

# Venous malformation

Venous **malformations** (VMs) are a type of type of **vascular malformation** that results from **veins** that have developed abnormally, which stretch or enlarge over time. VMs can be extremely painful and sensitive. A VM usually looks like a bluish discoloration. It can be a single lesion or it may be one of many.

Gain-of-function **PIK3CA** pathogenic variants have been identified in overgrowth syndromes collectively termed “**PIK3CA-related overgrowth spectrum**” (PROS). There are no previously reported cases of cerebrovascular **venous malformations** in PROS syndromes, though somatic activating **PIK3CA** variants have been identified in extracranial venous malformation. This study was approved by the Institutional Review Board at Boston Children's Hospital. A 14-year-old female mosaic for the de novo p.R108H pathogenic variant in the **PIK3CA** gene was found to have a large tumor involving the **superior sagittal sinus** with mass effect on the **motor cortex** most consistent with a **parafalcine meningioma**. She underwent surgical resection with pathology demonstrating a venous malformation. **PIK3CA** pathogenic variants have been identified in nonsyndromic extracranial venous and lymphatic malformations as well in **brain tumors**, including **glioma** and **meningioma**. However, **PIK3CA** variants have not previously been identified in purely intracranial venous malformations. This distinction is relevant to treatment decisions, given that **mTOR inhibitors** may provide an alternative option for noninvasive therapy in cases of suspected **venous malformation** <sup>1)</sup>.

## Orbital Venous Malformation

<sup>1)</sup>

Filippidis A, Lidov H, Al-Ibraheemi A, See AP, Srivastava S, Orbach DB, Fehnel KP. Intracranial venous malformation masquerading as a meningioma in PI3KCA-related overgrowth spectrum disorder. Am J Med Genet A. 2021 Dec 2. doi: 10.1002/ajmg.a.62570. Epub ahead of print. PMID: 34854542.

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