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Rhombencephalosynapsis: Cerebellar Embryogenesis

Hidetsuna Utsunomiya, Koichi Takano, Tetsuzo Ogasawara, Takeo Hashimoto, Takeo Fukushima, and Masatoshi Okazaki

Summary: We describe two infants in whom rhombencephalosynapsis was diagnosed with MR imaging *in vivo*. In contrast to Dandy-Walker malformation, the vermian maldevelopment in this anomaly is characterized by an absence of the anterior vermis and a deficiency of the posterior vermis. The cerebellar hemispheres are fused. In an attempt to identify the pathogenesis of these anatomic manifestations, we question the traditional concept of the embryologic development of the cerebellar primordium.

Rhombencephalosynapsis is an extremely rare cerebellar malformation that involves vermian agenesis or severe hypogenesis, fusion of the cerebellar hemispheres, and apposition or fusion of the dentate nuclei; it is presumably due to maldevelopment of the rhombic lips in the early fetal period. We found thirty cases of this anomaly reported, either pathologically (1–9) or radiologically, including 11 cases revealed by magnetic resonance (MR) imaging (10–15).

We report two cases of rhombencephalosynapsis detected at MR imaging. We also question the traditional view of the development of the cerebellar primordium to explain the anatomic manifestations of this anomaly.

Case Report

Case 1

A 4-year-old boy was born by a normal delivery after a 35-week gestation. At birth, no dysmorphism was noted. Routine chromosomal examination was normal (46,XY). He had generalized hypotony with a poor sucking reflex but was prone to arch his back when crying. During infancy, he demonstrated delayed motor milestones. A neurologic examination at age 3 years 1 month revealed global developmental delay and hypotonic extremities with mild scoliosis. Cranial MR imaging at this age (Fig 1) showed a flat-based cerebellum without cerebellar vallecule, apposition or fusion of the dentate nuclei, and a fused cerebellar peduncle, which indicated hypogenesis of the cerebellar vermis with fusion of the cerebellar hemispheres. However, the nodulus appeared to be formed because the fastigium of the fourth ventricle was present. Flocculi were also observed. The superior colliculi were close, but separated. The corpus callosum, although thinned and stretched by hydrocephalus, was fully formed. The anterior commissure appeared to be hypoplastic. Forniceal fusion with a deficient septum pellu-

cidum was also evident. The pituitary gland, infundibulum, and optic chiasm were normal.

At 4 years 1 month, the child was unable to sit by himself and his neck was still unstable. Although no nystagmus was evident, he had severe convergent strabismus; speech was still severely delayed.

Case 2

Hydrocephalus was diagnosed in utero in this now 3-year-old boy, who was born by a cesarean delivery after a 34-week gestation. Routine chromosomal examination was normal (46,XY).

A ventriculoperitoneal shunt was placed 9 days after birth. Cranial MR imaging at 2 months showed diffuse thinning of the corpus callosum due to hydrocephalus (Fig 2A). The lower cerebellum was extended downward to the foramen magnum, and protuberance of the nodulus was seen below the fastigium of the narrowed fourth ventricle.

At 2 years 3 months, his extremities were hypotonic and his neck was still unstable. Psychomotor developmental milestones were severely delayed. MR imaging at 3 years 2 months revealed a grossly dysmorphic brain. The ventricles were decompressed and posterior parietal cortical infolding had developed; these findings were believed to be a consequence of the shunt. The corpus callosum, although distorted by hydrocephalus, appeared to be fully formed. The tectum appeared fused and pointed posteriorly. The aqueductal flow void was not apparent on any sequence. Infratentorially, the middle cerebellar peduncle appeared fused around the narrowed fourth ventricle. A flat-based cerebellum without cerebellar vallecule was evident. The cerebellar folia were also oriented transversely (Fig 2B). The superior part of the cerebellum was herniated upward through the tentorial incisura, most likely due to hydrocephalic decompression.

Discussion

Rhombencephalosynapsis is a cerebellar anomaly with vermian hypogenesis or agenesis that is not associated with disconnected cerebellar hemispheres, such as in Dandy-Walker malformation, Joubert syndrome, or tectocerebellar dysraphia. Instead, it shows fusion of the cerebellar hemispheres, the middle cerebellar peduncles, and the dentate nuclei (10–12). The vermian maldevelopment in this anomaly is characterized by an absence of the anterior (rostral) vermis and a deficiency of the posterior (caudal) vermis; the nodulus tends to be formed (1–5, 9, 11, 13). This finding is in contrast to Dandy-

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From the Departments of Diagnostic Radiology (H.U., M.O.) and Neurosurgery (T.F.), Fukuoka (Japan) University Hospital; the Department of Diagnostic Radiology, Fukuoka University Chikushi Hospital (K.T.); and the Departments of Neuroradiology (T.O.) and Neonatology (T.H.), St Mary's Hospital, Kurume, Japan.

Address reprint requests to Hidetsuna Utsunomiya, MD, Fukuoka University Hospital, 7-45-1, Nanakuma, Jonan-ku, Fukuoka, 814-80, Japan.

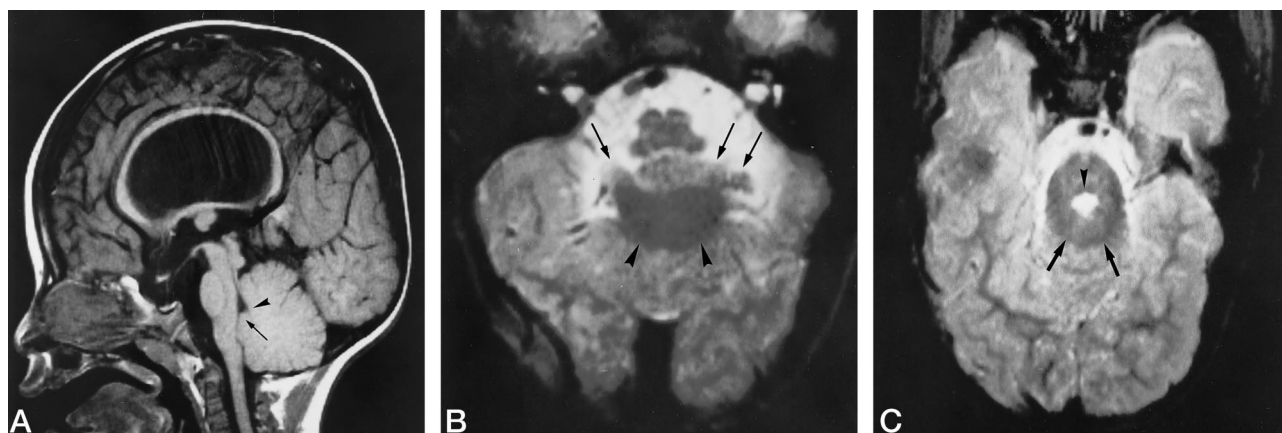


FIG 1. Case 1

A, Midsagittal T1-weighted image (600/15/4 [repetition time/echo time/excitations]) shows caudal displacement of the lower cerebellum with an absent cisterna magna. Note a narrowed fourth ventricle with formation of the fastigium (arrowhead). The protuberance below the fastigium represents the nodulus (arrow). Other vermian components are not seen clearly. The corpus callosum is thinned and stretched.

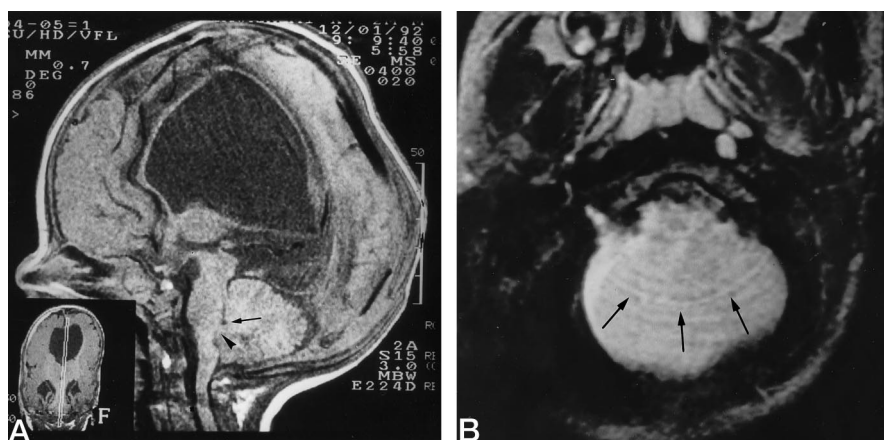
B, Axial T2-weighted image (3000/90/1) through the upper medulla oblongata shows vermian hypogenesis with formation of the flocculonodular lobe (arrows). Note the large corpus medullare (arrowheads).

C, Axial T2-weighted image through the pons shows apposition or fusion of the middle cerebellar peduncles and dentate nuclei (arrows). Note the deformity of the fourth ventricle, which appears to be diamond-shaped (arrowhead).

FIG 2. Case 2

A, Midsagittal T1-weighted image (400/20/2) shows a thinned and stretched corpus callosum due to hydrocephalus. The aqueduct is not patent. Note the narrowed fourth ventricle with formation of the fastigium (arrow) and protuberance of the nodulus (arrowhead).

B, Axial T2-weighted image (2500/80/1) shows a fused cerebellum with transversely oriented folia, which cross the midline (arrows).



Walker malformation, in which the posterior vermis tends to be defective (12, 14).

The first description of rhombencephalosynapsis was published by Obersteiner in 1914 and was based on a postmortem examination of a 28-year-old man who died by suicide (1). Eighteen additional cases, identified pathologically, have been described in the literature up to 1995 (2–9). Since 1991, 11 cases of this unusual anomaly have been revealed by MR imaging during life (10–15).

The MR features of rhombencephalosynapsis are characteristic. A diamond-shaped fourth ventricle, instead of the normal crescent shape, is seen on axial sections (11). This abnormal configuration of the fourth ventricle indicates vermian hypogenesis or agenesis, since fusion or apposition of the dentate nuclei and middle cerebellar peduncles can be observed behind a pointed fourth ventricle (10). The nodulus may be formed if the fastigium is present in a sagittal section (Figs 1A and 2A). Other distinctive features are a flat and uninterrupted continuity of the base of the cerebellar hemispheres, transversely oriented folia in the inferior cerebellum, and large corpus medullare, all of which represent fused cerebellar hemispheres (10–12). Deficiency or absence of the septum pellucidum (3, 6, 7, 9–11, 14–16), dysgenesis of the corpus callosum and the anterior commissure (2, 3, 7, 9–11, 13), fused fornices (10), and fused thalami (4) may also be present. These supratentorial

anomalies, which often dominate the clinical features, may represent malformations in ventral induction, which occur at the time the cerebellum is developing (10). In the midbrain, variable fusion of the colliculi is usually a complication (2–4, 10, 13). Furthermore, aqueductal stenosis, which may cause congenital hydrocephalus, is commonly associated with this anomaly (4, 6, 8–10). Therefore, rhombencephalosynapsis should be suspected in infants in whom both congenital hydrocephalus and a flat-based cerebellum without formation of the cerebellar vallicula are observed by MR imaging.

In the traditional view of embryonic development, the cerebellum arises from two distinct embryonic primordium known as the rhombic lips. At the beginning of 8 weeks of gestation, proliferating neuroblasts from symmetric alar plates form paired rhombic lips that thicken, project farther into the fourth ventricle, and extend progressively toward the midline. At the end of 12 weeks of gestation, the rhombic lips on the two sides fuse in the midline beginning rostrally, and the anterior vermis is thereby formed before the posterior vermis (11, 16). On the basis of this concept, the Dandy-Walker malformation is believed to result from a broad insult to the alar plate involving the dorsal fourth ventricle and the rhombic lips after the anterior vermis has already formed (12). However, this concept is not useful for describing the genesis of rhombencephalosynapsis, because the anterior vermis is defective and the cerebellar

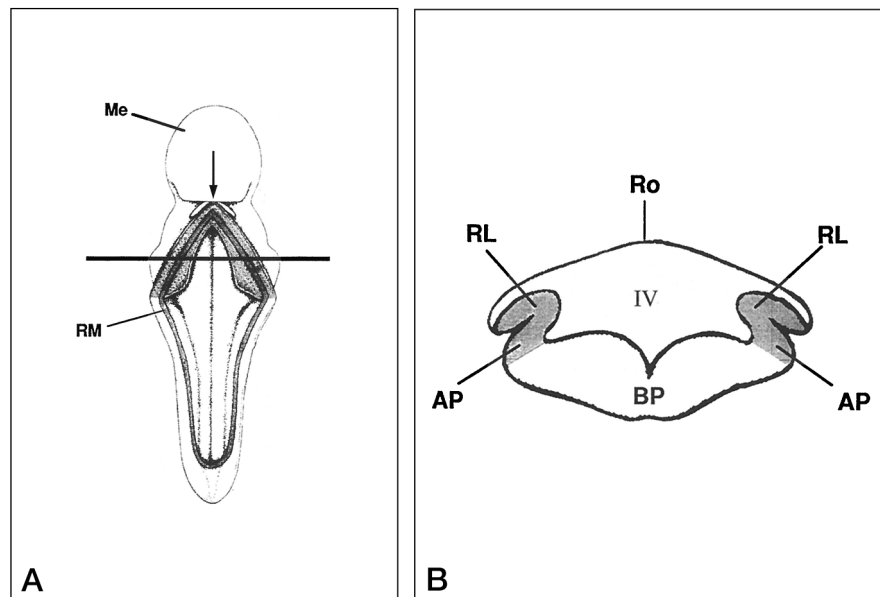


FIG 3. Schematic drawing of the cerebellar primordium at 6 weeks' gestation.

A, Dorsal view: the cerebellar primordium (dark area) essentially continues and straddles the midline in the shape of an inverted V (arrow). RM indicates attachment of the rhombencephalic membrane; Me, mesencephalon.

B, Transaxial section through the line illustrated in A: the cerebellar primordium is observed as a pair of unconnected structures, so-called rhombic lips (RL). This observation may lead to the misconception that the rhombic lips of the two sides fuse in the midline beginning rostrally. Ro indicates roof of the rhombencephalic cavity; IV, fourth ventricle; AP, alar plate; and BP, basal plate.

hemispheres are fused; there is no disconnection of the cerebellar hemispheres. These anatomic characteristics raise the question of whether fusion of the developing cerebellum across the midline has occurred.

Sidman and Rakic (17) reported that the cerebellar primordium arises from the tuberculum cerebelli, which consists of a band of tissue in the dorsal part of the first neuromere and straddles the midline in the shape of an inverted V (Fig 3A). They also emphasized that, from its appearance on a transverse section, the cerebellar primordium might easily be misinterpreted as a pair of unconnected, bilaterally symmetric structures growing in the lateral wall of the fourth ventricle (Fig 3B). Thus, they suggested that the cerebellar primordium is essentially unpaired.

During the third gestational month, the midline component shows accelerated growth and begins to fill the previously acute angle between the limbs of the inverted V. Beginning at about the 12th week of gestation, cerebellar fissures begin to form in the transverse axis of the cerebellum. The first to appear is the posterolateral fissure, which separates the flocculonodular lobe from the rest of the cerebellum; the flocculonodular lobe is believed to be the most primitive phylogenetically (12). Consequently, by 13 weeks, the primary fissure has formed and divides the medial part of the cerebellum into the anterior vermis and posterior vermis. By 16 weeks, the precentral fissure has marked off the central lobule from the lingula. Thus, vermian differentiation occurs first in the most posterior (caudal) part of the primordium and develops toward the anterior (rostral) part.

Considering this notion of embryonic development, rhombencephalosynapsis may represent a case in which vermian differentiation has failed to occur in the more rostral part of the midline cerebellar primordium during the third gestational month, and the fused cerebellar hemispheres may be explained by the fact that the cerebellar primordium is essentially unpaired. Furthermore, this concept indicates that fusion of the hemispheres is not secondary to a primary maldevelopment of the vermis, but rather results from a primary failure of vermian differentiation; in this anomaly, the term undivided cerebellar hemispheres may be more appropriate than fused hemispheres.

Thus, this unusual anomaly throws doubt on the traditional concept of the embryonic development of the cerebellar primordium. If the cerebellum originates from a single anlage, the Dandy-Walker malformation may represent secondary degeneration or regression of the median part of the cerebellar primordium, probably due to hydrocephalus of the fourth ven-

tricle. Further investigation is needed to interpret the genesis of these cerebellar anomalies.

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