

DIAGNOSTIC YIELD OF PRENATAL ULTRASOUND IN IDENTIFYING CONGENITAL FETAL ANOMALIES IN A TERTIARY CARE SETTING

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ABSTRACT

Background: Congenital anomalies have emerged as a major contributor to neonatal morbidity and mortality. Globally congenital fetal anomalies affect 2-3% of births and constitute fifth largest cause of neonatal deaths. Ultrasonography remains the primary tool for fetal imaging in the antenatal period. The objective is to estimate the prevalence and spectrum of congenital fetal anomalies detected prenatally by ultrasonography in pregnant women visiting tertiary care setting. **Materials and Methods:** This descriptive cross-sectional study was carried out in the Department of Radio-diagnosis, PES Institute of Medical Sciences and Research, Kuppam, during the period 2024–2025. The pregnant women in the second trimester were referred from the outpatient and inpatient departments of Obstetrics and Gynaecology. Those pregnant women who underwent second trimester routine ultrasound during this period were included in the study. **Result:** Routine second-trimester ultrasound examinations of 4,088 women revealed 125 cases of fetal anomalies. The most observed fetal anomalies were in the order of cardiovascular system followed by central nervous system, genitourinary, musculoskeletal, others, digestive and respiratory system. Cardiovascular system defects were the commonest (44%), of which ventricular septal defects was the most frequently encountered. The antenatal prevalence of congenital fetal anomalies was 3.06 % in our study. **Conclusion:** Antenatal ultrasound is the cost-effective modality to diagnose the congenital malformation. It is the safe, accurate and non-invasive method. Congenital malformation, one of the important causes of infant mortality and morbidity can be reduced by proper preconception care and routine second trimester anomaly scan. Even the treatment and rehabilitation of these anomalous children is a challenging task. So regular antenatal visits and prenatal diagnosis by early trimester screening test are recommended for prevention, timely intervention and even planned termination. We found cardiovascular and neurological defects as commonest malformation which can easily be prevented by pre-conceptional folic acid and vit-B12.

INTRODUCTION

Prenatal US has been shown to be an effective primary imaging modality for depiction of congenital fetal anomalies.^[1,2]

A US examination to delineate fetal anatomy is generally recommended between 18 and 22 weeks gestation as a part of normal obstetric care.^[3]

This period allows for optimal early evaluation of fetal anatomy, soft anatomic markers that suggest aneuploidy, diagnosis of multiple gestation and chorionicity, reassessment of gestational dating, and presence of any abnormal placentation. Fetal anatomy components of a standard examination at 18 to 20 weeks involve measurements taken in an orderly manner of the head, face, neck, heart, abdomen, spine, extremities, and genital regions.

Evaluation of the maternal pelvic anatomy includes cervical length.^[4]

Congenital anomalies are the structural or functional anomaly (e.g. metabolic disorder) that occurs during intrauterine life and can be identified prenatally, at birth or later in life. These defects of prenatal origin result from defective embryogenesis or intrinsic abnormalities in the development process.^[5]

Congenital anomalies can involve a single organ or multiple organs and have been associated with various etiological factors. It is estimated that in around half of the congenital anomalies, aetiology cannot be identified and in those where known aetiology is present, 20-25% are multifactorial.^[6]

The sensitivity of detection of fetal anomalies, before the 24th week of gestation was 93% for the central nervous system, 45.2% for the circulatory system, 85.2% for the digestive system, 85.7% for the urinary system, 84.6% for the musculoskeletal system and 95.2% for other anomalies. Therefore, it is suggested that ultrasonography between the 20th and 22nd weeks of pregnancy can detect the majority of congenital anomalies.^[7]

MATERIALS AND METHODS

This is a descriptive cross-sectional study carried out in the department of Radiology at PES Institute of medical sciences and research, Kuppam. Pregnant women in second trimester referred to radiology department from the outpatient and inpatient department of Obstetrics and gynaecology department of the same hospital between August 2024 to August 2025 were included in the study. The radiologists performed all transabdominal ultrasonography on a SAMSUNG HS 70 and GE machines using curvilinear probe after taking verbal consent from the patient. The request form for antenatal ultrasound contains patient's name, age, gravida, parity and last menstrual period.

RESULTS

During the study period of August 2024 to August 2025, a total of 4088 second trimester prenatal ultrasound was done. The gestational age was between 18 to 24 weeks. Total 125 fetal congenital anomalies were detected among the pregnant women scanned. There were 55 anomalies related to CVS followed by 22 anomalies of CNS, 16 anomalies of genitourinary system, 10 anomalies of extremities, 9 anomalies were categorized as miscellaneous, 8 anomalies of musculoskeletal system and 3 anomalies of gastrointestinal tract and 3 anomalies of respiratory system.

Of the total 55 anomalies of cardiovascular system, majority were Ventricular septal defects. Of 22 anomalies of central nervous system, posterior fossa anomalies were more common. Of 16 genitourinary system anomalies, urinary tract dilatation (UTD) was most common. Of 12 anomalies involving limbs, majority were CTEV.

The antenatal prevalence of congenital fetal anomalies was 3.06 % in our study.

Of the women having congenital fetal anomalies, majority were between the age of 18-35 years old.

System wise anomalies: [Table 1 and Figure 1] represent the distribution of system-wise anomalies observed in the study sample. The Cardiovascular System (CVS) accounts for the highest number of anomalies (55 cases, 44.0%), followed by the Central Nervous System (CNS) with 22 cases (17.6%), Musculoskeletal system (MSK) 18 cases (14.4 %) and the Genitourinary Tract (GUT) with 16 cases (12.8%). Other systems anomalies such as Craniofacial anomalies show lower incidence rates 9 cases (7.2 %), while anomalies related to the Gastrointestinal Tract (GIT) and Respiratory System (RS) are least prevalent (each 2.4%). This tabulated overview facilitates comparison of anomaly frequency and proportion across major body systems.

Table 1: Showing system wise Distribution of congenital fetal anomalies.

System involved	Number	Percentage (%)
Cardio Vascular System	55	44.0%
Central Nervous System	22	17.6%
Musculoskeletal System	18	14.4%
Genito Urinary System	16	12.8%
Craniofacial / Head and Neck	9	7.2%
Gastro Intestinal System	3	2.4%
Respiratory System	3	2.4%

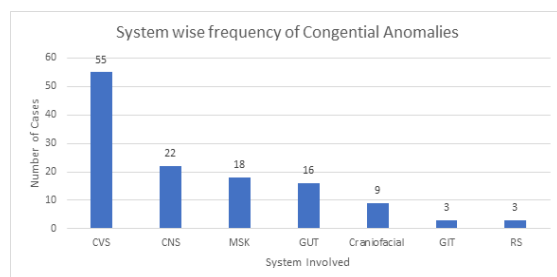


Figure 1. Represents the frequency of system-wise anomalies observed in the study sample.

Distribution of CVS anomalies:

There were 55 cardiovascular system anomalies as shown in [Figure 1].

[Figure 2] illustrates the proportional distribution of various congenital cardiovascular anomalies seen in this study.

- The largest segment by far represents Ventricular Septal Defects (VSD) [Figure 5,6], accounting for 83% (45 cases) of all cases. This

indicates that VSD is by far the most common congenital defect in the dataset.

- The remaining 17% of cases are distributed among several less frequent heart anomalies:
- Persistent left superior vena cava makes up 4% (2 cases) of the total [Figure 10].
- Hypoplastic left heart syndrome accounts for 3% (2 cases) [Figure 12-16].
- Tetralogy of Fallot constitutes 2% (1 case) [Figure 17].
- Total Anomalous Pulmonary Venous Connection (TAPVC) contributes 2% (1 case) [Figure 8,9].
- ARSA (Aberrant Right Subclavian Artery) also represents 2% (1 case) [Figure 11].

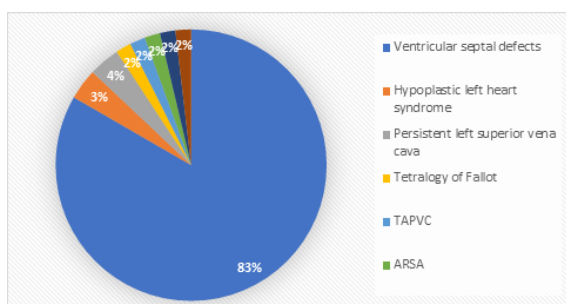


Figure 2: Illustrates the proportional distribution of various congenital heart defects observed in the sample.

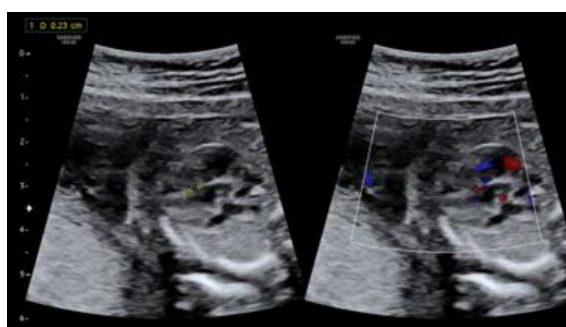


Figure 3: Small muscular ventricular septal defect measuring 2.3 mm.



Figure 4: Inlet ventricular septal defect measuring 3.3 mm.



Figure 5:

Dilated extrahepatic intraabdominal umbilical vein with direct connection between right atrium and umbilical vein. This is a case of Umbilicosystemic shunt with Ductus agenesis.



Figure 6: Axial plane showing supra cardiac TAPVC



Figure 7: On 3 vessel view: Additional vessel (Left brachiocephalic vein) is seen adjacent to the pulmonary artery which is joining with the SVC in a case of Supra cardiac TAPVC.



Figure 8: Axial section showing Persistent Left Sided Superior Vena Cava.



Figure 9: Axial section showing Aberrant right subclavian artery.



Figure 10: Axial section showing hypoplastic left ventricle in a case of left heart hypoplastic syndrome.



Figure 11: Axial section showing Hypoplastic left ventricle, hypertrophied right ventricle and ventricular septal defect seen in a case of left heart hypoplastic syndrome.



Figure 12: Axial plane showing left axis deviation with angle > 45 degree in a case of left heart hypoplastic syndrome.



Figure 13: Axial section showing pericardial effusion in a case of left heart hypoplastic syndrome.



Figure 14: Axial image showing dilated pulmonary artery in a case of left heart hypoplastic syndrome.

Figure 15 Pink type Tetralogy of Fallot.

Distribution of CNS anomalies:

There were 22 central nervous system anomalies as shown in [Figure 1].

[Figure 3] illustrates the proportional distribution of various congenital central nervous anomalies seen in this study.

- Posterior fossa anomalies account for 29%, forming one of the largest segments [Figure 18-22].
- Mega cisterna magna also constitutes 29%, equal in proportion to posterior fossa anomalies [Figure 26].
- Corpus callosal agenesis represents 14% of cases [Figure 23].
- Acrania–anencephaly sequence accounts for 9% [Figure 25].
- Ventriculomegaly also makes up 9%.

- Craniosynostosis contributes 5%.
- Holoprosencephaly is the least common, at 5% [Figure 24].

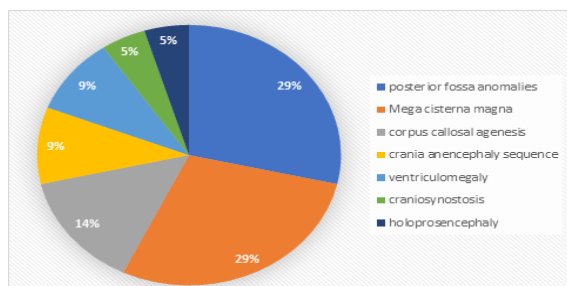


Figure 15: Illustrates the proportional distribution of various congenital cranial defects observed in the sample.



Figure 16: Axial section showing dilated lumbar vertebra with bony spur in midline – Likely diastematomyelia. This is seen in case of Arnold Chiari Type II Malformation.



Figure 17: Axial section of brain showing abnormal skull shape and reveals bilaterally symmetric indentation over the frontal bone giving it a typical appearance called lemon skull sign in a case of Arnold Chiari Type II Malformation.



Figure 18: Axial section of the brain showing cerebellum smaller than normal for gestational age and it is abnormal in shape (curved/banana shaped), so called banana cerebellum in a case of Arnold Chiari Type II Malformation.



Figure 19: Axial section showing occipital encephalocele. This is a case of occipital encephalocele with herniation of posterior fossa content, occipital horns of lateral ventricles, thalamus and occipital cortex.



Figure 20: Mid sagittal plane showing Tegmentovermian angle of 36° and a small vermis and abnormal fastigium in a case of vermian hypoplasia.



Figure 21: Mid sagittal plane shows partial agenesis of corpus callosum.

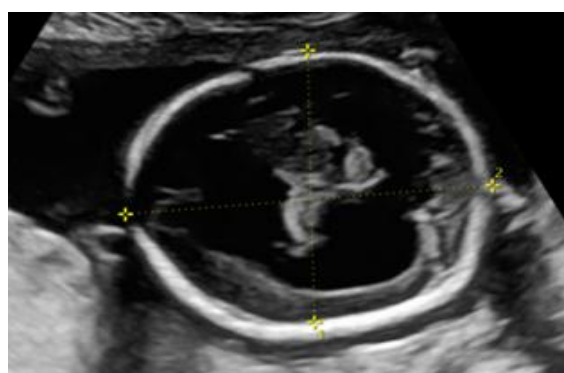


Figure 22: Axial section showing normal skull shape, no midline falx, fused thalami. This is a case of alobar holoprosencephaly.



Figure 23: Plane showing prominent eyes with normal nose and lips giving Frog Eye appearance. No cranial vault, skull bones and loss of brain tissues. This is seen in a case of Acrania- Anencephaly sequence.



Figure 24: Axial posterior fossa view showing Mega Cisterna Magna.

Distribution of Musculoskeletal system:

There are 18 musculoskeletal system anomalies observed in the sample as shown in [Figure 1].

[Figure 4] illustrates the proportional distribution of various musculoskeletal anomalies seen in this study.

- CTEV (Congenital Talipes Equinovarus) constituted the largest proportion with 10 cases (55%) [Figure 42-45]
- Lethal skeletal dysplasia accounted for 5 cases (28%), representing the second most frequent category [Figure 32-41].
- Non-lethal skeletal dysplasia was the least common, with 3 cases (17%) [Figure 35,36].

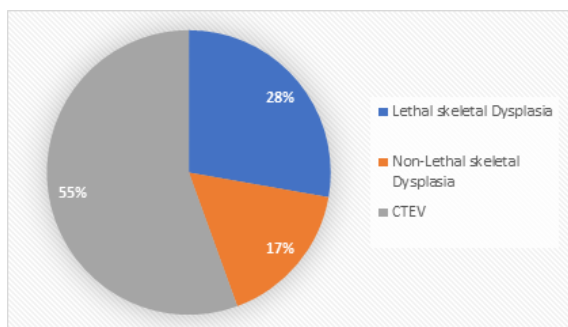


Figure 25: Illustrates the proportional distribution of various musculoskeletal anomalies observed in the sample.



Figure 26: Showing severely shortened long bones. This is a case of Skeletal Dysplasia - Osteogenesis Imperfecta.



Figure 27: Showing severely shortened long bones. This is a case of Skeletal Dysplasia - Osteogenesis Imperfecta.

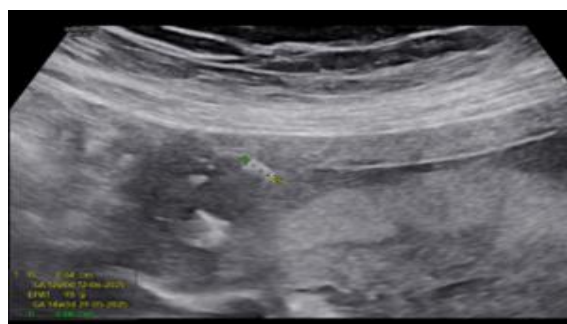


Figure 28: Showing severely shortened long bones. This is a case of Skeletal Dysplasia - Osteogenesis Imperfecta.

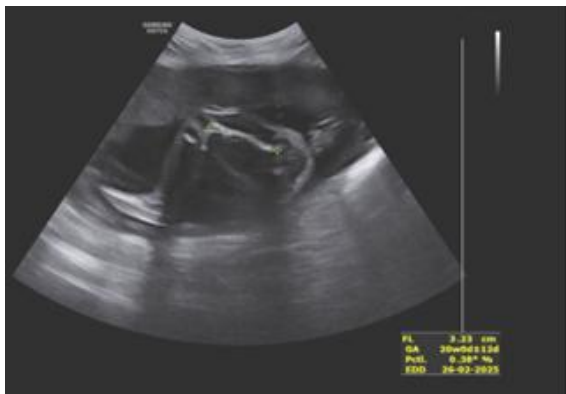


Figure 29: Showing short femur with bowing. This is a case of Non-lethal skeletal dysplasia – Achondroplasia.



Figure 30: Showing short femur with bowing. This is a case of Non-lethal skeletal dysplasia – Achondroplasia.



Figure 31: Inferior facial angle measures 39 degrees. This is a case of micrognathia with Rhizomelia.



Figure 32: Large cystic hygroma in the posterior aspect of neck, upper thorax, covering the head.

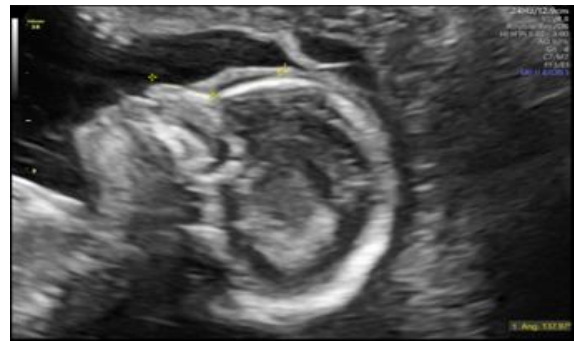


Figure 33: Mid sagittal section showing Flat nasal bridge with midfacial hypoplasia. Facial angle measured 138 degrees. This is a case of Chondrodysplasia punctata.

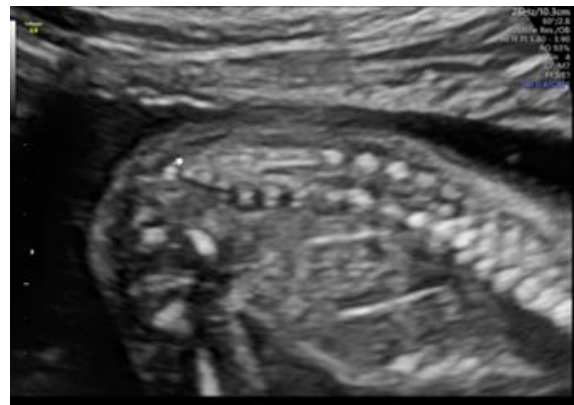


Figure 34: Showing premature ossification of coccyx. This is a case of Chondrodysplasia punctata.

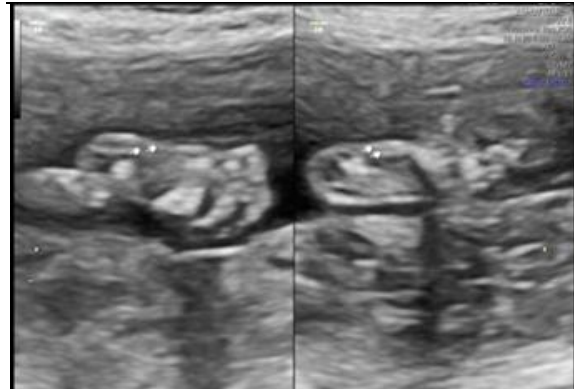


Figure 35: Showing premature ossification of coccyx. This is a case of Chondrodysplasia punctata.



Figure 36: Showing Polydactyly/syndactyly.



Figure 37: Showing Hypertrophied great toe.



Figure 38: Showing bilateral congenital talipes equinovarus (CTEV)/ club foot.

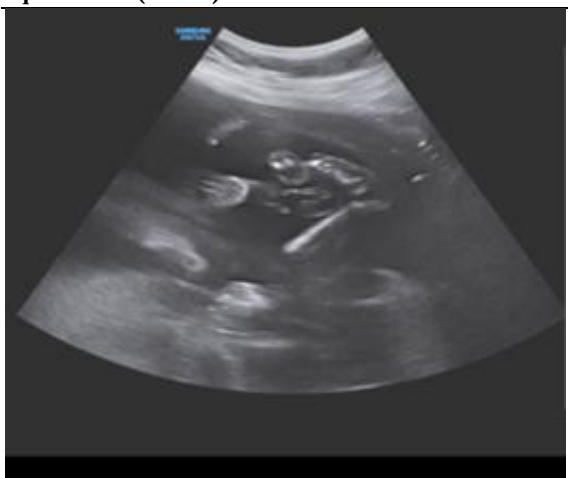


Figure 39: Showing Rocker bottom Foot.

Distribution of Genito urinary system:

There are 16 genitourinary system anomalies observed in the sample as shown in [Figure 1].

- Urinary tract dilatation (UTD) accounts for 57% forming the largest segment [Figure 29]
- Pelviectasis constitutes 19% of cases [Figure 31]
- Lower urinary tract obstruction represents 6% of cases [Figure 30]
- Duplex kidney contributes 6% of cases [Figure 28].
- Renal agenesis contributes 6% of cases [Figure 27].
- Multicystic dysplastic kidney is the least common, accounting for 6% of cases.

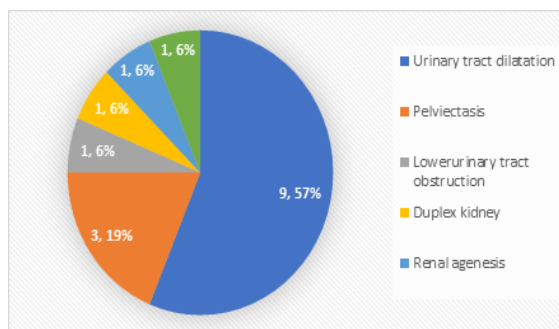


Figure 40: Illustrates the Distribution of Genitourinary system anomalies observed in the sample.

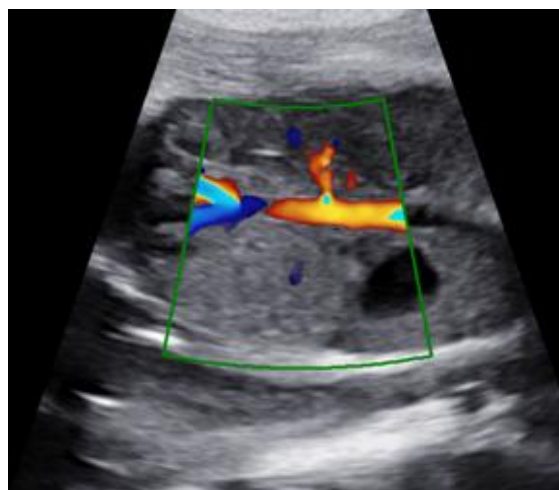


Figure 41: Coronal section showing absent renal artery flow in case of left renal agenesis.



Figure 42: Gross dilatation and bifid pelvicalyceal system with parenchymal thinning. This is a Duplex kidney.



Figure 43: Large ureterocoele. This is a case of Urinary tract dilatation (UTD A2-3).

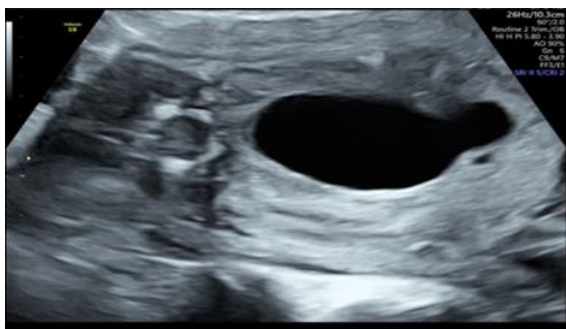


Figure 44: Showing keyhole sign in a case of lower urinary tract obstruction.



Figure 44: Showing left renal pelviectasis. This is a case of Urinary tract dilatation (UTD A2-3).

Other anomalies:



Figure 45: Showing Cleft lip.

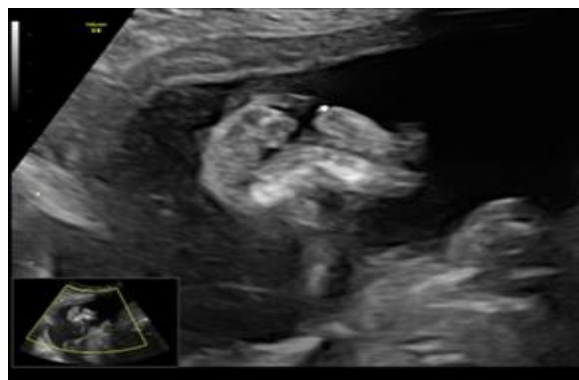


Figure 46: Showing cleft lip and palate.



Figure 47: Showing Microphthalmia.



Figure 48: Showing hyperechoic lesion with multiple cystic spaces within seen in the right hemithorax. This is a case of Congenital Pulmonary Airway Malformation (Type II).



Figure 49: Showing hyperechoic lesion with multiple cystic spaces within seen in the right hemithorax. This is a case of Congenital Pulmonary Airway Malformation (Type II).

DISCUSSION

Congenital fetal anomalies, also commonly referred to as birth defects, congenital disorders or congenital malformations. These anomalies usually develop before birth and are present at the time of birth and they can affect infant's health which can be life threatening.

Congenital malformation varies from region to region. Prevalence of congenital malformation in our study is 3.06 %. Worldwide prevalence is 3-7%.

Its prevalence depends on geographic distribution, socioeconomic condition, ethnicity, region of study. Studies in different part of world shows difference prevalence results. One study showed prevalence of congenital anomalies in Oman found 2.46% while it is 4.3% in Taiwan. In India, its prevalence is 1.7%.

Prevalence of congenital malformation according to the systems is different in different region. For example, one study is Iran showed that musculoskeletal anomalies (27.5%) are the most occurring anomalies followed by skin anomalies (19.7%) and genitourinary system anomalies (15.8%). However, the respiratory system anomalies have the lowest prevalence (1.82%). Another study that was done in Saudi Arabia showed that the CNS anomalies are the most occurring anomaly followed by musculoskeletal and then renal system anomalies. One study that was done in Brazil showed that the congenital anomalies are more prevalent in male as compared to female (59% male newborn and 41% in female newborn).

Fetal wellbeing monitoring is necessary to assess the status of fetus. So routine use of antenatal ultrasound has been standardized. The rationale for routine second trimester screening is to make an approach securing the health of embryo or fetus. It also detects congenital anomalies, placental abnormalities, fetal growth disorder and multiple pregnancies. Antenatal ultrasound is also done to establish the gestational age and viability of fetus. Ultrasound machines have been developed more efficiently with their software advancements. All these changes made ultrasound machine very reliable and safer modality that can manipulate the images for accurate interpretations. However, training of personnel is necessary to gather and interpret images. All these factors may result in diagnosing virtually the 100% congenital anomalies before birth. Prenatal diagnosis affects the society and patient in many ways. It improves the affected

newborns outcome. First it prepares the family to accept the newborn with malformation. It also helps the family to prepare for long term care or need for urgent surgical correction. Second, it may allow for in utero intervention. Last, antenatal diagnosis of malformation may also prepare the hospital, particularly the neonatology team in advance to care the newborn. Risk factors for congenital malformation have also been scientifically known like obesity, infections and toxic agents. Some protective measures are also scientifically proven like glycemic control in diabetic women. Folic acid supplementations before conception and during pregnancy plays very important role in preventing congenital defects (Neural Tube Defects).

Embryologic and Anatomic Correlation with Prenatal Sonographic Findings

The pattern of congenital anomalies observed in the present study can be explained by the timing and complexity of embryologic development of the affected organ systems. Most major structural anomalies arise during the organogenesis period (3rd to 8th week of gestation), when critical processes such as cellular differentiation, migration, and septation occur. Disruption during this period results in permanent structural defects that can be detected on prenatal ultrasonography.

The predominance of cardiovascular system anomalies, which accounted for 44% of detected malformations, reflects the intricate embryogenesis of the fetal heart. Cardiac development begins in the third week of gestation and involves sequential processes including cardiac looping, septation of the atria and ventricles, and formation of the outflow tracts. Abnormalities in endocardial cushion formation and septation result in defects such as ventricular septal defects, which were the most frequently encountered cardiac anomaly in this study. These defects are typically visualized on prenatal ultrasound as discontinuities in the interventricular septum on the four-chamber view.

Conotruncal anomalies arise due to defective neural crest cell migration and abnormal partitioning of the truncus arteriosus, leading to conditions such as tetralogy of Fallot, truncus arteriosus, and transposition of the great arteries. Prenatal sonographic evaluation using left and right ventricular outflow tract views and the three-vessel view plays a crucial role in identifying abnormal great vessel relationships, thereby improving detection of these anomalies.

Table 2: Illustrates the embryologic defect, resulting congenital anomalies and expectant ultrasound finding in cardiovascular system.

Embryologic Defect	Affected Developmental Process	Resulting Congenital Heart Defect	Key Prenatal Ultrasound Finding
Incomplete atrioventricular septation	Failure of endocardial cushion fusion	Atrial septal defect (ASD), Ventricular septal defect (VSD), Atrioventricular septal defect (AVSD)	Defect in interatrial or interventricular septum on four-chamber view
Abnormal neural crest cell migration	Faulty conotruncal septation	Tetralogy of Fallot, truncus arteriosus, transposition of great arteries	Overriding aorta, single great vessel, or parallel great vessels on outflow tract views

Abnormal cardiac looping	Malposition of ventricles and great vessels	Double outlet right ventricle, transposition of great arteries	Abnormal alignment of ventricles and great vessels
Defective septum primum or secundum development	Atrial septation abnormality	Atrial septal defect	Discontinuity in atrial septum
Defective interventricular septum formation	Ventricular septation abnormality	Ventricular septal defect	Gap in interventricular septum
Abnormal development of aortic arch derivatives	Persistence or regression of arch segments	Coarctation of aorta, interrupted aortic arch	Narrowed or interrupted aortic arch on sagittal or three-vessel trachea view
Endocardial cushion defect	Failure of AV valve and septal formation	AV canal defect	Common atrioventricular valve, atrial and ventricular septal defects

Central nervous system anomalies, the second most common group in the present study, are largely attributable to disturbances in neural tube closure and brain vesicle development, which occur between the 3rd and 5th weeks of gestation. Failure of neural tube closure results in open neural tube defects, while

abnormalities in ventricular development lead to ventriculomegaly and hydrocephalus. These conditions are readily identified on prenatal ultrasound through characteristic findings such as altered cranial morphology, abnormal ventricular size, and associated spinal defects.

Table 3: Illustrates the embryologic defect, resulting congenital anomalies and expectant ultrasound finding in central nervous system.

Embryologic Defect	Developmental Stage Affected	Resulting CNS Anomaly	Key Prenatal Ultrasound Findings
Failure of neural tube closure	3rd–4th week (neurulation)	Anencephaly	Absent calvarium, exposed brain tissue, polyhydramnios
Incomplete closure of caudal neuropore	3rd–4th week	Spina bifida (open NTD)	Lemon sign, banana sign, ventriculomegaly, spinal defect
Abnormal dorsal induction	3rd–5th week	Encephalocele	Herniation of brain tissue through skull defect
Abnormal brain vesicle development	5th–6th week	Holoprosencephaly	Single ventricle, absent midline structures, fused thalami
Abnormal ventriculogenesis or CSF circulation	6th–8th week	Hydrocephalus	Dilated lateral ventricles (>10 mm), dangling choroid plexus
Partial or complete agenesis of corpus callosum	8th–12th week	Agenesis of corpus callosum	Colpocephaly, absent cavum septipellucidi, teardrop ventricles
Abnormal posterior fossa development	6th–8th week	Dandy–Walker malformation	Enlarged posterior fossa, vermian agenesis, cystic dilatation of 4th ventricle
Abnormal rhombencephalon development	6th–8th week	Arnold–Chiari II malformation	Small posterior fossa, banana-shaped cerebellum, associated spina bifida

Genitourinary anomalies result from disruptions in the development of the ureteric bud and metanephric blastema, which normally interact to form the kidneys and collecting system. Sonographic findings such as renal agenesis, multicystic dysplastic kidney, and hydronephrosis reflect underlying defects in

renal induction, differentiation, or urinary tract patency. Similarly, musculoskeletal anomalies arise from abnormal mesenchymal differentiation and ossification, and are detected as limb shortening, skeletal deformities, or abnormal thoracic configuration on ultrasound.

Table 4: Illustrates the embryologic defect, resulting congenital anomalies and expectant ultrasound finding in genitourinary system.

Embryologic Defect	Developmental Process Affected	Resulting Genitourinary Anomaly	Key Prenatal Ultrasound Findings
Failure of ureteric bud formation	Absent induction of metanephric blastema	Renal agenesis	Absent kidney, non-visualized renal artery, oligohydramnios (bilateral)
Abnormal interaction between ureteric bud and metanephric blastema	Defective nephron differentiation	Multicystic dysplastic kidney	Enlarged kidney with multiple non-communicating cysts, absent renal pelvis
Abnormal ascent of kidneys	Failure of cranial migration	Ectopic kidney	Kidney located in pelvis or lower abdomen
Abnormal fusion of metanephric blastema	Fusion anomaly	Horseshoe kidney	Renal fusion across midline, low-lying kidneys, abnormal renal axis
Ureteropelvic junction obstruction	Impaired urine drainage	Hydronephrosis	Dilated renal pelvis ± calyces, normal ureter
Posterior urethral valve formation	Abnormal urethral development	Posterior urethral valves	Distended bladder, bilateral hydroureteronephrosis, “keyhole sign”
Failure of mesonephric duct incorporation	Abnormal ureteric insertion	Vesicoureteral reflux (indirect)	Hydronephrosis with dilated ureter (suggestive)

Fate of congenital anomalies is variable. Some defects are completely cured by surgery while others may require subsequent surgeries of long-term disabilities which is quite stressful for family also a burden for hospital resources. Some can be life threatening and some can achieve the improvement in functions and quality of life. World Health Organization and other international bodies address congenital malformation as prioritizing public health issue. Especially in developing countries, early recognition of congenital malformation is necessary with proper preventive measures and management. These measures can improve the overall status of the newborn. Congenital malformations may result in fetal death or still birth. If congenital malformation is diagnosed early, it will definitely help the further management of fetus or new born.

CONCLUSION

Antenatal ultrasound is the cost-effective modality to diagnose the congenital malformation. It is the safe, accurate and non-invasive method. Congenital malformation, one of the important causes of infant mortality and morbidity can be reduced by proper preconception care and routine second trimester anomaly scan. Even the treatment and rehabilitation of these anomalous children is a challenging task. So Regular antenatal visits and prenatal diagnosis by early trimester screening test are recommended for

prevention, timely intervention and even planned termination. We found cardiovascular and neurological defects as commonest malformation which can easily be prevented by pre-conceptual folic acid and vit-B12.

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