## Mucolipidosis IV (MCOLN1)

Mucolipidosis type IV is a disease caused by defects in the MCOLN1 gene, which encodes for the protein mucolipin 1. This protein is found in the membranes of lysosomes and endosomes, and is involved in the transport of various molecules. Mucolipin 1 is essential for the development and maintenance of the brain and retina. Infants with the disease typically develop poor motor skills, being slow to crawl, and rarely learning to walk or speak properly. Sufferers may have muscle weakness and difficulties swallowing. Visual impairment gradually advances, usually leading to complete blindness before the age of 10. Iron deficiency may occur as well. A small number of sufferers, about 5% of the total, develop a milder form of the disease, where they may be able to walk and talk. People with mucolipidosis type IV may live for many decades, although they tend to have a shortened lifespan.

Overall, it's estimated that about 1 in 625,000 people suffer from mucolipidosis type IV, although the figure rises to 1 in 37,000 among those of Ashkenazi Jewish descent, where about 1 in 100 may be carriers. The faulty gene is inherited in an autosomal recessive manner, which typically requires both parents to be asymptomatic carriers of the faulty gene copy.

### Sources

NIH, Genetics Home Reference: MCOLN1 gene.
See http://ghr.nlm.nih.gov/gene/MCOLN1

NIH, Genetics Home Reference: Mucolipidosis Type IV.
See http://ghr.nlm.nih.gov/condition/mucolipidosis-type-iv

Recombine Website: Mucolipidosis Type IV.
See https://recombine.com/diseases/mucolipidosis-type-iv

Schiffmann, R. *et al*. (2005), "Mucolipidosis IV," in Pagon, R.A. et al., editors, *GeneReviews* [Internet]. See http://www.ncbi.nlm.nih.gov/books/NBK1214/