

QUESTIONS & ANSWERS

English Version

WHAT IS LOWE SYNDROME?

Lowe Syndrome (LS) is a rare genetic condition that causes physical and mental handicaps and medical problems. Also called the oculo-cerebro-renal (OCRL) syndrome, it was first described in 1951 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-bisphosphate. This enzyme is essential to normal metabolic processes that take place in a certain 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 in LS including cataracts and kidney and brain problems. How the enzyme deficiency leads to these defects is not yet completely understood.

WHAT ARE THE COMMON FEATURES OF LOWE SYNDROME?

- Cataracts in both eyes, found at birth or shortly after
- Glaucoma (in about half the cases)
- Poor muscle tone and delayed motor development
- Cognitive delays, ranging from borderline to severe
- Seizures (in about half the cases)
- Severe behavior problems (in some cases)
- Kidney involvement ("Leaky" kidneys, or renal tubular acidosis)
- Short stature
- Tendency to develop rickets, bone fractures, scoliosis and joint problems

WHAT ARE BOYS WITH LS LIKE?

Generally, they are affectionate and sociable, love music, and have a great sense of humor.

HOW IS LS TREATED?

There is no cure, but many of the symptoms can be effectively treated 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 are continuing to investigate the function of the gene and the complicated biochemistry and cellular mechanisms of LS. Other areas that researchers have investigated in recent years include behavior problems and clinical care.

CAN LS BE PREVENTED?

In families in which a case of LS has occurred, testing to determine carrier status of at-risk females can be done. Various family planning options are available, including prenatal testing. Families should consult with a geneticist to learn more about their options.

WHERE CAN I GO FOR MORE INFORMATION?

Contact the Lowe Syndrome Association (LSA), an international non-profit organization made up of parents, friends, relatives, and professionals. The LSA provides information, fosters communication among families, and supports research. It publishes a newsletter as well as this web site, and sponsors conferences.

Membership Information: lowesyndrome.org/membership