DISEASES SUPPORTED

- Alpha-Mannosidosis
- Aspartylglucosaminuria
- Beta-Mannosidosis
- Fucosidosis
- Galactosialidosis
- Mucolipidosis II Alpha/Beta
- Mucolipidosis III Gamma
- Schindler Disease
- Sialdisosis

375

Affected Members Supported Worldwide

Helena & Andrea - Fucosidosis

WHAT ARE GLYCOPROTEIN STORAGE DISEASES?

- They are very rare, progressive and largely untreatable inherited genetic defects.
- Glycoprotein diseases are a subset of Lysosomal diseases, characterized by impaired degradation of glycoproteins within the lysosomes.
- Seven of these diseases are caused by deficiency of enzymes that function within the lysosome and two are due to impaired trafficking of enzymes to the lysosome.
- The course of these diseases means they affect multiple systems, with clinical symptoms which may vary from patient to patient and even among affected siblings.
- For most patients the implications are eventual loss of mental and/or physical function and reduced life expectancy. For those who live into adulthood there are often severe physical and/or neurological symptoms.

Ivan- Galactosialidosis

SUPPORTING FAMILIES WITH GLYCOPROTEIN STORAGE DISEASES

ISMRD

Contact ISMRD Today!

info@ISMRD.org
www.ISMRD.org
P.O. Box 683
Turnersville NJ 08012, USA

Dr. D'Azzo & Annie - Sialidosis

OUR MISSION

ISMRD is the leading advocate for families worldwide affected by Glycoprotein Storage Diseases.

Through partnerships built with medicine, science and industry; we seek to detect and cure these diseases, and to provide a global network of support and information.

OUR VISION

We seek a future in which children with Glycoprotein storage diseases can be detected early, treated effectively and go on to live long, healthy and productive lives.

Anna - ML III

OUR HISTORY

Founded in 1999 by Paul and Debora Murphy, ISMRD was born out of a desperate need for support and information for families facing the challenges of Glycoprotein Storage Diseases. Their daughter, Taryn, was diagnosed with Alpha-Mannosidosis, a rare and debilitating condition. Driven by love and determination, the Murphys established ISMRD to connect families, raise awareness, and foster research collaborations to find effective treatments and cures.

Through unwavering dedication, ISMRD has grown into a leading global organization providing invaluable support to individuals and families impacted by these rare diseases. They have successfully advocated for increased research funding, facilitated international collaborations among researchers and clinicians, and developed vital resources for families.

Disclaimer: This information is for general knowledge and informational purposes only and does not constitute medical advice. For diagnosis and treatment of any medical condition, consult a qualified healthcare professional.

Tim - Alpha-Mann

Oliver - Beta-Mann

OUR ACHIEVEMENTS

1999 ISMRD established

2003 1st Scientific/Family conference held in Washington D.C.

2005 "Crossing Oceans for a Cure" workshop and fundraiser held in Michigan

2007 2nd Scientific/Family Conference held in Michigan

2011 ISMRD wins Patient Advocate Leaders Award for Innovation from Genzyme

2012 3rd Scientific/Family Conference held in Charleston, South Carolina

2013 Chase Community Giving grants $10,000 to ISMRD

2015 4th Scientific/Family Conference in St Louis, Missouri

2017 5th Scientific/Family Conference in Rome, Italy; Vatican letter of blessing

2019 6th Scientific/Family Conference in Atlanta, Georgia

2023 ISMRD-Sponsored ongoing research into Bone Marrow Transplant and Alpha-Mannosidosis

2024 ISMRD-Sponsored Feline Mucolipidosis completed research projects: Gene Therapy Research in Mucolipidosis and GNPTAB-related Disorders and ISMRD becomes founding member of the Global LSD Collaborative

Check out our website for a full list of achievements!
www.ISMRD.org