

Key Facts about Lowe Syndrome

Lowe syndrome is a rare genetic syndrome that causes physical and intellectual/learning disabilities.

It is estimated that Lowe syndrome occurs in 1 in 500,000 people.

Lowe Syndrome is nearly exclusively present in males however exceptionally rare cases have been documented in females.

Diagnosis is made based on [physical characteristics](#) as well as [genetic testing](#).

Lowe syndrome is characterised by problems with the eyes, central nervous system and kidneys. All individuals with Lowe syndrome have impaired vision.

The severity of Lowe syndrome varies. Some boys with Lowe syndrome are mildly affected and able to attend mainstream schools with special needs help, while others are severely affected with loss of sight and mobility.

Almost all affected males have some degree of intellectual disability; 10%-25% function in the low-normal or borderline range, approximately 25% in the mild-to-moderate range, and 50%-65% in the severe-to-profound range of [intellectual disability](#).

Delayed language development is evident in early childhood. Most individuals learn to communicate verbally to some extent by age seven years; some eventually become quite talkative.

The most common [vision problem](#) is clouding of the lenses of the eyes (cataracts), which is present from birth.

Kidney (renal) abnormalities, most commonly a condition known as renal Fanconi syndrome, typically develop in individuals with Lowe syndrome and will eventually cause renal failure.

Some people with Lowe syndrome may display challenging behaviours and repetitive behaviours. It is important to consider the cause and function of these behaviours, and ask for support from a psychological service when considering reducing these.

Temper outbursts are common in Lowe syndrome and research is currently underway to understand why

these outbursts occur.

A number of support groups are available for individuals with Lowe syndrome and their families.

The research recently conducted with the help and funding of the Lowe Syndrome Trust.
