Case 14736

Romboencephalosynapsis
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Section: Neuroradiology
Area of Interest: Neuroradiology brain
Procedure: Diagnostic procedure
Technique: MR
Technique: Catheter arteriography
Special Focus: Congenital Case Type: Clinical Cases
Authors: Huapaya Torres Janice, Donato Angel, Figueroa Ramon E.
Patient: 22 months, male

Clinical History:

22-month-old male patient with developmental disability and delayed walking.

Imaging Findings:

Brain MRI:
Dysplastic and truncated appearance of the posterior body and splenium of the corpus callosum, and apparent absence of the rostrum. There is abnormal contour and configuration of the superior cerebellar cistern and irregular orientation of the parietal occipital fissure. There is apparent fusion of the cerebellar hemispheres (rhombencephalosynapsis). The torcular herophili is in normal position. There is a focal defect in the posterior body and splenium of the corpus callosum to the left of midline. There is a small cavum septum pellucidum. Partial absence/dysgenesis of the posterior body and splenium of the corpus callosum. Apparent absence of the rostrum. Rhombencephalosynapsis. Delayed pattern of myelination within the periatrial white matter and white matter adjacent to the occipital horns.

Discussion:

Rhombencephalosynapsis is a rare malformation of the cerebellum, characterized by dorsal fusion of the cerebellar hemispheres, agenesis or hypogenesis of the vermis, fusion of dentate nuclei and superior cerebellar peduncles [1, 4].
Clinical manifestations include mild dysmorphism and hand and spine anomalies. Presentation ranges from mild truncal ataxia and normal cognitive abilities to severe cerebral palsy and mental retardation. Disturbance in the development of the vermis between 28 and 41 days is pathogenetic (2).
 Syndromes associated with rhombencephalosynapsis are Gómez-López-Hernández (rhombencephalosynapsis with parietal/temporal alopecia, trigeminal anaesthesia, towering skull, and dysmorphism) and vertebral abnormalities, anal atresia, cardiovascular anomalies, trachea–oesophageal fistula, renal anomalies, and limb defects association. Associated mid-brain abnormalities in rhombencephalosynapsis include aqueductal stenosis and midline fusion of the tectum. Forebrain abnormalities include absent olfactory bulbs, dysgenesis of the corpus callosum, and atypical forms of holoprosencephaly [2, 3].

Brain MRI shows abnormal midline sagittal view of cerebellum, no normal fastigial point, no primary vermian fissure, posterior pointing of 4th ventricle creating keyhole or diamond configuration, no open defect as in Dandy-Walker continuum. Multiple other intracranial findings were described such as 50% hydrocephalus, 71%
dysgenesis/agenesis of corpus callosum, 62% absent cavum septi pellucidi, 24% pontine hypoplasia [4]. Small, single-lobed cerebellum is the hallmark of rhombencephalosynapsis [4].

Detailed evaluation of posterior fossa structure is mandatory in any fetus with severe ventriculomegaly [3]. Careful postnatal evaluation by paediatric neurologist and endocrinologist. Gómez-López-Hernández syndrome can easily be missed. Hydrocephalus may require shunt placement [4].

**Differential Diagnosis List:** Romboencephalosynapsis, Cerebellar hypoplasia, Unilateral cerebellar disruption, Vermian agenesis: Partial or complete

**Final Diagnosis:** Romboencephalosynapsis

**References:**

Description: Brain MRI: There is apparent fusion of the cerebellar hemispheres (rhombencephalosynapsis) with continuous appearance of vermis and both cerebellar hemispheres. There is a small cavum septum pellucidum. Origin: Augusta university, Georgia, USA.
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Figure 2

Description: Brain MRI shows dysplastic and truncated appearance of the posterior body and splenium of the corpus callosum, and apparent absence of the rostrum. Origin: Augusta University
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