Case 7605

Rhombencephalosynapsis
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Section: Neuroradiology
Case Type: Clinical Cases
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Patient: 12 years, female

Clinical History:
A 12 year old girl with congenital aqueductal stenosis presented with nausea, vomiting and headache and neck pain.

Imaging Findings:
MRI revealed tonsillar herniation with hypoplasia of corpus callosum and hydrocephalus. Typical findings of rhombencephalosynapsis were also seen, absent vermis with continuous transverse folia with fusion of dentate nuclei and cerebellar hemispheres. Fourth ventricle was box shaped instead of normal crescent shaped. Primary fissure was absent with roundening of fastigial recess (normally it is sharp).

Discussion:
Rhombencephalosynapsis is a rare anomaly characterized by agenesis or hypoplasia of cerebellar vermis with fusion of cerebellar hemispheres and dentate nuclei. It was first described by Obersteiner in 1914 [1].
In the traditional concept, rhombencephalosynapsis was considered as an abnormal development of the vermis with subsequent fusion of the hemispheres [2]. However, Utsunomiya et al [3] suggested that this anomaly is more likely due to a failure of vermian differentiation with undivided development of hemispheres, thus doubting the traditional concept.
Aqueductal stenosis is commonly associated with this anomaly [4]. Other associated anomalies are corpus callosum dysgenesis, holoprosencephaly, fused thalami and craniosynostosis.
Common clinical findings are ataxia, gait abnormalities, developmental delay, cerebral palsy, psychiatric disorders. Typical MRI findings on axial images are vermian agenesis, fusion of cerebellar hemispheres and dentate nuclei, continuous transverse folia. Diamond shaped 4th ventricle instead of normal crescent shape has also been described on axial images [5]. On sagittal images, roundening of fastigial recess of 4th ventricle and absent primary fissure can be seen [6].
Joubert syndrome (vermian agenesis or hypogenesis) is included in the differential but typically cerebellar hemispheres are not fused.

Differential Diagnosis List: Rhombencephalosynapsis

Final Diagnosis: Rhombencephalosynapsis

References:
Utsunomiya H, Takano K, Ogasawara T, Hashimoto T, Fukushima T, Okazaki M (1998) Rhombencephalosynapsis:
Figure 1

Description: Continuous transverse folia can be seen. Origin:
Description: Vermis is absent with box shaped fourth ventricle. Fusion of dentate nuclei can be seen (black arrows). Cerebellar hemispheres are fused. Origin:
Description: Fastigial recess of fourth ventricle (black arrow) is round in shape. Normally it is sharp. Tonsillar herniation can be seen. Origin:
Description: Corpus callosum is hypoplastic with tonsillar herniation. Origin: