



# Together We Are Strong

**ISMARD's International Glycoproteinoses Forum**

**In Minneapolis, MN and From Around the World!**

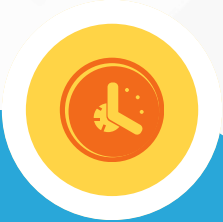


**Friday AUG 8th  
& Saturday AUG 9th**

This is a Preliminary Program Only. All times and speakers will be subject to change. Be sure to check back as updates are made.

More Information:  [www.ismrd.org](http://www.ismrd.org)

# Program



**THURSDAY, AUGUST 7TH**

## ***TOGETHER WE ARE STRONG***

ISMRD is delighted to introduce you to its 7th International Glycoproteinoses Family and Scientific Forum. The theme of our forum is in keeping with ISMRD's mission of seeking to detect and cure the Glycoprotein Storage Diseases, and to provide a global network of support and information, through partnerships built with medicine, science and industry. Working together is our GREATEST STRENGTH!

The Forum will combine both in-person and remote presentations as well as webinar access for families around the globe.

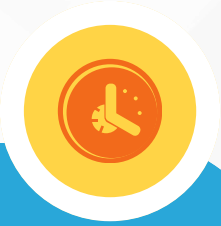
ISMRD WELCOMES YOU ALL!

4:00 PM -6:00 PM

- REGISTRATION CHECK-IN



# Program



**FRIDAY, AUGUST 8TH**

7:00 AM - 8:30AM

## ***BREAKFAST***

8:00 AM - 8:05 AM

- **Welcome** *Carolyn Paisley-Dew, ISMRD President*

8:10 AM - 8:15 AM

- Opening of the Forum *Marc Patterson, M.D.*

8:20 AM - 8:25 AM

- **Our Mission** - *Chester B. Whitley, Ph.D., M.D.*

8:25 AM - 8:30 AM

- Overview “Think Tank” - White Paper & NIH Grant

8:30 AM - 8:35 AM

- The Status of Patient Needs, Treatment, The Future

8:35 AM - 8:40 AM

- Outcome Measures *Chester B. Whitley, Ph.D., M.D.*

8:45 AM - 8:55 AM

- Patient Voice: Unmet Medical Needs  
*Chester B. Whitley, Ph.D., M.D.*

## ***ALPHA-MANNOSIDOSIS***

9:00 AM - 9:05 AM

- Overview *Karolina M Stepien, M.D., Ph.D.*

9:10 AM - 9:20 AM

- Treating the Whole Brain in Large Animal Homologue of Alpha-Mannosidosis *John H Wolfe, VMD, Ph.D.*

9:25 AM - 9:35 AM

- Options of Therapy in Alpha-Mannosidosis  
*Robert Šáhó, M.D.*

9:40 AM - 9:50 AM

- Patient Voice: The Journey that Changed Everything  
*Sarah Forsman, ISMRD Communications Officer*

9:55 AM - 10:05 AM

- Long-Term Effects of BMT in Alpha-Mannosidosis  
*Julie B Eisengart, Ph.D., LP*

10:10 AM - 10:20 AM

- Patient Voice Discussion: Unmet Medical Needs

10:20 AM - 10:35 AM

## ***Coffee Break***

# Program



**FRIDAY, AUGUST 8TH** *continued*

## ***ASPARTYLGLUCOSAMINURIA (AGU)***

10:35 AM - 10:40 AM

- Overview *Karolina M Stepien, M.D., Ph.D.*

10:40 AM - 10:50 AM

- Clinical Manifestations and Long-Term Outcomes in Adults with AGU *Karolina M Stepien, M.D., Ph.D.*

10:50 AM - 11:00 AM

- Patient Voice: Unmet Medical Needs  
*Troy Lund, Ph.D., M.D.*

11:05 AM - 11:15 AM

- Outcome Measures of HSCT for AGU  
*Troy Lund, Ph.D., M.D.*

11:20 AM - 11:30 AM

- Patient Voice Discussion: Unmet Medical Needs

## ***BETA-MANNOSIDOSIS***

11:35 AM - 11:40 AM

- Overview *Virginia Kimonis, M.D., M.R.C.P.*

11:40 AM - 11:50 AM

- How to Fast-Track Through FDA Approval for ERT & Natural History Study *Virginia Kimonis, M.D., M.R.C.P.*

11:55 AM - 12:05 PM

- Beta-Mannosidosis Revisited  
*Angela Martin Rios M.D., MSC*

12:10 PM - 12:20 PM

- Patient Voice: Beta-Mannosidosis Parent and Patient Advocate, The Lost Enzyme Project  
*Laurel Gregier, ISMRD Board Member*

12:25 PM - 12:35 PM

- Patient Voice Discussion: Unmet Medical Needs

12:40 PM - 1:40 PM

***LUNCH***

# Program



**FRIDAY, AUGUST 8TH** *continued*



1:40 PM - 1:45 PM

1:45 PM - 1:55 PM

2:00 PM - 2:10 PM

2:15 PM - 2:25 PM

2:30 PM - 2:40 PM

2:45 PM - 2:55 PM

3:00 PM - 3:15 PM

3:15 PM - 3:20 PM

3:20 PM - 3:25 PM

3:30 PM - 3:35 PM

3:40 PM - 3:45 PM

3:45 PM - 3:55 PM

4:00 PM - 4:10 PM

4:15 PM - 4:30 PM

4:30 PM - 5:30 PM

5:30 PM

## **FUCOSIDOSIS**

- Overview *Karolina M Stepien, M.D., Ph.D.*
- Long-Term Outcomes of Fucosidosis in Adults  
*Karolina M Stepien, M.D., Ph.D.*
- HSCT for Fucosidosis *Troy Lund, Ph.D., M.D.*
- Patient Voice: Fucosidosis Parent and Patient Advocate  
*Carolyn Paisley-Dew, ISMRD President*
- Evaluation of the Effects of JR-471  
*Mathias Schmidt, PD, Ph.D.*
- Patient Voice Discussion: Unmet Medical Needs

## **BREAK**

## **SCHINDLER DISEASE**

- Overview *R.J. Desnick, M.D., Ph.D.*
- Current Approaches to Treatment *R.J. Desnick, M.D., Ph.D.*
- Patient Voice: Unmet Medical Needs  
*Chester B. Whitley, Ph.D., M.D.*
- Outcome Measures *R.J. Desnick, M.D., Ph.D.*
- Patient Voice Discussion: Unmet Medical Needs
- End of Day Recap  
*Chester B. Whitley, Ph.D., M.D. & Marc Patterson, M.D.*

- **GROUP PHOTO**

- **POSTER SESSION**

## **CLOSE OF DAY 1**

# Program



**FRIDAY, AUGUST 8TH** *continued*



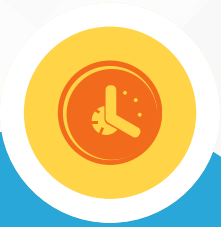
4:30 PM - 5:30 PM

## POSTERS

- **AAV-based gene therapy as a treatment strategy in a Fucosidosis mouse model** *Hannah Bäumges, MS*
- **A novel homozygous variant in MAN2B1 causing Alpha-Mannosidosis** *Gabriela Ponte de Mattos*
- **Outcome of haemopoietic stem cell transplantation in 21 patients with Alpha-Mannosidosis** *Robert Šáhó, M.D.*
- **Evaluation of the effects of JR-471, a transferrin receptor-targeted alpha-L-fucosidase, on the disease phenotypes of a murine model of Fucosidosis** *Mathias Schmidt, PD, Ph.D.*
- **Real world reflections of the patient odyssey in Alpha-Mannosidosis: Insights and Challenges in diagnosis** *Sonia Sgrò*
- **The diagnostic odyssey of a biochemically confirmed case of ML II: a case-report of the first western patient with multiple dysplasia type Ain-Naz** *Professor Fernanda Sperb Ludwig*
- **Hormonal dysfunction as a long-term complication of Glycoproteinoses** *Karolina M Stepien, M.D., Ph.D.*
- **Quality of life and burden of caregivers of patients with Alpha-Mannosidosis: Results from an international survey** *Karolina M Stepien, M.D., Ph.D.*
- **Secondary mitochondrial dysfunction in Glycoproteinoses: Alpha-Mannosidosis and Fucosidosis** *Karolina M Stepien, M.D., Ph.D.*
- **Improving blood-brain barrier penetration in Hurler syndrome using an IDUA-ApoE fusion enzyme delivered via the PS Gene Editing System** *Chester B. Whitley, Ph.D., M.D.*
- **Penetrating the blood-brain barrier: Utilizing the PS gene editing system to encode a novel fusion  $\beta$ -galactosidase for the treatment of GM1-gangliosidosis** *Chester B. Whitley, Ph.D., M.D.*



# Program



**SATURDAY, AUGUST 9TH**

7:00 AM - 8:30 AM

8:00 AM - 8:05 AM

## ***BREAKFAST***

- Recap Our Mission *Marc Patterson, M.D.*

## ***GALACTOSIALIDOSIS***

8:10 AM - 8:15 AM

8:15 AM - 8:25 AM

- Overview *Leigh Fremuth, Ph.D.*
- The Journey to Therapy in Galactosialidosis  
*Leigh Fremuth, Ph.D.*
- Patient Voice: Galactosialidosis Patient Advocate,  
Galactosialidosis Network  
*Çağdaş Canbolat, ISMRD Board Member*
- Outcome Measures *Marc Patterson, M.D.*
- Patient Voice Discussion: Unmet Medical Needs

8:30 AM - 8:40 AM

8:45 AM - 8:55 AM

9:00 AM - 9:10 AM

## ***SIALIDOSIS***

9:15 AM - 9:20 AM

9:20 AM - 9:30 AM

- Overview *Leigh Fremuth, Ph.D.*
- Understanding Mechanisms of Pathogenesis to Treat  
Sialidosis *Leigh Fremuth, Ph.D.*
- Patient Voice: The Path to Gene Therapy  
Sialidosis Patient Advocate *Dan Peach, Cure Mucopolipidosis*
- Outcome Measures *Marc Patterson, M.D.*
- Patient Voice Discussion: Unmet Medical Needs

9:30 AM - 9:40 AM

9:45 AM - 9:55 AM

10:00 AM - 10:10 AM

10:15 AM - 10:30 AM

## ***Coffee Break***

# Program



**SATURDAY, AUGUST 9TH** *continued*

## **MUCOLIPIDOSIS**

10:30 AM - 10:40 AM

- Overview of GNPTAB-related (ML) disorders  
*Richard Steet, Ph.D.*

10:40 AM - 10:50 AM

- Mechanisms of Drug Resistance in ML *Richard Steet, Ph.D.*

10:55 AM - 11:05 AM

- Patient Voice: Patient Survey Mucopolipidosis Diseases  
*Jackie James, Cure Mucopolipidosis President*

11:10 AM - 11:20 AM

- Preclinical research in ML II/III a/b *Patricia Dickson, M.D.*

11:25 AM - 11:35 AM

- Patient Voice: Mucopolipidosis Patient Advocate  
*Jackie James, Cure Mucopolipidosis President*

11:40 AM - 11:50 AM

- Outcome Measures *Patricia Dickson, M.D.*

11:55 AM - 12:55 PM

- **Recap and Next Steps - Our Mission: The ISMRD Project**  
*Chester B. Whitley, Ph.D., M.D. & Marc Patterson, M.D.*

1:00 PM - 1:20 PM

- Patient Voice: *Jeanine Jarnes, PharmD, BCOP, BCPS*

1:25 PM - 1:30 PM

- Thank You from ISMRD *Danielle Forsman, Alpha-Mannosidosis Parent, ISMRD Treasurer*

- **CLOSE OF FORUM**