Lowe syndrome

Lowe syndrome (LS) or oculo-cerebro-renal (OCRL) syndrome was first described in 1951 by Dr. Charles Lowe and colleagues. LS is a congenital, rare, genetic condition that primarily affects the eyes, brain, and kidneys.

Diagnosis is confirmed through genetic testing. The LS gene is located on the X-chromosome-essentially affecting males only. Females who have the LS gene are carriers for the genetic defect. However in some cases, LS is the result of an original mutation and the mother is not a carrier.

Lowe syndrome is caused by a defective gene that results in a deficiency of an enzyme called phosphatidylinositol 4,5-biphosphate. 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 including cataracts, kidney and brain problems. How the enzyme deficiency leads to these defects is not yet completely understood.

Common features

- Cataracts in both eyes, typically found at birth
- Glaucoma (in about half of the cases)
- Poor muscle tone and delayed motor development

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LOWE SYNDROME and the Lowe Syndrome Association

- Cognitive impairment – ranging from borderline to severe
- Seizures (in about half of the cases)
- Behavioral problems – ranging from mild to significant
- Kidney impairment (“leaky” kidneys or renal tubular acidosis)
- Short stature
- Various dental challenges- including cysts
- Tendency to develop rickets, bone fractures, scoliosis and joint problems
- Optimum life span of about 35-40 years primarily due to progressive kidney failure though deaths occur at both earlier and later ages.

Lowe syndrome individuals

While every child has their unique challenges and personality, generally speaking individuals with LS are affectionate, loving and sociable. They often love music and have a great sense of humor.

Treatments

Treatment involves removal of cataracts fitting with glasses; surgical and/or medical management of glaucoma; early infant therapy, preschool intervention and individualized education programs; behavior modification plans; anticonvulsant therapy. Treatment of renal tubular dysfunction includes oral supplements of sodium and potassium bicarbonate or citrate to correct acidosis and hypokalemia, and oral phosphate and oral calcitriol to correct hypophosphatemia and renal rickets; treatment of ESRD with chronic dialysis and renal transplant in selected individuals. There currently is no cure for Lowe syndrome.

Prevention

In families in which a case of LS has occurred, various family planning options are available, including pre-natal testing and a special eye examination to help determine carrier status of at risk females. Knowing the specific abnormality on the LS gene via genetic testing of the individual with LS makes testing an individual for carrier status more effective. Families should consult with a geneticist to learn more about their options.

The Lowe Syndrome Association

The Lowe Syndrome Association (LSA) is registered as a not-for-profit corporation in the US. The LSA is international, representing approximately 25 countries from a multitude of ethnic, demographic and socio-economic backgrounds.

The LSA is governed by a Board of Directors comprised of caretakers, parents, professionals and friends who are passionate about implementing LSA’s mission. Board members are dedicated to helping families and children with Lowe syndrome through education, research, and community engagement and consult with a professional Medical & Scientific Advisory Board on matters of clinical and research related interests. The LSA maintains good working relationships with Lowe syndrome patient groups in the UK and Italy and provides links to additional groups on its website.

The LSA is officially incorporated in Chicago Ridge, Illinois and our Board of Directors are located throughout the United States.

Research

A key part of the LSA’s mission is to commission medical research so that at least one meaningful treatment can be identified by 2029. Research, nationally and internationally, into Lowe syndrome is modest at best. The LSA has supported research that includes mapping the OCRL1 gene, carrier detection, genetic testing, development of renal tubular cell lines, studies of the OCRL1 protein, exploring the physiological function of ptdins[4]p generation in the nucleus, genetic suppressors of 5-phosphatase mutants in c. elegans, studying the role of OCRL-1 in endocytic pathways, the cell biology of the Lowe syndrome protein, the study of rho-gtpase signaling abnormalities, the development of iPS cells from Lowe syndrome patients, and the development and characterization of animal models, including mouse models, in Lowe syndrome. LSA has funded 26 research initiatives between 1998-2021.

Other research is taking place on a limited basis around the world. The LSA is in the unique position of having connections with researchers and clinicians combined with the vision and drive to broaden this network and continues to taking strides in this direction

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LS medical experts

The LSA has a volunteer Medical Science Advisory Board (MSAB) consisting of clinicians and researchers. This body exists to aid caregivers in answering medical questions, screen requests for research funding, and assist in directing the LSA in its search for a cure.

Please visit our website www.lowesyndrome.org