



UNIVERSITY of  
**ROCHESTER**  
MEDICAL CENTER

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### DEPARTMENT OF IMAGING SCIENCES

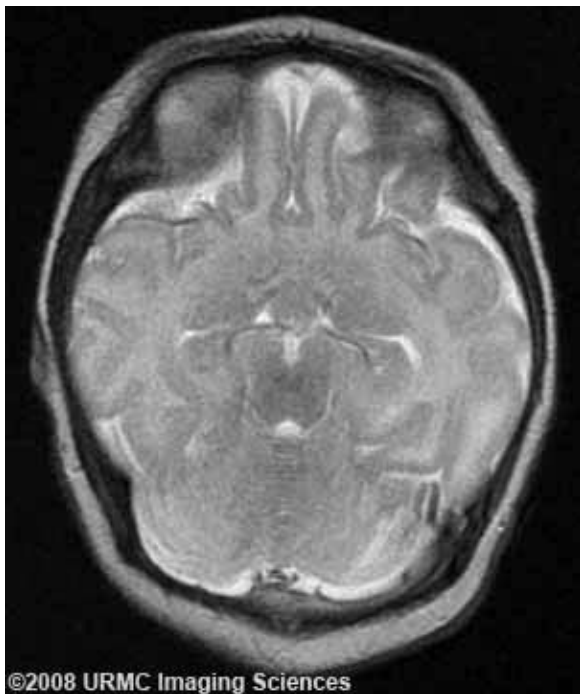
## Imaging Sciences Interesting Cases

### CASE 259

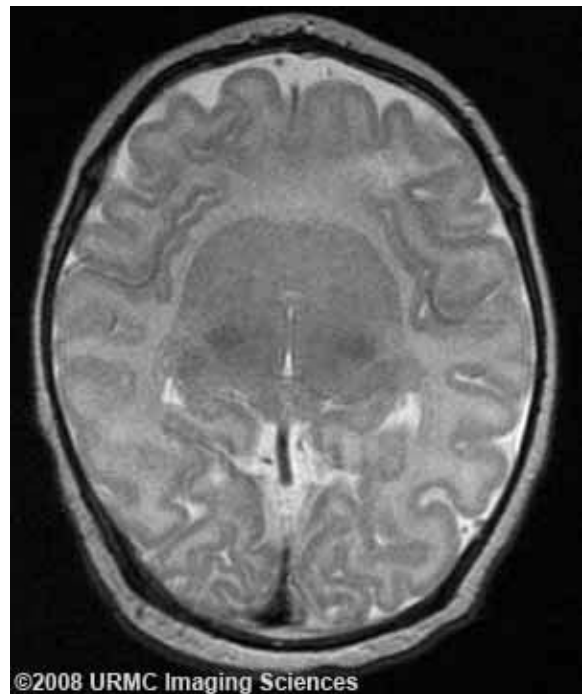
**Parul Patel, MD**

**CLINICAL PRESENTATION:** Patient is a 2-week-old female with cleft lip and cleft palate questionable for holoprosencephaly.

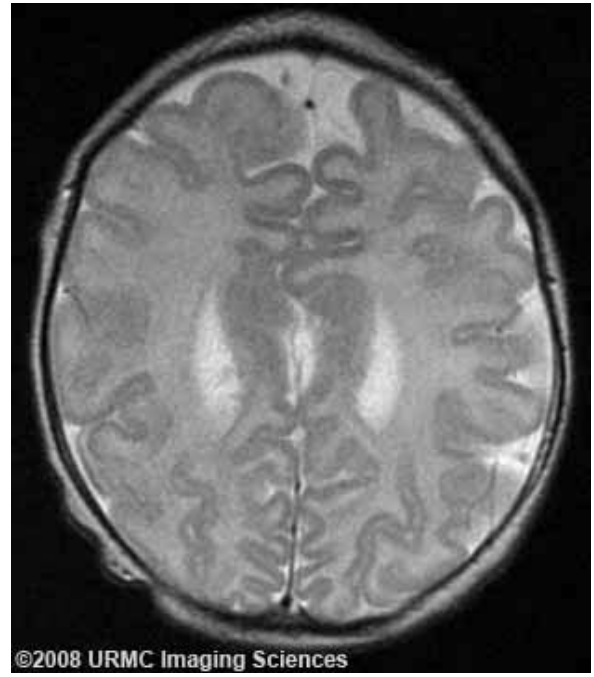
**IMAGING FINDINGS:** Partial fusion of the frontal lobes with absence of the falx and septum pellucidum in this region, hypoplasia of the genu and body of the corpus callosum, absent frontal horns of the lateral ventricles is consistent with a mild form of semilobar holoprosencephaly. Bilateral thalami are appropriately separated.



**Figure 1:** MRI T2 axial image at the base of the frontal lobe demonstrates the frontal lobe to be partially fused more posteriorly on this image.



**Figure 2:** MRI T2 axial image slightly more superior demonstrates the frontal lobes to again be partially fused. The basal ganglia are also fused. The thalami however are clearly separated.



**Figure 3:** MRI T2 axial image more superior demonstrates that there is partial cleavage of the frontal lobes. This image also demonstrates the presence of an interhemispheric fissure.

### **DIAGNOSIS: Semilobar holoprosencephaly**

**DISCUSSION:** Holoprosencephaly is a disorder characterized by the failure of the prosencephalon (the forebrain of the embryo) to develop. During normal development the forebrain is formed and the face begins to develop in the fifth and sixth weeks of human pregnancy. Holoprosencephaly is caused by a failure of the embryo's forebrain to divide to form bilateral cerebral hemispheres (the left and right halves of the brain), causing defects in the development of the face and in brain structure and function.

The estimated incidence of holoprosencephaly is 1 per 13,000 live births. The more severe forms of holoprosencephaly can usually be diagnosed prenatally by intrauterine ultrasound. There is no sex predilection. Most cases are thought to be sporadic. Risk factors associated with the development of holoprosencephaly include maternal diabetes, toxoplasmosis, syphilis, rubella and fetal alcohol syndrome. There is also an association of holoprosencephaly with various chromosomal anomalies. Most frequently, trisomies 13 and 15 are associated with holoprosencephaly. Patients with holoprosencephaly often have associated facial and cranial anomalies. For example, our patient presented with cleft lip and cleft palate. More severe associated deformity is cyclopia (single eye). Also, associated visceral abnormalities occur in about 75% of cases. They include cardiac, skeletal, genitourinary, and gastrointestinal anomalies.

There are several forms of holoprosencephaly. Alobar holoprosencephaly is the severest form. Most affected children are severely abnormal, manifesting seizures, abnormal neonatal reflexes and abnormalities of tone from the time of birth. These patients also have severe facial deformities and hypertelorism. In the extreme cases, the orbits and globes are fused, resulting in cyclopia. In these patients, the hypothalami and basal ganglia are fused and reduced in volume and the third ventricle is absent. These patients lack an interhemispheric fissure, falx cerebri, and corpus callosum. In most cases, no sylvian fissure is identified and if present, they are usually in the most anterior portion of the brain, close to the midline. The septum pellucidum is absent in all forms of holoprosencephaly.

In semilobar holoprosencephaly, the brain is less dysmorphic and the facial anomalies are mild or absent. Imaging studies reveal the interhemispheric fissure and falx cerebri to be partially formed as they were in our patient. They are usually well formed posteriorly and less well formed anteriorly in the frontal area. The deep cerebral nuclei are typically partially separated, but the hypothalami, caudate heads and thalami remain partly unseparated. In our patient the basal ganglia were partly fused and the thalami were separated.

Lobar holoprosencephaly is the least severe form. Affected patients typically present with mild or moderate developmental delays. The frontal lobes are more fully developed and more separated than the semilobar form. The sylvian fissures usually look near or completely normal. In these patients the interhemispheric fissure and falx cerebri extend into the frontal region of the brain. The falx however may be hypoplastic anteriorly.

**REFERENCES:**

1. Ball WS Jr. Pediatric Neuroradiology. Lippincott Williams & Wilkins; Philadelphia, 1997.
2. Barkovich JA. Pediatric Neuroimaging. 3rd. Ed. Lippincott Williams & Wilkins, Philadelphia, 2000.