A Case Report on Syntelencephaly - Middle Interhemispheric Holoprosencephaly Variant

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Abstract: Holoprosencephaly is a rare congenital brain malformation. It has been traditionally classified in to three types: alobar, semi lobar and lobar forms. Syntelencephaly is a lesser known variant of holoprosencephaly, Middle Interhemispheric Fusion (MIH) variant. We present a case of Syntelencephaly along with a review of imaging findings and the structural abnormalities of the brain in Syntelencephaly, compare these features with those of classic holoprosencephaly (HPE) and consider its embryogenetics. [Ankita P NJIRM 2017; 8(6):102-105]

Key Words: Syntelencephaly, Middle Interhemispheric Holoprosencephaly Variant

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Case Report: A one year old female child presented with a history of developmental delay with delayed achievement of head holding and unable to sit without support. There was no significant peri natal history. No history of any familial disorder or similar complaints in siblings. No history of in utero exposure to infections, drugs, or other teratogens was elicited. Neurologic examination revealed normal pupillary light reflex, normal deep tendon reflexes, mildly increased muscle tone in four extremities.

MR imaging was performed with a 1.5-T superconducting magnet : Axial T1W, T2W, FLAIR, DW & T2* images were obtained. Sagittal T1W & T2W images were obtained. Coronal T2W, FLAIR, T1W & T2W IR sequences were obtained.

MR Imaging Findings:

1.Interhemispheric fusion of the cerebral hemispheres: both gray matter and white matter were fused in the posterior frontal and anterior parietal lobes with an absence of the interhemispheric fissure (IHF) Figure 1(a) . Fused crossing white matter tracts were identified under the fused cortex – best appreciated in T1W IR & T2W sequences Figure 1(b) & 1(c).The gyral pattern was normal. IHF was present frontally and occipitally.

Figure 1(a)

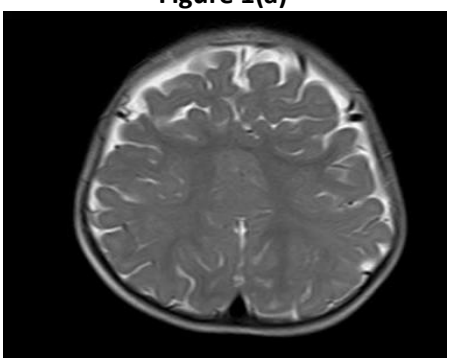


Figure 1(b)

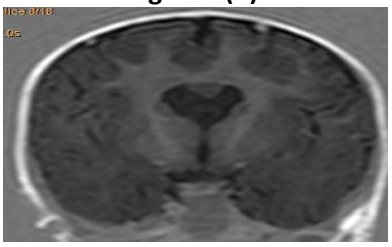
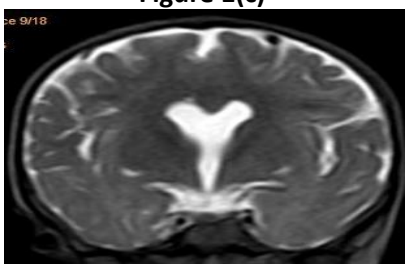


Figure 1(c)



2. Absent septum pellucidum: with fused bodies of both lateral ventricles. However normal differentiation of ventricular horns was noted Figure 2(a)&(b).

Figure 2(a)

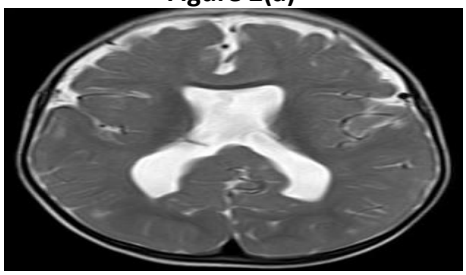
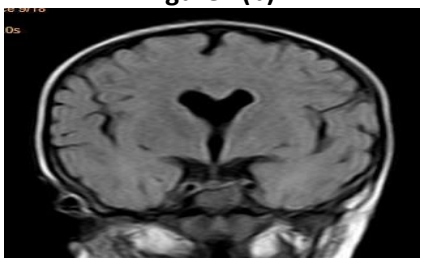
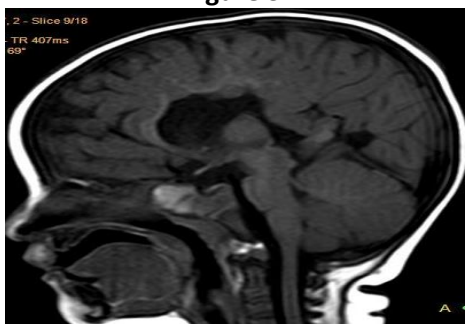


Figure 2(b)



3. Dysgenesis/ Partial agenesis of corpus callosum: normal rostrum, genu and splenium were present with absent intervening body Figure 3.

Figure 3



4. The sylvian fissures: were high riding with oblique orientation and abnormally connected across the midline over the vertex Figure 4(a), 4(b) & 4(c).

Figure 4(a)

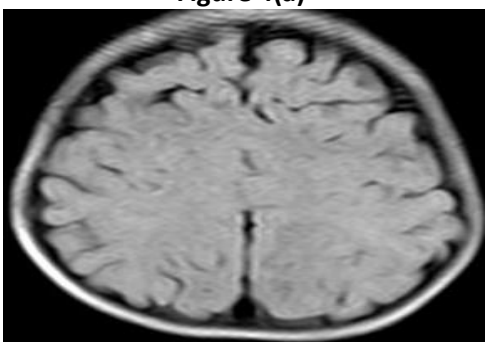


Figure 4(b)

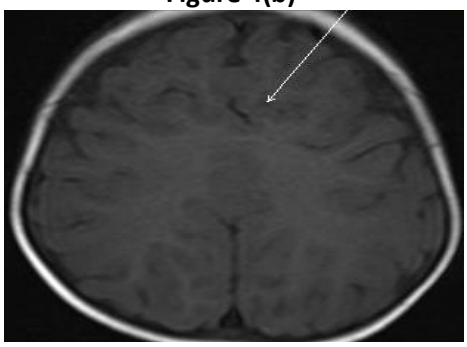
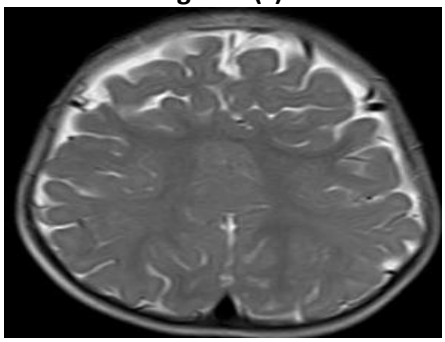


Figure 4(c)



5. Grey matter nodule: noted perching upon the dorsal aspect of fused lateral ventricle Figure 5(a), 5(b) &5(c).

Figure 5(a)

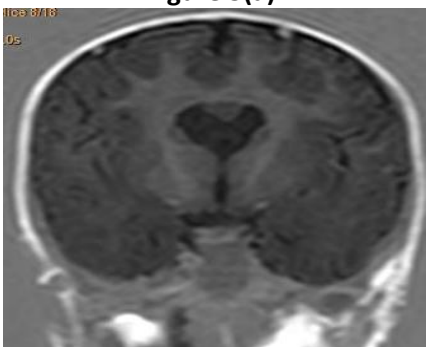


Figure 5(b)

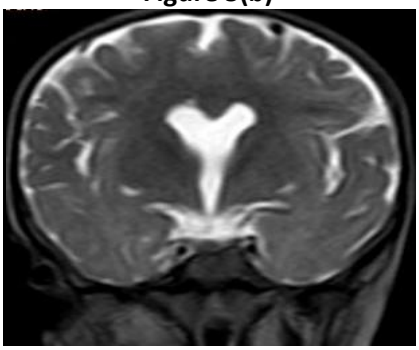
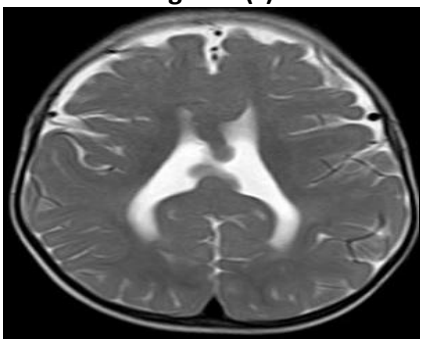
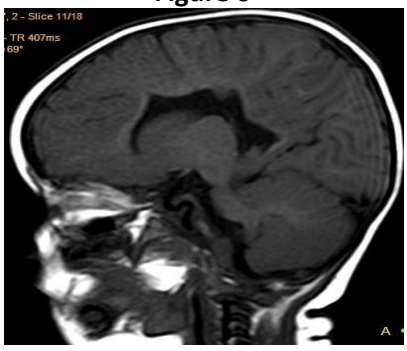


Figure 5(c)



6. The border of lateral ventricle appeared irregular due to overlying grey matter Figure 6.

Figure 6



Normal Findings Were:

- Normal Third and fourth ventricles seen.
- Optic tract and optic radiations were normal.
- No abnormal gyral pattern or grey matter heterotopias noted.
- Basal ganglia, hypothalamus and thalami were normally separated.
- Brain stem and cerebellum were normal.

Based on the classic imaging appearances, a diagnosis of MIH variant of holoprosencephaly was made.

Discussion: HPEs have traditionally been classified according to the system of DeMyer and coworkers into alobar, semi lobar, and lobar forms[1]. A fourth subtype, called the middle interhemispheric variant (MIH) of holoprosencephaly or syntelencephaly, was first identified in 1993 by Barkowich[2].

Neuroimaging in MIH reveals presence of anterior and posterior inter hemispheric fissure without separation of cerebral hemispheres in the posterior frontal and parietal regions. The sylvian fissures have an abnormal orientation and appear to connect across the midline in majority of the patients. Associated hetrotropic gray matter and cortical dysplasia are also common, including abnormal thickening of the cortex lining the anterior IHF[3].

The thalami are the most common deep nuclei affected in MIH and their abnormalities may be associated with dorsal midline cysts. The corpus callosum is malformed with variable presence of splenium& genu and absent intervening body-Only brain malformation with such distribution of corpus callosum . There may also be an associated azygous cerebral artery in MIH patients which was not seen in this patient.

Associated chiari malformations, cerebellar hypoplasia, cephaloceles and polymicrogyria have also been reported in MIH.

MIH differs from classic HPE in certain key aspects: Although MIH is considered a variant of HPE -The sites of involvement in this malformation differ from those seen in classic HPE. In contrast to HPE, instead of the most severe involvement being in the most rostral basal midline, patients with MIH have a relative sparing of the basal forebrain, with a well formed anterior and posterior interhemispheric fissure and normal or nearly normal caudates, hypothalami and basal ganglia. However, they can have more severe involvement of the thalami and the IHF in the region of the posterior frontal lobes and parietal lobes[4].

This translates into a slightly different clinical profile of patients with MIH as compared to HPE. Absence of endocrinopathy is noted in MIH as compared with the classic subtype which likely correlates with the lack of hypothalamic abnormalities. Choreoathetosis in MIH is also lower than that for semi lobar HPE, likely secondary to lack of caudate and lentiform nuclei abnormalities. Mobility, upper extremity function, and language have similarly been correlated with the degree of non separation of the caudate, lentiform and thalamic nuclei, and grade of HPE5[6]

Various forms of HPE: Are differentiated mainly by midline fissure, corpus callosum differentiation of cerebrum and lateral ventricle[7,8]

Alobar HPE:

- Most severe form of holoprosencephaly
- No interhemispheric fissure, sylvian fissure, falx or sagittal sinus
- No separation of cerebral hemispheres and no identifiable lobes of cerebrum-pancake brain with cresentric monoventricle.
- Absent /atrophic optic/olfactory tracts.

Semilobar HPE:

- Fused basal ganglia and thalami
- Interhemispheric fissure and falx are rudimentary
- Only temporal horns of lateral ventricles may be differentiated.
- Splenium of corpus callosum seen mostly, genu and body are absent

Lobar HPE:

- Absent/dysplastic frontal IHF-rest IHF normal and fused frontal hemispheres
- Absent rostrum and genu of corpus callosum with normal body and splenium.
- Dysmorphic frontal horns of lateral ventricles and normally developed temporal and occipital horns.

However, both MIH and classic HPE share a fundamental similarity:

- Non-cleavage of a substantial portion of the supratentorial brain into two separate hemispheres.

- Absent septum pellucidum with fused both lateral ventricles.

Septo-optic dysplasia[7,8]:

- Absence/hypoplasia of septum pellucidum with squared off frontal horns.
- Optic nerve and chiasma hypoplasia with visual impairment
- Completely separated cerebral hemispheres and rest of lateral ventricles.

Embryology: Mutations in ZIC2 on Chromosome 13 cause classic HPE as well as MIH. This provides evidence that MIH is a variant of HPE9. In MIH, impaired induction or expression of genetic factors appears to influence the embryonic roof plate, whereas in classic HPE, induction or expression of the embryonic floor plate seems to be affected. It results into a primary defect in basal forebrain patterning during the first 4 weeks of embryogenesis. This defect results in incomplete separation of the cerebral hemispheres and ventricles.

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