ES Phelan-McDermid ASOCIACIÓN SÍNDROME

THE SYNDROME

Phelan-McDermid syndrome is a **rare genetic disorder.** It is caused by the loss of genetic material from chromosome 22 and the absence or mutation of the SHANK3 gene, during cell division.

This can cause a wide range of **medical, intellectual, and behavioral challenges** that vary in severity from person to person based on the size and severity of their genetic change. Some people with Phelan-McDermid syndrome **lose essential skills,** and most require **long-term care and medical attention.**

The first warning sign is an unexpected **developmental delay,** either by the presence of low or weak muscle tone (**hypotonia**), failure to acquire **psychomotor milestones** (gait, fine motor skills...), and **absent or delayed speech**.

In the absence of a physical phenotype to facilitate diagnosis, **genetic testing is necessary.** This mutation usually appears spontaneously and is **not inherited.**

DIAGNOSIS

When the results point to a possible genetic origin, the diagnosis is usually confirmed by the **following tests:**

- **Chromosomal Microarray Analysis** (CMA)
- **Conventional chromosome analysis** (karyotyping)
- **Fluorescence in situ hybridization** (FISH), which can detect large deletions
- **Whole exome sequencing** (WES) and **whole genome sequencing** (WGS)

LIVING WITH 22Q13

After receiving the diagnosis, daily life with the syndrome in the family environment is a l**ong-distance race.**

To help families in this process, the **Phelan-McDermid Syndrome Association** was created in Spain in 2013.

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THE ASSOCIATION

The **Phelan-McDermid Syndrome Association,** based in Madrid, Spain, is composed of parents and relatives of the persons affected by this rare disease. It was created in 2013 and was classified Entity of Public Interest in 2016.

OUR MISSION

We aim at **improving the quality of life** of people affected by the syndrome by accelerating research, supporting families and raising awareness. Through our organization in Spain, we offer **reliable and up-to-date information** about the syndrome, a **community of support for families** and we seek **funding to support research** about the syndrome.

OUR WORK

We organise periodically **European Conferences on Phelan-McDermid Syndrome** and we inform about scientific and medical progress of the research that we finance.

Our **website** and the **PhelanApp** offer useful resources and information for families and affected people.

We finance medical studies that help us gather more data about the syndrome and its symptomatology.

We offer an **Information and Orientation Service** as well as a **Psychological Support Service** to attend to families and affected people.

We organize solidarity races **RUN LIKE A HERO** to collect funds and raise awareness about the syndrome.

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E-mail: info@22q13.org.es
Web: www.22q13.org.es
Phone: +34 686 222 978

BECOME A MEMBER AND COLABORATE

Scan this QR Code and discover how to collaborate with the Association!

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