DEVELOPMENTAL DISORDERS

RHOMBENCEPHALOSYNAPSIS SPECTRUM OF SEVERITY

Investigators at University of Washington, Seattle, University of Southern California, and Children's Hospital Los Angeles evaluated neuroimaging findings in 42 patients (17 female, 25 male; age range, 2 days to 44 years) with rhombencephalosynapsis (RES). RES is defined as a partial or complete absence of the cerebellar vermis and midline fusion of the cerebellar hemispheres. A spectrum of RES severity is proposed, ranging from mild (partial absence of nodulus and vermis), to moderate (absence of posterior vermis) to severe (absence of posterior and anterior vermis), to complete (absence of entire vermis including nodulus). Severity of RES correlates with fusion of the tonsils, midbrain abnormalities including aqueductal stenosis and midline fusion of the tectum. RES is also associated with forebrain abnormalities including absent olfactory bulbs, dysgenesis of corpus callosum, absent septum pellucidum and rarely, atypical holoprosencephaly. In other patients with aqueductal stenosis at the U Washington, 9% were identified with RES. Subjects with more severe RES have more severe neurodevelopmental outcome. (Ishak GE, Dempsey JC, Shaw DWW, et al. Rhombencephalosynapsis: a hindbrain malformation associated with incomplete separation of midbrain and forebrain, hydrocephalus and a broad spectrum of severity. **Brain** 2012 May;135:1370-1386). (Respond E-mail: ishakg@u.washington.edu; ddoher@uw.edu).

COMMENT. RES occurs alone or in combination with other congenital malformations, such as Gomez-Lopez-Hernandez syndrome (RES plus parietal scalp alopecia, tower skull, and trigeminal anesthesia) and VACTERL (vertebral anomalies, anal atresia, cardiovascular anomalies, trachesophageal fistula, renal anomalies, and limb defects). Ataxia is the most frequent manifestation of RES, and cognitive outcome may be normal. A dorsal-ventral patterning defect is one hypothesis of the etiology (Sarnat HB. Molecular genetic classification of central nervous system malformations. **J Child Neurol** 2000;15:675-687). The cerebellar fusion is comparable with holoprosencephaly in the forebrain, an associated defect in some cases of RES. The absence of the vermis in RES may be compared also with Joubert syndrome (JS). The vermis in JS is shortened whereas in RES it is narrowed (Barth PG. **Brain** 2012 May;135:1346-1347).

Classification update of CNS malformations. Researchers at the University of California at San Francisco and international centers review and propose a modified classification of malformations of cerebral cortical development, common causes of neurodevelopmental delay and epilepsy. A major change in the group with cortical dysgeneses with abnormal cell proliferation is a new classification of focal cortical dysplasias (FCDs), a common cause of refractory epilepsy often amenable to surgery. (Barkovich AJ, Guerrini R, Kuzniecky RI, Jackson GD, Dobyns WB. A developmental and genetic classification for malformations of cortical development: update 2012. **Brain** 2012 May;135:1348-1369). (Respond: E-mail: james.barkovich@ucsf.edu).