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Leukodystrophy (Adult Onset)

An information sheet for the person who has been diagnosed with a leukodystrophy, their family, and friends.

'Leukodystrophy' and the related term 'leukoencephalopathy' refer to a group of conditions that affect the myelin, or white matter, of the brain and spinal cord.

Leukodystrophies are neurological, degenerative disorders, and most are genetic. This means that a person's condition is caused by a change to one of the genes that are involved in the development of myelin, leading to deterioration in many of the body's neurological functions. The pattern of symptoms varies from one type of leukodystrophy to another, and there may even be some variation between different people with the same condition, however all are described as progressive. This means that although there may be periods of stability, the condition doesn't go into 'remission' as may be seen in some other neurological conditions, and over time the condition worsens.

While most leukodystrophies occur in childhood, some appear for the first time in adolescence or adulthood.

How is it diagnosed?

Some adult-onset leukodystrophies first show up as changes in memory or thought processes, personality changes, or physical symptoms such as loss of balance or speech problems. As a result of these symptoms, people may experience breakdown in relationships, loss of employment, and/or they may seek to manage their symptoms through using alcohol or other substances. Although looking back it may be recognized that these are
connected to the underlying condition, at the time it may be difficult for the person affected, their family and friends, and even their doctor, to understand what is going on. For some people, the first contact with a doctor happens following an accidental head injury or seizure which appears to be 'out of the blue', but which perhaps followed other changes that had been less noticeable.

For others, early symptoms affect mainly the lower body – weakness, stiffness, cramps, unusual sensation in the legs, possibly also with urinary symptoms and related problems. The person may notice they trip more easily, particularly on uneven ground or steps.

Other symptoms that people with adult-onset leukodystrophy may experience include sensitivity to extremes of temperature, such as difficulty tolerating hot summer weather; pain or abnormal sensation, particularly in the legs; shaking or tremors; loss of vision and/or hearing; headaches, and difficulty with coordination.

People with leukodystrophy often experience long delays before receiving a correct diagnosis. This is partly because the symptoms can be quite vague and associated with many different disorders. Leukodystrophies are rare, and it is routine medical practice to rule out more common and treatable causes before testing for rarer conditions. There are many different tests that can be undertaken, but MRI (Magnetic Resonance Imaging), blood tests, urine tests and specific genetic tests are most commonly ordered.

As mentioned earlier, the term 'leukodystrophy' covers a range of conditions; some of these may be diagnosed on a single blood test, while others may remain 'unclassified' (ie no specific cause found) even after many tests over a long period of time. A family history is an important part of the medical assessment to search for clues about possible inheritance of the condition.

Is there a cure?

Developments in genetic and other therapies provide hope for the future, but currently there is no cure. However, it is important to remember that there are things that can be done to manage the condition, and the fact that there is no cure does not mean there is no hope.

For some people whose condition is diagnosed prior to the appearance of symptoms (such as when screening occurs following the diagnosis of leukodystrophy in another family member) bone marrow transplant may be an option. This procedure is also extremely risky, however, and is

not suitable for all individuals or conditions. Treatment for leukodystrophy is generally aimed at providing comfort and management of symptoms. The life expectancy of adult-onset leukodystrophies varies enormously – while for some people the condition progresses over months or a few years, others have many years of productive life ahead, with varying levels of impact from symptoms.

Can it be passed on?

Leukodystrophies are not infectious. Some leukodystrophies are inherited from the parent/s, even though there may have been no sign of the condition for several generations, while others occur for the first time in the affected person.

Genetic conditions can be passed on to future generations. For this reason, once a diagnosis of a genetic condition has been made, doctors and genetic counsellors will discuss the issues that may face other family members, including those who have no symptoms. This can be a complex and sensitive area – different forms of leukodystrophy have different 'patterns of inheritance' and the information you are given needs to be relevant to your individual and family situation. While some family members may be reassured that they are unaffected by the genetic condition, others may be faced with important decisions about their own health or future family planning. For this reason, contact with a genetic health service is appropriate.

What can family and friends do to help?

A diagnosis of leukodystrophy is naturally distressing and frightening. Dreams and hopes for the future are suddenly challenged, and a whole new world of therapies and services opens. The most common questions often have no answers, and many people say that living with this uncertainty is one of the hardest things to deal with.

However, it is important to remember that people affected by leukodystrophy, and their families, have the same needs as anyone else for friendship, fun and a 'normal life'. As a friend or family member, you can help the person by learning all you can about their condition. Don't be afraid to ask them if they are willing to talk to you about it. Showing an interest and a willingness to continue your friendship, even if some of the things you do together change, can make a huge difference. All calls are confidential.

What about support for family and friends?

As with any genetic condition, a diagnosis of leukodystrophy affects the extended family and friends as well as the individual. This is partly because people affected by leukodystrophy have complex support needs, and their family, friends and other carers also need support to be there for the affected person while meeting their own needs for health and wellbeing. In addition, family and friends may have their own feelings of fear and grief. It's important that family and friends also have someone to talk to about how they are feeling, and 'time out' to recharge their own batteries.

How can Leukodystrophy Australia help?

Leukodystrophy Australia employs two Family Advocates (one full-time and one part-time) to assist people affected by leukodystrophy and their immediate supports. More information about leukodystrophy, leukoencephalopathy and support services can be obtained from the Family Advocates. Discussions with the Family Advocate are confidential unless otherwise agreed. Please feel free to contact our Family Advocates on:
0418 790 059, 0418 755 994 or 1800141400
or through email: advocate@leuko.org.au

LEUKODYSTROPHY .org.au AUSTRALIA

PO Box 248, Wahroonga NSW 2076
Ph: 1800 141 400 **M:** 0418 755 994
info@leuko.org.au | www.leuko.org.au

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