Welcome to Neuroradiology

CASE # 5049762318

Titulo: Fetal MRI Alobar Holoprosencephaly
Case: 5049762318
Sections: Brain
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Patient: F, 29 year(s)

CLINICAL HISTORY

29 year old female with abnormal fetal ultrasound examination.

IMAGING FINDINGS

Fetal MRI shows a borderline small intracranial volume with a T2 low signal behavior brainstem and midline fused thalamic nuclear mass centrally (Figure 1), surrounded by T2 hyperintense faintly visualized cerebellar hemispheres in the infratentorial compartment (Figure 2) and T2 hyperintense cerebral hemispheres in the supratentorial compartment. There is abnormal loss of fetal hemisphere definition, suggesting a bilateral global monoventricle with preservation of superficial cortical surface (Figure 3). The T2 hypointense basal nuclei are otherwise poorly defined, such as seen in fused thalami with no recognizable morphology. There are no definable lateral and third ventricles with common space in keeping with a large monoventricle. There is poor development of the facial structures and orbits with a rudimentary ocular structure centrally positioned (Figure 4) at the root of the proboscis (Figure 5) suggesting a cyclops morphology.

DISCUSSION

In the early intrauterine life after the primary neurulation, the cephalic end of the neural tube forms the prosencephalon, the mesencephalon and the rhombencephalon (1). The prosencephalon cleaves into the telencephalon and diencephalon. The telencephalon forms the cerebral hemispheres, putamen, and caudate nuclei while the diencephalon forms the thalamus, hypothalamus, globus pallidus and optic vesicles.
Holoprosencephaly (HPE) results from abnormal cleavage of the prosencephalon in the five week gestational age.Â HPE is classically divided in alobar, semilobar, and lobar forms. Other conditions included in the spectrum are the middle interhemispheric variant (Syntelencephaly) and septo-optic dysplasia.(2)
The spectrum starts with aprosencephaly (absence of the prosencephalon) and atelencephaly (absence of the telencephalon) They are united in one entity call aprosencephaly / atelencephaly or AP/AT, the most severe expression of the prosencephalon development.
Alobar HPE consist of a primitive monoventricule with no interhemispheric fissure or falx cerebri, fused thalami, basal ganglia and hypothalamus, with absence of the third ventricle. The brain can be described as a pancake, cup or ball shaped. Olfactory bulbs and tracts are missing. There is dysgenesis of the optic nerves. The corpus callosum (CC), the anterior commissure, and the septum pellucidum are not visible. The orbits and globes are fused resulting in cyclopia or partially fused globes (synophthalmia). There is no nose, rather a proboscis, an elongated appendage seen superior to the orbit.
Semilobar HPE main characteristic is incomplete interhemispheric fissure definition with a primordium of the falx cerebri. Monoventricle acquires a hemispheric form but has no septum pellucidum. There is still frontal lobe fusion in more than 50% approx. Thalami are partially fused, you can see a dorsal cyst.
Lobar HPE show complete formation of the interhemispheric fissure with separation of the cerebral hemispheres, with dysplastic frontal horns, formed third ventricle. Splenium and posterior body of the CC are formed. The septum pellucidum is not formed.
Middle interhemispheric variant (Syntelencephaly) is a subtype of HPE in which the posterior frontal and parietal lobes are fused, whereas the more frontal, occipital and temporal polar areas of the cerebrum are normally cleaved. Portions of the genu and splenium are developed.
The imaging study of choice for the diagnosis of HPE is MRI. Prenatal MRI is done with rapid sequences (single shot fast spin-echo T2) which allows for fast scanning to compensate for fetal motion(3). Fetal imaging is an important tool in patient counseling and pregnancy management.

FINAL DIAGNOSIS

Alobar Holoprosencephaly

DIFFERENTIAL DIAGNOSIS LIST

HydranencephalyFetal hydrocephalusOpen lip schizencephaly

REFERENCES

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[3]Griffiths PD, Jarvis D (2016)Â In Utero MR Imaging of Fetal Holoprosencephaly: A Structured Approach to Diagnosis and Classification. AJNR Am J Neuroradiol 2015 Nov 12

CITACION
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