



Matthew 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, characterised 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.

ISMRD Supports The Following Diseases

- Alpha Mannosidosis
- Fucosidosis
- Mucopolipidosis III
- Beta Mannosidosis
- Galactosialidosis
- Schindler Disease
- Aspartylglucosaminuria
- Mucopolipidosis II
- Sialidosis



Taryn Alpha Mannosidosis



The International Advocate for Glycoprotein Storage Diseases



Supporting families with Glycoprotein storage diseases



Jonalin I-Cell disease



Our Mission

ISMRD is the leading advocate for families world wide 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.



Sammy and Hudson ML II/III

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.



Brooke ML II

Our History

ISMRD was incorporated as the International Society for Mannosidosis & Related Diseases in Baltimore, Maryland, USA on March 10, 1999. Paul and Debora Murphy, parents of a child with Alpha-Mannosidosis, developed the organisation to fill a void they perceived existed both for affected families and scientists and physicians with an interest in research and treatment modalities.

In 2003 at a scientific/family workshop supported by the NINDS, ISMRD included Mucopolipidosis Type II and Type III into its network and the organisation developed a new focus on the 9 diseases, which all have Glycoprotein storage as their central problem.



Alex Sialidosis



Autumn
Mucopolipidosis III

Our Achievements

- 1999** ISMRD was established
- 2000** Website launched
- 2003** 1st Scientific/Family conference held in Washington
- 2004** Penguin Cafe was launched
- 2005** "Crossing Oceans for a Cure" workshop and fundraiser held in Michigan bringing together the largest group of ML families ever
- 2006** ISMRD partnered with the Greenwood Genetic Centre to develop a Natural History Study for our 9 diseases
- 2007** 2nd Scientific/Family Conference held in Michigan
- 2008** Consensus development meeting on bone disease in Mucopolipidosis held in New Zealand
- 2008** AMP scholarship gained to extend the Natural History study to New Zealand and Australian patients
- 2009** Successful support of \$250,000 application to NIH for Natural History study continuation

“ISMRD now supports over 175 families worldwide.”



Ethan ML II