

# ABOUT MCAP/M-CM, A PROS CONDITION

**M**egalencephaly-  
**C**apillary  
**M**alformation  
(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

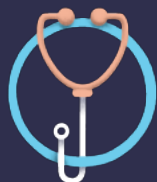
### What are features of MCAP/M-CM?

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](http://www.m-cm.net)\*

▶ [understandingpros.com](http://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.