



ISMRD ANNUAL REPORT FOR 2023

OUR MISSION

ISMRD is the leading advocate for families worldwide affected by a Glycoprotein Storage Disease. 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, be treated effectively, and go on to live long, healthy and productive lives.

BOARD OF DIRECTORS (All non-salaried)

Carolyn Paisley-Dew	President	Australia
Danielle Forsman	Treasurer	USA
Sarah Forsman	Communications Officer	USA
Darko Jamnik	Board Member	Slovenia
Patricia Gribel	Board Member	USA
Laurel Gregier	Board Member	USA
Lama Khalil	Board Member	Saudi Arabia
Tareq Qashou	Board Member	Saudi Arabia
Hussein Peeran	Board Member	USA
Cagdas Canbolat	Board Member	UK
Abby Lindenberg	Board Member	USA

This Board is more international than it has ever been before, and represents more of our diseases than ever before. We are proud of the strong and dedicated members of our Board.

ISMRD Professional Advisory Board (All non-salaried)

Prof Richard Steet: Scientific Chair	USA	Sara Cathey	USA
Steve Walkley	USA	Dag Malm	Norway
Alessandra d’Azzo	USA	Charles Vite	USA
Marc Patterson	USA	Amelia Morrone	Italy
Thomas Braulke	Germany	Vish Koppaka	USA
Enrico Moro	Italy	Jenny Klein	USA

Our Professional Advisory Board is an important source of scientific and medical advice and information for the Board and the President. We would like to thank each and every one of the PAB members.

OUR ACTIVITIES FOR 2023

Chiesi Alpha-Mannosidosis FDA approval

Chiesi Global Rare Diseases successfully gained FDA approval in February 2023 to introduce Lamzede, a non-central nervous system enzyme replacement therapy for Alpha-Mannosidosis adults and children, into the USA. ISMRD has been working hard to get the word out to Alpha-Mannosidosis families in the US, including assisting with a webinar in June 2023. Several young American children benefitted from Lamzede in 2023 as they approached bone marrow transplants, and in the months that followed the transplant.

JCR Pharmaceuticals and Fucosidosis

JCR is developing a new drug candidate, JR-471, a blood-brain barrier (BBB)-penetrating α -L-fucosidase for the treatment of patients with Fucosidosis.

Medipal Holdings Corporation is helping to fund the global commercialization of JR-471 outside Japan, including research, development, manufacturing, and marketing.

A meeting was held in February 2023 between ISMRD, Fucosidosis families from around the world, JCR Pharmaceuticals and Medipal Holdings. The meeting was both face-to-face and virtual, and held during WORLDSymposium 2023.

ISMRD continues to assist JCR Pharmaceuticals as it lays the foundations for developing an enzyme replacement therapy for Fucosidosis.

PGT-M testing for Sialidosis families in the UK

Following on the heels of successful approval for the licensing of Fucosidosis for use in preimplantation genetic testing for monogenic disorders (PGT-M) in the UK in 2022, ISMRD sought patient testimony for PGT-M among the international Sialidosis community. Sialidosis has now been licensed for PGT-M in the UK. This is a wonderful step forward for this and future Sialidosis families in the UK.

ISMRD-Sponsored Research into the longterm effects of Bone Marrow Transplant on Alpha-Mannosidosis

Thanks to the generous Al-Ansari Grant from an Alpha-Mannosidosis family, ISMRD funded in 2023 a project looking at the long-term effects of Bone Marrow Transplant on Alpha-

Mannosidosis. There will be an interim Report after 12 months, and a Final Report after 24 months, at the end of August 2025.

ISMRD-Sponsored Feline Mucopolipidosis Research

Gene Therapy Research in Mucopolipidosis: *To Evaluate AAV Gene Therapy in the Feline Model of ML II*

This research is ongoing, and a final report is expected at the end of August 2024.

GNPTAB-related Disorders

This research is ongoing, and a final report is expected at the end of August 2024.

Beta-Mannosidosis

Laurel Gregier is the first Beta-Mannosidosis parent to join the ISMRD Board. Laurel worked very hard during 2023 to look for options for Beta-Mannosidosis. This was a difficult task, given that we knew of only four Beta-Mannosidosis patients in the world, all in the US. By the end of 2023, five more Beta-Mannosidosis patients had been located, in the UK, and Laurel had been instrumental in the emergence of the Lost Enzyme Project, in collaboration with Kimonis Lab at the University of California, Irvine. The first step of this project will be a pre-clinical study into Beta-Mannosidosis enzyme replacement therapy using JCR Pharmaceuticals' J-Brain Cargo technology. This study will require \$520,925, and Laurel and the Lost Enzyme Team are fundraising for this amount.

Galactosialidosis

Clara Lorin Canbolat was diagnosed in early 2023 with Galactosialidosis. Her father, Cagdas Canbolat, embarked on finding a treatment for the disease. He joined the ISMRD Board and has been building connections with pharmaceutical companies, hospitals and leading researchers around the world, exploring therapies ranging from Gene Therapy to drug repurposing. During 2023, he also identified and engaged with many living patients and their clinical teams across the globe, and began monitoring and incorporating Galactosialidosis symptoms in more detail. He is supporting the repurposing of a drug which may stop the progression of the condition, visiting Germany to meet with the team behind the work. It is expected that the drug will be made available once legal proceedings are finalised. Through intensive networking across the world, he has also been the catalyst for the establishment of a team who have pulled together a project and submitted funding applications for the manufacturing of the AAV8 virus gene therapy.

Fundraising and Donations

Giving Season Fundraiser

ISMRD held a Giving Season fundraiser for the month of December 2023, with a campaign of emails and Facebook posts featuring some of the glycoproteinoses and ISMRD families. The campaign exceeded its goal of \$5,000, successfully raising over \$6,000.

Facebook Birthday fundraisers

Facebook offers a very simple and effective way to fundraise for ISMRD. On your birthday, Facebook asks if you would like to hold a birthday fundraiser. Once you say yes, you can nominate ISMRD as your chosen charity, and the rest is easy. This seems to be a fairly new Facebook feature. One of our Board members raised over \$500 for her birthday in December 2023.

CouponBirds

CouponBirds is a useful and regular fundraiser for ISMRD. Donations are small, but steady, and we hope that they will increase as more of our members begin to utilise CouponBirds. Join CouponBirds [here](#).

Donations

Donations totalling \$92,006 were made to the ISMRD during 2023. Much of this was donated by the Al-Ansari Alpha-Mannosidosis family for research into the long-term effects of bone marrow transplant for Alpha-Mannosidosis. Other donations were from family members and friends, their workplaces, churches, fundraisers, and through Facebook, AmazonSmile and CouponBirds. There were family members who held birthday fundraisers, and others who raised funds in memory of a loved one. We also have Board members and ex-Board members who pay for various ongoing expenses for ISMRD.

ISMRD is very grateful to these considerate individuals and organizations and thanks them profusely for their kind and generous donations.

TechSoup and Quickbooks

In addition to fundraising, ISMRD constantly looks for ways to reduce its operating costs. Our hardworking Treasurer, Danielle Forsman, in 2023 gained access for ISMRD to Techsoup, a platform geared to help support non-profits by providing extensive donated or discounted software, hardware and technology services. Using TechSoup, Danielle switched to Quickbooks for our recordkeeping and tax reporting needs, at a cost savings of almost \$500 per year.

Online Presence

Continuing to develop our online presence is a priority for ISMRD, as it is an important source of new memberships, and requests from researchers, pharmaceutical companies and medical experts as the go-to organization for information about the glycoproteinoses, and from newly diagnosed families.

In line with this, in 2023 Board member Sarah Forsman was appointed ISMRD Communications Officer, and Kyrsten Lawless became our Promotions Officer. Sarah is very energetic and enthusiastic, with many ideas, and is spearheading the work to help propel ISMRD into 2024 with a stronger online presence. This has included a fundraising campaign during the Giving Season.

We feature an increasing number of partnership logos on our website, including the NORD Platinum membership seal, Candid's Silver Seal of Transparency, the Galactosialidosis Network, the Lost Enzyme Project, AngelFlight and Patient Worthy.

ISMRD would like to thank Chiesi Global Rare Diseases for its generous grant to update our website. Sarah will be lead on this project, to commence in 2024.

ISMRD's Facebook pages continue to flourish and provide important support and information for its members, and the wider community. They are also an important source of new memberships. The main page, the ISMRD Group page, is open to families only, to provide privacy as they discuss personal issues. All other pages are open to the public.

Conferences and Other Development Attended

President Carolyn Paisley-Dew and Board Member Laurel Gregier attended the WORLDSymposium Orlando Florida 20 Feb – 2 March 2023.

Carolyn also attended the Lysosomal Disease Summit that was held In Melbourne, 27-29 October 2023 Australia.

Board member Darko Jamnik attended the October 2023 Training Workshop for patient representatives and advocates on leadership and communication skills in Gdański, Poland. This course was jointly organised by EURORDIS (Rare Diseases Europe) and the Medical University of Gdański.

Attendance at conferences and courses such as these is vital for keeping up-to-date with new developments and research, for networking and making important contacts in the rare disease arena and for development of Board members.

Financial Statement

Income Statement for the 2023 calendar year
Balance sheet for the end of the year (Dec. 31, 2023)

Attachment A
Attachment B

ISM RD CONTACT DETAILS

Address:

3150 Almaden Expressway Ste 103
San Jose, CA 95118
USA

Mailing Address:

PO Box 683
Turnersville, NJ 08012
USA

E-mail: info@ismrd.org

Website: www.ismrd.org

Income Statement (Profit and Loss)

International Society for Mannosidosis & Related Diseases

For the year ended December 31, 2023

2023

Income

A. Mann Research Fund	82,955.00
Donation - Amazon Smile	205.70
Donation - Recurring payment	1,456.41
Donation- Unrestricted.	7,049.14
Facebook Donations	340.55
Total Income	92,006.80

Gross Profit

92,006.80

Operating Expenses

Bank Service Charges	5.00
Charity Registration	25.00
Conferences/Meetings	3,903.79
Consulting & Accounting	314.82
Contractors & Professional fees	279.03
Family Scholarship	66.73
Grants to Other Organisations	82,969.00
Membership Fees	150.00
ML Reserach Grant paid	6,000.00
Postage & Delivery	194.00
Total Operating Expenses	93,907.37

Operating Income

(1,900.57)

Net Income

(1,900.57)

Balance Sheet

International Society for Mannosidosis & Related Diseases As of December 31, 2023

DEC 31, 2023

Assets

Current Assets

Cash and Cash Equivalents

California Account	88,802.38
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Total Cash and Cash Equivalents	88,802.38
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Total Current Assets	88,802.38
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Total Assets	88,802.38
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Liabilities and Equity

Equity

Current Year Earnings	(1,900.57)
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Retained Earnings	90,702.95
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Total Equity	88,802.38
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Total Liabilities and Equity	88,802.38
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