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Chapter

Antenatal Diagnosis of Congenital Anomalies on Ultrasound Screening

Callen Kwamboka Onyambu and Norah Mukiri Tharamba

Abstract

Congenital fetal anomalies contribute to the global burden of disease in children. Various screening programs have been used for antenatal screening of these anomalies. Screening targets low risk population and is usually done in the second trimester though some are done at the mother's first antenatal visit especially in resource constrained setting. Mother's who have had a previous pregnancy with congenital anomaly are given targeted elaborate screening. Early diagnosis of this anomalies can lead to early intervention and better outcomes. Diagnosis of the malformations also leads to clinical decision making on mode of delivery thereby avoiding birth related trauma to the mother and the baby. In case of lethal congenital anomalies early diagnosis aids in clinical decision making on the management of the pregnancy.

Keywords: Ultrasound screening, pregnancy, fetal anomalies, fetal well being

1. Introduction

According to the World Health Organization (WHO), the term congenital anomaly includes any morphological, functional, biochemical or molecular defects that may develop in the embryo and fetus that is present at birth, whether detected at that time or not [1].

Major congenital anomalies impair function or greatly interfere with cosmetic value.

They may be life threatening and therefore need immediate management. Major anomalies could have a negative impact on a child's well-being and development, if early corrective surgery is not done. Minor anomalies, on the other hand, may cause little or no functional effects. They do not cause distress in the newborn and hence there is no urgency for their correction in the neonatal period [2].

Congenital anomalies occur in 2–3% of all births and are an important cause of perinatal morbidity and mortality accounting for 20–30% of perinatal deaths.

In a study conducted by Nayab Alia in Madina Teaching Hospital, Faisalabad on gray scale ultrasound, antenatal prevalence of congenital anomalies was 29.75 per 1000 and 2.97% [3]. A similar study in Saudi Arabia showed the antenatal prevalence of congenital anomalies to be 27.96 per 1000. The median maternal age at diagnosis was 27.5 years and the median gestational age at diagnosis was 31 weeks [4].

However, the actual numbers of these anomalies vary among different countries with prevalence of anomalies reported to be 2% in England and 1.49% in South Africa [5]. The reason for the regional difference of congenital anomalies might be attributed to variable factors, such as: maternal risk factors, environmental exposures, ecological, economical and ethnic factors [6, 7].

1.1 Pattern of congenital anomalies

The patterns of congenital anomalies may be different between regions and the actual numbers may vary significantly between countries [8]. In different countries, people have varied cultural and religious practices including exposure during antenatal period to various environmental factors. This may contribute to varied patterns of congenital anomalies. In some instances a common exposure to teratogenic factors or a hereditary condition with variable penetrance, may lead to high numbers of some anomalies, where severely affected individuals were not observed because of fetal/infant mortality [9]. Certain population groups are also regarded as "high risk groups" for congenital anomalies such as those living in heavily polluted industrial zones [10].

According to one study, frequency of congenital anomalies was more in males than females, with CNS anomalies being the most common. The anomalies were more common in gestation age of 29–32 weeks [11]. In a retrospective study of 200 cases of congenital anomalies carried out in Jos, Nigeria, the highest incidence was reported in the gastrointestinal system 61 cases. No association was found between the occurrences of the various congenital anomalies [12].

In yet another study conducted among South African live born neonates at Kalafong Hospital, Pretoria, in which the incidence of congenital anomalies was 11.87 per 1,000 live births, the most commonly affected system was the central nervous system (2.30 per 1,000 live births) [13].

1.2 Risk factors for congenital anomalies

According to WHO, approximately 50% of all congenital anomalies cannot be associated with a specific cause. However, there are some known risk factors which include socioeconomic factors with an estimated 94% of severe birth defects occurring in middle and low income countries. This is because mothers are more susceptible to macronutrient and micronutrient malnutrition and may also have increased exposure to agents that cause or increase the incidence of abnormal prenatal development, especially infection and alcohol. Other known factors are genetic and environmental factors [14, 15]. This is reaffirmed by a study that was conducted in Tanzania, that showed significant association between congenital anomalies and lack of periconceptional use of folic acid, maternal age above 35 years, exposure to pollutants and high birth order above [16].

Women with uterine anomalies have also been found to be at risk for particular CAs. In one study, the risk for some specific defects such as nasal hypoplasia, omphalocele, limb deficiencies, teratomas, and anencephaly was four times higher among offspring of mothers with a bicornuate uterus [17].

1.3 Ultrasound imaging IN congenital anomalies

Many congenital anomalies are identified prenatally on usual work up which includes detailed ultrasound and amniocentesis.

The diagnostic ability of ultrasound is well established by several studies with detection rate dependent on a number of factors which include the type of

abnormality, sophistication of equipment and experience of operator [18]. In a study conducted by Shama Munim at Aga Khan University Hospital (AKUH), Karachi on the accuracy of ultrasound in the diagnosis of congenital abnormalities, antenatal ultrasound successfully diagnosed fetal abnormalities in 48.8% of cases and more than 90% Central Nervous system defects and renal abnormalities [18].

However, a 1997 Report of the Royal College of Obstetricians and Gynecologists Working Party on Ultrasound Screening for Fetal Abnormalities identified that one of the problems with screening scans was the variable way in which they were conducted because there are no clear guidelines about what should, or should not, be examined. In a multicenter study, comparison was made of the precision of sonographic examinations done before 24 weeks gestational age at tertiary ultrasound laboratories contrasted with nontertiary ones. All the institutions were amply furnished with up-to-date equipment and supported with didactic and practical in-service training as required. The study found global sensitivity for sonographically demonstrable fetal abnormalities was 35% in tertiary centers, significantly higher compared to 13% in community hospitals. This further emphasizes that operator experience, competence, and training are vital determinants [19].

Ultrasound imaging is now routinely used in most countries for the purpose of screening pregnancies for fetal malformations but modalities, reliability and value of such screening is controversial [14, 19].

Regarding the time in gestation at which sonographic screening should be done, it is worthy noting that majority of the structural abnormalities are increasingly detected with advancing gestation. In early pregnancy, it is possible to diagnose with confidence certain categories of fetal anomalies, such as anencephaly, which can be reliably demonstrated at 10–14 weeks gestational age [20]. In certain instances, omphalocele and extremity malformations are also detectable using sonography in the first trimester, whereas other structural defects, like urinary system anomalies, are demonstrable later in pregnancy. However, a routine anomaly scan should be performed between 18 and 22 weeks of gestation. This period compromises between dating the pregnancy and the timely detection of major congenital anomalies [21].

Ultrasound examination at 10–14 weeks includes measurement of nuchal translucency, which is the maximum thickness of the subcutaneous translucency between the skin and the soft tissue overlying the cervical spine of the fetus. An increased nuchal translucency is associated with aneuploidy and cardiac malformations [22].

Ultrasound at around 20–21 weeks is done to screen fetuses for morphological anomalies. The utility of second trimester sonographic scan for detection of chromosomal anomalies was first recommended in 1985 [23]. Chromosomal aberrations were increasingly found to be associated with certain ultrasound findings, including biometric parameters (e.g., shortened femur and humerus, pyelectasis, thickened nuchal fold, dilated ventricles, fetal growth retardation) and morphologic features.

Ultrasound is the main diagnostic tool in the prenatal detection of congenital anomalies. It allows examination of the external and internal anatomy of the fetus. Even though a number of women are at increased risk of fetal malformations, either as a result of family history or owing to exposure to teratogens like infection and some drugs, the great majority of fetal abnormaitlies arise in the low risk category. As a result, sonographic evaluation ought to be offered routinely to all pregnant mothers. This is typically performed at 18–23 weeks of gestation, and should be done to a high level of precision. The scan should comprise systematic evaluation of the fetus for the detection of any defects.

2. Systemic review of congenital anomalies on ultrasound

2.1 Central nervous system (CNS)

The frequency of central nervous system anomalies varies according to geographic area and race. It is approximately 1–2:1000 newborns. Survivors are often severely disabled, necessitating long-term care.

The fetal brain undergoes major developmental changes during pregnancy. At 7 weeks of gestation, a sonolucent region is demonstrable in the cranial pole representing the fluid-filled rhombencephalic vesicle. At 9 weeks, demonstration of the convoluted pattern of the three primary cerebral vesicles is possible. The most prominent structures from 11 weeks are the echogenic choroid plexuses which fill the lateral ventricles. In the early second trimester, the lateral ventricles and choroid plexuses decrease in size relative to the brain mass.

Effective ultrasound screening for CNS anomalies can be carried out by examination of two important axial planes through the fetal brain; the transventricular and transcerebellar planes. Therefore, familiarity with the normal appearance of the fetal brain in these planes and at different gestational ages is vital for prompt identification of congenital anomalies.

The transventricular plane is at the level of the ventricular atria, with the echogenic choroid plexus being the dominant landmark. Measurements of atrial diameter made perpendicular to the walls should not normally exceed 10 mm.

This plane is obtained by axial sonogram at the level of the cavum septum pellucidum and shows the lateral margins of the frontal horns, the medial and lateral limits of the posterior horns of the lateral ventricles, and the choroid plexuses. It is used for fetal biometrics and quantification of the ventricular width (**Figure 1**).

The second crucial axial plane is the transcerebellar plane, which allows the examination of the midbrain and posterior fossa. The anatomic landmarks are the inferior portion of the third ventricle and the cerebellar hemispheres, which are outlined by fluid in the cisterna magna. The normal cisterna magna measures 2 to 11 mm in width (**Figure 2**).

A small cisterna magna (<2 mm) suggests a Chiari II malformation. However, it may also occur in massive ventriculomegaly. A large cisterna magna (>11 mm) may be a normal variant (megacisterna magna) or indicate a variety of anomalies. These

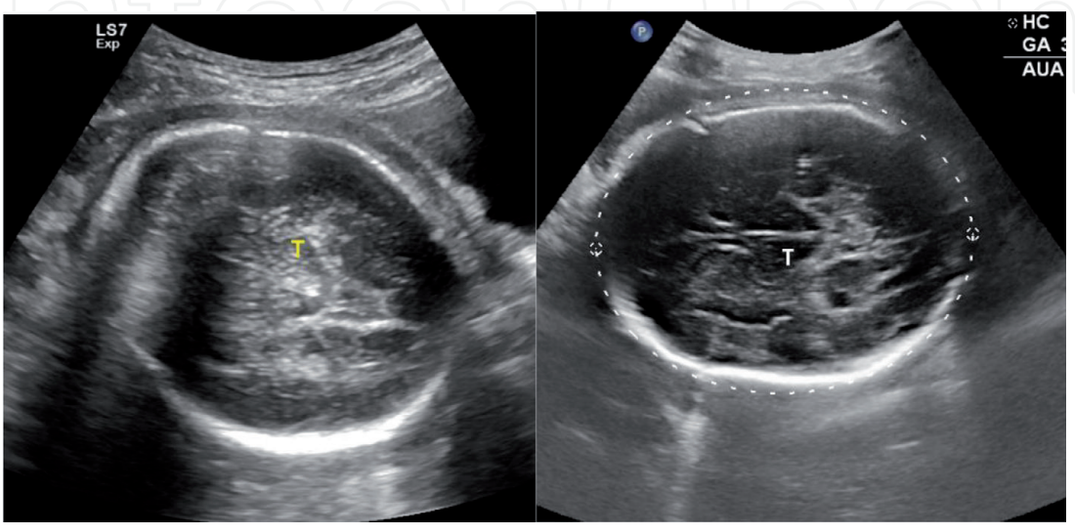

Figure 1.
Obstetric ultrasound showing biometric measurements. Fetal head: *Sonograms of the fetal head at the level of the thalami (T) showing measurement of the head circumference.*

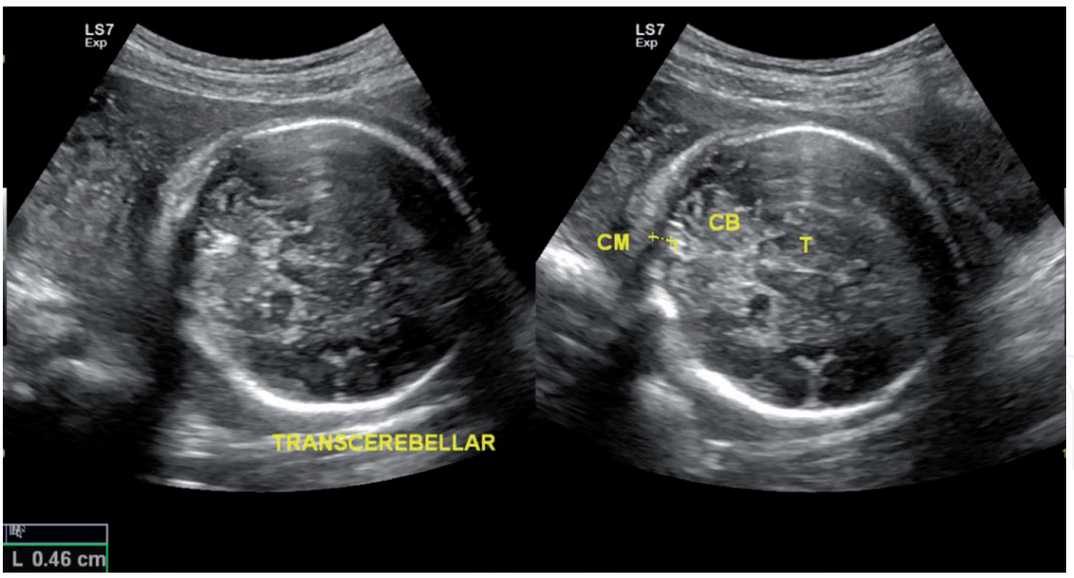

Figure 2.
Obstetric ultrasound; trans-cerebellar plane: This shows the cerebellar hemispheres (CB), cisterna magna (CM, between the two calibers) and thalami (T).

include Dandy-Walker malformation, arachnoid cyst, and cerebellar hypoplasia. When these two planes are anatomically normal, the risk of CNS anomaly is minute (0.005%).

Additional scanning planes along different orientations may be required occasionally, when one needs better definition of intracranial anatomy detail.

A sagittal and/or coronal view of the entire fetal spine should be obtained. In the sagittal plane, the normal spine has a 'double railway' appearance and one can appreciate intact soft tissue above it. In the coronal plane, the three ossification centers of the vertebra are visualized and should tether down into the sacrum. These views are used for assessment of vertebral integrity which rules out spina bifida. The presence and regularity of the whole spine from these views rules out sacral agenesis and scoliosis.

2.2 Cardiovascular system

The heart is the first functional organ in the embryo. It is prominently located and has visible contractions, which has made it the object of study for centuries. However, as recently as the late 1970s and early 1980s, the field of heart development was still in its infancy. In 1984, Dr. Constance Weinstein and colleagues at the National Institutes of Health (NIH) organized a meeting to bring together both cardiologists and basic scientists to summarize what was known about the development of the heart.

Congenital Heart Disease (CHD) is the most common birth defect comprising over 30 types of defects, ranging from mild to severe. It is found in 5–10 per 1,000 live births and in about 30 per 1,000 still births. Some forms of CHD resolve spontaneously, others are fully correctible, while others are life-threatening. Nearly 3 children out of every thousand live births have severe CHD requiring early surgery, while another 13/1000 requires surgery or catheter-based intervention later in childhood.

Precise US diagnosis of fetal heart abnormalities often requires specialized equipment and a high level of expertise.

The examination of the fetal heart begins with the assessment of the disposition of abdominal and thoracic organs. Abnormal disposition is associated with complex

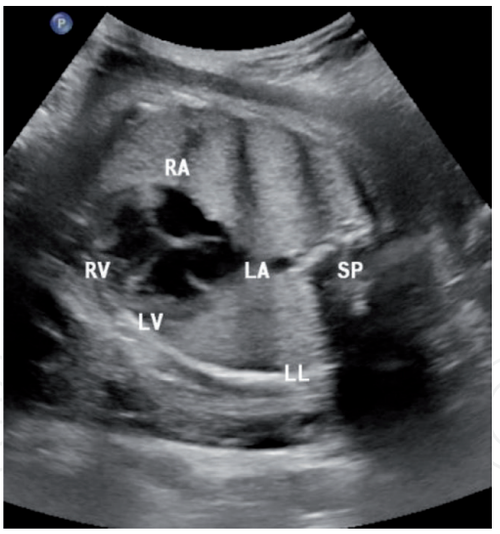

Figure 3.
Obstetric ultrasound; 4-chamber heart. Normal four-chamber view of the heart. Transverse scan through the fetal heart showing right ventricle (RV) anteriorly, left ventricle (LV) on the left, right atrium (RA) on the right and the left atrium (LA) as the most posterior. The spine (SP) and left lung (LL) are demonstrated.

cardiac anomalies. A transverse scan of the upper abdomen allows demonstration of the situation of the liver, stomach and great abdominal vessels. An axial section of the chest demonstration of a four-chamber view of the fetal heart. The heart covers about one third of the thorax. It is shifted to the left part of the thorax with the apex directed to the left.

About 90% of ultrasonographically detectable fetal cardiac defects demonstrate some abnormalities in the four-chamber view. Heart rate and rhythm are assessed subjectively. M-mode is useful for the evaluation of abnormal cases though is of little help in assessing morphology of the heart (**Figure 3**).

2.3 Pulmonary abnormalities

The lungs are interrogated in the same section used for the four-chamber view of the heart. The fetal lungs are uniformly echogenic. At 18–23 weeks, the middle third of the thoracic area at the level of the four-chamber view is occupied by the heart and the remaining two thirds by the lungs.

2.4 Anterior abdominal wall

The prevalence of anterior abdominal wall defects in sub Saharan Africa is not known as there are no population based studies. This could be due to non-availability of prenatal diagnosis and poor outcome of care post-delivery [24]. In a study carried out in a Nigerian tertiary hospital, omphalocele was the most common anterior abdominal wall defect seen [25].

Normal development of the anterior abdominal wall depends on the fusion of four folds (cephalic, caudal and two lateral). Developmental midgut herniation occurs at 8–10 weeks with subsequent retraction into the abdominal cavity at 10–12 weeks.

The integrity of the abdominal wall should always be adequately demonstrated. This is achieved via transverse scans which demonstrate the insertion of the umbilical cord. The urinary bladder should be visualized within the pelvis which rules out bladder exstrophy.

A study conducted to determine the ability of routine obstetric ultrasound to detect and accurately describe fetuses with anterior abdominal wall defects

demonstrated examination between 16 and 22 weeks gestation detected 60% of defects with a false positive rate of 5.3%. Fetuses with gastroschisis were incorrectly assigned as exomphalos in 14.7% of cases recognized before 22 weeks gestation. The diagnosis was completely accurate in 71.6% of cases. Therefore, problems of diagnostic accuracy should be put into consideration when counseling couples with a fetal anomaly.

2.5 Gastrointestinal tract

The fetal abdomen differs from the abdomen of the older child or adult. The fetal abdomen is large in relation to its body dimension compared with the adult. The liver is larger with the left lobe is bigger than the right owing to its greater supply of oxygenated blood. The umbilical vein is an important US landmark. Half the blood it carries goes directly to the inferior vena cava via the ductus venosus. The remainder perfuses the liver via the left portal vein. The gall bladder is visualized as an ovoid cystic structure below the intrahepatic portion of the umbilical vein. The spleen may be demonstrated in a transverse plane posterior and to the left of the stomach. The adrenal glands are up to 20 times larger in relative size because of the presence of a fetal zone. The pelvis is small with the pelvic organs extending into the lower abdomen. Swallowing commences at 11 to 12 weeks gestational age (GA). The fetal stomach is visible from 9 weeks of gestation as a sonolucent cystic structure in the upper left quadrant of the abdomen. It should be filled with swallowed fluid by 18 weeks GA. The small bowel is moderately echogenic and centrally located. Peristalsis in small intestine loops is usually demonstrable by the third trimester. The visualized small gut usually measures just below 6 mm in width and below 15 mm in length. The large bowel is seen after 20 weeks of intrauterine life as a tubular organ in the periphery of the abdominal cavity. It gradually fills up with meconium but does not usually surpass 23 mm in caliber.

The abdominal circumference should be measured in a scan of the abdomen demonstrating the stomach and the portal sinus of the liver. The visceral situs should also be evaluated.

This is done by demonstrating the relative location of the stomach, hepatic vessels, abdominal aorta and inferior vena cava (**Figure 4**).

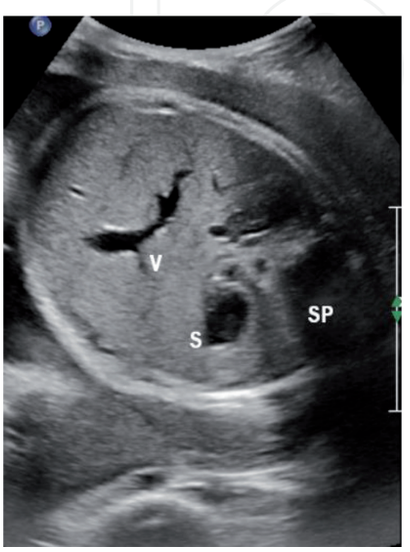

Figure 4.
Obstetric ultrasound showing the fetal abdomen. Fetal abdomen: The stomach (S) and intrahepatic portion of the umbilical vein (V) are demonstrated. The spine (SP) is seen posteriorly.

A case series of gastrointestinal abnormalities in fetuses with echogenic bowel detected during the antenatal period revealed that prenatal diagnosis of bowel abnormalities is challenging owing to the varying appearance of the bowel throughout pregnancy [26].

A related study showed that the prenatal ultrasound scan is unreliable in the detection or exclusion of fetal gastrointestinal malformations (GIM). Therefore clinicians involved in prenatal sonography or counseling should exercise caution in making such diagnoses. In this study, there were 220 confirmed cases of GIM, of which only 35 (16%) had been correctly identified prenatally. However, prenatal ultrasound was quite reliable in the detection of duodenal obstruction with 55% confirmed cases identified prenatally [27].

2.6 Kidneys and urinary tract

Detection of congenital urinary system anomalies is an important aspect of the prenatal ultrasound examination. Prenatal diagnosis of urinary tract abnormalities known to precipitate neonatal urosepsis and sequel such as renal scarring has made it possible to commence early intervention. A complete workup of the infants can be initiated early and before life-threatening complications occur.

The kidneys are visualized on sonography from as early as nine weeks of gestation and in all cases from twelve weeks. Echogenicity is high at nine weeks but reduces with advancing gestational age.

In a longitudinal scan, the kidneys are seen as elliptical structures while on axial sonograms, they are seen as rounded structures on either side of the spine. At 20 weeks, they show a hyperechoic capsule and the cortical area is slightly more echogenic than the medulla. Fat tissue normally accumulates around the kidneys as gestation progresses which enhances the borders of the kidneys in contrast with other organs. Normal ureters are rarely visualized in the absence of distal obstruction or reflux. The fetal bladder can be seen from the first trimester in more than 90% of subjects by 13 weeks (**Figure 5**).

A retrospective review of 56 children with urinary tract abnormalities detected by prenatal ultrasound revealed that more than half of the abnormalities were isolated hydronephrosis or multicystic dysplasia of the kidney [28].

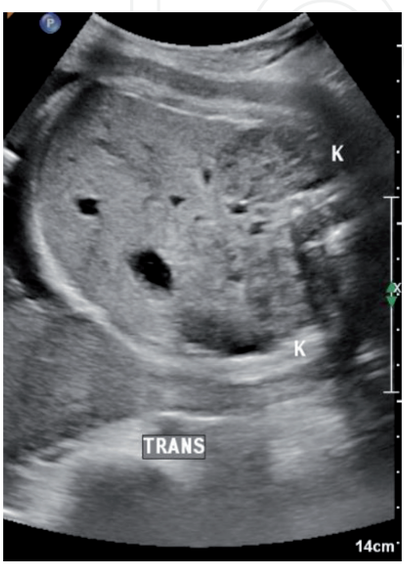

Figure 5.
Obstetric ultrasound showing fetal kidneys. Fetal abdomen at the level of the kidneys: Both kidneys (K) are seen on either side of the spine in this transverse sonogram.

The most frequent causes of hydronephrosis in the antenatal period are ureteropelvic junction (UPJ) obstruction, ectopic ureterocele, and posterior urethral valves (PUV). Renal pelvis of more than 10 mm in anteroposterior diameter or more than half of the anteroposterior diameter of the kidney in transverse section are conclusive evidence of significant hydronephrosis.

2.7 Skeleton

Detection of fetal anomalies is satisfactory for most organ systems but remains poor for cardiac, skeletal, and craniofacial anomalies. In a study to assess the accuracy of the prenatal diagnosis of skeletal dysplasias by Barbara V. Parilla et al., the antenatal diagnosis was correct in 20 (65%) of 31 cases [29]. This suggests that precise antenatal diagnosis of skeletal dysplasia is challenging. However, the antenatal prediction of lethality was highly accurate.

In general, skeletal dysplasias are uncommon. They affect 1 in every 4,000–5,000 births, even though the incidence may be higher since the features may not be apparent until early childhood, at what time short stature, joint abnormalities or other complications become apparent.

Skeletal dysplasias comprise a heterogeneous group of disorders of skeletal growth that result in bones of abnormal size and shape.

Normally, all long bones are consistently seen from 11 weeks and the limbs move about readily at this gestation.

At the 18–23 week scan, the 3 segments of each limb should be visualized. It is nevertheless only necessary to measure the length of one femur. The relationship of the leg and foot should also be evaluated to rule out club foot.

US findings that are highly associated with the presence of a generalized skeletal dysplasia include shortening of extremity bones, fractures, bowing of long bones, demineralization, and a small thorax. A ratio of femur length to foot length of less than 0.9 and femur length–abdominal circumference ratio of less than 0.16 suggest a skeletal dysplasia.

3. Conclusion

Antenatal ultrasound can allow for early detection of fetal anomalies and therefore early intervention as well as appropriate management of the pregnancy.

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