

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?

p arm (short arm)

q arm (long arm)

CHROMOSOME 22

22q13.3 region

SHANK3 is in this region

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 (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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