

Incidence and Magnetic Resonance Imaging

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Mini Review

Holoprosencephaly (HPE) is a developmental anomaly featured by a failure in differentiation and cleavage of the prosencephaly resulting in incomplete separation of the two hemispheres. In almost 80% of individuals it is accompanied by specific craniofacial anomalies. Both genetic and teratogen factors are responsible for the development of HPE [1]. Rare in absolute terms, HPE is the most common brain abnormality and is seen in 1 per 8000-16,000 live births [2-4]. In an analysis of 21 HPE epidemiologic articles Orioli IM and Castilla EE [5] found that the pregnancy outcomes had relevant impact on the incident rate of HPE, being lower than 1 per 10,000 in live and stillbirth and between 40-50 per 10000 in aborted embryos. In a large series of 4,157,224 births, the same authors observed 370 infants with suspected holoprosencephaly (0.009%) stressing that isolated HPE was homogeneous among the 11 sampled countries, increasing from 0.5/10,000 births to 1/10,000 births between 1967 and 2000% [6]. The early embryonic occurrence may be even higher with prevalence of 1:250 in embryos [7] but may not be detected due to most fetuses aborting in early gestation [2]. In our small series of 4000 MRI explorations in children of different ages (from newborns to 15 years old) only three cases harboring HPE were diagnosed, accounting for almost 0.05%. Even though, the holoprosencephaly has been divided into three categories (alobar, semilobar and lobar) and a clear distinction between them does not exist. However, another two categories have been added to the previous one: the middle interhemispheric fusion variant (MIHF/MIHV or syntelencephaly [8] and a septopreoptic type [9]. Alobar HPE is a rare and the most severe congenital malformation, usually diagnosed by prenatal ultrasound, rarely postnatally by CT or MRI, because the infant is most often stillborn. There is no separation of cerebral hemisphere with only one large ventricle and failure of transverse cleavage into diencephalon and telencephalon. Semilobar HPE, less dysmorphic than alobar (HPE), has both the frontal and parietal lobes completely fused and interhemispheric fissure exist posteriorly. The concomitant anomalies might be microcephaly, macrocephaly, motor abnormalities such as choreoathetosis or lower extremity spasticity.

In lobar HPE, which is less severe than the previous two subtypes, both hemispheres and lateral ventricles are clearly defined but the most rostral aspect of the frontal lobes are fused especially ventrally. Mild or moderate developmental delay, pituitary dysfunction, or visual problems may harbor the patient with above mention anomaly. Diagnosis of HPE can be established by CT or MRI. Imaging by MRI is the study of choice and can determine the clinical subtypes and associates anomalies. The presentation of the imaging characteristics of HPE's subtypes are given below. In middle interhemispheric fusion

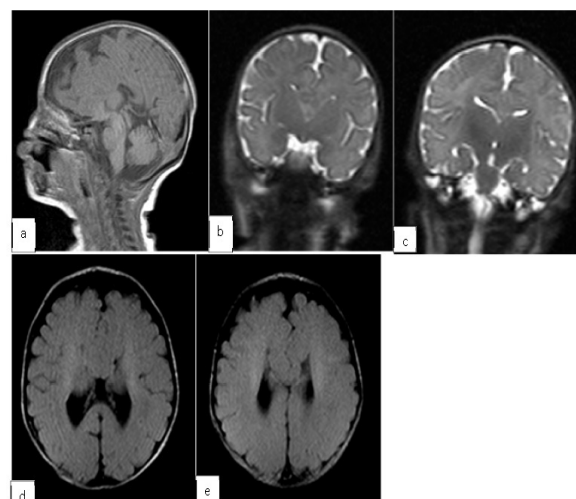


Figure 1: A case of semilobar holoprosencephaly (a 12 year old girl who underwent MRI examination after the first episode of seizures. She has been under antiepileptic drugs up to now. a) Sagittal T1W SE tomogram shows absence of rostrum, genu, anterior and part of posterior body of corpus callosum. Only small part of posterior body and splenium of corpus callosum are visible. b) and c) Coronal T2W SE tomograms: There is a fusion of thalami and also small part of frontal lobes around anterior commissure; frontal horns of the lateral ventricles are present, slightly dysplastic and opened laterally; the sylvian fissures look normal. e) Axial T2 FLAIR tomograms, the frontal lobes are more fully developed than in semilobar holoprosencephaly; the interhemispheric fissure and falx cerebri extend into the frontal area of the brain, although the anterior falx is hypoplastic. The septum pellucidum is absent.

variant (MIHF/MIHV or syntelencephaly) there is no separation of the posterior frontal and parietal lobes, absence of the body of the corpus callosum followed by different variations in the cleavage of the basal ganglia and thalami. In a septopreoptic type of HPE (very rare anomaly) only the septal and/or preoptic regions are not separated. Other entities may include septo-optic dysplasia, central incisor syndrome, non-specific midline dysplasia, frontonasal dysplasia, agnathia-otocephaly (Figures 1-3).

We are presenting MRI characteristics of our cases below.

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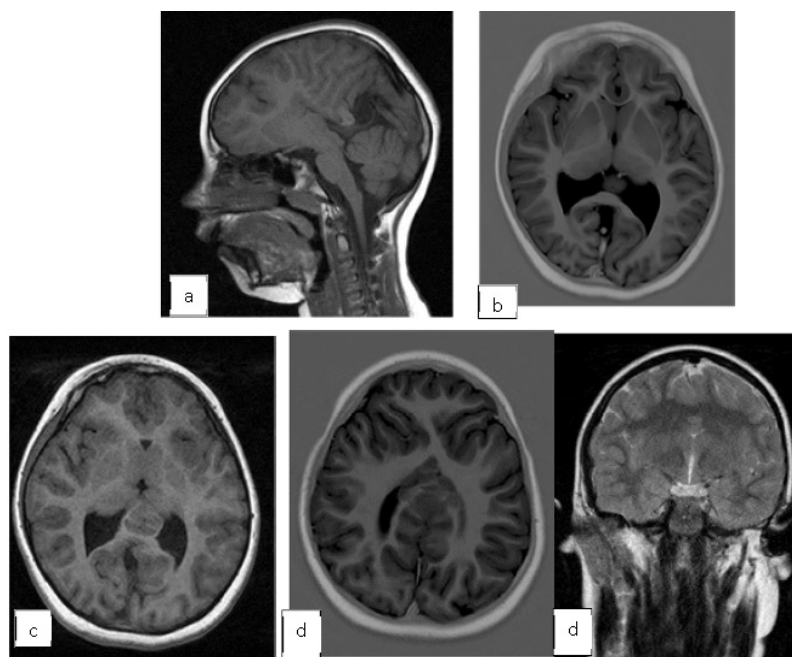


Figure 2: A case of lobar holoprosencephaly in neonatus one- month old who underwent MRI exam after ultrasound exploration. Almost all tomograms are degraded by motion artifacts in spite of sedation. The child is under close observation by paediatrician and neurologist. a) Sagittal T1W SE tomogram shows absence of rostrum, genu, anterior and part of posterior body of corpus callosum. Only small part of posterior body and splenium of corpus callosum are visible. b) and c) Coronal T2W SE tomograms: There is a fusion of thalami and also small part of frontal lobes around anterior commissure; frontal horns of the lateral ventricles are present, slightly dysplastic and opened laterally; the sylvian fissures look normal. e) Axial T2 FLAIR tomograms. The frontal lobes are more fully developed than in semilobar holoprosencephaly; the interhemispheric fissure and falx cerebri extend into the frontal area of the brain, although the anterior falx is hypoplastic. The septum pellucidum is absent.

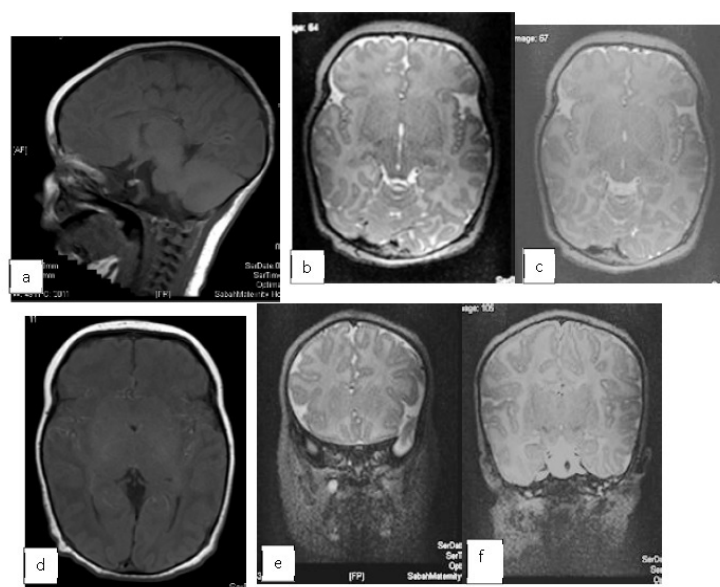


Figure 3: A case of mild lobar holoprosencephaly in one month old girl with multiple congenital anomalies (Cleft hard palate and bifid uvula, retro-micrognathia, calvarial middle bone defect) MRI was performed to rule out brain malformation. There is no defined corpus callosum on sagittal T1W SE images. It is only visible posterior thin part which could be consider at least as thin splenium/posterior body but it correlates with hippocampal commissure. a) and c) axial T2W SE, and d) axial T1W SE: There is partial fusion of basal ganglia in a part of thalami and also in hypothalamic region. Frontal horns of both lateral ventricles are almost invisible, present but dysplastic, very thin and displaced laterally. The third ventricle is also present but smaller than usual. Sylvian fissures look normal. e) and f) coronal T2W SE tomograms confirm lack of frontobasal lobes separation with incomplete interhemispheric fissure and anterior falx and hallmark of examination which is partial fusion of deep gray nuclei.

References

1. Barkovich AJ (2005) Pediatric Neuroimaging. (4th edn), LippincottWilliams and Wilkins, USA.
2. Raam MS, Solomon BD, Muenke M (2011) Holoprosencephaly: A guide to diagnosis and clinical management. Indian Pediatr 48: 457-466.
3. Dubourg C, Bendavid C, Pasquier L (2007) Holoprosencephaly. Orphanet J Rare Dis 2: 1-8.
4. Winter TC, Kennedy AM, Woodward PJ (2015) Holoprosencephaly: A survey of the entity, with embryology and fetal imaging. Radiographics 35: 275-290.
5. Orioli IM, Castilla EE (2010) Epidemiology of holoprosencephaly: Prevalence and risk factors. Am J Med Genet C Semin Med Genet 154C: 13-21.
6. Orioli IM, Castilla EE (2007) Clinical epidemiologic study of holoprosencephaly in South America. Am J Med Genet A 143A: 3088-3099.
7. Matsunaga E, Shiota K (1977) Holoprosencephaly in human embryos: Epidemiologic studies of 150 cases. Teratology 16: 261-272.
8. Barkovich AJ, Quint DJ (1993) Middle interhemispheric fusion: an unusual variant of holoprosencephaly. AJNR Am J Neuroradiol 14: 431-440.
9. Hahn JS, Barnes PD, Clegg NJ, Stashinko EE. (2010) Septopreoptic holoprosencephaly: A mild subtype associated with midline craniofacial anomalies. AJNR Am J Neuroradiol 31: 1596-601.