# Things to know about Phelan-McDermid syndrome

pmsf
Phelan-McDermid Syndrome
FOUNDATION

## What is Phelan-McDermid syndrome?

Phelan-McDermid syndrome is a rare genetic disorder involving chromosome 22 that can affect many critical functions in a person's body — from learning and communicating to eating and sleeping.

This results in intellectual and physical disabilities that vary from person to person. Some people with Phelan-McDermid syndrome lose essential skills, and most require long-term care and medical attention.

## Common symptoms:

- Low muscle tone
- Delay in reaching milestones such as sitting, walking, and rolling over
- Intellectual disability
- Delayed or absent speech
- Difficulty making eye contact
- Repetitive/obsessive behaviors and limited interests

ADDITIONAL MEDICAL SYMPTOMS MAY APPEAR LATER. THEY MAY ALSO ONLY APPEAR IN A SUBSET OF PEOPLE. LEARN MORE AT PMSF.ORG.

## How rare is Phelan-McDermid syndrome?

Phelan-McDermid syndrome is estimated to impact 1 in 10,000 births. The PMSF has the largest community of diagnosed individuals, with over 3,600 members worldwide.

The estimated prevalence suggests this number should be much higher, highlighting the challenges of rare disease diagnosis and support!

## What treatments are available?

**Currently, there are no approved treatments for Phelan-McDermid syndrome's root cause.**

**Care focuses on managing symptoms and risks, often with a team of specialists.**

**Therapy is common, and clinical trials for new treatments are increasing.**

Read about recent developments in treatment and research on pmsf.org.

## PMSF is here to help.

The Phelan-McDermid Syndrome Foundation's mission is to make today better and the future brighter for everyone living with Phelan-McDermid syndrome — from the moment of diagnosis to the delivery of treatments and cures.

### HERE'S HOW WE DO IT:

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

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