MYHRE SYNDROME FOUNDATION

JANUARY 2023

ANNUAL REPORT

PREPARED AND PRESENTED BY

KATE WEARS & THE MYHRE SYNDROME FOUNDATION BOARD OF DIRECTORS

WELCOME

Being diagnosed with a rare syndrome can be a lonely experience, especially when you might be the only person or family in the entire country that is affected.

The Myhre Syndrome Foundation was founded so that the members of the international Myhre community don't feel alone, that they have someone to turn to.

One of our core values is Hope.

Giving hope is funding our own research into Myhre syndrome. Giving hope is providing the community with treatment guidelines. Giving hope is being there to listen and to be guided on what the community needs.

We are indebted to our supporters, who enable us to give hope, who help us in our mission to find treatments and, one day, a cure.

In 2022, MSF achieved a huge milestone of funding Myhre research and publishing the first Myhre Syndrome Patient and Family Handbook. This is just the start as we now look ahead to 2023.

We are grateful you are part of our journey.

Kate Wears,
Executive Director, MSF
Proud Mom to Josh who has Myhre

ABOUT MYHRE SYNDROME FOUNDATION

HOW IT STARTED

Myhre Syndrome Foundation (MSF) was founded in 2019, by two families whose children have Myhre syndrome.

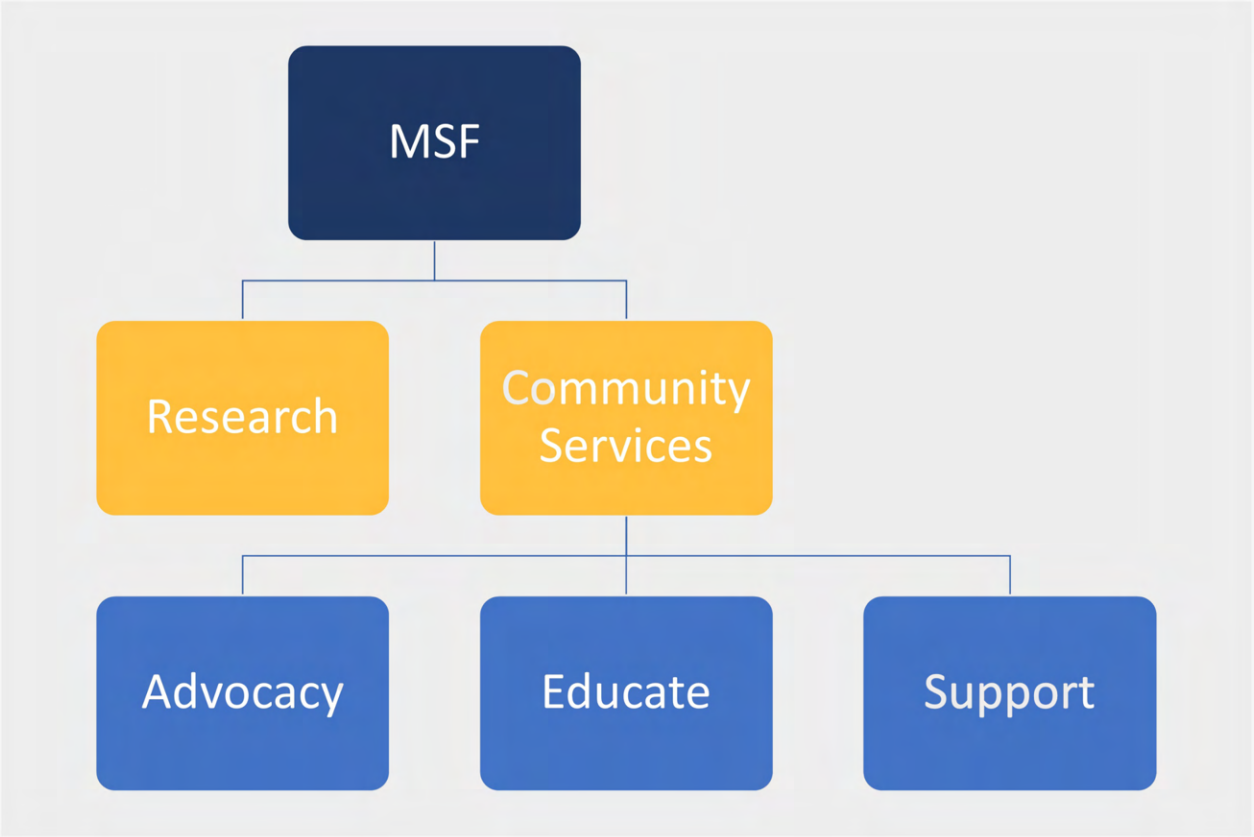
Since then MSF has grown into a global community and network of individuals, families, physicians and care providers who all aim to make the lives of those living with Myhre easier.

We are indebted to the Smith and Young families for working so hard to establish the foundation.

OUR MISSION

MSF is a patient advocacy organization dedicated to providing hope and improving the lives of those impacted by Myhre syndrome.

We foster collaboration among all relevant stakeholders to build a strong global community in order to advance **research**, as well as **support**, **educate** and **advocate** for those impacted by Myhre syndrome.



RESEARCH

THE GOALS OF MYHRE SYNDROME RESEARCH ARE MULTIFACETED.

NOT ONLY IS THERE A NEED TO UNDERSTAND THE PHYSICAL PRESENTATION OF SYMPTOMS (PHENOTYPE), THERE IS A NEED TO UNDERSTAND THE UNDERLYING CAUSES OF THE SYMPTOMS (MECHANISMS). THIS KNOWLEDGE WILL ENABLE SPECIALIZED TREATMENT PLANS, THERAPEUTICS AND A CURE.

In 2022, MSF began work on a Research Roadmap.

The MSF Research Roadmap is a patient and family-led effort that establishes our research objectives and goals over the next five years.

It prioritizes funding for research to achieve life-improving treatments as quickly and efficiently as possible, whilst working towards our longer-term goal of finding a cure.

It was devised following consultation with patients and families via surveys and focus groups, as well as engagement with industry experts, including members of the MSF Professional Advisory Board.

The research roadmap will help us:

- Direct our limited financial resources in a direction that is most impactful for patients and families
- Incentivize researchers to work collaboratively on projects that are most aligned with patient and family priorities
- Attract governmental or industry funding to accomplish the most resource-intensive tasks

RESEARCH (CONT'D)

THE RESEARCH ROADMAP WILL COMPLEMENT ONGOING RESEARCH, INCLUDING RESEARCH FUNDED BY MSF.

IN 2022, MSF AWARDED TWO RESEARCH GRANTS TO FUND THE FOLLOWING MODELS.

MOUSE MODEL
BOSTON, USA

A population of mice was created with a genetic variant that causes Myhre syndrome, under the direction of Dr. Mark Lindsay (Assoc. Prof. of Medicine, Harvard Medical School). These mice will be studied to understand the internal mechanisms causing Myhre syndrome symptoms.

This effort will complement the ongoing natural history study being led by Dr. Angela Lin (Prof. of Pediatrics, Harvard Medical School) at the MGH Myhre Syndrome Clinic.

Housing both the mouse model and natural history study under one roof will enable rapid comparison of the mouse phenotype to the human phenotype, which will accelerate progress toward therapeutics and open potential avenues to new funding.

ZEBRAFISH MODEL
GHENT, BELGIUM

Due to its complexity, the cellular mechanisms in Myhre syndrome are still poorly understood.

Two zebrafish models have been developed harboring disease-associated variants comparable to those identified in individuals with Myhre syndrome. This effort is led by Dr. Bert Callewaert (Ghent Univ. Center for Medical Genetics).

These models will add to our understanding of what the specific disease mechanisms are and which factors determine the variability of the clinical symptoms. Additionally, zebrafish are excellent, low-cost candidates for screening existing drugs, which will enable efficient identification of pharmacological targets.

RESEARCH

THE MSF PROFESSIONAL ADVISORY BOARD (PAB) IS A NETWORK OF RESEARCHERS AND CLINICIANS WHO COLLABORATE TO ADVANCE MYHRE RESEARCH AND PROVIDE CRUCIAL GUIDANCE FOR MYHRE DOCTORS AND PATIENTS.

In 2022, The PAB:

- Updated GeneReviews and published the new findings
- Assisted in the creation of the Myhre Syndrome Patient and Family Handbook
- Provided new and updated content for the Summer Seminars program
- Worked with dozens of clinicians around the world to knowledge share for better treatment options for Myhre patients
- Collaborated with the MSF Board on the Research Roadmap
- Continued to work on research papers and the collaborative process that will enable Evidence Based Clinical Guidelines to be published in the future
- Represented Myhre syndrome at conferences and events to educate colleagues and raise awareness

COMMUNITY SERVICES

MSF IS DEDICATED TO EDUCATING, ADVOCATING AND SUPPORTING THE INTERNATIONAL MYHRE COMMUNITY.

We are proud to represent the Myhre community and are passionate about supporting them at each step.

We support the community from the first point of diagnosis through to updating on latest clinical findings and treatment options.

We assist the Myhre community in three main areas:

- **Education**
- **Support**
- **Advocacy**

EDUCATION

PROVIDING KNOWLEDGE ON MYHRE FOR BETTER HEALTH OUTCOMES

Myhre Syndrome Patient and Family Handbook

In 2022, we created the first ever handbook that details:

- What Is Myhre Syndrome?
- How Is It Diagnosed & What Do The Genetics Mean?
- Diagnosis & Beyond
- Thinking Ahead
- Myhre Syndrome Foundation Support
- The Myhre Syndrome Clinic

- Patient and Family Interpretation of GeneReviews 2022
  - Summary
  - Diagnostic Testing of Myhre Syndrome
  - Clinical Characteristics/ Treatment/Management of Symptoms
  - Prevention of Secondary Complications

- Resources
  - Appointment Notes Template
  - Overview Of Care Team Template
  - Medical & Radiology Terminology
  - Patient Registry Data

It is available in English, Spanish, French, Italian, German and Dutch.

Website Translation

We also translated our website into English, Spanish, French, Italian, German and Dutch, increasing the reach and knowledge of information on Myhre to our international community of patients, families, researchers, physicians and caregivers.

The handbook has been requested over 150 times
64% of requests are from outside the US
30% of requests are for non-English language versions
25 handbooks have been sent to medical professionals

EDUCATION (CONT'D)

EVENTS HELP US REACH OUR COMMUNITY TO SHARE KNOWLEDGE

Summer Seminars

Following the success of the 2021 MSF International Virtual Conference, in 2022 we held a Summer Seminars program, releasing new videos and updated content every Saturday throughout July.

Content included new speakers, as well as updated clinical findings covering these areas:

- ENT
- Skin
- Genetics
- Endocrine
- Pulmonology
- Brain & Spine
- Autism Spectrum Disorder
- Developmental Disabilities
- Nephrology & Hypertension
- Neuro-Ophthalmology
- Clinical Updates from the Myhre Syndrome Clinic in Boston, from Dr. Angela Lin

The Summer Seminars program was a free event, with all content available on demand and available in closed captions for over 100 languages. Find these at myhresyndrome.org/videos

Dr. Angela Lin presenting at the seminars

"PROVIDING EVERYONE WITH UP-TO-DATE INFORMATION ON HOW THE SYNDROME PROGRESSES IS VITAL.

WE WANT TO ENSURE NO MATTER THEIR LOCATION, OUR COMMUNITY HAS ACCESS TO INFORMATION THAT WILL IMPROVE TREATMENT OPTIONS."

Ines White, MSF President

SUPPORT & ADVOCACY

BRINGING TOGETHER AND REPRESENTING THE MYHRE COMMUNITY MEANS WE ARE LISTENING TO THEIR NEEDS AND RAISING AWARENESS OF THESE NEEDS TO THE WIDER RARE DISEASE AND MEDICAL NETWORKS.

Coffee Hours
Each month we bring the international Myhre community together by hosting a virtual coffee hour.

Each month we discuss a new topic, from sleep, food, behavior to what we love to do in our spare time. It's a place to ask questions, share experiences and get to know each other.

Advocating for the Myhre Community
Patti Schultz, Patient Advocacy lead and Board Member, spoke at the Mayo Clinic in Rochester, Minnesota at The Rare Storyteller's Event. Over 300 doctors were in attendance and Patti spoke about her journey to a diagnosis and educated them all on Myhre syndrome.

Kate Wears (right), Executive Director, was invited to speak on a panel at the Milken Institute Future of Health Summit in Washington, D.C., raising awareness of the work we do, and championing the needs of rare disease communities.

IN REMEMBRANCE

WE WILL NEVER FORGET THOSE WE HAVE LOST TO MYHRE SYNDROME. EACH DAY WE WILL DO OUR BEST TO RAISE AWARENESS, RAISE FUNDS AND CONTINUE IN OUR MISSION TO FIND A CURE.

Jax, 7 years old, from Queensland, Australia, passed away after a short illness in November 2022.

Jax's parents honored their little boy.

"Jax was the light in all our eyes, a beloved son, brother and friend, grandson and nephew who will be deeply missed by our family and all who came to know our sweet, dear Jaxxy Boy. Always laughing, joking, smiling and full of positivity, Jax's memory will forever continue to beat in our hearts.
We miss you darling boy. XOXO"

Ellie, 8 years old, from California, lost her battle with Myhre in November 2022. Ellie's family said...

"Ellie was the youngest of six children. For eight years she was adored, cherished and everyone's go to for hugs and cuddle time. The name Ellie means bright shining light and that is what she brought to our family. She truly was a beacon of light that lit up our lives. Every giggle and laugh was infectious, we all adored her great sense of humor. She loved learning, singing, dancing, cooking, books, swimming, and drinking ice water. She loved her family fiercely. Ellie showed us how to find joy in every day! We miss her greatly and are deeply thankful for the time she spent with us. No amount of time with her would have ever been enough."

BUDGET

FISCAL YEAR 1 JULY TO 30 JUNE, ALL FIGURES IN USD ($)

WE ARE INDEBTED TO MSF SUPPORTERS WHO ENABLE US TO FUND RESEARCH AND SUPPORT THE MYHRE COMMUNITY.

ITEM	2020/2021	2021/2022
REVENUE		
Government		
Foundations & Organizations		
Individuals	105,626	112,578
Miscellaneous		
Total	**105,626**	**112,578**
EXPENSES		
Personnel		2,363
Professional Fees		18,117*
Administrative Expenses	8,215	8,153**
Fundraising		2,878
Grant Funding to Advance Myhre Research		150,000
Total	**8,215**	**181,511**
ASSETS		
Balance beginning of the year - 1 July	20,288	117,699
Balance end of the year - 30 June	117,699	48,766

*Strategic consultants for capacity building to develop five year strategic and development plan

**Fees assosiated with the running of the organizaion, including but not limited to website hosting, event platform, bank fees, design resource, insurance.

OUR 2023 VISION

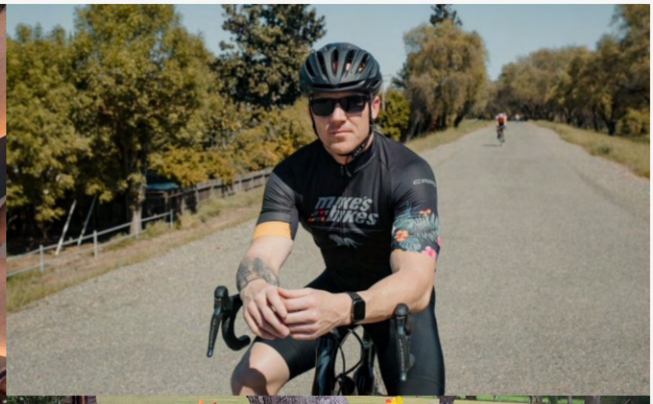
WE HAVE JUST BEGUN, JOIN US FOR THE JOURNEY

At MSF we are energized and ready to realize the opportunities 2023 presents us.

Key projects for this year include:

- **Publishing the MSF Research Roadmap**
- **Hosting our second MSF International Virtual Conference - Fall 2023**
- **Extending our Patient Advocacy programs**
- **Raising $200,000 to support our research objectives**
- **Applying for grants to help us further our mission and goals**
- **Attending events to increase awareness of Myhre syndrome**

THANK YOU TO OUR SUPPORTERS



We wouldn't exist without our community of MSF champions.

From educating about our cause, creating a fundraiser, or simply donating, our supporters make the work we do possible.

Thank you for helping us move forward research and for supporting the Myhre community as we seek to provide life-improving treatments and, one day, a cure.