MYHRE Syndrome is an extremely rare genetic disorder, caused by a mutation in the SMAD4 gene. This mutation is referred to as a de novo mutation because it happens by chance. It’s not caused by anything a parent did or didn't do.

Myhre Syndrome affects fewer than 200,000 people. Yet, there are less than 200 documented cases of Myhre Syndrome worldwide, and the true numbers are unknown. Currently, there’s no cure.

WHAT DOES MYHRE Syndrome CAUSE?
Because it’s a connective tissue disease, Myhre Syndrome evolves over time causing fibrosis through many systems of the body. Other common characteristics of Myhre Syndrome patients may include the following:

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

Unfortunately, some of these problems gradually get worse and can lead to potentially life-threatening complications.

IMPACT ON FAMILIES
Because Myhre Syndrome is a rare disease, the lack of scientific knowledge and quality information about it can lead to delays in diagnosis and difficulties accessing appropriate treatments. As a result, families are left isolated, resulting in heavy social burdens.

On top of that, there's a huge financial impact. A patient must somehow pay for medical bills (diagnosis, treatment, and medication), as well as multidisciplinary therapies throughout his or her life.

HOW IS IT TREATED?
The treatment of Myhre Syndrome is directed toward the specific symptoms that are present in each individual. In addition, treatment requires comprehensive, coordinated efforts of a team of specialists that include, but are limited to, primary care physicians, cardiologists, pulmonologists, geneticists, orthopedists, and ophthalmologists.

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

MYHRE Syndrome FOUNDATION
Myhre Syndrome Foundation (MSF) was founded in 2019 as a grassroots effort aimed at providing hope, resources, and advocacy for Myhre families and to support research funding and education within the medical community. By maintaining minimum operating expense, we're committed to ensuring donors have the highest level of confidence when making contributions to the foundation.

MSF is an IRS accredited 501(c)(3) non-profit charitable organization, and all donations are tax-deductible.

GIVE THE GIFT OF HOPE
Funds raised will be used to provide grants for basic science and translational research, to support the development of a patient registry and to continue expanding awareness and education throughout international patient and healthcare communities.

To make a donation or get involved, visit myhresyndrome.org/donate.