MYHRE SYNDROME

An Introduction

Myhre Syndrome, first classified in 1981, is in the ultra-rare category of genetic disorders, with around 200 known cases in the world.

Raising awareness for this little-known condition was one of the motivations for a group of Myhre parents who, in 2019, set up the Myhre Syndrome Foundation.

The Myhre Syndrome Foundation is dedicated to serving Myhre Syndrome families by providing a network of hope and support and collaborating with scientific and medical communities to encourage and fund promising research.

Our goals are simple:

- To educate the public and the medical community to ensure early diagnosis and proper treatment.
- To promote safe and effective treatments through advocacy, research funding and a comprehensive patient registry.
- To provide a caring and supporting community for Myhre patients and families.

Resource Library

After a diagnosis the volume of information can often be overwhelming, but on the MSF website you'll find research papers that have been categorized with short abstract introductions to guide you through each paper. **myhresyndrome.org/library**

Common Characteristics Include:

- Problems with lungs and airways
- Problems with heart and blood vessels
- Hearing loss
- Intellectual disability and/or autism
- Short stature
- Characteristic facial features, such as small eyes, small mouth, or prominent chin
- Limited joint mobility
- Thickened skin

Early diagnosis and intervention and regular follow up are essential to ensuring children and adults with Myhre Syndrome can live their fullest lives.

Professional Advisory Board

MSF is proud to be supported by an international Professional Advisory Board (PAB) who are a network of physicians that all have experience in treating Myhre patients.

They meet monthly to progress discussions on research, treatment plans and clinical findings. They also provide topical updates that can be found on the MSF website.

Patient Registry

The Myhre Syndrome Patient Registry is an online, confidential database of standardized information about individuals with Myhre syndrome.

By sharing diagnostic and treatment history, you can help provide researchers with data necessary to conduct informed research and clinical trials.

This will ultimately identify improved treatment options and outcomes for patients. We believe this is the single most important thing we can all do to move forward research and one day find a cure.
myhresyndrome.org/patient-registry

Myhre Syndrome Clinic – Boston

The Myhre Syndrome Clinic at the Massachusetts Hospital has now seen 35 Myhre patients. Dr Angela Lin co-ordinates your visit, lasting 2-4 days, being seen by a multidisciplinary team supporting people of all ages and their families who have Myhre.

For more information, to make an appointment or talk to the genetic team visit: **massgeneral.org/children/myhre-syndrome**

Community Support

At MSF we want to support you. We're here to answer your questions, help tell your story and push forward greater knowledge and understanding of this complex condition.

We welcome the chance to talk to you so please do get in touch: **myhre@myhresyndrome.org**

You can get to know the Myhre community by reading our articles or listening to our podcasts – you'll find these and lots more on the Myhre Community section of the MSF website: **myhresyndrome.org/meet-our-community**

If you'd like to connect to a parent and Myhre adult support group, you'll find these on **Facebook** – just search for **Myhre Syndrome.**