



What is LCC ?

LCC is a type of leukodystrophy.

Leukodystrophies are a group of genetic conditions that mainly affect the white matter of the brain and spinal cord. The white matter is the wiring network of the brain that connects different brain regions and links the brain to the spinal cord and rest of the body.

LCC is characterized by abnormal white matter on brain imaging (MRI) with cerebral calcification and cysts. LCC causes progressive damage or inflammation of the white matter, at multiple sites in the brain.

LCC can present at any age from early childhood to late adulthood.

What are the symptoms of LCC?

Symptoms vary and depend on the location of the cysts which can cause specific impairments related to the part of the brain that is affected.

Symptoms include:

- Developmental delay and cognitive decline.
- Progressive muscle stiffness (spasticity).
- Seizures.
- Weakness of one side of the body (hemiplegia).
- Involuntary muscle contraction (dystonia).
- Involuntary quivering of muscles (tremors).
- Uncoordinated muscular movements (ataxia).
- Gait abnormalities.
- Speech impairment (dysarthria).

What causes LCC?

LCC is caused by changes in the *SNORD118* gene. This gene is necessary for the processing of ribosomal RNA and splicing of mRNA. Changes in this gene impact the process of DNA replication and synthesis.

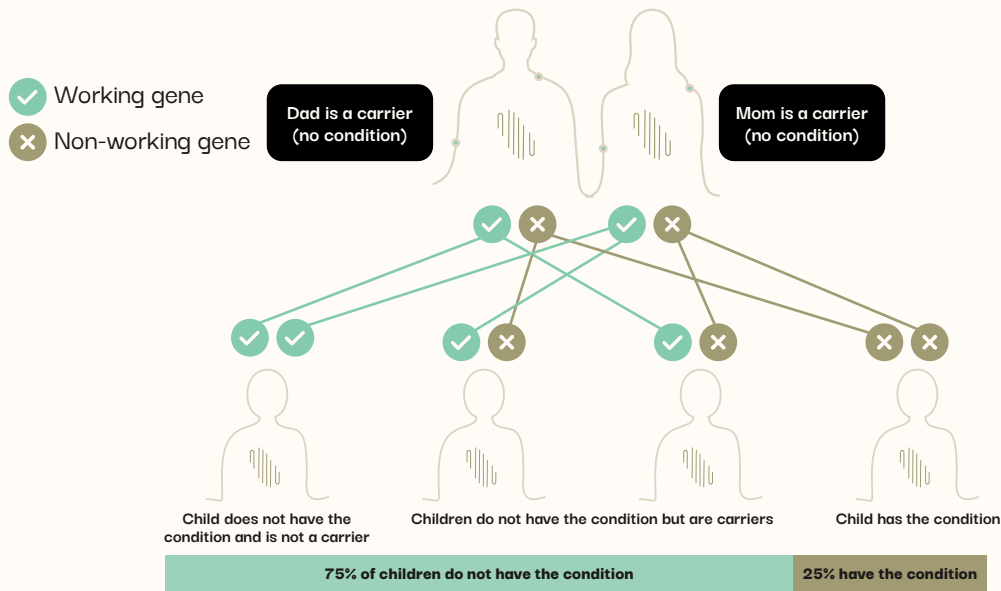
The exact nature of its effects on white matter is unknown.

How is LCC diagnosed?

- MRI brain suggestive of leukodystrophy, cysts, and intracranial calcifications.
- Neurological impairments specific to the location of the cysts.
- Molecular testing of the *SNORD118* gene.

How is LCC inherited?

LCC is inherited in an autosomal recessive pattern. This is where two copies of the altered gene causes LCC. Each parent passes on one copy of the altered gene change. This means that in each of the subsequent pregnancies the couple has a 1 in 4 chance of having an affected child. This also means that siblings of the parents can also be carriers.



Can LCC be treated?

There are currently no specific treatments available. Symptomatic management of epilepsy, brain swelling (oedema) and raised intracranial pressure is often required. Surgical treatment is sometimes needed to treat raised intracranial pressure.

Support and resources:

- **Leukodystrophy Australia** leuko.org.au
- **Mission Massimo Foundation** missionmassimo.com
- **United Leukodystrophy Foundation** ulf.org/leukodystrophies/adrenoleukodystrophy
- **Hunter's Hope** huntershope.org/familycare/leukodystrophies/adrenoleukodystrophy

Research:

- **Australian Leukodystrophy Clinical and Research Program** leukonet.org.au
- **Clinical trials** clinicaltrials.gov
- **Global Leukodystrophy Initiative** theglia.org

References:

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