

The Role of Ultrasound in Anencephaly Diagnosis

Anencephaly

When the neural tube does not close correctly, amniotic fluid reaches undeveloped organs and nervous system which causes the cerebrum and cerebellum to deteriorate. The fetus is born missing its forebrain and large parts of the skull. The majority of these pregnancies result in miscarriage, and if the child is born it is most likely stillborn or will die within a few hours.¹

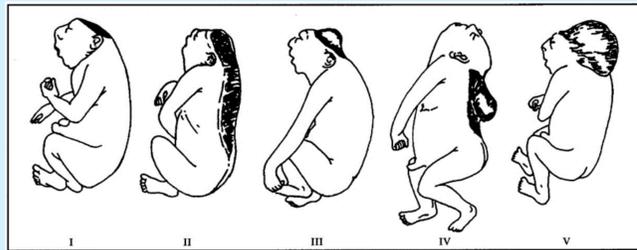


Figure 1. Classification of the different types of anencephaly.²

There are five types of anencephaly (see Figure 1):

- Type I- Classic anencephaly with symmetric absence of cranial bones above the orbits and no cerebrovascular matter.
- Type II- An extended deficit in the spine where the neural tube is open part way down the back.
- Type III- An exophytic stroma above the first cervical vertebrae.
- Type IV- Cervical lordosis with cerebrovascular tissue covered with dura mater.
- Type V- Extrusions of the brain tissue.²



Figure 2. Infant born with anencephaly.³

Gestation Timeline

10-12 Weeks Gestation

Acrania: flat bones in the cranial vault are absent. 'Mickey Mouse' sign present.



12-14 Weeks Gestation

Crown-rump length is reduced from normal gestational age.



18-22 Weeks Gestation

Absence of cerebral hemispheres and cranial vault. Appearance of frog eyes.



Figure 3. Acrania.⁴



Figure 4. Mickey Mouse Sign.⁵



Figure 5. Crown-rump length.⁶



Figure 6. Frog eyes.⁴

Diagnosis

Ultrasound is commonly used to diagnose neural tube defects because it is non-invasive and reliable. It has been seen to have 98% sensitivity and 100% specificity for the detection of neural tube defects.⁷ Anencephaly is often diagnosed during routine pregnancy ultrasound appointments. The baby's head appears flattened because of the abnormal development which can first be seen at the end of the first trimester.⁸ Once into the second trimester, more characteristics of anencephaly are prominent and a definite diagnosis can be made.⁷

Radiographic Appearance

The first finding that suggests anencephaly is acrania which is the absence of cranial bones (see Figure 3). The coronal view of the head shows some cerebral tissue that still exists as two semi-circles above the face, known as the 'Mickey Mouse' sign (see Figure 4). Crown-rump length is decreased by an average of 2 weeks less than expected for the gestational age and continues to degenerate (see Figure 5). Symmetric absence of the cranial vault is a constant feature in anencephalic fetuses. The cerebrum deteriorates resulting in no tissue present above the orbits which creates the appearance of frog eyes (see Figure 6).⁹

References

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