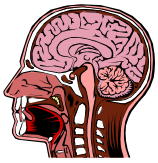
Disability Awareness Begins With You



Lowe Syndrome

Q: What is Lowe syndrome?
A: Lowe syndrome (LS) is a rare genetic condition affecting males that causes physical and mental handicaps and medical problems. Also called the oculo-cerebro-renal syndrome of Lowe (OCRL), it was first described in 1952 by Dr. Charles Lowe and colleagues.

What causes Lowe syndrome?
Lowe syndrome is caused by a defective gene that results in the deficiency of an enzyme called phosphatidylinositol 4,5-biphosphate 5 phosphatase. This enzyme is essential to normal metabolic processes that take place in a small part of the cell called the Golgi apparatus. Because of the enzyme deficiency, cell functions that are regulated by the Golgi are abnormal, leading to various developmental defects including cataracts and problems in the brain and kidneys. How the enzyme deficiency leads to these defects is not yet completely understood.

Why can't the missing enzyme just be replaced?
Scientists must first better understand the subtle imbalance caused by the biochemical defect. It is possible that overcorrection could be just as harmful as the original lack of the enzyme. In addition, there is currently no method available to target therapies to the Golgi apparatus, the small subcompartment of the cell where the LS enzyme is located.

How is LS inherited?
The LS gene is located on the X chromosome. Only males can actually have the condition. Females who have the LS gene are carriers. In some cases, LS is the result of an original mutation and the mother is not a carrier.

Where are diagnostic tests done?
To diagnose LS, a small skin sample is taken and sent to the Biochemical Genetics Laboratory at Baylor College of Medicine in Houston, Texas. Prenatal diagnosis is also provided at this lab. Physicians may make arrangements for these tests by calling
1-800-246-2436 or 713-798-4982 or through e-mail at: bioc@bcm.tmc.edu.

What are the common features of LS?

- Cataracts in both eyes, found at birth or shortly after
- Glaucoma (in about half the cases)
- Poor muscle tone and delayed motor development
- Mental retardation, ranging from borderline to severe (in a few cases intelligence may be normal)
- Seizures (in about half the cases)
- Significant behavior problems (in many, but not all, cases)
- Kidney involvement ("leaky" kidneys, or renal tubular acidosis)
- Short stature
- Tendency to develop rickets, bone fractures, scoliosis, and joint problems
- Maximum life span of about 35-40 years due to progressive kidney failure, although deaths have occurred at earlier ages due both to renal failure and to other causes. Life expectancy may increase as knowledge increases and new treatments are developed.

How is LS treated?
There is no cure, but many of the symptoms can be treated effectively through medication, surgery, physical and occupational therapies, and special education.

What about research?
In 1992 the gene that causes LS was found. In 1995 researchers discovered that the gene defect causes an enzyme deficiency. Researchers continue to investigate the function of the gene and the complicated biochemistry and cellular mechanisms of LS. Other areas of research in recent years include behavior problems and clinical care.

How is development and education?
Boys with Lowe syndrome are not all alike. The presence and severity of symptoms varies from child to child, even in the same family. As a result, the age of achieving major developmental milestones varies. However, some general statements can be made about the development and educational needs of boys with Lowe syndrome based on the observations of families in the Lowe Syndrome Association (LSA) as well as on recent research findings. Because of their physical and mental handicaps, boys with Lowe syndrome are usually significantly delayed in most areas of development. While development generally follows the normal sequence, the level of development in one area (language, for instance) may be more advanced than in another (gross motor skills, for instance). Various therapies and educational programs can help affected boys develop to their maximum potential.

In general, children with Lowe syndrome, even those with behavior problems, are known to have "bubbling" personalities. They are loving and sociable and have a wonderful sense of humor. Many have a special feel for music. When healthy and in a stimulating environment, they are often happy and alert. With help, they can grow and develop into functioning members of their families and communities and can be enjoyed and appreciated for their unique personalities and accomplishments.

What about adulthood?
Throughout life, most boys with Lowe syndrome live at home with their families. A few families, however, have decided that placement in a residential setting is appropriate due to the complexities of medical management, educational needs, or severe behavior problems. As adults, some individuals have successfully made the move into a group home while a few live independently. Some work in sheltered workshops or participate in other special programs for adults with disabilities.

*Based on information from: **L**iving with Lowe Syndrome: A Guide for Parents, Friends & Professionals (©2000)*