Support for Physicians

Alström Syndrome International is dedicated to finding a cure for this devastating disease and to developing and promoting the best treatments for those already suffering.

We support the research and medical community with:

Research
ASI sponsors research clinics and scientific collaboration to enable physicians to deliver the best possible care to their patients. It also maintains a comprehensive network for communicating with patients and families when new recommendations or clinical therapies become available.

Education
ASI collects and organizes information for researchers and physicians, and develops educational tools to keep them abreast of the latest developments.

Physician Support
ASI facilitates medical networking internationally and seeks to support patient-based research that will benefit all patients.

Clinical Features of Alström Syndrome

Alström Syndrome is a rare, recessive genetic disorder. Considerable variability exists in the expression of the features in Alström Syndrome.

Manifestations:

- Nystagmus and photophobia in early infancy
- Progressive degeneration of the retina in childhood, eventual blindness
- Mild to moderate bilateral hearing loss
- Congestive heart failure secondary to dilated cardiomyopathy
- Hyperinsulinemia, insulin resistance, diabetes mellitus, usually in childhood
- Childhood obesity
- Elevation of liver enzymes, liver dysfunction, portal hypertension
- Chronic obstructive pulmonary disease
- Progressive, chronic kidney failure
- Normal intelligence; sometimes delayed developmental milestones
- Normal extremities; no polydactyly or syndactyly
- Short stature, advanced bone age, scoliosis
- High triglycerides (hypertriglyceridemia)
- Hypothyroidism
- Hypertension
- Acanthosis nigricans (patchy darkening of the skin)
- Male hypogonadism
- Frequent episodes of otitis media (glue ear)
- Frequent urinary tract infections, incontinence
- Seizures

Support for Families

ASI was founded to support families and individuals living with Alström Syndrome, a rare, life-threatening, genetic disease. Our three program areas provide essential support and services to families.

Research, Education & Family Support
A team of Family Support Coordinators, composed of Alström parents, is pleased to answer your questions personally and offer individual support when you call or write. No question is too small or too complex. Through our extensive contact with families, physicians, and researchers associated with ASI, we have built a solid core of knowledge and understanding about Alström Syndrome.

Through its ongoing sponsorship of scientific and family conferences, it has garnered the universal interest, participation, and cooperation of those patients, parents, physicians and researchers touched by the disease.

The mission of Alström Syndrome International (ASI), a 501 (c) (3) non-profit charitable organization, is to provide support, information, and coordination world-wide to families and professionals in order to treat and cure Alström Syndrome.

www.alstrom.org

Since 1995, ASI has collected the largest data repository on the syndrome in existence.

Drop by Drop We Make a Lake

The story of Emma, a 4-years old

The story of Dillon, an 18-years old

The story of Phoebe, a 9-years old

Support for Families

Our International Conferences make it possible for families, care givers, and professionals to hear current information about Alström Syndrome presented by physicians and researchers. The meetings also offer a unique setting for mutual sharing and support. ASI triennial Conferences provide families with individual consultations, workshops, learning sessions, and countless opportunities to meet and socialize with others in similar situations.
Alström Syndrome besets children at a young age and is very often misdiagnosed. Physicians unfamiliar with the syndrome—the array of problems that Alström comprises—are most likely to identify only the medical problem that is first apparent, such as retinal degeneration.

Until recently, very few pediatricians and general practitioners would have had reason to suspect that their young patient was suffering from a rare genetic disease. That’s beginning to change, as ASI makes research and information sharing a priority.

ASI has helped to identify more than 1,000 children affected by the syndrome and has created an international network of physicians and scientists who are engaged in research and in sharing their knowledge with their peers.

Other critically important services for Alström Syndrome families include:

• Offering expertise for newly identified patients and their families to help understand, monitor, and cope with Alström Syndrome
• Facilitating genetic counseling and testing
• Providing information services that include a website, private forums for discussions, newsletters, and periodic regional and international conferences
• Responding to emergency medical situations
• Providing access to an international corps of leading physicians in the treatment of the syndrome
• Assisting patients and families to attend Alström Syndrome clinics around the world
• Engendering family to family friendships and support

ASI has collaborated with research scientists to identify and understand the function of the gene that causes Alström Syndrome, and now it is focused on developing effective treatments and ultimately finding a cure.

Even as physicians seek to treat the many complications involved—blindness, childhood obesity and diabetes, hearing loss, juvenile cardiomyopathy, and major organ failure—they are turning to ASI to help them learn from their peers and the latest research.

In the past year alone, research scientists have tested therapies to prevent or reverse vision loss, developed protocols that will expand the understanding of the cardiomyopathy in infants, and illuminated some of the unique mechanisms that cause the uncontrollable weight gain in children with Alström - new breakthroughs are on the horizon!

What lies ahead? Earlier interventions. Better treatments. And yes, maybe someday, even a cure. But until then, ASI remains a source of support and information for families and a willing collaborator for researchers and professionals.