

# Lowe Syndrome Association



## ABOUT THE LSA

The **Low Syndrome Association** is an international voluntary non-profit organization made up of parents, professionals and friends. The LSA is dedicated to helping children with Lowe syndrome and their families. The primary purposes of the LSA are to provide information, foster communication among families, and encourage and support medical research. Major activities include publishing a newsletter (*On the Beam*) and an informational booklet (*Living with Lowe Syndrome: A Guide for Families, Friends and Professionals*), sponsoring conferences every two years, and awarding medical research grants.

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Care today ... cure tomorrow!

## Low Syndrome Association

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## PARENT INFORMATION FORM

Parents, please provide information as indicated — print legibly and use extra paper if needed.

Print name and birth date of son(s) with Lowe syndrome						Family web site: <a href="http://www.lowesyndrome.org">http://</a>	
1	SON'S FIRST NAME	SON'S LAST NAME	BIRTH MO	BIRTH DAY	BIRTH YR	Languages spoken other than English:	
2	SON'S FIRST NAME	SON'S LAST NAME	BIRTH MO	BIRTH DAY	BIRTH YR	If you are not the biological parent of your child with LS, please check: <input type="checkbox"/> Adoptive Parent <input type="checkbox"/> Foster Parent <input type="checkbox"/> Other (Please specify):	
PERMISSION TO RELEASE NAME						<input type="checkbox"/> I do <input type="checkbox"/> I do not — give permission for the LSA to include my name, address, phone number, and son's name and birth date in the annual Parent Directory, and to share my name with other LSA parents as appropriate.	
RESEARCH ROSTER						<input type="checkbox"/> I do <input type="checkbox"/> I do not — give permission for the LSA to provide my name, address, phone number, and child's name and birth date to legitimate researchers who are working on LS. I understand I am under no obligation to participate in any research project about which I am contacted.	
AUTHORIZATION						YOUR SIGNATURE: _____  DATE SIGNED _____	

# QUESTIONS & ANSWERS ABOUT LOWE SYNDROME

## 1. 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.

## 2. 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. 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 and kidney and brain problems. How the enzyme deficiency leads to these defects is not yet completely understood.

## 2. How is it inherited?

The LS gene is 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.

## 4. 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
- 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
- Optimum life span of about 35-40 years due to progressive renal failure; however, many deaths have occurred at earlier ages due to renal failure, seizures, and other causes; life expectancy may increase in the near future as knowledge increases.

## 5. What are boys with LS like?

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

## 6. 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.

## 7. 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.

## 8. Can LS be prevented?

In families in which a case of LS has occurred, a special eye examination can help determine carrier status of at-risk females. Research currently underway may lead to a more definitive genetic test for carrier status. Various family planning options are available, including prenatal testing. Families should consult with a geneticist to learn more about their options.

## 9. 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 800-246-2436 or 713-798-4982 or contact by e-mail: bio@bcm.tmc.edu.

## 10. How can I learn more?

Contact the LSA for a copy of *Living with Lowe Syndrome: A Guide for Families, Friends and Professionals*. Single copies are free. More information can also be found on the LSA’s web site: [www.lowesyndrome.org](http://www.lowesyndrome.org).

### LSA MEMBERSHIP AND DONATION FORM Send to: LSA, 18919 Voss Road, Dallas, Texas 75287

Name (please print legibly):		Telephone:		Email address:	
Street address:		City:	State:	Zip Code:	Country:
PLEASE CHECK ONE:	<input type="checkbox"/> Parent of LS child (Parents, complete other side of form.) <input type="checkbox"/> Organization/Agency <input type="checkbox"/> Other: (please specify)				
CONTRIBUTION:	<input type="checkbox"/> \$15 Member <input type="checkbox"/> \$25 Contributor <input type="checkbox"/> \$50 Sustainer <input type="checkbox"/> \$100 Sponsor <input type="checkbox"/> \$250 Friend <input type="checkbox"/> \$500 Patron <input type="checkbox"/> \$1,000 Benefactor <input type="checkbox"/> Other: \$ _____ <input type="checkbox"/> Residential agency with client who has Lowe syndrome (free membership) <input type="checkbox"/> I am the parent of a child with Lowe syndrome and would like to join but cannot afford membership at this time.				
MEMORIAL OR SPECIAL OCCASION GIFT:	This donation is given <input type="checkbox"/> in memory of <input type="checkbox"/> in honor of:		Send acknowledgement of gift to:		
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The IRS recognizes the LSA as a Section 501(c)(3) non-profit charitable organization. All donations are tax-deductible to the extent provided by law.