Inheritest®
CARRIER SCREEN

Mucolipidosis type IV

What is mucolipidosis type IV?

Mucolipidosis type IV is an inherited disease characterized by poor growth, severe developmental delay, and progressive vision loss.[1] The symptoms of mucolipidosis type IV are attributed to an inability of the body to move lipids and other substances properly within cells, causing accumulation of these substances in cells and organs. Mucolipidosis type IV belongs to a group of diseases called lysosomal storage disorders.[2]

What are the symptoms of mucolipidosis type IV and what treatment is available?

Symptoms of mucolipidosis type IV begin within the first year of life and may include:[1,2]

- Intellectual disability
- Limited or absent word use
- Difficulty using hands and eating
- Weak muscle tone that progresses to abnormal muscle stiffness
- Inability to walk or crawl; inability to walk unaided
- Progressive retinal degeneration (breakdown of the light-sensitive layer at the back of the eye) and corneal (clear cover of the eye) clouding that leads to severe vision loss by early teens
- Iron deficiency

There is no cure for mucolipidosis type IV. Most children with mucolipidosis type IV live into adulthood.[1] Management of MLIV symptoms may include speech and physical therapy, eye surgery, eye medications, antiseizure medications, and iron supplements.[1]

An atypical form of mucolipidosis type IV with a milder clinical course has been seen in about 5% of children, usually of non-Ashkenazi Jewish descent.[1, 2]

How is mucolipidosis type IV inherited?

Mucolipidosis type IV is an autosomal recessive disease caused by mutations in the *MCOLN1* gene.[2] An individual who inherits one copy of a *MCOLN1* gene mutation is a carrier and is not expected to have related health problems. An individual who inherits two MCOLN mutations, one from each parent, is expected to be affected with mucolipidosis type IV.

If both members of a couple are carriers, the risk for an affected child is 25% in each pregnancy; therefore, it is especially important that the reproductive partner of a carrier be offered testing.

Who is at risk for mucolipidosis type IV?

Mucolipidosis type IV can occur in individuals of all ethnicities. The majority of individuals with mucolipidosis type IV (about 70%) are of Ashkenazi (Eastern European) Jewish ancestry.[2] Among Ashkenazi Jews, the carrier frequency is 1 in 89,[3] and the incidence is calculated to be approximately 1 in 31,700.

Having a relative who is a carrier or is affected can increase an individual's risk of being a carrier. Consultation with a genetics health professional may be helpful in determining carrier risk and appropriate testing.

What does a positive test result mean?

If a gene mutation is identified, an individual should speak to a physician or genetic health professional about the implications of the result and appropriate testing for the reproductive partner and at-risk family members.

What does a negative test result mean?

A negative result reduces, but does not eliminate, the possibility that an individual carries a gene mutation. The likelihood of being a carrier is also influenced by family history, medical symptoms, and other relevant test results.

Where can I get more information?

Mucolipidosis IV foundation: http://www.ml4.org/

Genetics Home Reference: http://ghr.nlm.nih.gov/condition/mucolipidosis-type-iv

References

1. Schiffmann R *et al.* Mucolipidosis IV. Available at http://www.ncbi.nlm.nih.gov/books/NBK1214/. Accessed: Feb 29, 2012.
2. Genetics Home Reference: Mucolipidosis type IV. Available at http://ghr.nlm.nih.gov/condition/mucolipidosis-type-iv. Accessed March 13, 2012.
3. Scott S *et al*. Experience with carrier screening and prenatal diagnosis for 16 Ashkenazi Jewish genetic diseases. *Hum Mut*. 2010; 31: 1-11.