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## Leukodystrophy

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### Summary

- Leukodystrophy refers to a group of inherited disorders that affect the white matter of the brain. It causes loss of normal brain functions.
  - Onset of symptoms is variable.
  - There is no cure, but supportive treatment can help manage some of the symptoms.
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Leukodystrophy is not a single condition, but refers to a group of conditions that mainly affect the white ('leuko') matter of the brain and the spinal cord. The primary leukodystrophies affect myelin production or breakdown. Myelin is the substance that surrounds and insulates the nerve fibres in the nervous system. When myelin is damaged, the underlying nerve tissue cannot operate in the regular way.

Leukodystrophies are usually progressive conditions, which means they get worse over time. They cause loss of normal brain functions. Life expectancy depends on the type of leukodystrophy.

### Symptoms of leukodystrophy

The symptoms can vary, depending on how quickly damage to the white matter occurs, which parts of the brain (or spinal cord) are affected and sometimes, how much the peripheral nerves are affected. The white matter has a large role in motor function (movement), so damage usually leads to problems in this area.

The onset of symptoms varies. They can include subtle or gradual changes to:

- muscle tone
- body movements
- walking style
- speech
- ability to chew and swallow food
- eye sight
- hearing
- mental ability
- behaviour.

### Types of leukodystrophy

Some of the many different types of leukodystrophy include:

- Adrenoleukodystrophy (ALD) – this is one of the more common of the leukodystrophies. Typically, it only affects boys. It is due to an X-linked gene. Symptoms of the childhood cerebral form usually begin before 10 years of age and may progress rapidly. The adrenal glands may also be affected, which means not enough cortisone is produced. The same genetic change can lead to adrenomyeloneuropathy in adults. This form affects the spinal cord and the adrenal glands. Female carriers of the gene may show similar, but usually much milder, symptoms. In some with the same genetic change, the adrenal gland may be affected in isolation.
- Metachromatic leukodystrophy (MLD) – this is another common leukodystrophy. It has a range of subtypes, some of which relate to the age when symptoms appear – for example, late infantile MLD, juvenile MLD and

adult MLD. In the infantile form, a loss of motor (movement) and verbal skill may be the first signs. Symptoms typically become progressively worse.

- Canavan's disease (spongy degeneration) – this form causes abnormal myelin growth. The typical form is characterised by poor head control, decreased tone, regression and a markedly enlarged head. Eventually, the decreased tone changes to stiffness. Life expectancy is limited.
- Krabbe disease (globoid leukodystrophy) – this affects the myelin of the central and peripheral nervous systems. About 90 per cent of those affected are babies, and symptoms usually show up before they are six months old. There is also a rare adult form.

### **Causes of leukodystrophy**

Genes come in paired sets, with one set inherited from each parent. The leukodystrophies are almost all inherited in either an X-linked or autosomal recessive way.

- Autosomal recessive – the child can only inherit the disorder if both parents are carriers of the gene. Boys and girls are equally affected. The carrier parents won't have any symptoms, but each of their children will have a one in four chance of developing leukodystrophy and a one in two chance of being a carrier themselves.
- X-linked – the gene is carried on the X chromosome, which is involved in determining the baby's sex. Men have an X and a Y chromosome and women have two X chromosomes. Because men only have a single copy of the genetic programs on the X chromosome, if there is a genetic change, there are no other genes to compensate. Boys and girls are therefore affected differently.
- Leukodystrophy is different to multiple sclerosis – the white matter can be affected as a result of (secondary to) a number of conditions, including ischaemia, infection, other metabolic conditions and multiple sclerosis.

Multiple sclerosis (MS) is a disease that inflames the myelin sheaths and causes plaques or lesions to appear. MS can progress in different ways: some people may become seriously disabled, while others may have a few episodes and then be symptom free.

### **Diagnosis of leukodystrophy**

The diagnosis of a specific leukodystrophy is often not easy. Frequently, it will involve the input of a number of specialists, including neurologists, geneticists and metabolic physicians. In around half the cases, a named diagnosis may not be possible.

Depending on the type of leukodystrophy, diagnostic methods can include:

- physical examination
- blood tests
- urine tests
- MRI scans
- nerve conduction tests
- neurocognitive tests
- nerve biopsy
- genetic tests.

### **Treatment for leukodystrophy**

Currently, leukodystrophies cannot be cured. Stem cell therapy and bone marrow transplantation have each been tried in some cases. However, the benefits depend on the timing, age of onset and severity of symptoms. Therapies such as gene therapy and enzyme replacement therapy are under investigation. Treatment is mainly supportive and may include:

- physical therapy
- occupational therapy

- psychological counselling
- family counselling (including genetic counselling)
- medications (for example, medications for seizures).

### **Where to get help**

- Your doctor
- Australian Leukodystrophy Support Group Inc. Tel. (03) 9845 2831

### **Things to remember**

- Leukodystrophy refers to a group of inherited disorders that affect the white matter of the brain. It causes loss of normal brain functions.
- Onset of symptoms is variable.
- There is no cure, but supportive treatment can help manage some of the symptoms.

### **This page has been produced in consultation with and approved by:**

Victorian Clinical Genetics Services (VCGS)

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