Prenatal ultrasound diagnosis of holoprosencephaly

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ABSTRACT
We describe a Pakistani woman who on prenatal grey scale ultrasound examination showed a fetus with a monoventricular cavity, fused thalami, absence of septum pellucidum and falx cerebri and hypotelorism. (Rawal Med J 2012;37:340-342).

Key words
Holoprosencephaly, monoventricular cavity, fused thalami, hypotelorism, polyhydramnios

INTRODUCTION
Holoprosencephaly is a complex developmental abnormality of the forebrain occurring in early embryonic life; incidence is 1:16,000 births.¹ This anomaly results from failure of cleavage of prosencephalon and categorized into three forms; alobar, semilobar and lobar. A spectrum of craniofacial malformations can occur which include a monoventricular cavity, fusion of the thalami and absence of the corpus callosum, falx cerebri, optic tracts and olfactory bulb. Facial defects may affect the orbits, nose and upper lips.² Associations are trisomy 13, 18, polyhydramnios, renal and cardiac anomalies.¹ We report a case who on antenatal ultrasound examination showed monoventricle, fused thalami, absence of septum pellucidum and falx cerebri and hypotelorism along with polyhydramnios.

CASE REPORT
A 36 years old female patient presented with gestational amenorrhea of 30 weeks. She had three live issues and two previous abortions. After two first trimester abortions, the following three pregnancies were uneventful. She was referred to radiology department of our hospital for ultrasound examination for the first time.

A gray scale ultrasound examination revealed a monoventricular cavity, fused thalami, absence of septum pellucidum and falx cerebri and hypotelorism (Fig 1 and 2). Polyhydramnios was also identified; the amniotic fluid index was 33 (Fig 3).

Fetal spine and heart were normal. No abdominal abnormality was seen. On the basis of above mentioned findings, sonological diagnosis of holoprosencephaly was suggested. She developed premature labor pains and there was rupture of membranes at gestational age of 31 weeks.

Fig 1. Axial ultrasound image of fetal brain showing fused thalami.

Fig 2. Ultrasound image of the fetal brain showing hypotelorism.
Fig 3. Antenatal ultrasound showing polyhydramnios

The baby born was a stillbirth and midline facial defects including hypotelorism and midline cleft lip were identified which confirmed the diagnosis of holoprosencephaly.

DISCUSSION
Holoprosencephaly denotes an incomplete or absent division of the embryonic forebrain (prosencephalon) into distinct lateral cerebral hemispheres. DeMyer roughly categorized holoprosencephaly into 3 types (from most severe to least severe), alobar, semilobar and lobar holoprosencephaly. In alobar holoprosencephaly, there is a complete absence of midline forebrain division, resulting in a monoventricle and fused cerebral hemispheres. Semilobar holoprosencephaly is characterized by an incomplete forebrain division, resulting in partial separation of the cerebral hemispheres, typically posteriorly. In lobar holoprosencephaly, there is complete ventricular separation, with focal areas of incomplete cortical division or anterior falx hypoplasia present.

Distinctive midline facial malformations occur in most cases. Typical facial anomalies are correlated with the degree of holoprosencephaly and have prognostic significance. From most severe to least severe, these include cyclopia, in which a single, midline, fused eye exists in a single orbit below a proboscis; ethmocephaly, in which ocular hypotelorism is present with an interorbital proboscis; ceboccephaly, in which ocular hypotelorism is present with a single-nostril nose; ocular hypotelorism and midline clefting; and more subtle facial dysmorphic features may also be present. These include a flat nasal bridge and tip; a single, midline, upper incisor; a bifid uvula; absent nasal bones and nasal septum; and congenital nasal pyriform aperture stenosis (decreased width of the nasal pyriform aperture at the level of the interior meatus). Microcephaly is the rule, and macrocephaly, if present, is suggestive of hydrocephalus. Virtually all surviving individuals with the more severe forms of holoprosencephaly have some developmental delay, often persisting as mental retardation. In general, this finding is directly correlated with the severity of holoprosencephaly.

Also not uncommon are seizures, hypotonia and/or hypertonia, extrapyramidal features, such as dystonia and/or chorea; hypothalamic and brainstem dysfunction leading to autonomic dysfunction and swallowing difficulties; pituitary dysfunction, which can manifest as partial or complete panhypopituitarism with resultant endocrine deficiencies; and feeding difficulties, which can lead to aspiration pneumonia and failure to thrive.

Prenatal evaluation by means of transabdominal or transvaginal ultrasonography can be used to identify most cases of alobar or semilobar holoprosencephaly. Prenatal MRI can be helpful in cases where the infant's head is not easily accessible at the time of ultrasonography or where the anatomy is not satisfactorily delineated at prenatal ultrasonography. The advent of high-resolution real-time ultrasound imaging has allowed detection on the group of holoprosencephalies, but lack of familiarity with uncommon forms may lead to diagnostic confusion. Coronal sonograms of the fetal head, in addition to standard axial projections, should be performed whenever an intracranial cystic abnormality is identified. The degree of facial dysmorphism tends to parallel the severity of holoprosencephaly and therefore, sonographic evaluation of facial morphology may aid in prenatal diagnosis.
REFERENCES


