maternal diabetes and hypertension and more prevalent cause of congenital anomalies in this area. This study will further guide us to improve existing health facilities for providing basic reproductive health practices, as well as medical genetic screening and counselling.

Methodology:

This Institutional based retrospective observational study was conducted under the department of radio diagnosis, Rajiv Gandhi Institute of Medical Sciences, Adilabad. Rajiv Gandhi Institute of Medical Sciences is the only tertiary care institute catering to the population of Adilabad and surrounding districts of Asifabad, Nirmal, Mancheeryal located in Telangana state. There about 60% tribal populations in this area. The department of radio diagnosis of Rajiv Gandhi Institute of Medical Sciences provides services to all this population.

After obtaining the institutional ethical committee permission, all the case records of antenatal mothers from July 2017 to March 2022, who came for ultrasound scan to the department of radio diagnosis, were reviewed. The details of antenatal mothers who were detected to have a

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fetus with structural congenital anomalies on ultra sound scan were noted. Antenatal mothers with gestational age less than 10weeks were excluded.

The Demographic and clinical information, ultrasound imaging findings were noted from the records. The Data was entered into Microsoft Excel version 10.0 and was analyzed using SPSS version 20.0. Descriptive summary using frequencies, proportions, graphs, and cross tabs was used to display study results.

The mothers who were carrying a fetus with congenital anomalies, referred to department of Obstetrics and Gynecology for further management.

Results:

A total of 38,303 antenatal ultrasound scans were done during the study period (July 2017 to March 2022). Among them, 558 antenatal mothers found to be carrying babies with structural anomalies. The overall percentage of fetuses with visible congenital malformation in the present study is 1.45%.

In present study, it was found that a majority of mothers with fetal congenital anomalies belong to age 18-27 years (Table 1)

Table 1: Age group wise distribution of pregnant mothers

Age groups	Frequency	Percentage
<18	NIL	0 %
18-22	242	43.4 %
23-27	246	44 %
28-32	55	9.9 %
33-37	12	2.2 %
>37	3	0.5 %
Total	558	100 %

In the present study, the most common organ system anomaly was central nervous system and involved in 149(26.8%) patients. Other systems involved are the following, genitourinary system141 (25.3%), multiple systems 90 (16.2%), cardio vascular system 58 (10.3%), musculo skeletal system 56(10%), facial 36 (6.5%), gastrointestinal system in 10 (1.7%), lymphatic system in 15 (2.6%), Respiratory system in 3 (0.5%) individuals (figure 1).

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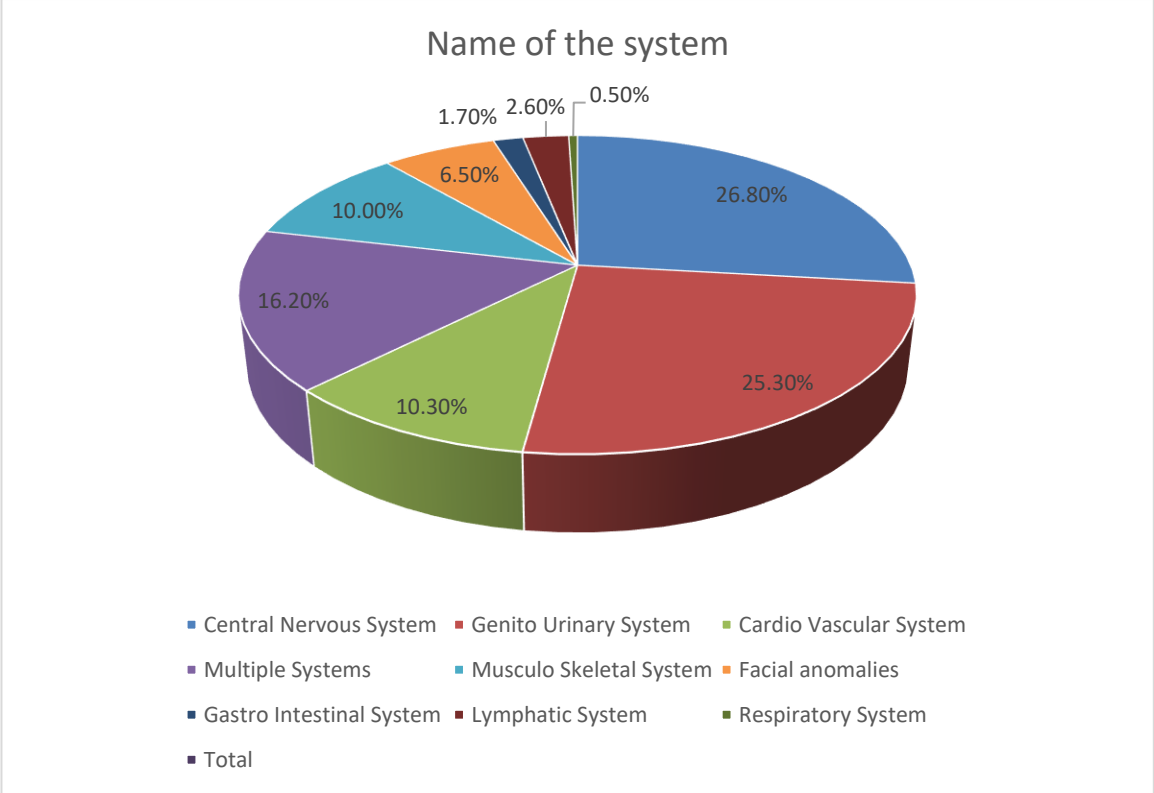

Figure 1: Different systems associated with anomalies

System specific anomalies are presented from table 2 to 9.

Table 2: Specific CNS anomalies

CNS anomalies	Frequency	Percentage (%)
Anencephaly	31	20.8 %
Hydrocephalus	24	16.2 %
Dilated ventricles and mega cistern magna	22	14.7 %
Arnold chiary malformation	18	12 .2%
Meningocele	16	10.7 %
Meningo myelocele	16	10.7 %
Encephalocele	8	5.4 %
Corpus callosal agenesis	7	4.3 %
Spina bifida	4	2.6 %
Arachnoids cyst	1	0.6 %
Dandy walker malformation	1	0.6 %
Microcephaly	1	0.6 %
Total	149	100 %

Table 3: Specific Renal congenital anomalies

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Renal anomalies	Frequency	Percentage
Hydro nephrosis	62	43.9 %
Bilateral PUJ obstruction	31	21.9 %
Non visualized kidney	17	12.3 %
Ectopic kidney	16	11.3 %
Multi cystic dysplastic kidney	12	8.5% %
Autosomal ressive polycystic kidney disease	1	0.7 %
Absent urinary bladder	2	1.4 %
Total	141	100 %

The Central nervous system, gastro intestinal system, musculoskeletal system and cardiovascular system are the most common systems involved in multiple system anomalies.

Table 4: Specific Cardio vascular congenital anomalies

Cardio Vascular Anomalies	Frequency	Percentage
Ventricular Septal Defect	26	44.8 %
ASD with other defects	9	15.5 %
Hypoplastic left atrium	5	8.6 %
ASD & VSD	4	6.8 %
Dilated rt atrium	4	6.8 %
Small rt ventricle	4	6.8 %
Dilated left atrium	3	5.1 %
Hypoplastic heart	2	3.4 %
Transposition of great arteries	1	1.72 %
Total	58	100 %

Table 5: Specific musculoskeletal anomalies

Musculo skeletal anomalies	Frequency	Percentage
Club foot	42	75.8 %
Short limbs	12	20.6 %
Absent bilateral thighs	1	1.8 %
Kyphoscoliosis	1	1.8 %
Total	56	100 %

Table 6: Specific Facial anomalies

Facial anomalies	Frequency	Percentage
Cleft lip	20	55.6%
Cleft lip and palate	16	44.4 %
Total	36	100 %

Table 7: Specific Gastro Intestinal anomalies

Gastro Intestinal anomalies	Frequency	Percentage
Congenital diaphragmatic hernia	4	40 %
Gastroschisis	4	40 %
Umbilical hernia	2	20 %
Total	10	100 %

Table 8: Specific lymphatic system anomalies

Lymphatic system anomalies	Frequency	Percentage

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Cystic hygroma	15	100 %
Total	15	100 %

Table 9: Specific Respiratory system anomalies

Respiratory system anomalies	Frequency	Percentage
Cystic malformation of lungs	2	66.3 %
Small pulmonary artery	1	33.7 %
Total	3	100 %

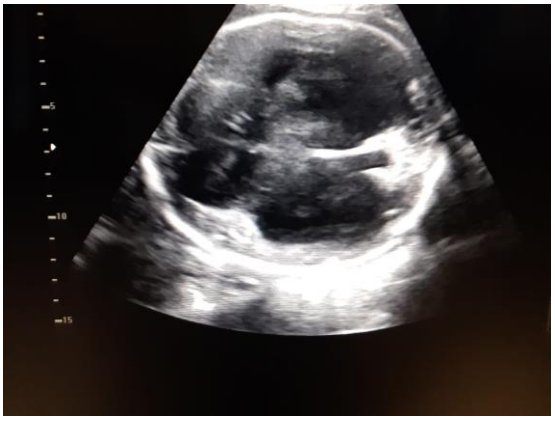

Image 1: Gross dilatation of lateral ventricles (Hydrocephalus)

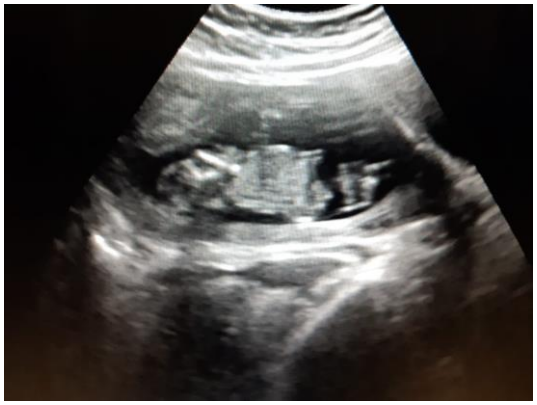

Image 2: Anencephaly

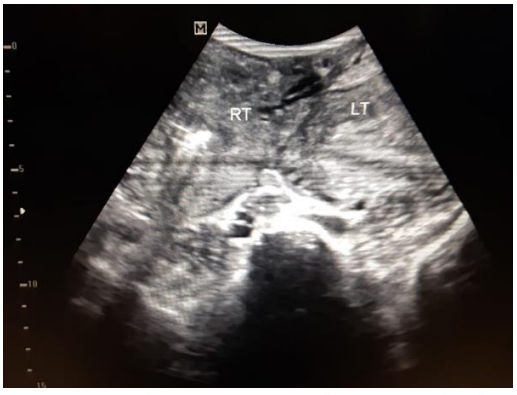

Image 3: Autosomal recessive poly cystic kidney disease

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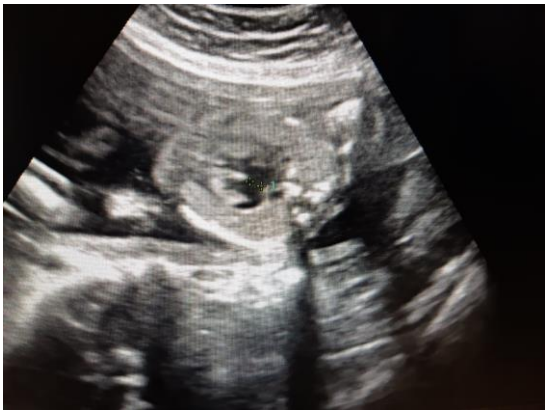

Image 4: Ventricular septal defect

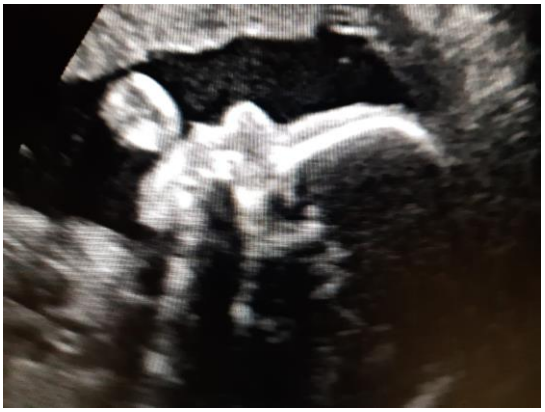

Image 5: Absent nasal bone

Consanguineous marriages are reported to play a major role in the occurrence of gross congenital anomalies. It was observed in 27.5% of antenatal mothers. History of previous anomalies found in 4.2% of mothers followed by other risk factors as presented in table 10

Table 10: Associated risk factors for congenital anomalies

Risk factor	YES	NO
Consanguinity	153 (27.5%)	405 (72.5%)
Previous anomalies	23 (4.2%)	535 (95.8%)
Fever	42 (7.5%)	516 (92.5%)
Rash	12 (2.1%)	546 (97.8%)
Drug intake	15 (2.6%)	543 (97.4%)
Thyroid disorders	66 (11.8%)	498 (89.2%)
DM	14 (2.5%)	544 (97.5%)
HTN	57 (10.2%)	501 (89.2%)
TB	1 (0.2 %)	557 (99.8%)

Discussion:

In our research, we observed an overall incidence of fetal structural anomalies at 1.45%, which, interestingly, appeared to be relatively low when compared to the general population[1]. This lower incidence rate could potentially be attributed to the unique setting of our study, which was conducted in a tertiary care hospital. It's worth noting that not all expectant mothers attending this hospital received ultrasound scans during their antenatal care, which might have

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contributed to the lower incidence we observed. In contrast, our findings differed from those reported in other studies where incidence rates were documented.

The present study revealed that central nervous system anomalies were the most common type of anomaly, occurring as both single system and multiple system malformations. This observation aligns with findings from other study by Sozan K. Ameen et al from Iraq[10]. The prevalence of central nervous system anomalies may be attributed to several factors, including lack of access to folic acid -fortified foods, insufficient conventional intake of folic acid, in adequate consumption of folic – acid rich foods like vegetables as well as suboptimal care[10]. In certain studies, the most frequently observed congenital anomalies are associated with the musculoskeletal system, with central nervous system anomalies ranking as the second most common[11].

In the central nervous system category, the predominant anomalies identified were anencephaly and hydrocephalus with other defects following in terms of frequency as showed in the result section. These findings are similar with a study conducted by Sozan K Ameen[10], and the results diverge from other studies that were conducted at the time of birth[11,12].

In the current study, the highest number of patients with hydronephrosis and bilateral PUJ obstruction were identified within the genitourinary system category. In contrast, in a study done by Vinod SL et al., the incidence was reported at 1.1%, and in the study carried out by Sarkar S et al., it was noted as 1% [12,13]. These findings demonstrate inconsistency between the current study and the earlier research.

Among the cardiovascular anomalies, ventricular septal defect is most common followed by other defects. In the gastrointestinal system defects the most common defect is congenital diaphragmatic hernia and gastroschisis. In other studies, the most common gastrointestinal anomalies were tracheoesophageal fistula and gastroschisis[11,12,13] and the findings are not consistent.

In the musculoskeletal system category, the present study revealed a higher prevalence of congenital talipes equinovarus. These findings are consistent with studies conducted by Sarkar S et al and Vinodh SL et al.

Conclusion:

Despite its retrospective nature and limited enrollment to women at RIMS Adilabad seeking antenatal ultrasound, this study aimed to investigate congenital malformation patterns and maternal risk factor associations. To enhance newborn health and reduce congenital malformations, we must provide second trimester ultrasound scans, counselling, identify high-risk pregnancies, manage maternal illness promptly, improve pregnancy care, prioritize maternal health and empower women.

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