

# Diagnosis of Prenatal Abnormalities by Ultrasound

## Introduction

Ultrasounds play a very large role when imaging a fetus; that is why prenatal ultrasounds are primarily done to ensure that any possible abnormalities are addressed and treated sooner, rather than later. There are multiple different types of ultrasounds. However, each type of ultrasound varies. Even though certain types are still able to aid one another in the discovery of abnormalities, the different types of ultrasounds still show a variety of findings and are of a specific importance.

## First Trimester

During the first trimester of pregnancy serious abnormalities can be found on a fetus. However, just like any other study that may be performed, important characteristics may also go undetected. Common places for anomalies to be discovered during the first trimester include the brain, face, neck, chest, abdomen, and spine. Though abnormalities can be found in the chest, abdomen, and spine during this first trimester, these types of anomalies are commonly missed. Listed below are some of the common abnormalities that may be discovered throughout the first trimester of pregnancy. See Table 1 below for a detailed selection of abnormalities that do not get missed, may be missed, and will be missed during the first trimester.

## Common Abnormalities for First Trimester

- Omitted brain and skull fragments
- Cleft lip
- Cystic Hygromas
- Chest masses
- Abnormal alignment of the heart
- Ventral wall defects
- Hernias
- Enlarged bladder
- Spina bifida
- Large masses

**First Trimester**  
**Table 1 (10-13 weeks)**

|              | Don't miss  | You might miss  | You will miss   |
|--------------|---|---|---|
| Brain/ skull | <ul style="list-style-type: none"> <li>• Anencephaly/acrania</li> <li>• Holoprosencephaly</li> <li>• Large cephalocele</li> <li>• Cystic hygroma</li> </ul>                         | <ul style="list-style-type: none"> <li>• Smaller caphaloceles</li> </ul>  | <ul style="list-style-type: none"> <li>• Agenesis of the corpus callosum</li> <li>• Posterior fossa abnormalities</li> <li>• Ventriculomegaly</li> <li>• Destructive lesions</li> </ul> |
| Face         | <ul style="list-style-type: none"> <li>• Hypotelorism (severe)</li> <li>• Micrognathia (severe)</li> <li>• Large median or bilateral clefts</li> <li>• Large facial mass</li> </ul> | <ul style="list-style-type: none"> <li>• Micrognathia (mild)</li> <li>• Small facial mass</li> </ul>                                  | <ul style="list-style-type: none"> <li>• Hypotelorism (mild)</li> <li>• Unilateral clefts</li> </ul>  |
| Chest        | <ul style="list-style-type: none"> <li>• Abnormal situs</li> <li>• Mediastinal shift</li> </ul>   | <ul style="list-style-type: none"> <li>• Most chest masses</li> </ul>   | <ul style="list-style-type: none"> <li>• Structural cardiac abnormalities</li> </ul>  |
| Abdomen      | <ul style="list-style-type: none"> <li>• Omphalocele (large)</li> <li>• Gastroschisis</li> <li>• Limb-body-wall-complex</li> <li>• Megacystis</li> </ul>                            | <ul style="list-style-type: none"> <li>• Omphalocele (small)</li> </ul>   | <ul style="list-style-type: none"> <li>• Bowel atresia</li> <li>• Renal abnormalities</li> <li>• Abnormal genitalia</li> </ul>  |
| Spine        | <ul style="list-style-type: none"> <li>• Large neural tube defects</li> <li>• Large soft-tissue masses</li> </ul>   | <ul style="list-style-type: none"> <li>• Small soft-tissue masses</li> </ul>  | <ul style="list-style-type: none"> <li>• Most spina bifida</li> <li>• Hemivertebra</li> </ul>   |
| Extremities  | <ul style="list-style-type: none"> <li>• Large limb reduction defect</li> <li>• Severe skeletal dysplasia</li> </ul>  | <ul style="list-style-type: none"> <li>• Small limb reduction defects</li> <li>• Joint contractures</li> <li>• Polydactyly</li> </ul> | <ul style="list-style-type: none"> <li>• Mild skeletal dysplasia</li> <li>• Syndactyly</li> </ul>   |

**Second Trimester**  
**Table 2 (18-20 weeks)**

|               | Don't miss*  | You might miss   | You will miss   |
|---------------|--|--|---|
| Brain/ skull  | <ul style="list-style-type: none"> <li>• Ventriculomegaly</li> <li>• Dandy-Walker malformation</li> <li>• Arnold-Chiari II malformation</li> <li>• Agenesis of the corpus callosum</li> </ul>  | <ul style="list-style-type: none"> <li>• Partial agenesis of the corpus callosum</li> <li>• Vascular malformations</li> <li>• Destructive lesions</li> </ul> | <ul style="list-style-type: none"> <li>• Lissencephaly</li> <li>• White matter lesions</li> <li>• Small infarcts</li> </ul> |
| Face          | <ul style="list-style-type: none"> <li>• Cleft lip</li> <li>• Micrognathia</li> <li>• Large masses</li> </ul>  | <ul style="list-style-type: none"> <li>• Superficial cleft lip</li> <li>• Mild micrognathia</li> <li>• Retrognathia</li> <li>• Ear abnormalities</li> </ul>  | <ul style="list-style-type: none"> <li>• Isolated cleft palate</li> </ul>   |
| Neck          | <ul style="list-style-type: none"> <li>• Nuchal edema/ cystic hygroma</li> <li>• Large neck mass</li> </ul>  | <ul style="list-style-type: none"> <li>• Small neck masses</li> </ul>  | <ul style="list-style-type: none"> <li>• Thyroid abnormalities</li> <li>• Laryngeal obstruction</li> </ul>                  |
| Chest         | <ul style="list-style-type: none"> <li>• Diaphragmatic hernia</li> <li>• Large chest mass</li> <li>• Pleural effusion</li> </ul>   | <ul style="list-style-type: none"> <li>• Small chest masses</li> </ul>   | <ul style="list-style-type: none"> <li>• Diaphragmatic eventration</li> </ul>   |
| Heart         | <ul style="list-style-type: none"> <li>• Ventricular disproportion</li> <li>• Large ventricular septal defect</li> <li>• Tricuspid/ mitral valve atresia</li> <li>• Arrhythmia</li> <li>• Tetralogy of Fallot</li> <li>• Truncus arteriosus</li> <li>• Transposition of great vessels</li> </ul> | <ul style="list-style-type: none"> <li>• Smaller ventriculoseptal defects</li> <li>• Aortic coarctation/ stenosis</li> </ul>                                 | <ul style="list-style-type: none"> <li>• Atrial septal defects</li> <li>• Anomalous pulmonary venous drainage</li> </ul>    |
| Abdomen       | <ul style="list-style-type: none"> <li>• Gastroschisis</li> <li>• Omphalocele</li> <li>• Choledochal cyst</li> <li>• Ascites</li> </ul>  | <ul style="list-style-type: none"> <li>• Esophageal atresia</li> <li>• Extrapulmonary bronchopulmonary sequestration</li> </ul>                              | <ul style="list-style-type: none"> <li>• Intestinal atresia</li> <li>• Anal atresia</li> </ul>                              |
| Genitourinary | <ul style="list-style-type: none"> <li>• Single umbilical artery</li> <li>• Renal agenesis</li> <li>• Multicystic/ dysplastic kidneys</li> <li>• Hydronephrosis</li> <li>• Megacystis</li> <li>• Bladder exstrophy</li> </ul>  | <ul style="list-style-type: none"> <li>• Unilateral renal agenesis</li> <li>• Ectopic kidney</li> <li>• Ambiguous genitalia</li> </ul>                       | <ul style="list-style-type: none"> <li>• Hypospadias</li> </ul>   |
| Spine         | <ul style="list-style-type: none"> <li>• Spina bifida</li> <li>• Sacrococcygeal teratoma</li> </ul>  | <ul style="list-style-type: none"> <li>• Hemivertebra</li> <li>• Spina bifida occulta</li> </ul>   | <ul style="list-style-type: none"> <li>• Tethered cord</li> </ul>   |
| Extremities   | <ul style="list-style-type: none"> <li>• Limb reduction defects</li> <li>• Micromelia (short limbs)</li> <li>• Fractures/ bowing</li> <li>• Joint contractures</li> <li>• Absent thumb</li> </ul>  | <ul style="list-style-type: none"> <li>• Polydactyly</li> <li>• Syndactyly</li> <li>• Soft-tissue abnormalities</li> </ul>                                   | <ul style="list-style-type: none"> <li>• Mild skeletal dysplasia</li> </ul>   |

## Conclusion

Though there are multiple different types of ultrasounds, that may show similar anomalies, no two are the same. Each different type of ultrasound is of its own importance. Studies and scientists are proving that as people learn more, ultrasounds are constantly improving. In turn, this is how medical professionals are coming up with more ways for abnormalities to be discovered, diagnosed, and treated. Do to the constant ways of finding methods to improve, ultrasounds, along with other imaging modalities, are continually changing and are proving to be a large asset in the medical field.

## References

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## Second Trimester

Once the second trimester has begun, a large portion of the fetus' anatomy can be viewed. Abnormalities in parts of the fetus that were once hard to see in the first trimester, may now have a greater chance of being detected. More anomalies are likely to be found during the second trimester because the fetus has had more time to fully develop. Along with the better visualization of the chest, heart, abdomen, and spine, the genital area has also begun to develop and can now be visualized. Below are some common abnormalities that may be found during the second trimester. Table 2, to the left, gives a detailed selection of abnormalities that do not get missed, may be missed, and will be missed during the second trimester of pregnancy.

## Common Abnormalities for Second Trimester

- Improper heart alignment
- Ventricle abnormality
- Cleft lip
- Spina bifida
- Pleural effusion
- Masses

## Forms of Ultrasounds

| 2D  | 3D  | 4D  |
|---|---|---|
| <ul style="list-style-type: none"> <li>• Around the longest</li> <li>• Seen in two dimensional plane</li> <li>• Commonly done with 3D ultrasounds</li> <li>• Substituted with fetal MRIs</li> </ul> | <ul style="list-style-type: none"> <li>• First used in the 1900's</li> <li>• Seen in three dimensional plane</li> <li>• Commonly done with 2D ultrasounds</li> <li>• Also utilized for reading MRI and CT images</li> </ul> | <ul style="list-style-type: none"> <li>• Commonly images heart to see moving 3D images</li> <li>• May be obtained using spatiotemporal image correlation (STIC)</li> <li>• Also done by using matrix-array transducers</li> </ul> |