

ABOUT MCAP/M-CM, A PROS CONDITION

Megalencephaly-
Capillary
Malformation
(MCAP or M-CM)

What is MCAP/M-CM?

- ▶ MCAP, sometimes referred to as M-CM, is a PROS condition that is generally characterized by overgrowth throughout the body and brain, developmental delays, and low muscle tone, known as hypotonia
- ▶ Most people with MCAP/M-CM are born with the condition or have features of MCAP/M-CM during prenatal scans. MCAP/M-CM is not hereditary

Different Conditions, One Common Cause – PROS

- ▶ PROS, or PIK3CA-Related Overgrowth Spectrum, is a wide-ranging spectrum of disorders caused by a mutation in the PIK3CA gene
- ▶ PROS conditions are rare and diverse, and are typically characterized by atypical growths and anomalies in the blood vessels and lymphatic system
- ▶ PROS conditions can look different from each other in size, shape, and type of growth or malformation based on where in the body the mutation is found

What are features of MCAP/M-CM?

MCAP/M-CM is different for each person and ranges in severity. Most people living with MCAP/M-CM do not have every sign associated with the condition.

These features may include:

- Brain irregularities due to irregular widening of the sac-like spaces (or ventricles) of the brain, which contain cerebrospinal fluid (CSF)
- Excessive accumulation of CSF may lead to hydrocephalus, one of the potentially serious complications of MCAP or M-CM
- Growth irregularities, such as an atypically large head and asymmetric growth pattern, where one side of the body is clearly larger than the other
- Vascular anomalies, like skin mottling spread over the trunk, limbs, and face, known as capillary malformations
- Body asymmetry, such as webbed toes or fingers

MCAP/M-CM Diagnosis



- ▶ Because MCAP/M-CM has observable features, it often can be diagnosed through a physical exam, imaging, and/or medical history
- ▶ Additionally, there are tissue tests to identify the presence of a PIK3CA mutation
- ▶ However, a negative test result does not rule out having a PROS condition as mutations can be difficult to detect

MCAP/M-CM Management



- ▶ Managing MCAP/M-CM can be challenging and typically requires collaboration from a multidisciplinary team
- ▶ Like other PROS conditions, there are few management options for MCAP/M-CM
- ▶ Some options, like physical therapy and surgery, address symptoms and manifestations rather than the root cause of the disease

Find resources and support for people with MCAP/M-CM and their families by visiting:

▶ www.m-cm.net*

▶ understandingpros.com

* This link takes you to an external website that is independently operated and not managed by Novartis Pharmaceuticals Corporation. Novartis assumes no responsibility for the site.