

FACT SHEET: VAN DER KNAPP SYNDROME

What Causes van der Knapp Syndrome?

Van der Knapp Syndrome was identified in 1995, and scientists continue to study this disease. Mutations in a gene called MLC1 seem to be the cause of at least some of the cases of this disease, although some reports have suggested that there some cases may have a different cause. The function of MLC1 in the body is not yet understood.

What are the symptoms of van der Knapp Syndrome?

The symptoms of van der Knapp Syndrome vary, as does age of the first presentation of symptoms of the disease, which can be anywhere between birth and 10 years of age. Symptoms may include the following:

- Seizures
- Ataxia: loss the ability to coordinate muscular movement
- Macrocephaly: an abnormally large head
- Spasticity: muscle spasms
- Difficulties with learning
- Mental decline

How is van der Knapp Syndrome diagnosed?

Van der Knapp Syndrome is currently diagnosed by a combination of clinical signs and symptoms and the results of an MRI, which shows a very distinct pattern. Please see our MRI fact sheet for more information on the MRI. The MLC1 gene can be sequenced, but since there are some cases of van der Knapp Syndrome that appear to have normal MLC1 genes, it is not definitive.

How is van der Knapp Syndrome treated?

There is currently no cure for this disease. Treatment for van der Knapp Syndrome is symptomatic and supportive.

How is research on van der Knapp Syndrome progressing towards better treatments?

Research continues on van der Knapp Syndrome, and many scientists are studying MLC1, the gene responsible for at least some cases of the disease. A better understanding of the role of this gene in the body can help scientists to identify potential new treatments for the disease.

Are there other names for van der Knapp Syndrome?

Other names for van der Knapp Syndrome include:

- Vacuolating Megalencephalic Leukoencephalopathy with Subcortical Cysts
- LVM
- VL
- Leukoencephalopathy with swelling and
- Megalencephalic Leukoencephalopathy with subcortical Cysts (MLC)

