

CONTEMPORARY REVIEW ARTICLE

10-14 Weeks Scan

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INTRODUCTION

With the advent of high-frequency transducers the image quality has improved and a distinct description of the embryonic morphology has now become possible.

The description of embryonic anatomy, the normal anatomic relations and development of abnormalities as seen by ultrasound is now termed as sonoembryology.

Knowledge of the normal embryonic development and the appearances of the normal embryo on ultrasound is important.

It is important in order to confirm the presence of normal anatomy or deviation from normal anatomy to diagnose anomalies.

The chronological development of the human embryo is now well-described and is extremely useful in identifying fetal malformations by ultrasound using transabdominal and transvaginal multi-frequency 3-10 MHz transducers (Fig. 1).



Fig. 1: USG machine and probe

EMBRYOLOGIC DEVELOPMENT AS SEEN ON ULTRASOUND

Gestational age 09-10 weeks¹

- Skeletal anatomy including fingers and toes can be seen with high frequency ultrasound transducers (Fig. 2).

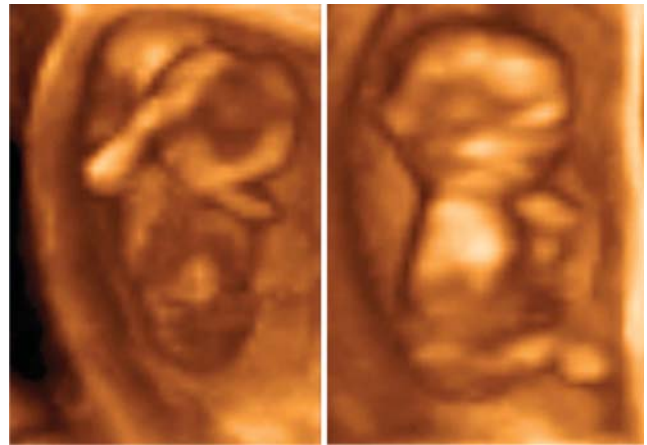


Fig. 2: Skeletal anatomy including fingers and toes can be seen with high frequency ultrasound transducers

- The cerebellar hemispheres can be easily seen and there is a physiological deficiency in the vermis at this stage and the defect in the vermis can be delineated (Fig. 3).
- Echogenic choroids plexuses can be seen filling the lateral ventricles (Fig. 4).
- Differentiation of the spine can now be seen (Fig. 5).
- The umbilical cord can be seen connecting the fetus and the placenta on 2D and color flow mapping (Fig. 6).

Gestational age 10 weeks and more

- Fetal profile can be better seen with proper fetal posture (Fig. 7).
- Limbs can be much better delineated.
- Calvarium can be seen with delineation of the hemispheres, choroid plexuses and posterior cranial fossa structures.
- Spine can be much better appreciated.
- Cardiac rate, rhythm and configuration can be assessed.
- The gut is seen herniating during the 10th week, which is back in the abdomen by the 11th week³ (Fig. 8).



Fig. 3: The cerebellar hemispheres can be easily seen and there is a physiological deficiency in the vermis at this stage and the defect in the vermis can delineated



Fig. 4: Echogenic choroids plexuses can be seen filling the lateral ventricles



Fig. 5: Differentiation of the spine can now be seen

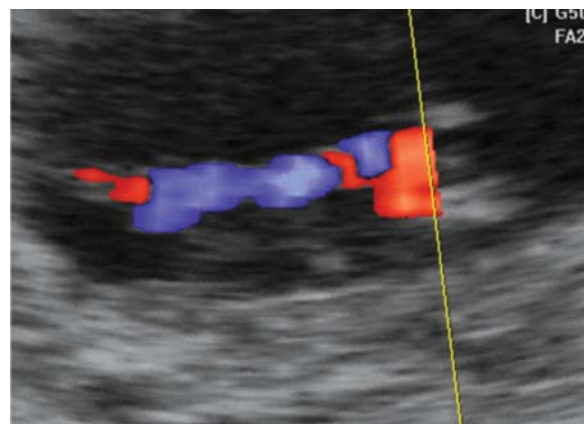


Fig. 6: The umbilical cord can be seen connecting the fetus and the placenta on 2D and color flow mapping



Fig. 7: Fetal profile can be better seen with proper fetal posture



Fig. 8: The gut is seen herniating during the 10th week, which is back in the abdomen by the 11th week

- Kidney can also be seen adjacent to the fetal spine as echogenic structures.
- Fetal bladder filling can be seen in many cases by the 12th week (Fig. 9).

NUCHAL TRANSLUCENCY

In the first trimester, the term translucency is used because this is the ultrasonographic feature that is observed; during the second trimester, the translucency usually resolves and, in a few cases it may evolve into either nuchal edema or cystic hygroma.

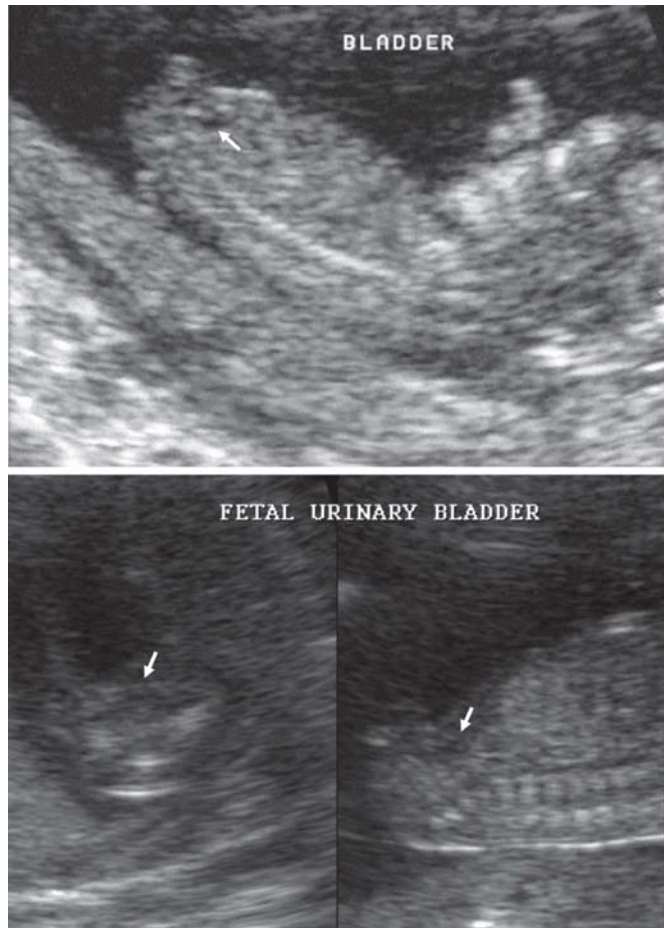


Fig. 9: Fetal bladder filling can be seen in many cases by the 12th week

- In 74-77% of trisomy 21 fetuses the fetal nuchal translucency is increased with a low false positive rate.
- Sensitivity for detection of chromosomal abnormalities is extremely high by a combined screening of maternal age, fetal nuchal translucency and maternal biochemistry.
- The translucency (subcutaneous) between the skin and soft tissue posterior to the cervical spine has to be measured (Fig. 10).
- Nuchal translucency thickness usually increases with gestational age with 1.5 mm and 2.5 mm being the 50th and 95th percentile respectively for gestational ages between 10 and 12 weeks. 2.0 mm and 3.0 mm are the 50th and 95th percentile respectively for gestational ages between 12 and 14 weeks.²
- An increased nuchal translucency thickness not only indicates increased suspicion of chromosomal abnormalities but also indicates a possibility of multiple structural defects especially of the fetal heart and abdomen. Therefore, a



Fig. 10: The translucency (subcutaneous) between the skin and soft tissue posterior to the cervical spine has to be measured



Fig. 11: An increased nuchal translucency thickness not only indicates increased suspicion of chromosomal abnormalities but also indicates a possibility of multiple structural defects of the fetal heart and abdomen. Therefore, a normal karyotype does not in any way normalcy of the fetus

normal karyotype does not in any way ensure normalcy of the fetus (Fig. 11).

- A thickened nuchal translucency with spontaneous resolution and a normal nuchal skin fold thickness does not exclude a karyotypic abnormality.
- High risk patients for chromosomal abnormalities and cardiac defects should definitely be subjected to an ultrasound between 10-14 weeks for measurement of nuchal translucency thickness.

In trisomy 21, the medial nuchal translucency thickness is about 2 mm above the normal medial for 2 mm above the normal medial for crown rump length. The corresponding values for trisomies 18 and 13, triploidy and Turner's syndrome are 4 mm, 2.5 mm, 1.5 mm and 7 mm respectively.^{6,7}

PRENATAL DIAGNOSIS

“The palpebral fissure is narrow. The nose is small. The skin has a slight dirty yellow tinge and is deficient in elasticity giving the appearance of being too large for the body”.

The above is an extract from the paper “observations on an ethnic classification of idiots” by Langdon down published in 1866. These observations by down has made the basis of trying to make an ultrasound diagnosis of mental retardation (Down's syndrome) and other chromosomal anomalies in the first trimester.

Today prenatal diagnosis has become an important aspect of antenatal ultrasound as many fetal anomalies can be diagnosed due to our better understanding of sono-embryology.⁹ But only a few anomalies are common enough for screening and these are:

1. Down's syndrome.
2. Neural tube defects.
3. Hemoglobinopathies.
4. Tay Sach's disease.
5. Cystic fibrosis.
6. Fragile 'X' carriers.
7. Duchenne's muscular dystrophy.

The incidence of detectable anomalies is neural tube defects 2/1000 births, Down's syndrome 1/900 births, other chromosomal anomalies 1/2000-3000 births.⁴ The aims of prenatal diagnosis and for a screening test is to select a woman at a higher risk enough to warrant an invasive test. Individual risk assignment by age, family history, markers is made and a cut off point is selected, which will determine whether the result is screen positive or screen negative and the test is judged by the detection rate and false positive rates.

Today an anomaly scan by a high resolution transvaginal scan offers a reliable screening method for chromosomal anomaly detection. A late first trimester 10-14 weeks scan is an ideal time.⁵

THE 10-14 WEEKS SCAN

The 10-14 weeks fetal scan should be done by a transvaginal probe with facilities for color Doppler and 3D/4D incorporated. The scan should include documentation of:

1. Viability.
2. Fetal number.
3. Implantation site and placentation.
4. Growth measurement.
5. Liquor assessment.
6. Detailed anomaly scan.
7. Nuchal anomaly scan.
8. Other marker study (nasal bone and iliac angle).
9. Doppler of umbilical and ductus venosus and fetal echocardiography.

10. 3D for surface anatomy.
11. 4D for maternal fetal bonding.

COMMON ANOMALIES THAT CAN BE DETECTED ON ULTRASOUND

Most fetuses with chromosomal defects have structural anomalies, some of these can be recognized at the 10-14 weeks scan.⁸

NEURAL TUBE DEFECTS

- Acrania (exencephaly) shows a deformed cephalic end of the fetus with a thin membranous covering superior to the face with part of the brain also seen (Fig. 12).
- Anencephaly can be diagnosed in the first trimester with the characteristic appearance of nothing (brain and bone) seen superior to the orbits (Fig. 13).
- Depending on the size of the herniated contents of the brain and the defect in the bone encephaloceles can be diagnosed in the late first trimester. Cystic structures can be seen herniating from the defect with an associated microcephaly an abnormally shaped head.
- Inadequate visualization of the posterior cranial fossa structures with a disorganization of the parallel configuration of the fetal spine can be seen in the late first trimester.

CYSTIC HYGROMA (FIG. 14)

- Can be seen in the longitudinal section of the spine at the cranio-vertebral junction and cervical area.
- Can be seen as a localized lesion in the cervical area or can be seen as a diffuse lesion on the thorax and abdomen.
- Simple nuchal edema is usually associated with cardiac defects or trisomies.
- Septated extensive fluid collections are usually associated with Turner's syndrome.

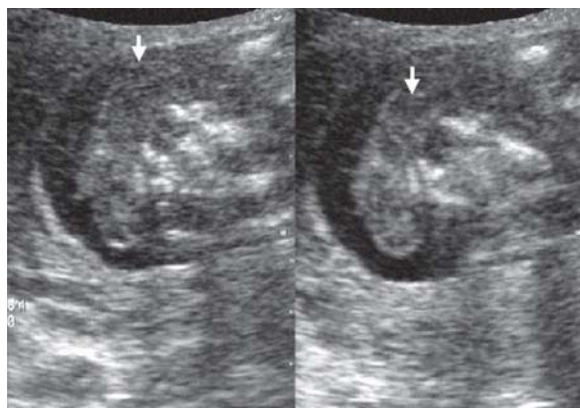


Fig. 12: Acrania (exencephaly) show a deformed cephalic end of the fetus with a thin membranous covering superior to the face with part of the brain also seen

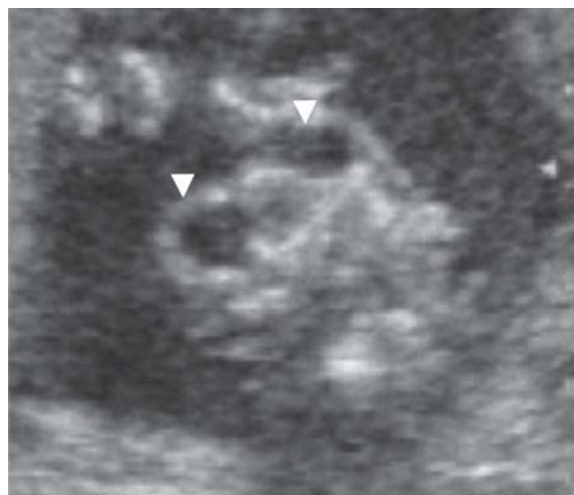


Fig. 13: Anencephaly can be diagnosed in the first trimester with the characteristic appearance of nothing (brain and bone) seen superior to the orbits



Fig. 14: Cystic hygroma

FACIAL ANOMALIES (ESPECIALLY ORBITS)

- Proboscis can be delineated on ultrasound as early as 12 weeks (Fig. 15).
- Severe hypotelorism can also be delineated as orbits can be delineated from 11 weeks onwards.
- Single orbit, anophthalmia can also be diagnosed.
- Holoprosencephaly occurs in 3%.

HEART DEFECTS

- Very gross anomalies of cardiac configuration or M-mode tracings indicating abnormal heart-rate and rhythm can be diagnosed at the end of the first trimester. These definitely have to be followed with a detailed evaluation of the cardiac



Fig. 15: Proboscis can be delineated on ultrasound as early as 12 weeks

configuration and connections at an appropriate time (Fig. 16).

BODY-WALL DEFECTS

- Because of the fact that there is a physiological defects in the abdominal wall as seen on ultrasound, one should be



careful in diagnosing omphalocele before 12 weeks (Fig. 17).

- Ectopia cordis and limb body wall complex can be diagnosed on ultrasound with their associated skeletal defects and anomalies.

GASTROINTESTINAL ANOMALIES

- Esophageal or duodenal atresia can be suspected in the first trimester but for a definitive diagnosis re-evaluation is to be always suggested (Fig. 18).

GENITOURINARY ANOMALIES

- Nonvisualization of kidneys with a minimal oligohydramnios or even normal liquor amnii can raise a suspicion of renal agenesis.
- Visualization or nonvisualization of the renal arteries on color flow mapping can also diagnose renal agenesis (unilateral or bilateral).
- Dilated/overdistended urinary bladder can be diagnosed after 12 weeks (Fig. 19).

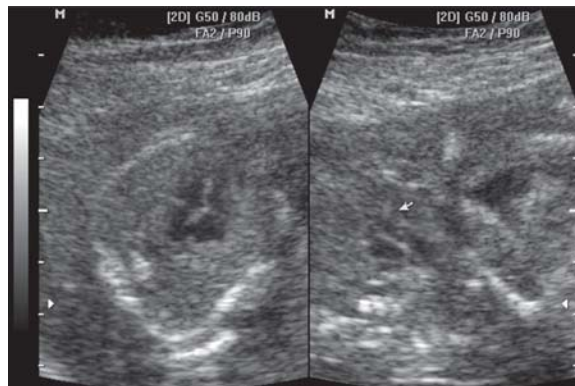


Fig. 16: Very gross anomalies of cardiac configuration or M-mode tracings indicating abnormal heart rate and rhythm can be diagnosed at the end of the first trimester. These definitely have to be followed with a detailed evaluation of the cardiac configuration and connections at an appropriate time



Fig. 17: Because of the fact that there is a physiological defects in the abdominal wall as seen on ultrasound, one should be careful in diagnosing omphalocele before 12 weeks



Fig. 18: Esophageal or duodenal atresia can be suspected in the first trimester but for a definitive diagnosis re-evaluation is to be always suggested



Fig. 19: Dilated/overdistended urinary bladder can be diagnosed after 12 weeks

FETAL GROWTH

An impaired fetal growth could be associated with chromosomal anomalies. Drugan et al reported that a fetal CRL smaller than 7 mm of expected indicates a three time higher risk of chromosomal anomalies.

MUSCULOSKELETAL ANOMALIES

- Abnormalities in shape, size and proportions with cranium can raise suspicion of reduction defects or skeletal dysplasias, which can be diagnosed as early as 11 weeks.
- Defects in spinal curvature can also be delineated at around 12 weeks.

ABSENT NASAL BONE

Absence or delay in development of nasal bone is an important marker for trisomy 21. Can be easily recognized by ultrasound in late first trimester (Fig. 20).⁹

FETAL ILIAC ANGLE

Widened fetal iliac angles (haring) is an important marker for trisomy 21. 2D sonography and now 3D ultrasound can demonstrate iliac angles at 11-14 weeks.^{10,11}

MEGA CYSTIS

First trimester megacystis has a frequency of about 1 in 1633 and in 60% it resolves. If megacystis persists it is a marker for chromosomal anomalies.¹²

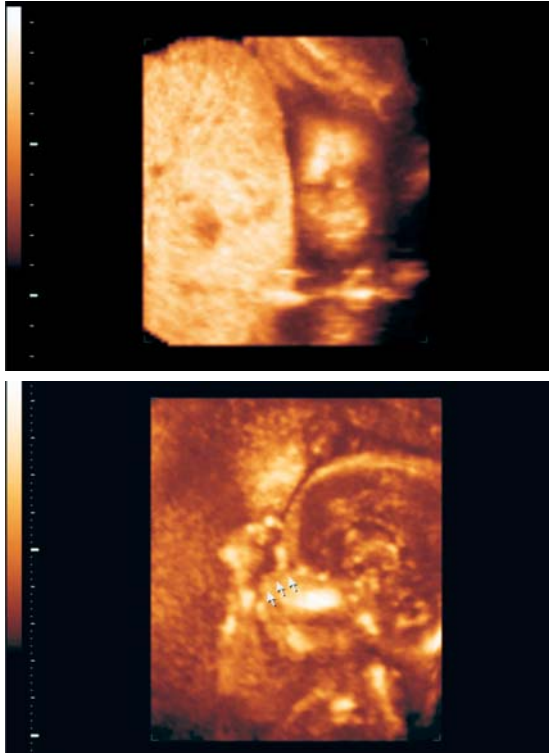


Fig. 20: Absence or delay in development of nasal bone is an important marker for trisomy 21. Can be easily recognized ultrasound in late first trimester

UMBILICAL CORD DIAMETER

An increased umbilical cord diameter over 95th centile of reference is more frequently seen with abnormal fetal karyotype.¹³

UMBILICAL CORD DOPPLER

A higher than normal pulsatility of umbilical artery in the first trimester and in fetuses with increased NT has been noted by various workers (Martinez et al)¹⁴ how ever others have not noted a significant differences (Brown¹⁵ and Jauniaux¹⁶) combined with the presence of other markers umbilical artery pulsatility make be taken as a sign.

DUCTUS VENOSUS VELOCITY

Congenital heart defects are common in chromosomal anomalies and these may present with an abnormal fetal hemodynamics, which are best reflected in ductus venosus velocities in the 11-14 weeks scan.

Absence or inversion of forward velocity in atrial contraction (ACV) is seen in 70-90% of fetuses with chromosomal anomalies.¹⁷

FUTURE TRENDS

With three-dimensional ultrasound making its entry into the first trimester, it is possible to analyze the recorded ultrasound in multiple planes through the embryo giving a more precise diagnosis (Fig. 21).

The step by step development, normal physiological processes and also the maldevelopment of organs can thus be assessed leading to an accurate diagnosis in early pregnancy itself reducing the torment of the parents.

CONCLUSION

By 10 weeks onwards it is possible to offer a safe and accurate technique for prenatal diagnosis of chromosomal anomalies using transvaginal high resolution screening and subjecting only screen positive patients to an invasive testing of CVS or early amniocentesis.

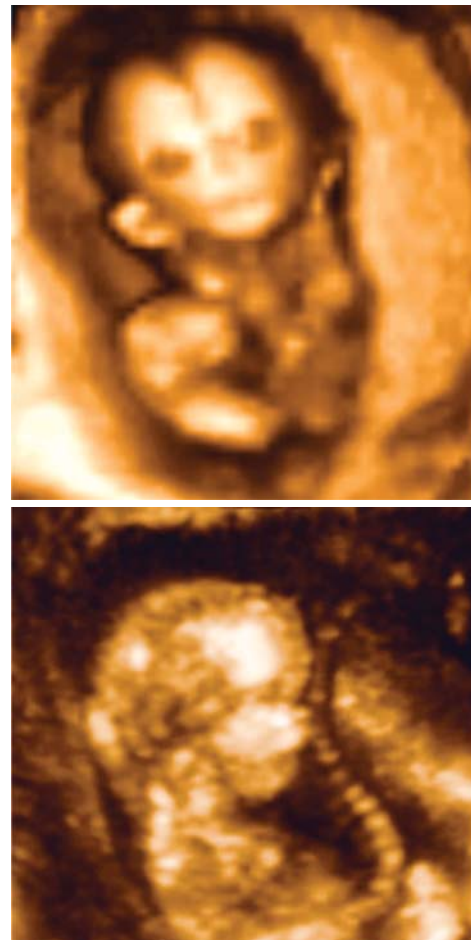


Fig. 21: With three-dimensional ultrasound making its entry into the first trimester, it is possible to analyse the recorded ultrasound in multiple planes through the embryo giving a more precise diagnosis

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