Myhre Syndrome Foundation Patient Registry at CoRDS

March 2022 Update

MYHRE SYNDROME FOUNDATION

SANFORD RESEARCH

The Myhre Syndrome Foundation Patient Registry at CoRDS includes participants from **5 continents** and **13 countries**, including Argentina, Australia, Canada, France, Germany, Israel, Italy, Netherlands, Norway, Spain, United Kingdom and United States.

sample size: 67

55% of registry participants reported having one or more **heart conditions**. Of those responding:

- 56% have "arterial stenosis" (nonspecific term)
- 50% have hypertension
- 47% have a congenital heart defect
- 6% have cardiomyopathy
- 3% have pericardial disease

sample size: 58

57% of registry participants reported **respiratory conditions** including:

- Asthma
- Larynx and/or tracheal stenosis
- Obstructive sleep apnea
- "Restrictive pulmonary disease" (nonspecific term)
- Subglottic stenosis

sample size: 56

Of the registry participants:

61% Female
39% Male

sample size: 62

People with Myhre Syndrome reported the following **skeletal features/abnormalities**:

88% short stature

82% limited range of motion

71% small hands and feet

57% brachydactyly

39% shortened long bones

25% webbing of hands and/or feet

14% thick calvarium

10% scoliosis

10% fused vertebrae

6% hemivertebrae

sample size: 51

Myhre syndrome is caused by a change in the SMAD4 gene. Of the respondents that knew their **gene variant**:

- 51.4% had Ile500Val
- 31.4% had Arg496Cys
- 11.4% had Ile500Thr
- 5.7% had Ile500Met

sample size: 35

Age of Diagnosis:

0-5: 45.1%
6-10: 29.4%
11-20: 19.6%
21-30: 2.0%
31+: 3.9%

sample size: 51

Thank you to all registry participants!
To learn more, go to:
www.myhresyndrome.org/patient-registry

Almost 2/3rds of registry participants reported **hearing loss**. Sensorineural, conductive and mixed hearing loss have all been reported in people with Myhre Syndrome.

sample size: 55

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Myhre syndrome is caused by a change in the SMAD4 gene. There are four gene changes or variants that have been reported. Of the respondents that knew their variant:

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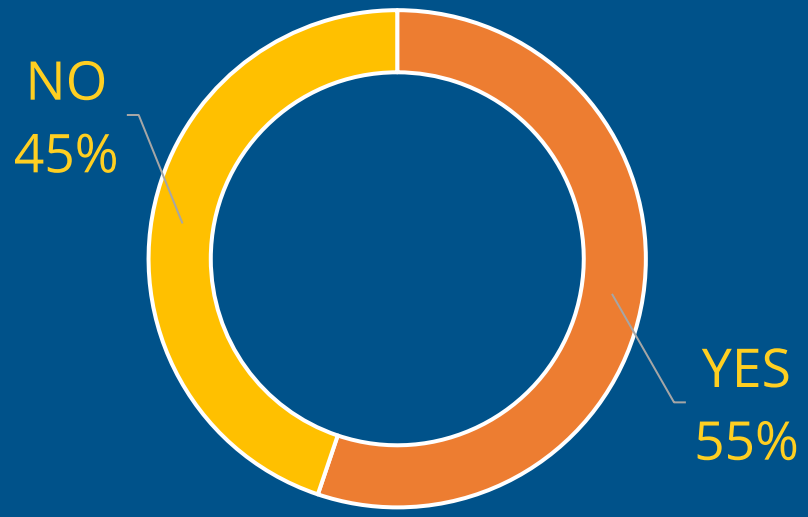
HEART CONDITIONS

55% of registry participants reported having a cardiovascular condition



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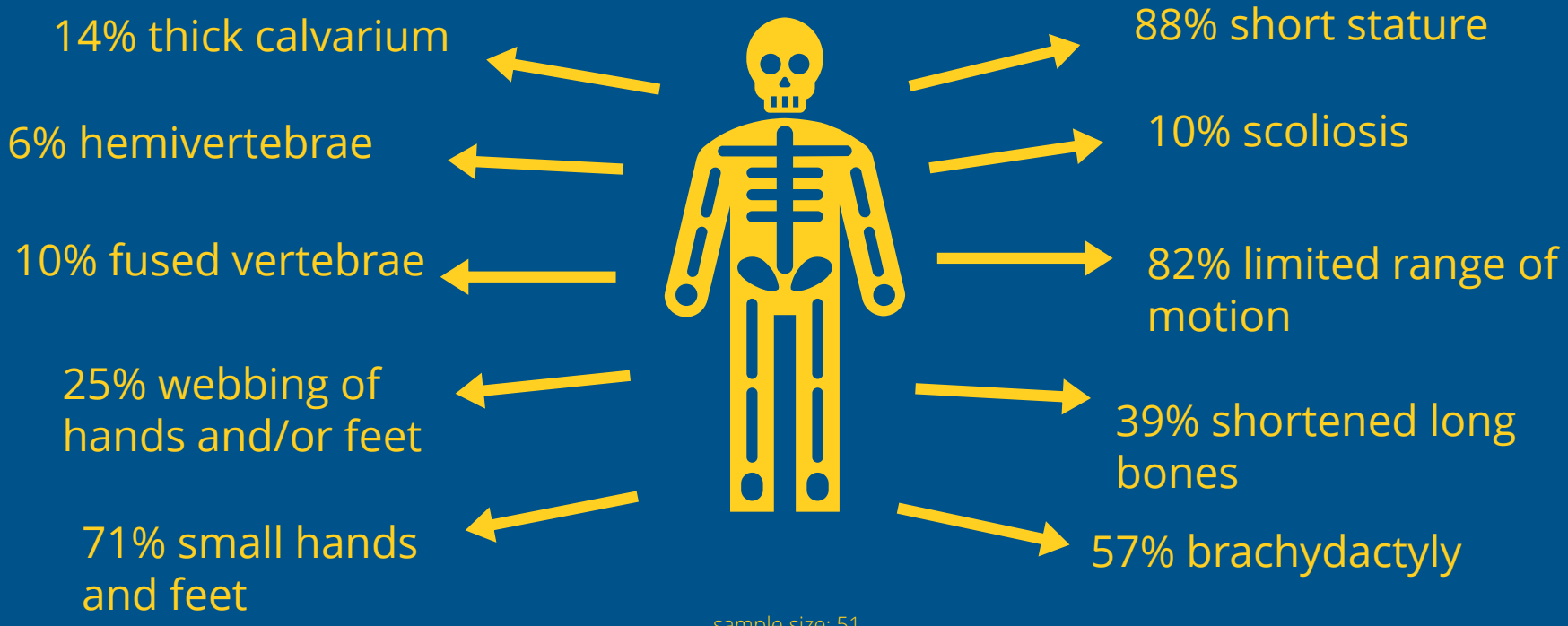

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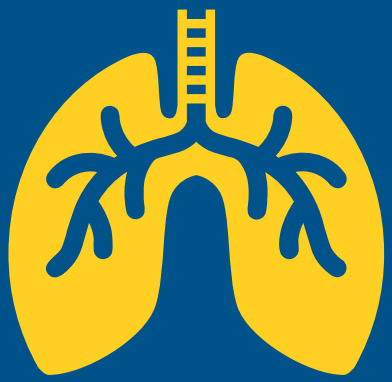

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