

Picture of the Month

Holoprosencephaly at 10 weeks 2 days (CRL 33 mm)

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The increasing relevance of sonoembryology in the management of pregnancies at risk of developmental anomalies has already been recognized in previous issues of this Journal^{1–3}. The latest paper from the Trondheim group, published in this issue provides, in addition, evidence of the significant role which 3-D reconstruction can play in the diagnostic process in the first trimester⁴.

We are now becoming familiar with features that remained unseen by earlier generations of equipment and operators and the Picture of the Month demonstrates this fact with impressive clarity. The image (Figure 1) demonstrates the three large brain cavities of the early fetus, namely the diencephalon, mesencephalon and rhombencephalon. Closer observation below the normal brain cavities reveals an abnormal smaller cystic area, the holosphere, which lies immediately superior to a finger-like projection of a proboscis. These two features strongly suggest the diagnosis of alobar holoprosencephaly.

Towards the end of the sixth postmenstrual week growth and differentiation of the cranial end of the neural tube

produces the three primary brain cavities, the forebrain or prosencephalon, the mid brain or mesencephalon and the hind brain or rhombencephalon. In normal embryological development the prosencephalon divides, during the sixth postmenstrual week, into two cavities, the telencephalon and the diencephalon. The cerebral hemispheres and the lateral ventricles are derivatives of the former while the thalami and the third ventricle derive from the latter. Complete failure of the telencephalon to divide during the seventh week⁵ results in a single cavity or holosphere rather than two lateral ventricles typical of normal development. The holosphere is diagnostic of alobar holoprosencephaly. The less severe forms of holoprosencephaly, namely semi-lobar and lobar, arise from incomplete division of the telencephalon.

The size of the holosphere varies considerably and may be small as in the case demonstrated here or may be far larger as typified by the cases reported at 10 weeks by both Wong and Turner^{2,3}. There is therefore no 'typical' appearance of alobar holoprosencephaly, based on the size of the holosphere, rather the diagnosis is made by failure to demonstrate both and separate lateral cerebral ventricles.

Incomplete cleavage and development of the forebrain also results in the facial anomalies frequently associated with holoprosencephaly and as demonstrated by the Picture of the Month. The proboscis, which can be clearly seen in this sagittal view, commonly accompanies cyclopia where the eyes or their precursor anlagen are fused or partially fused into a midline structure within a single orbit. Coronal views of the fetal face would be required to confirm the presence of the cyclopia.

All these features can be identified using 2-D imaging providing adequate resolution of the equipment and optimal presentation of the late embryo/early fetus. The advantage of the 3D techniques is that such appearances can be obtained and a diagnosis made when the fetus is not in such a position, thus reducing stress and anxiety to the parents and maximizing use of operator time for the clinical department.

T. Chudleigh and S. Campbell

REFERENCES

- 1 Blaas H-G. The examination of the embryo and early fetus: how and by whom? *Ultrasound Obstet Gynecol* 1999; 14: 153–7



Figure 1 Sagittal view of fetus at 10 weeks 2 days of gestation (CRL 33mm) with holoprosencephaly. The holosphere and proboscis are clearly demonstrated.

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- 3 Turner C, Silva S, Jeanty P. Prenatal diagnosis of alobar holoprosencephaly at 10 weeks of gestation *Ultrasound Obstet Gynecol* 1999; 13: 360–2
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- 5 Blaas H-G, Eik Nes SH, Kiserud T, Hellevik LR. Early development of the forebrain and midbrain: a longitudinal study from 7 to 12 weeks of gestation. *Ultrasound Obstet Gynecol* 1994; 4: 183–192