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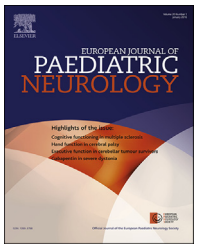

Case Study

"Acquired" Dandy–Walker malformation and cerebellar hemorrhage: Usefulness of serial MRI

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ABSTRACT

Background/purpose: Neuroimaging plays a fundamental role in the diagnosis of Dandy Walker malformation (DWM), a posterior fossa anomaly that is usually associated with genetic abnormalities, but may rarely be ascribed to acquired causes. Here, we report the clinical history and neuroimaging studies of a child with a complex cardiac malformation, developmental delay, and oculomotor anomalies whose neuroimaging findings were consistent with an acquired form of DWM.

Methods/results: Fetal MRI at gestational weeks 27 and 31 showed cerebellar and vermis hypoplasia and fourth ventricle enlargement, together with hemosiderin deposits on the cerebellar hemispheric surface, but without significant vermian rotation. Postnatal MRIs at 5 days and 13 months revealed progressive counter-clockwise rotation of the hypoplastic cerebellar vermis with cystic dilation of the fourth ventricle, eventually leading to a full-blown DWM.

Conclusion: This case strengthens the opinion that DWM is a heterogeneous condition, and may support the hypothesis that acquired meningeal abnormalities in the form of cortico-pial hemosiderosis may play a role in the development of DWM. This case also demonstrates that serial neuroimaging plays a key role in the correct diagnosis of posterior fossa malformations, whose prognosis is difficult to establish on second trimester fetal MRI and requires longer clinical follow-up.

Nonstandard abbreviations: DWM, Dandy–Walker malformation; FFE, fast field echo; MRI, Magnetic Resonance Imaging.

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1. Introduction

The term Dandy–Walker malformation (DWM) refers to a group of posterior fossa malformations characterized by hypoplasia and upward rotation of the cerebellar vermis and cystic dilation of the 4th ventricle, which extends posteriorly to almost completely fill the entire posterior fossa.[1] Elevation of the tentorium, hydrocephalus, and enlargement of the posterior fossa are not consistent, and they are not part of the diagnostic criteria.[1] Affected individuals often display episodic apnea, hypotonia, developmental delay, seizures, and cerebellar signs; about half have mental retardation. It is possible that the malformation remains totally asymptomatic.[2] The etiology of DWM is still poorly understood but is presumed to be multifactorial, with genetic forms accounting for the majority of patients. Here, we report the clinical and imaging follow-up of a pediatric patient with a complex cardiac malformation, developmental delay, and oculomotor anomalies, whose neuroimaging findings were suggestive for an acquired form of DWM with progressive vermian rotation.

2. Case study

The patient is the first female sibling of healthy non-consanguineous parents of Italian origin. Pregnancy was uneventful until a second trimester ultrasound scan revealed a complex heart defect (atrioventricular canal) and cerebellar hypoplasia. At 27 weeks gestation, magnetic resonance Imaging (MRI) confirmed global cerebellar hypoplasia, with regular biometrics of the pons and no signal alteration (Fig. 1A, E). At 31 weeks, follow-up MRI showed reduced cerebellar volume, enlarged fourth ventricle, and a mildly reduced pons volume (Fig. 1B, F). Hemosiderin deposits were suspected on the irregular surface of the cerebellar hemispheres (Fig. 1F), arising the possibility of a clastic etiology. Funicolocentesis and standard kariotype came out normal.

The child was born at term with natural delivery and good pre-perinatal period. Auxological parameters were all under the 3rd percentile, Apgar score was 9\10 (1st minute) and 9\10 (5th minute). Because of the complex cardiac malformation, the baby was treated with ACE inhibitors and underwent heart surgery in the first months of life. Renal, abdominal, and ocular malformations were ruled out by appropriate tests. No facial dysmorphism was noted. In the 5th day of life, a neonatal MRI scan was performed, which confirmed global cerebellar hypoplasia (Fig. 1C) with superficial hemosiderin deposits on fast field-echo (FFE) images (Fig. 1G), mild pons hypoplasia, and further fourth ventricle enlargement. Neither hydrocephalus nor additional brain malformations were reported. The child began a neurological follow-up that documented global psychomotor delay in milestones acquisition.

We first saw the patient at age 12 months. Neurological assessment confirmed global developmental delay with global quotient of 67 on Griffiths Developmental Scale and worse performances on locomotor and oculomanual coordination tasks, brisk patellar reflexes with normal muscle tone, and oculomotor abnormalities including esotropia, alternating fixation (with dominant right eye), discontinuous smooth pursuit, and difficult to evoke saccades. Fundoscopy and

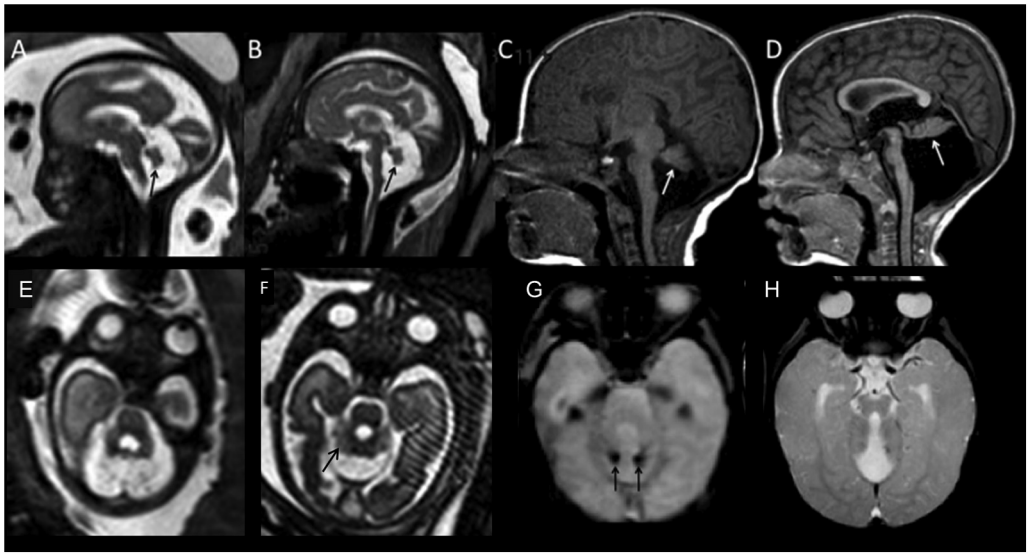

Fig. 1 – A–D sagittal MR images obtained at gestational weeks 21 (A) and 30 (B), and at postnatal 5 days (C) and 12 months (D) show progressive upward displacement of a hypoplastic vermis (arrows) and enlargement of the posterior cranial fossa, eventually resulting in an abnormality that is virtually indistinguishable from a classical Dandy–Walker malformation. Also notice small pontine protuberance. E, F axial T2-weighted images obtained at gestational weeks 21 (E) and 30 (F) show global cerebellar hypoplasia, involving both vermis and cerebellar hemispheres; note the irregular surfaces of the cerebellar hemispheres (arrow). G axial gradient-echo T2*-weighted image obtained at 5 days postnatally (G) shows hemosiderin deposits over the inferior surfaces of the hypoplastic cerebellar hemispheres. H axial gradient-echo T2*-weighted image obtained at age 12 months (H) shows resorption of the hemosiderin deposits.