THINGS TO KNOW ABOUT

Phelan-McDermid syndrome

*pronounced **FAY-luhn mick-DUR-mid***

A rare genetic disorder that changes how a person's brain and body develop — affecting learning and communication as well as moving, eating and sleeping.

Disabilities vary widely from person to person. Most people with Phelan-Mcdermid syndrome require long-term care.

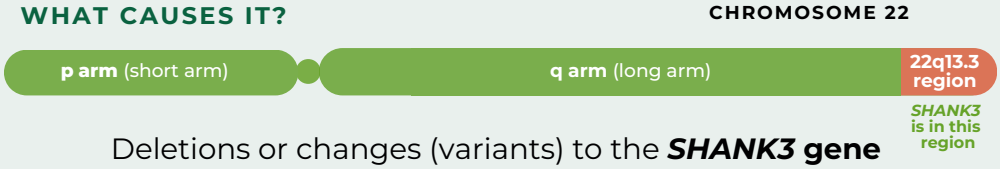
HOW RARE IS IT?

Estimated to affect about

1 in 7,300

That could mean up to
50,000 in the U.S. · 1.1 million worldwide

WHAT CAUSES IT?



Deletions or changes (variants) to the ***SHANK3* gene** or **22q13 region** at the end of chromosome 22.

Common symptoms

- **Moderate to severe intellectual disability**
- **Sleep disturbance**
- **Low muscle tone**
- **Loss of previously acquired skills**
- **Delayed or absent speech**
- **Autism or autism-like symptoms**
- **Seizures & epilepsy**
- **Chronic constipation, feeding & swallowing difficulties**

How is it diagnosed?

Phelan-McDermid syndrome is only diagnosed through genetic testing: whole-exome sequencing (WES), whole-genome sequencing (WGS), or chromosomal microarray. You should consider genetic testing if your loved one has developmental delay or intellectual disability, especially with speech delay, low muscle tone, autism or autism-like symptoms, or loss of skills. Additional chromosome testing may be recommended if a deletion is found.

What treatments are available?

No FDA-approved treatments are available yet, though several clinical trials are currently active. Clinical consensus guidelines help families and clinicians manage symptoms and risks.

CARE GUIDELINES + CLINICAL TRIALS | PMSF.ORG

PMSF is here to help.

The Phelan-McDermid Syndrome Foundation supports the largest global community of people diagnosed with Phelan-McDermid syndrome. Our mission is to ensure that every person with Phelan-McDermid syndrome is supported, valued, and treated with dignity while making today better and the future brighter, from the moment of diagnosis to the delivery of treatments and cures.

- Supporting and connecting families
- Improving medical care
- Driving research breakthroughs

Learn more. Find support. Get involved.

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