

# Panventriculomegaly

Panventriculomegaly (PaVM) defined by a wide [foramen of Magendie](#) and large [cisterna magna](#).

Clinical and brain imaging data from 28 PaVM patients (including 10 patients from 5 families) were retrospectively studied. Five children were included. In adult patients, the age at onset was  $56.0 \pm 16.7$  years. Tetraventricular dilation, aqueductal opening with flow void on T2-weighted images, and a wide foramen of Magendie and large cisterna magna (wide cerebrospinal fluid space at the fourth ventricle outlet) were essential MRI findings for PaVM diagnosis. 3D fast asymmetrical spin echo sequences were used for visualization of cistern membranes. Time-spatial labeling inversion pulse examination was performed to analyze cerebrospinal fluid movement. Copy number variations were determined using high-resolution microarray and were validated by quantitative polymerase chain reaction with breakpoint sequencing.

Adult patients showed gait disturbance, urinary dysfunction, and cognitive dysfunction. Five infant patients exhibited macrocranium. Patients were divided into 2 subcategories, those with or without downward bulging third ventricular floors and membranous structures in the prepontine cistern. Patients with bulging floors were successfully treated with endoscopic third ventriculostomy. Genetic analysis revealed a deletion in DNAH14 that encodes a dynein heavy chain protein associated with motile cilia function, and which co-segregated with patients in a family without a downward bulging third ventricular floor.

Panventriculomegaly with a wide foramen of Magendie and a large cisterna magna may belong to a subtype of [congenital hydrocephalus](#) with familial accumulation, younger age at onset, and symptoms of normal pressure hydrocephalus. In addition, a family with PaVM has a gene mutation associated with dysfunction of motile cilia <sup>1)</sup>.

<sup>1)</sup>

Kageyama H, Miyajima M, Ogino I, Nakajima M, Shimoji K, Fukai R, Miyake N, Nishiyama K, Matsumoto N, Arai H. Panventriculomegaly with a wide foramen of Magendie and large cisterna magna. J Neurosurg. 2015 Dec 4;1-9. [Epub ahead of print] PubMed PMID: 26636390.

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