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5TH GLYCOPROTEINOSES INTERNATIONAL CONFERENCE

Rome, Italy
November 1-4 2017

EMBRACING INNOVATION ADVANCING THE CURE

PROGRAM
& ABSTRACTS

5TH GLYCOPROTEINOSES INTERNATIONAL CONFERENCE

ROME, ITALY NOVEMBER 1-4 2017

EMBRACING INNOVATION ADVANCING THE CURE

ISMRD would like to say *a very special thank you* to the following organizations and companies who have very generously given donations to support the 5th International Conference on Glycoproteinoses.

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SANOFI GENZYME

EVERYLIFE FOUNDATION FOR RARE DISEASES

Amicus Therapeutics®

THE WAGNER FOUNDATION

ISMRD is *very grateful for all the help and support* that Symposia has given us in the organization of our Conference on-the-ground support in Rome.

symposia®

Dedicated to helping patients in the rare disease community with unmet medical needs

Ultragenyx Pharmaceutical Inc. is a clinical-stage biopharmaceutical company committed to creating new therapeutics to combat serious, debilitating diseases.

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Contents

Welcome to Rome!

On behalf of ISMRD, I would like to welcome you all to the Fifth Glycoproteinoses International Conference on **'Embracing Innovation and Advancing the Cure'**.

ISMRD is thrilled to bring our International Conference to Europe allowing us to connect with families, researchers, clinicians, support groups and others who work in the field of rare diseases. But, more importantly to help make the invaluable connections for families who perhaps have never met another family with their particular disease.

Over the next few days you will have the opportunity to learn about the advancements toward therapies for some of these diseases. You will hear from families about what it's like to live with one of these very rare conditions, and hear about the exciting advances towards Enzyme Replacement Therapy and Gene Therapy.

It takes many hours and hard work to bring together a meeting of this nature. I would like to thank our Primary Investigator Alessandra d'Azzo and her Scientific Committee who have put together a really strong scientific program with some amazing speakers from around the world.

Of course, our conference would not take place without significant charitable donations. I would like to thank the following companies and foundations, **Ultragenyx**, **EveryLife Foundation**, **Sanofi/Genzyme**, **Amicus** and **The Wagner Foundation**. I would also like to extend our very grateful thanks to Symposia who are event planners, who have worked alongside us here in Rome and have helped you check in at registration. They will be on site throughout the meeting offering support and answering any questions you may have.

To the families who are a part of the conference: welcome! It can be a lonely journey when you are told your child has a very rare genetic condition, especially when you might be the only family for miles around! These meetings help make all those important connections, creating a network of support, information and knowledge. I know there will be some lifelong friendships made this weekend. To families who have been to our meetings in the past please reach out to those who are new. I hope over the next few days you will have the confidence to approach the researchers and clinicians attending this meeting and ask all your questions. I know from experience that they look forward to this

aspect of the conference.

My hope for each family attending is that you will leave this conference knowing that you have an ISMRD family that stands behind you, and that you are not alone in this journey.

Jackie James
PRESIDENT ISMRD

On behalf of the Scientific Committee of ISMRD, it is my honor and great joy to welcome the patients, families, clinicians, and scientists who have traveled to Roma to attend the Fifth International Conference on Glycoproteinoses. This is the first ISMRD conference to be held in Europe. We changed the venue of this gathering to raise global awareness of these rare lysosomal disorders and increase the visibility of ISMRD as a family-centric organization that not only supports patients and their families but also funds scientific research and meetings on glycoproteinoses worldwide.

We designed this year's scientific program to bring together basic scientists and clinicians with patients and their families to share their latest discoveries in the areas of pathophysiology, preclinical therapy development, and clinical trials for these diseases. Patients and their family members have been included in the program to offer a unique opportunity to young researchers to learn first-hand about the experiences, challenges, and concerns of patients with glycoproteinoses and their expectations for the future.

Our vision for this conference is that it will stimulate the exchange of ideas, encourage new collaborations among investigators and clinicians with different expertise, spark interest in these diseases among postdoctoral research fellows and graduate students, strengthen connections among affected families around the world, and foster national and international partnerships to advance therapies for affected children and adolescents.

Together, we will keep advancing treatment and advocating for patients and families affected by these disorders.

Enjoy the conference!

Alessandra d'Azzo Ph.D.
CHAIR SCIENTIFIC COMMITTEE

ISMRD Mission & Governance

ISMRD is a U.S. 501 (c)3 charity that is governed by an all-volunteer organization led by a Board of Directors whose backgrounds span nations, diseases and experience. Each member of the Board serves a two-year term, which can be renewed upon the approval of the remaining members. We actively seek out others whose experience and background enhance our ability to carry out our Mission, and whose passion for that Mission enables us to reach our goals. 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; a future where doctors and other clinicians are knowledgeable of and able to detect these genetic defects efficiently and with accuracy. In our vision the public at-large will have a general knowledge and understanding of these diseases, and will actively strive to prevent their occurrence. Ultimately, we envision a world where there will no longer be a need for our organization or others like it to exist.

ISMRD BOARD OF DIRECTORS

Jackie James
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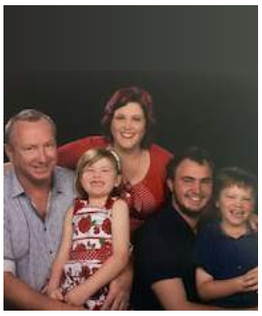


Juanita Van Dam
Australia

ISMRD ADVISORY BOARD

ISMRD's Board of Directors is assisted in the execution of its mission and goals by the following distinguished members of the international scientific and medical community.

Steven Walkley, D.V.M., Ph.D.
Albert Einstein College of Medicine, USA

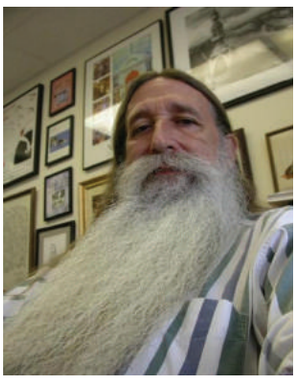
Barbara Burton, M.D.
Children's Memorial Hospital, Chicago, USA

Sara Cathey, M.D.
Greenwood Genetic Centre South Carolina, USA

Alessandra d'Azzo, Ph.D.
St Jude Children's Research Hospital, USA

Dag Malm, M.D., Ph.D.
University Hospital Tromsoe, Norway

Marc Patterson, M.D.
Child Neurology
Mayo Clinic, Rochester, USA



Mark Haskins, V.M.D., Ph.D.
University of Pennsylvania Veterinary Hospital, USA

Richard Steet, Ph.D.
University of Georgia Athens Georgia, USA

John Hopwood, Ph.D.
Women & Children's Hospital, Adelaide, Australia

Thomas Braulke, Ph.D.
University of Hamburg
Hamburg, Germany

Charles Vite, Ph.D.
School of Veterinary Medicine
Philadelphia, USA

ISMRD General Information

SPEAKER PRESENTATIONS

Please go to Registration during Welcome Reception on 1st November to get your presentations loaded onto the computer prior to the commencement of the conference.

If you missed the above please go to the Registration desk between 7:30am and 8:00am, or see your presentation chairperson 10-15 minutes prior to the commencement of the session.

NAME TAGS

Name tags are to be worn at all times to allow entry into the meeting and social functions.

MOBILE PHONES AND MOVEMENT BETWEEN MEETINGS

Participants are asked to ensure that all mobile phones are switched off during conference sessions.

To minimise disturbances in the session rooms whilst presenters are speaking we ask that you remain seated during presentations.

GENERAL QUESTIONS

ISMRD is thrilled to have the support of Symposia. Their staff are multilingual and are available to answer any questions you may have about the meeting.

ISMRD Board members are also available to help you.

CATERING

Welcome Reception:
1st November 6:30pm - 8:30pm
Buffet style food will be available

Breakfast is included in your Accommodation:
Go to the Sapori Dal Mondo Restaurant

Conference Breaks:
Tea, coffee and snacks will be served in the Conference Foyer

Lunch:
Lunch will be served in the Sapori Dal Mondo Restaurant at the International cooking stations.

REGISTRATION DESK TIMES

Wednesday 1st November
4:30pm - 6:30pm

Thursday 2nd November
7:30am - 4:30pm

Friday 3rd November
8:00am - 12noon

ISMRD Conference Functions

WELCOME RECEPTION

Wednesday 1st November

Time: 6:30pm – 8:30pm

Room: Foyer of the Conference Center

Drinks: A cash drinks station is available for all drinks.

Don't miss a great opportunity to meet your colleagues, meet old friends and make new ones before the conference begins at 8:30am the next day. An evening of local food and wine not to be missed.

AWARDS DINNER

Friday 3rd November

Time: 6:30pm for seating at 7pm

Room: The Conference Center

Join us for one of the highlights of our conference, this year's Awards Dinner will be something a little special.

Pre-dinner drinks will be available at a cash drinks station before the commencement of a 3-course dinner. The food, wine and entertainment promise to give you a great night of fun and relaxation.

Eating Out in Rome

The Restaurants below are a small sample of what is available within a few kilometres of the A. Roma LifeStyle Hotel. However if you want to go further into the city please ask the Hotel Staff to recommend a restaurant to you.

You may find it easier to catch a taxi to your destination. You can expect to pay approximately €20 per trip, depending on your destination.

BAR VITTORIA

5 minute walk from A.Roma Lifestyle Hotel

Piazza Biagio Pace 6,
00164 Rome
Phone 06 6615 7531

REVIEW: *"We stayed at the A.Roma Hotel and ate here most nights. The food is very good value and the service is fantastic! Always a good sign when a place is frequented by local residents as is the case here. Really added to our enjoyment of a great week in Rome. Thanks."*

CASETTE DI CAMPAGNA

1.6km from A.Roma Lifestyle Hotel

Via di Affogalasino, 40,
00148 Rome
Phone 06 6574 3230

REVIEW: *"Great food, top quality service and beautiful location."*

PERDINCIBACCO

2.1km from A. Roma LifeStyle Hotel

Via delle Fornaci 5,
00165 Rome
Phone 06 632 527

REVIEW: *"Delicious and inexpensive. The spaghetti with garlic and chilli was amazing. Antipasti misto was fab too."*

RISTORANTE COUSINE RESTAURANT

5 minute walk from A.Roma Lifestyle Hotel

Via Camillo Serafini 47,
00164 Rome
Phone 06 9259 9077

CUISINES: Italian, Pizza, Mediterranean

REVIEW: *"They make a good fresh cuisine, classic dishes prepared and presented well .... try."*

OSTERIA PALMIRA

1.8km from A.Roma Lifestyle Hotel

Via Abate Ugone 29
Phone 06 5820 4298

CUISINES: Italian, Mediterranean

REVIEW: *"If you are considering Osteria Palmira, go! It has a great atmosphere, good service, great food and great value!"*

Suggested Tour Options

Enquires can be made at the hotel with the concierge.

The Coliseum

The Vatican

The Trevi Fountain

Pantheon

Borghese Art Gallery

"Rome is the city of echoes, the city of illusions, and the city of yearning."

Giotto di Bondone
(Italian painter and architect. Died 1337)

Scientific Program

DAY 2: NOVEMBER 2ND 2017		
Chair: Alessandra d'Azzo		
8:30am	Welcome and introduction to the meeting	*Jackie James - USA, Alessandra d'Azzo - USA*
8:45am	**Keynote Presentation:** Glycoproteins and Glycoprotein Storage Diseases - An Overview	*Stuart Kornfeld - USA*
9:30am	Cell Biology and Pathophysiology of Lysosomal Storage Diseases	*Fran Platt - England*
10:00AM MORNING BREAK		
Session 1 – Alpha Mannosidosis		Chair: Dag Malm
10:20am	a-Mannosidosis Historical and Current Aspects	*Dag Malm - Norway*
10:40am	Diagnosis and monitoring of patients with glycoproteinoses disorders by novel UPLC-MS/MS Oligosaccharide analysis	*Sara Cathey - USA*
11:00am	Lysosomal alpha Mannosidase and alpha Mannosidosis	*Tommaso Beccari - Italy*
11:20am	Hematopoietic Cell Transplant for Glycoproteinoses	*Troy Lund - USA*
11:50am	Long-term Efficacy and Safety of Velmanase Alfa (Human Recombinant alpha-mannosidase) Long-term Enzyme Replacement Therapy for Alpha-Mannosidosis	*Line Borgwardt - Denmark*
12:15pm	Enhanced Phosphorylation of Lysosomal Enzymes Mediated by an Engineered GlcNAc-1-phosphotransferase	*Balraj Doray - USA*
12:25pm	A Patient's View: Living with Mannosidosis - A shared presentation	*John Forman - New Zealand Dag Malm - Norway*
		Chair: Maurizio Scarpa
12:40pm	General Discussion: Engaging with families, patients, Clinicians, Geneticists, genetic counsellors on clinical and molecular diagnosis, clinical care person experience with individual cases, Therapies and Clinical biomarkers for assessing outcomes etc.	*Panelists: Christina Lampe, Rossella Parini, Sara Cathey*
1:00PM LUNCH		
		Chair: Stuart Kornfeld
2:00pm	Lysosomal membrane proteins and their functions	*Paul Saftig - Germany*
2:30pm	Efficacy of BMT and AAV – mediated therapy in Krabbe disease	*David Wenger - USA*
Session 2: Mucolipidosis II and III		
3:00pm	Mannose-phosphorylation in health and disease	*Thomas Braulke - Germany*
3:30PM BREAK		

3:50pm	Activity Base Profiling In Live Zebrafish Reveals TGFß Regulation of Cathepsin Activation During MLII Pathogenesis	*Heather Flannagan-Steet - USA*
4:10pm	Functional Analysis of GNPTAB and GNPTG Null Cells Identifies Reactive Oxygen Species (ROS)-Dependent Increases in c-Met Activity	*Richard Steet - USA*
4:30pm	Zebrafish model of lysosomal disorders associated with skeletal defects: Challenging an old paradigm of disease pathogenesis	*Enrico Moro - Italy*
5:00pm	A Natural History Study and AAV-mediated Gene Therapy Approach for Feline Mucolipidosis II	*Allison Bradbury - USA*
5:20pm	Coatomer participates in the Golgi localization of GlcNAc-1-phosphotransferase	*Lin Liu - USA*
5:30pm	A Patient's View: Living with ML II and ML III – a shared presentation	*ML II – Paul Wagner - USA* *ML III – Jenny Noble - New Zealand*
		Chair: Sara Cathey
5:50pm	General Discussion: Engaging with families, patients, Clinicians, Geneticists, genetic counsellors on clinical and molecular diagnosis, clinical care person experience with individual cases, Therapies and Clinical biomarkers for assessing outcomes etc.	*Panelists: Christina Lampe, Rossella Parini, Gepke Visser, Agata Fiumara, Elena Procopio*

DAY 3: NOVEMBER 3RD 2017		
		Chair: Generoso Andria
8:30am	Heat Shock Protein-based therapies as clinical candidates for sphingolipidoses	*Thomas Kirkegaard Jensen - Denmark*
Session 3: Sialidosis (ML I) and Galactosialidosis		
9:00am	Sialidosis (ML I) and Galactosialidosis: Historical and clinical overview	*Generoso Andria - Italy*
9:30am	Molecular pathophysiology in Sialidosis: Links to adult conditions of aging	*Alessandra d'Azzo - USA*
10:00am	Lymphatic pathway in glycoproteinoses	*Noelia Escobedo - USA*
10:20am	Factors regulating transcription of the lysosomal sialidase NEU1	*Ida Annunziata - USA*
10:40AM BREAK		

11:00am	Enzyme replacement therapy for galactosialidosis, Towards the Clinical Trial	*Vish Koppaka - USA*
11:20am	Myoclonus is a key symptom of the adult form of Sialidosis Type I	*Laura Canafoglia - Italy*
11:30am	Molecular diagnosis of glycoproteinoses	*Amelia Morrone - Italy*
11:50am	A Patient's View: Living with Sialidosis Type I	*Daniel Peach - New Zealand*
		Chair: Bruno Bembi
12:10pm	General Discussion: Engaging with families, patients, Clinicians, Geneticists, genetic counsellors on clinical and molecular diagnosis, clinical care person experience with individual cases, Therapies and Clinical biomarkers for assessing outcomes etc.	*Panelists: Camilo Toro, Andrea Dardis, Gepke Visser, Alice Donati, Sara Cathey, Laura Canafoglia, Agata Fiumara, Amelia Morrone*
12:40pm	Afternoon free for networking or taking one of the tour options in Roma	

AWARDS DINNER 6:00PM FOR 7:00PM FOR SEATING

DAY 4: 4TH NOVEMBER 2017

		Chair: Alessandra d'Azzo
8:30am	Epilepsy in Lysosomal Storage Diseases	*Renzo Guerrini - Italy*
9:00am	Intracerebroventricular cerliponase alfa for children with CLN2 disease: Interim results from an ongoing Phase 2 extension study	*Angela Schulz - Germany*
Session 4: Aspartylglucosaminuria, Fucosidosis, Schindler Disease		**Chair: Alessandra d'Azzo**
9:30am	Personalized therapy approaches for AGU	*Ritva Tikkanen - Germany*
10:00am	AAV-based therapy in aspartylglucosaminuria mice	*Xin Chen - USA*
10:30am	A Mouse Model for Fucosidosis	*Torben Lübke - Germany*
11:00AM BREAK		
11:20am	A Patient's View: Living with Fucosidosis	*Jean Leonard - England*
11:40am	ERN for metabolic disorders: An Overview	*Maurizio Scarpa - Germany*
12:10pm	GalNAcT/AGU double knockout mice experience accelerated disease over the single aspartylglucosaminuria mice	*Matthew Ellinwood - USA*
12:30pm	The genetic diagnosis: a long and complex case of Mucolipidosis III in Brazil	*Ida Schwartz - Brazil*
12:40pm	Meeting Summary	*Maurizio Scarpa - Germany*
1:00pm	Closing of Meeting	*Jackie James - President ISMRD*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Accelerating Biotech Innovation for Rare Disease Treatments Through Science-Driven Public Policy
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Family Program

This year we are trying something a little different. Rather than having presentations in your workshops, we thought you would benefit from having round table discussions sharing information and asking all the questions you ever wanted to ask of the Professionals.

Please join the scientific meeting for the first part of the morning on day 2 and then see below when your particular workshop is scheduled.

DAY 2: NOVEMBER 2ND 2017		
8:30am	Welcome and introduction to the meeting	*Jackie James - USA, Alessandra d'Azzo - USA*
8:45am	Keynote Presentation: Glycoproteins and Glycoprotein Storage Diseases an Overview	*Stuart Kornfeld - USA*
9:30am	Cell Biology and Pathophysiology of Lysosomal Storage Diseases	*Frances Platt - England*
10:00AM MORNING BREAK		
Aspartylglucosaminuria Workshop		
10:20am	General Discussion	
	Research - What is the future - What can be done	
	Medical Management	
1:00pm	General Discussion Q&A	
Fucosidosis Workshop		Chair: Carolyn Paisley-Dew
10:20am	General Discussion	
	Research - What is the future - What can be done	
	Medical Management	
1:00pm	General Discussion Q&A	

DAY 3: NOVEMBER 3RD 2017		
Alpha Mannosidosis Workshop		Chair: Mark Stark
8:30am	Alpha Mannosidosis - An Overview	*Dag Malm - Norway*
	A Parents View: Bone Marrow Transplant	*Martin Woolley - England*
	Medical Management	
10:40AM MORNING BREAK		
	A Parents View: Living with Alpha Mannosidosis	*Mark Stark - United States*
12:30am	General Discussion and Q&A	

Mucolipidosis Workshop		Chair: Jackie James
8:30am	Mucolipidosis an Overview - Where are we at and what's new	*Sara Cathey - United States*
	Research - What is the future for ML, What can ML Families do to help	
	Medical Management	
10:40AM MORNING BREAK		
	Bone Disease and Pain Management - What is being done - What can be done	
	A Parent/Patient's View: Living with Mucolipidosis III	*Shirley Jamil - England*
12:30pm	General Discussion and Q&A	

DAY 4: NOVEMBER 4TH 2017		
Sialidosis Workshop		Chair: Daniel Peach
8:30am	Sialidosis - An Overview	
	Research - Where are we at?	
	A Patients View: Living with Sialidosis	*Faith Peach - New Zealand*
	Medical Management - An Overview	
10:00am	Origin and Treatment of Neurological Symptoms in Sialidosis	*Camilo Toro - United States*
12:30pm	General Discussion and Q&A	
Galactosialidosis Workshop		Chair: Daniel Peach
8:30am	Sialidosis - An Overview	
	Research - Where are we at?	
	A Patients View: Living with Sialidosis	*Faith Peach - New Zealand*
	Medical Management - An Overview	
10:00am	Origin and Treatment of Neurological Symptoms in Sialidosis	*Camilo Toro - United States*
12:30pm	General Discussion and Q&A	

Children's Program Information

IMPORTANT NOTICES FOR PARENTS

Parents are requested to have their children at the Child Care room by 8am in the Conference Center.

It is important to ensure that you are on time on day one, as it takes time to meet your carer and get the children transferred onto buses.

Please make sure that your child has on their name tag and that they have all their personal needs for the day (eg spare nappies/diapers, sunscreen, spare clothes).

If your child is in a stroller or wheelchair please show your carer how to fold and unfold the chair.

Please go to the Registration desk and check in each day.

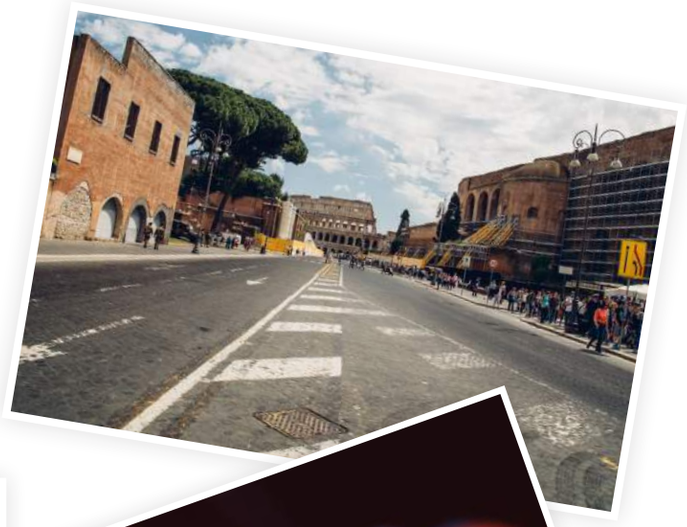
The children will be having Pizza or lunch on day one. Additional snacks and drinks will be in their backpacks.

For children on special diets please make sure you include this in their packs.

Parents – Please ensure that you are seated promptly before the commencement of the conference.

CAREGIVERS

We have some truly amazing carers all lined up for your children and adults affected by one of the 9 glycoprotein storage diseases. We want to thank them all for very generously giving up their time to care for our loved ones.



ISMRD is an internationally focused not-for-profit organization whose mission is to advocate for families and patients affected by one of the following disorders.

Alpha-Mannosidosis
Aspartylglucosaminuria
Beta-Mannosidosis
Fucosidosis
Galactosialidosis
Sialidosis (Mucolipidosis I)
Mucolipidosis II, II/III, III alpha/beta
Mucolipidosis III Gamma
Schindler Disease