



# Together We Are Strong

**ISMARD's International Glycoproteinoses Forum**

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

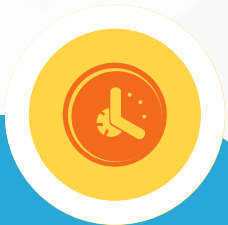


**Friday AUG 8th  
& Saturday AUG 9th**

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 OPENS



# Program



**FRIDAY, AUGUST 8TH**

## ***BREAKFAST***

7:00 AM - 8:00AM

8:00 AM - 8:05 AM

8:05 AM - 8:10 AM

8:10 AM - 8:20 AM

8:20 AM - 8:30 AM

8:30 AM - 8:40 AM

8:40 AM - 8:50 AM

8:50 AM - 9:00 AM

9:00 AM - 9:15 AM

9:15 AM - 9:30 AM

9:30 AM - 9:35 AM

9:40 AM - 9:55 AM

10:00 AM - 10:15 AM

10:20 AM - 10:35 AM

10:40 AM - 10:55 AM

11:00 AM - 11:10 AM

11:10 AM - 11:25 AM

● Welcome *Carolyn Paisley-Dew, ISMRD President*

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

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

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

● The Status of Patient Needs, Treatment, The Future

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

● Patient Voice: Unmet Medical Needs

*Jeanine Jarnes, PharmD, BCOP, BCPS*

● Therapy for Genetic Diseases: Overview and Considerations for the Glycoproteinoses *Marc Patterson, M.D.*

● Panel Discussion

## ***ALPHA-MANNOSIDOSIS***

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

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

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

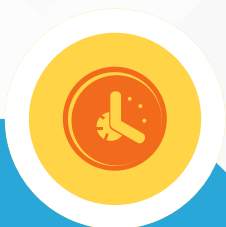
● A European Model-Enzyme Replacement Therapy  
*Robert Šáhó, M.D.*

● Alpha-Mannosidosis Patient Perspective  
*Sarah Forsman, ISMRD Communications Officer*

● Patient Voice: Unmet Medical Needs Panel

## ***Coffee Break***

# Program



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

## **ASPARTYLGLUCOSAMINURIA (AGU)**

11:30 AM - 11:35 AM  
11:35 AM - 11:45 AM  
11:45 AM - 11:55 AM

- Overview of AGU *Karolina M Stepien, M.D., Ph.D.*
- Clinical Experience in Adults with AGU
- Current Approaches to Treatment and Outcome Measures  
*Karolina M Stepien, M.D., Ph.D.*
- Patient Voice: Unmet Medical Needs Panel

## **LUNCH**

## **BETA-MANNOSIDOSIS**

1:15 PM - 1:20 PM  
1:20 PM - 1:35 PM

- Overview *Virginia Kimonis, M.D., M.R.C.P.*
- How to fast-track through FDA approval for ERT & Natural History Study *Virginia Kimonis, M.D., M.R.C.P.*
- Enzyme Replacement Therapy for Beta-Mannosidosis  
*Angela Martin-Rios M.D., MSC*
- Beta-Mannosidosis Parent and Patient Advocate, The Lost Enzyme Project *Laurel Gregier, ISMRD Board Member*
- Patient Voice: Unmet Medical Needs Panel

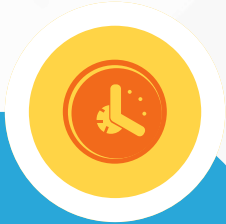
## **FUCOSIDOSIS**

2:30 PM - 2:35 PM  
2:40 PM - 2:55 PM  
3:00 PM - 3:15 PM  
3:20 PM - 3:35 PM  
3:40 PM - 3:55 PM  
4:00 PM - 4:10 PM

- Overview *R.J. Desnick, M.D., Ph.D.*
- HSCT for Fucosidosis *Troy Lund, Ph.D., M.D.*
- Clinical Experience in Patients with Fucosidosis  
*Karolina M Stepien, M.D., Ph.D.*
- Evaluation of the effects of JR-471 *Mathias Schmidt, Ph.D.*
- Fucosidosis Parent and Patient Advocate  
*Carolyn Paisley-Dew, ISMRD President*
- Patient Voice: Unmet Medical Needs Panel

# Program

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



4:15 PM - 4:20 PM

4:20 PM - 4:35 PM

4:35 PM - 4:45 PM

4:45 PM - 4:50 PM

4:50 PM - 5:30 PM

5:30 PM

## **SCHINDLER DISEASE**

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

## **POSTER SESSION**

## **CLOSE OF DAY 1**

# Program

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

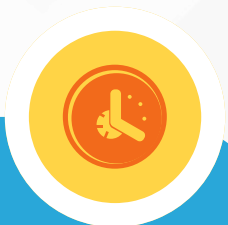


4:50 PM -  
5:30 PM

## POSTERS

- **AAV-based gene therapy as a treatment strategy in a Fucosidosis mouse model** *Hannah Bäumges, MS*
- **Hormonal dysfunction as a long-term complication of Glycoproteinoses** *Karolina M Stepien, M.D., Ph.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, 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.*
- **Real world reflections of the patient odyssey in Alpha-Mannosidosis: Insights and Challenges in diagnosis** *Sonia Sgrò*
- **A novel homozygous variant in MAN2B1 causing Alpha-Mannosidosis** *Gabriela Ponte de Mattos*
- **Secondary mitochondrial dysfunction in Glycoproteinoses: Alpha-Mannosidosis and Fucosidosis** *Karolina M Stepien, M.D., Ph.D.*
- **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*
- **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.*
- **Outcome of haemopoietic stem cell transplantation in 21 patients with Alpha-Mannosidosis** *Robert Šáhó, M.D.*

# Program



**SATURDAY, AUGUST 9TH**

7:00 AM - 8:00 AM

## ***BREAKFAST***

8:00 AM - 8:05 AM

8:05 AM - 8:15 AM

8:20 AM - 8:35 AM

8:40 AM - 8:55 AM

9:00 AM - 9:10 AM

9:15 AM - 9:20 AM

9:25 AM - 9:40 AM

9:45 AM - 10:00 AM

10:05 AM - 10:20 AM

10:20 AM - 10:30 AM

10:30 AM - 10:45 AM

## ***GALACTOSIALIDOSIS***

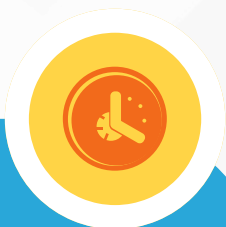
- Overview *Marc Patterson, M.D.*
- Clinical Experience in Patients *Leigh Fremuth, Ph.D.*
- Current Approaches to Treatment and Outcome Measures  
*David Wenger, Ph.D.*
- Galactosialidosis Patient Advocate Perspective:  
Galactosialidosis Network  
*Çağdaş Canbolat, ISMRD Board Member*
- Patient Voice: Unmet Medical Needs Panel

## ***SIALIDOSIS***

- Overview *Leigh Fremuth, Ph.D.*
- Therapeutic Approaches for Sialidosis  
*Leigh Fremuth, Ph.D.*
- Outcome Measures
- Sialidosis Patient Advocate Perspective:  
The Path to Gene Therapy  
*Dan Peach, Cure Mucopolidosis*
- Patient Voice: Unmet Medical Needs Panel

## ***Coffee Break***

# Program



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

## ***MUCOLIPIDOSIS II and III A/B***

### ***MUCOLIPIDOSIS III***

10:45 AM - 10:50 AM

10:55 AM - 11:10 AM

11:15 AM - 11:30 AM

11:35 AM - 11:50 AM

11:55 AM - 12:10 PM

12:15 PM - 12:30 PM

12:35 PM - 12:45 PM

12:45 PM - 12:50 PM

12:55 PM - 1:00PM

- Overview *Richard Steet, Ph.D.*
- Mechanisms of Drug Resistance in MLII  
*Richard Steet, Ph.D.*
- The Diagnostic Odyssey Case of ML II  
*Professor Fernanda Sperb Ludwig*
- Overview and Current Treatment Approaches in ML III  
*Ming Li, Ph.D*
- Current Approaches to Treatment and Outcome Measures  
*Marc Patterson, M.D.*
- Mucopolipidosis Patient Advocate Perspective,  
*Jackie James, President Cure Mucopolipidosis*
- Patient Voice: Unmet Medical Needs Panel
- **Recap and Next Steps - Our Mission: The ISMRD Project**  
*Chester B. Whitley, Ph.D., M.D. & Marc Patterson, M.D.*
- Thank You from ISMRD *Danielle Forsman, ISMRD Treasurer*

***CLOSE OF DAY 2***