Prenatal diagnosis of a new association of middle interhemispheric variant of holoprosencephaly (HPE) with di-mes-rhombencephalosynapsis (DMRS)

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The middle interhemispheric variant (MIHV) of holoprosencephaly is defined as failed separation of the posterior frontal and parietal lobes in the presence of anterior and occipital hemispheric division. Rhombencephalosynapsis is characterized by fusion of the cerebellar hemispheres and absent vermis. The terms mesencephalosynapsis and diencephalosynapsis describe an incomplete separation of the midbrain and diencephalon. The association of MIHV with DMRS has never been described in utero.

We report this unique association in 2 fetuses at 12.5 and 26.5 weeks.

The sonographic findings included:
• separated frontal lobes with confluence of the anterior horns
• fused parietal lobes over a large monoventricular cavity continuous with a large dorsal cyst
• separated occipital lobes displaced laterally by the cyst
• thalamic fusion obliterating 3rd ventr.
• small midbrain w/o an aqueduct
• small cerebellum with a rounded posterior contour
• in the older fetus: absence of the CSP, partial ACC, abnormal sulcation, thin brainstem, lack of the vermis, enlarged retrocerebellar space

Conclusion: The association of MIHV with DMRS has specific imaging features and probably share a common biological mechanism.