Background: Barth syndrome (BTHS) is a rare X-linked condition associated with male illness as a result of mutations in the Barth Syndrome Translocase (TSB4D) gene. While BTHS results in reduced energy production in all tissues, though particularly in those with greater energy needs such as the heart and skeletal muscles.

Additionally, altered mitochondrial function often affects the production and development of white blood cells. Barth syndrome (BTHS) is a rare X-linked condition characterised by the development of wide skeletal muscles.

Adolescents (n=18) results and symptoms: The most frequently reported symptom as important to improve with an effective treatment was muscle weakness (n=15, 100.0%), cardiomyopathy (n=13, 86.7%), and neutropenia (n=3, 20.0%) were the concepts included fatigue/tiredness (n=15, 100.0%), cardiomyopathy (n=13, 86.7%), muscle weakness (n=12, 80.0%), and neutropenia (n=3, 20.0%) were the concepts included fatigue/tiredness (n=15, 100.0%), cardiomyopathy (n=13, 86.7%), muscle weakness (n=12, 80.0%), and neutropenia (n=3, 20.0%) were the concepts included.

Cardiovascular signs and symptoms

- Low blood pressure (n=4, 26.6%)
- Reflux (n=2, 11.1%)
- Gagging (n=4, 22.2%)
- Vomiting (n=4, 22.2%)
- Low muscle tone (n=5, 27.8%)
- Muscle cramping (n=2, 11.1%)
- Balance/coordination problems (n=4, 22.2%)
- Inability to regulate body temperature (n=2, 11.1%)
- Mouth sores (n=5, 33.3%)
- Shortness of breath (n=5, 33.3%)
- Muscle weakness (n=12, 80.0%)

Muscle weakness and fatigue/tiredness were reported as the most important to improve in both subpopulations.

The following symptoms rated by two or more subjects were reported as the highest mean bother, worry, impact, and severity ratings:

Bother: 7 (1)
Impact: 5 (7)
Worry: 6 (1)
Severity: 4 (3)

Table 4: Adult signs and symptoms ratings

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Conclusions: Findings from the research suggest that the BTHS disease experience is characterised by frequent and impactful signs and symptoms.

In both subpopulations (i.e., adolescents and adults), the most frequently reported signs and symptoms included fatigue/tiredness, cardiomyopathy, and muscle weakness.

Barth syndrome (BTHS) is a rare X-linked condition resulting in reduced energy production in all tissues, though particularly in those with greater energy needs such as the heart and skeletal muscles.

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