OBJECT:
Rhombencephalosynapsis (RS) is a rare congenital posterior fossa malformation characterized by dorsal fusion of the cerebellar hemispheres, hypogenesis or agenesis of the vermis, and fusion of the dentate nuclei and superior cerebellar peduncles. The objective of this institutional study is to review the clinical conditions associated with RS and analyze the varied biological profile of this unique condition.

METHODS:
The study data were collected retrospectively from the medical records of patients at Rainbow Babies and Children's Hospital. After required institutional review board approval, the authors obtained information regarding the cases of RS reviewed by the Departments of Radiology, Genetics, and/or Pediatric Neurology. Medical charts were systematically reviewed, and 9 patients were analyzed in detail.

RESULTS:
The authors describe 6 cases of RS and 3 cases of partial RS. This case series demonstrates an association between RS and symptomatic hydrocephalus (7 of 9 patients) and RS and Chiari malformation (5 of 9 patients). Patients with symptomatic hydrocephalus underwent endoscopic third ventriculostomy or ventriculoperitoneal shunt insertion. One of the patients with an associated Chiari malformation underwent foramen magnum decompression.

CONCLUSIONS:
The authors present a large case series of RS. Patients with RS often had hydrocephalus and/or a Chiari Type I or II hindbrain malformation. Neuroimaging findings of RS are presented along with hypotheses to explain the embryopathology of this unusual condition.

PMID:
23331215
[PubMed - indexed for MEDLINE]