

# Holoprosencephaly at 9 weeks 6 days in a triploid fetus: 2D and 3D ultrasound findings

## OBJECTIVES

To report one of the earliest prenatally diagnosed cases of holoprosencephaly, in which 3D imaging was used to assist in the diagnosis.

## CASE REPORT

A transvaginal scan was performed at 9 weeks 6 days because of a suspected fetal anomaly detected at the booking scan. 2D and multiplanar 3D views revealed a singleton fetus with a crown-rump length of 34 mm. An anechoic area was clearly identified in the anterior aspect of the fetal brain, suggesting the diagnosis of alobar holoprosencephaly. A chorionic villus sampling was carried out one week later, at which time the pre-procedure abdominal scan revealed a single cerebral monoventricular cavity, fused thalami, midfacial hypoplasia, absent orbits, small omphalocele and an increased nuchal translucency thickness of 5.6 mm. The placenta appeared normal and the fetal growth interval was within normal range. Trophoblast culture revealed a 69,XXY karyotype. The patient miscarried at 13 weeks.

## CONCLUSIONS

The prenatal detection of holoprosencephaly before 10 weeks can be considered as an early manifestation of fetal triploidy, despite normal placental appearance and adequate growth interval. 3D ultrasound can assist in the first-trimester diagnosis by providing multiplanar views of the fetal head and face.



FIGURE 1a

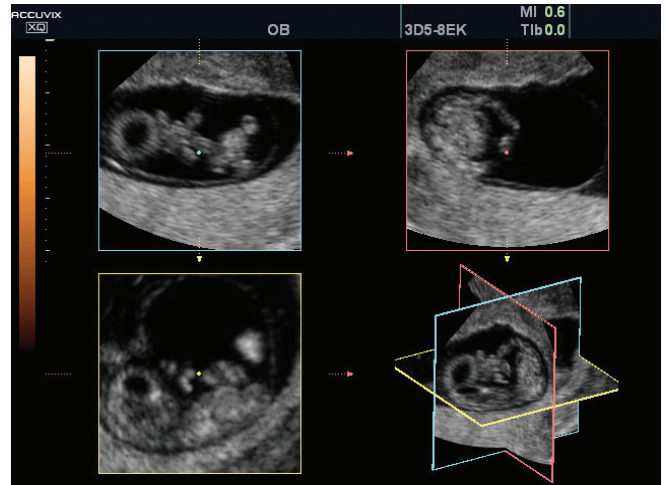


FIGURE 1c



FIGURE 1b

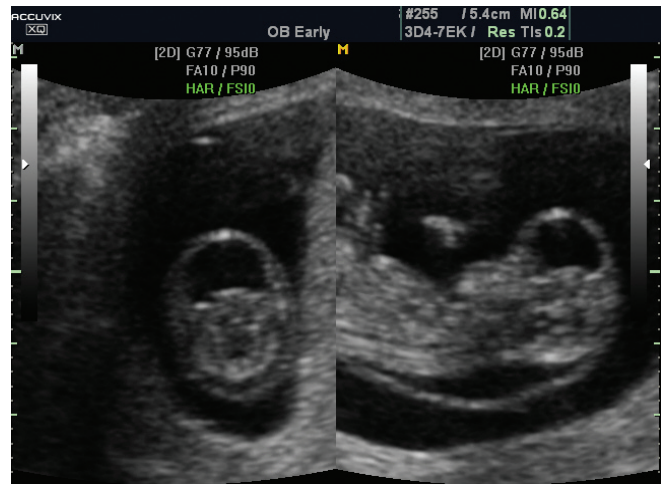


FIGURE 1d

FIGURE 1abcd. Transvaginal ultrasound findings at 9+6wks. 1a. Cross sectional view of the head shows monoventricle and fused thalami. 1b. Extended view of the fetus shows the brain anomaly. 1c. Multiplanar views and niche technique show holoprosencephaly. 1d. Findings at 10+6wks shows holoprosencephaly and increased NT.



FIGURE 2. Postmortem specimen shows abnormal facial features.