

FACT SHEET: MEGALENCEPHALIC LEUKOENCEPHALOPATHY WITH SUBCORTICAL CYSTS (MLC)

What Causes MLC?

Megalencephalic Leukodystrophy with subcortical cysts (MLC) 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 some cases may have a different cause. The function of MLC1 in the body is not yet understood.

What are the symptoms of MLC?

The symptoms of Megalencephalic Leukodystrophy with subcortical cysts (MLC) 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 of the ability to coordinate muscular movement
- Macrocephaly: an abnormally large head
- Spasticity: muscle spasms
- Difficulties with learning
- Mental decline

How is Megalencephalic Leukodystrophy with subcortical cysts (MLC) diagnosed?

Megalencephalic Leukodystrophy with subcortical cysts (MLC) 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 Megalencephalic Leukodystrophy with subcortical cysts (MLC) that appear to have normal MLC1 genes, it is not definitive.

How is Megalencephalic Leukodystrophy with subcortical cysts (MLC) treated?

There is currently no cure for this disease. Treatment for Megalencephalic Leukodystrophy with subcortical cysts (MLC) is symptomatic and supportive.

How is research on Megalencephalic Leukodystrophy with subcortical cysts (MLC) progressing towards better treatments?

Research continues on Megalencephalic Leukodystrophy with subcortical cysts (MLC), 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 Megalencephalic Leukodystrophy with subcortical cysts (MLC) ?

Other names for Megalencephalic Leukodystrophy with subcortical cysts (MLC) include:

- van der Knaap Syndrome
- Vacuolating Megalencephalic Leukoencephalopathy with subcortical cysts
- LVM
- VL
- Leukoencephalopathy with swelling and
- Megalencephalic Leukoencephalopathy with subcortical cysts (MLC)