

SYMPTOMS

Symptoms can vary in how often they appear and how strong they are. Each child may show different symptoms and not all children have the same signs.

Developmental delay: The child develops more slowly than others.

Intellectual disability / cognitive impairment: The child has difficulties with learning and thinking.

Low muscle tone (hypotonia): The muscles are weak or speaks less.

Delayed speech: The child learns to speak later or speaks less.

Epilepsy: The child may have seizures.

Behavioural difficulties: The child shows unusual or challenging behaviour.

Facial dysmorphisms: The face may look different, for example in the shape of the eyes, nose or head.

CONTACT

The **DMAP1 Syndrome** is committed to raising awareness, supporting research and helping families.

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Support Research and families:

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DMAP1 Syndrome

AWARENESS

NOT

IGNORANCE



WAT IS DMAP1 Syndrome?

The **DMAP1 syndrome** is a very rare genetic condition caused by changes (mutations) in the **DMAP 1 gene**. This gene plays an important role in regulating gene activity and repairing DNA.

If the **DMAP 1 gene** does not work properly, important processes inside the cells are disturbed. This especially affects brain development, growth and muscle tone.

The symptoms can vary greatly - even with the same genetic cause



PREVALENCE

DMAP1 syndrome belongs to the group of ultra-rare diseases - only a few cases have been documented worldwide. However, experts believe that many cases may go undetected, because the symptoms are nonspecific and only be identified through specialized genetic testing.

Despite the challenges, many children with **DMAP1 syndrome** make significant progress when they receive early support and loving care.

RESEARCH

Researchers around the world are trying to figure out exactly how the **DMAP1 gene** works. They are also looking for potential treatments. Self-help groups are very important as they allow affected individuals and families to share their knowledge and support each other.

TREATMENT

Despite the challenges, many Currently, there is no specific treatment for the genetic defect in the **DMAP1 gene**. Therapy focuses on alleviating symptoms and supporting affected individuals through:

- **Early intervention and special education** support to compensate for developmental delays.
- **Physiotherapy** to improve muscle tone and motor skills.
- **Occupational therapy** to support daily living skills.
- **Speech therapy** for difficulties with language and communication.

