MYHRE SYNDROME CLINIC

Myhre Syndrome: What You Need to Know About the Genetic Cause

Myhre syndrome is a rare, genetic condition that happens when there is a change in one copy of the SMAD4 gene. In this handout, you will learn about the signs and the cause of Myhre syndrome. You will also learn about research at MassGeneral Hospital for Children (MGHfC) and Massachusetts General Hospital.

WHAT IS MYHRE SYNDROME?
Myhre syndrome is a genetic condition that can be diagnosed at any age. Sometimes, the features can be overlooked or confused with another condition.

WHAT ARE THE SIGNS OF MYHRE SYNDROME?
Every person who has Myhre syndrome is slightly different. Some common characteristics include:

- **Short stature** (short in height)
- **Unique facial features**, such as small eyes, small mouth or prominent (larger) chin
- Intellectual disability in some people.
- **Learning challenges** in most people
- **Hearing loss**
- **Joint problems**
- Problems with **airways** (windpipe) and **lungs**
- **Unique heart problems**
- **Thickened skin**

WHAT CAUSES MYHRE SYNDROME?
Myhre syndrome is caused by a change in 1 copy of the SMAD4 gene. In our bodies, genes act like instruction manuals for proteins. The proteins help our bodies develop, grow and work properly.

Myhre syndrome happens by chance. It is not caused by anything a parent did or did not do When a gene changes by chance, it is called a de novo mutation.

IS THERE RESEARCH BEING DONE ON MYHRE SYNDROME AT MGHFC?
Yes. Researchers at MGHfC and Massachusetts General Hospital are very excited to learn more about this rare condition. Talk with your doctor to learn more about research on Myhre syndrome.

About the Myhre Syndrome Clinic
The Myhre Syndrome Clinic at MGHfC and Mass General aims to provide specialized, comprehensive care for people who have Myhre syndrome. The team of experts led by genetics and cardiology includes colleagues in radiology, pulmonary medicine, aerodigestive medicine and rheumatology. We also conduct research on Myhre syndrome and the role of the SMAD4 gene in the development of cardiovascular disease and other health conditions.

If you are interested in learning more or participating in our research, please contact us at 617-726-1561

Did you know?
Myhre syndrome is one of several medical conditions that can happen when there is a change in the SMAD4 gene. Myhre syndrome is unique because the change causes the SMAD4 gene to work more rather than slow it down. When this happens, it is called a gain-of-function mutation.