

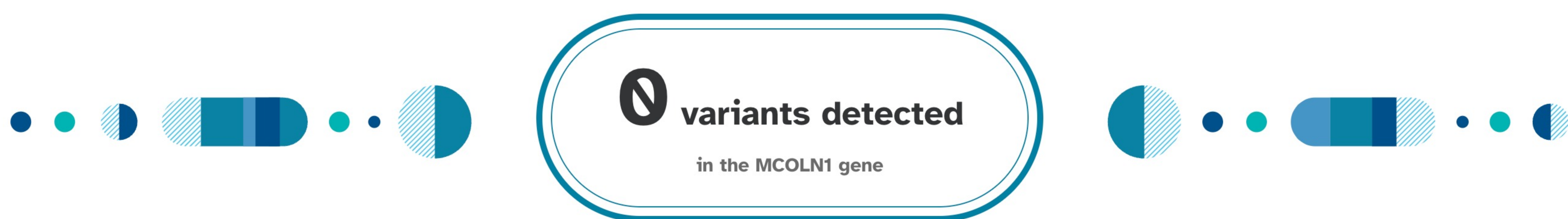
# Mucopolipidosis Type IV

Mucopolipidosis IV is a rare genetic disorder characterized by developmental delay and gradual vision loss in childhood. A person must have two variants in the MCOLN1 gene in order to have this condition.

Overview Scientific Details

### Jamie, you do not have the variant we tested.

You could still have a variant not covered by this test.



## How To Use This Test

**This test does not diagnose any health conditions.**

Please talk to a healthcare professional if this condition runs in your family, you think you might have this condition, or you have any concerns about your results.

[Review the Carrier Status tutorial](#)  
[See Scientific Details](#)

## + Intended Uses

- To test for the [IVS3-2A>G variant](#) in the [MCOLN1 gene](#).
- To identify [carrier](#) status for mucopolipidosis IV.

## - Limitations

- Does **not test** for all possible variants for the condition.
- Does **not report** if someone has two copies of a tested variant.
- Does **not cover** other types of mucopolipidosis.

## 🌐 Important Ethnicities

- This test is most relevant for people of [Ashkenazi Jewish](#) descent.
- This test does **not** include the second most common variant found in people of Ashkenazi Jewish descent.

### You are likely not a carrier.

This result may be less relevant for you because the variants that cause mucopolipidosis IV are rarely found in people of your ethnicity.

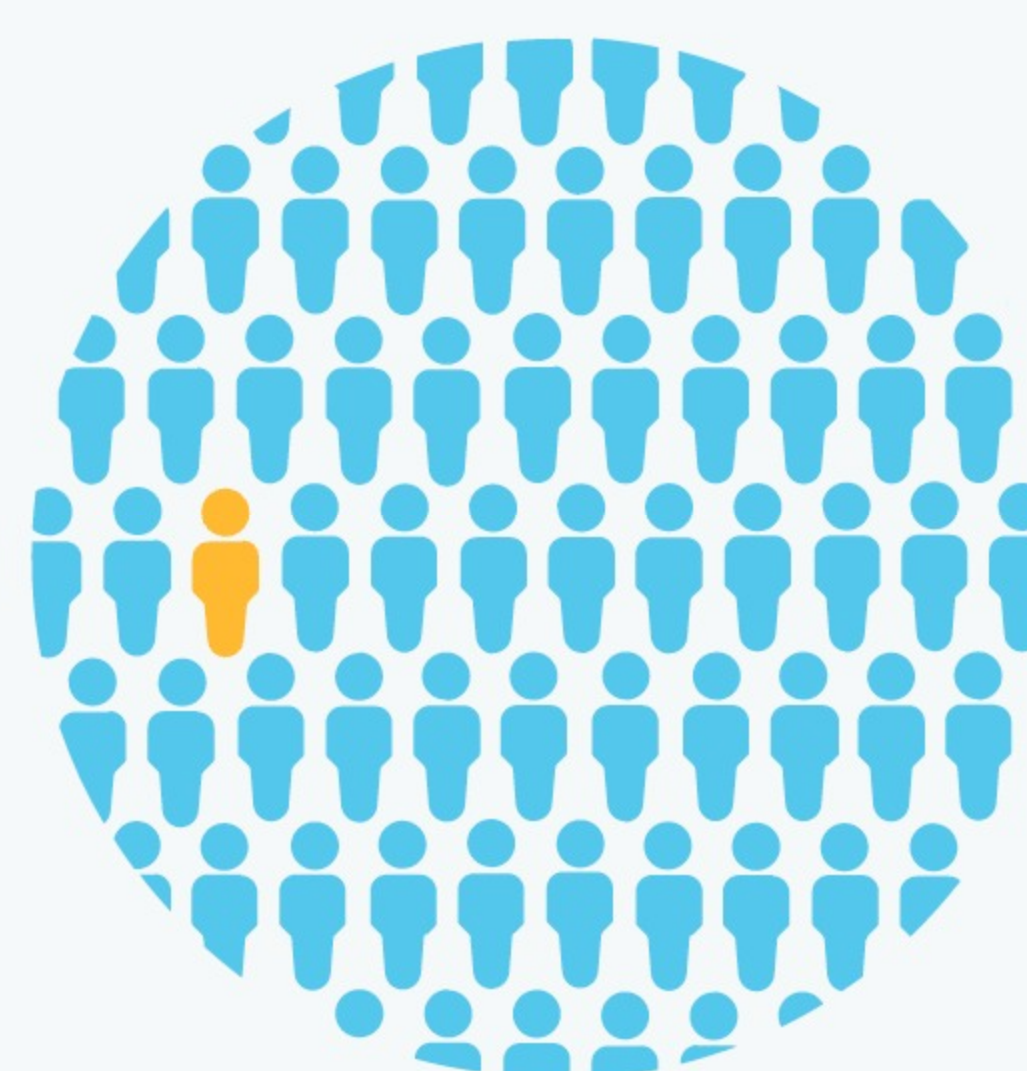


We ruled out the tested variant for mucopolipidosis IV.

This variant is most common in people of [Ashkenazi Jewish](#) descent.

### You still have a chance of being a carrier for mucopolipidosis IV.

We cannot estimate your chances because this condition is rare and not well studied in your ethnicity.



## About Mucopolipidosis Type IV

### 📅 When symptoms develop

Symptoms typically develop during infancy.

### 🌡️ Typical signs and symptoms

- Developmental disability
- Vision impairment that worsens over time
- Decreased muscle tone

### 👥 Ethnicities most affected

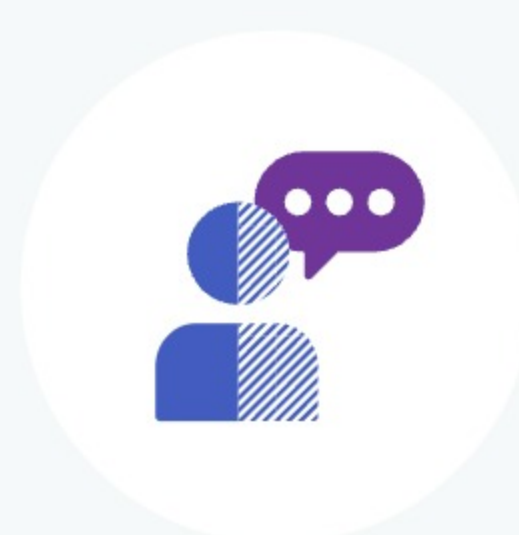
This condition is most common in people of [Ashkenazi Jewish](#) descent.

### 🩺 How it's treated

There is currently no known cure. Treatment focuses on managing symptoms and providing supportive care through speech, physical, and occupational therapy.

Read more at: [Genetics Home Reference](#) [GeneReviews](#) [National Organization for Rare Disorders](#)

### Consider talking to a healthcare professional if you are concerned about your results.



If you're starting a family, a genetic counselor can help you and your partner understand if additional testing might be appropriate.

[Connect with a GC](#)



Share your results with a healthcare professional.

[Print report](#)



Learn more about this condition and connect with support groups.

[Learn more](#)



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## Mucopolidosis Type IV

Mucopolidosis IV is a rare genetic disorder characterized by developmental delay and gradual vision loss in childhood. A person must have two variants in the MCOLN1 gene in order to have this condition.

[Overview](#)
[Scientific Details](#)

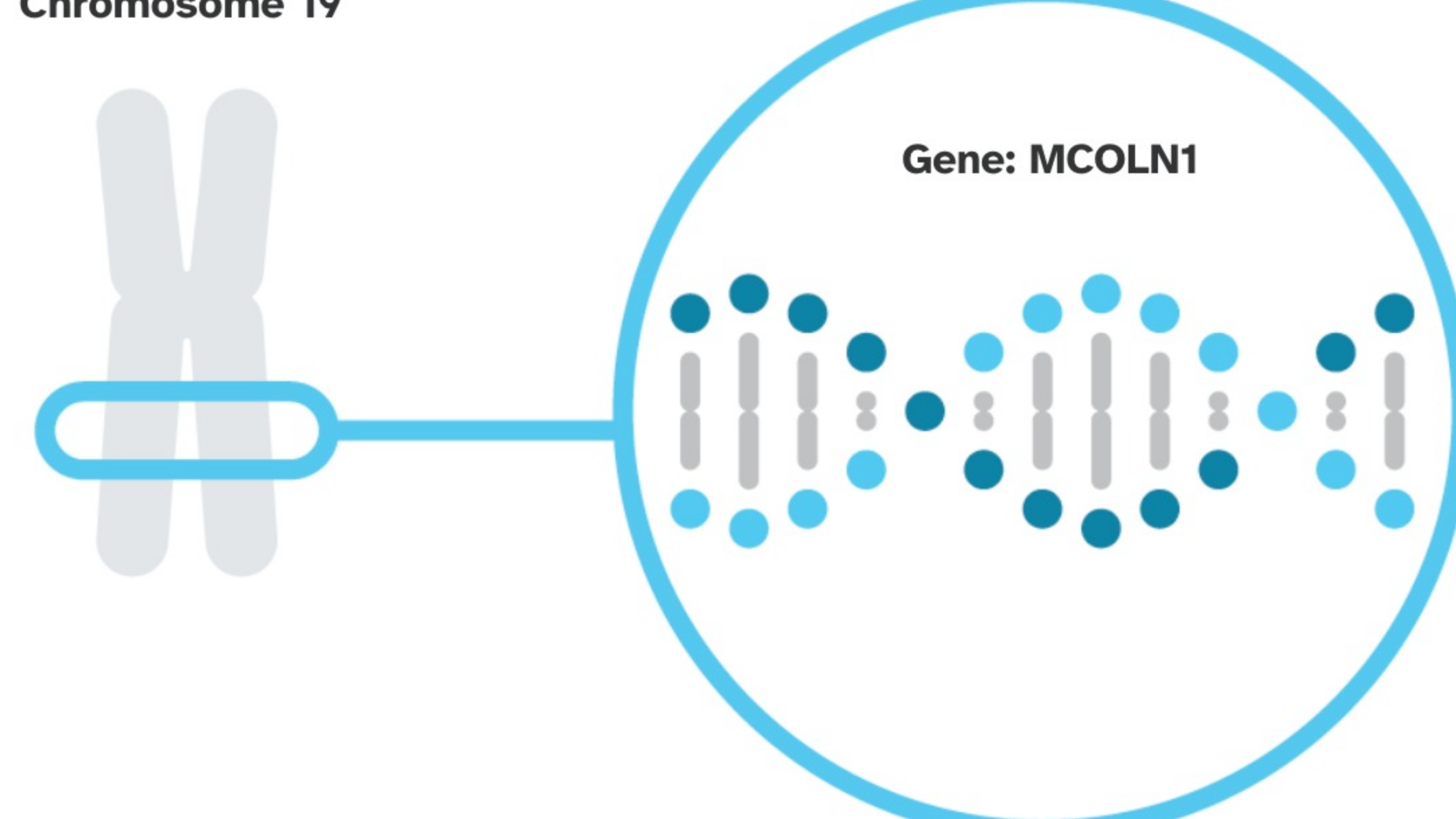
Mucopolidosis IV is caused by variants in the MCOLN1 gene.

MCOLN1

The MCOLN1 gene contains instructions for making a protein called mucopolipin-1, also known as mucopolipidin. This protein allows fats and proteins to be transported through the cell to where they can be broken down. Certain variants in MCOLN1 lead to a nonfunctional protein, causing fats and proteins to build up within affected cells.

Read more at [Genetics Home Reference](#)\*


Chromosome 19



You have no variants detected by this test.

Variants Detected

[View All Tested Markers](#)

Marker Tested	Your Genotype*	Additional Information
<b>IVS3-2A&gt;G</b> Gene: MCOLN1 Marker: <b>i4000425</b>	<b>A</b> Typical copy from one of your parents 	Typical copy from your other parent <ul style="list-style-type: none"> <li>✓ <b>Biological explanation</b></li> <li>✓ <b>Typical vs. variant DNA sequence(s)</b></li> <li>✓ <b>Percent of 23andMe customers with variant</b></li> <li>✓ <b>References [ 1 ]   <a href="#">ClinVar</a>*</b></li> </ul>

\*This test cannot distinguish which copy you received from which parent. This test also cannot determine whether multiple variants, if detected, were inherited from only one parent or from both parents. This may impact how these variants are passed down.

23andMe always reports genotypes based on the 'positive' strand of the human genome reference sequence (build 37). Other sources sometimes report genotypes using the opposite strand.

## Test Interpretation

This report provides an estimate of the chances of still being a carrier for people who do not have the variant(s) tested. This is known as the **post-test carrier risk**.

Post-test carrier risk is based on the average chance of being a carrier for a given ethnicity and the carrier detection rate of the test for a given ethnicity.

[View technical article on estimating post-test carrier risk.](#)

### Post-Test Carrier Risk

**This report provides an estimate of the post-test carrier risk for people of Ashkenazi Jewish descent only.**

- For people of partial Ashkenazi Jewish descent, post-test carrier risk is less than that for those who are fully Ashkenazi Jewish. The exact post-test risk depends on how much Ashkenazi Jewish ancestry a person has.
- Post-test risk for other ethnicities cannot be provided because sufficient data is not available.

#### Post-test carrier risk for relevant ethnicities

Ashkenazi Jewish	1 in 550	[ 1 ]
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## Test Details

### Indications for Use

The 23andMe PGS Carrier Status Test for Mucopolidosis Type IV is indicated for the detection of the IVS3-2A>G variant in the MCOLN1 gene. This test is intended to be used to determine carrier status for mucopolidosis IV in adults, but cannot determine if a person has two copies of a tested variant. The test is most relevant for people of Ashkenazi Jewish descent.

#### Special Considerations

- Carrier testing for mucopolidosis IV is recommended by ACMG for people of Ashkenazi Jewish descent considering having children. This test includes one of two variants recommended for testing by ACMG and does not include the second most common variant found in people of Ashkenazi Jewish descent.

### Test Performance Summary

#### Carrier Detection Rate & Relevant Ethnicities

The "carrier detection rate" is an estimate of the percentage of carriers for this condition that would be identified by this test. Carrier detection rate differs by ethnicity and is provided only where sufficient data is available.

Ashkenazi Jewish	77%	[ 1 ]
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#### Analytical Performance

Accuracy was determined by comparing results from this test with results from sequencing. Greater than 99% of test results were correct. While unlikely, this test may provide false positive or false negative results. For more details on the analytical performance of this test, refer to the package insert.

### Warnings and Limitations

- This test does not cover all variants that could cause this condition.\*
- This test does not diagnose any health conditions.
- Positive results in individuals whose ethnicities are not commonly associated with this condition may be incorrect. Individuals in this situation should consider genetic counseling and follow-up testing.
- Share results with your healthcare professional for any medical purposes.
- If you are concerned about your results, consult with a healthcare professional.

See the [Package Insert](#) for more details on use and performance of this test.

\* Variants not included in this test may be very rare, may not be available on our genotyping platform, or may not pass our testing standards.

## References

- Gross SJ et al. (2008). "Carrier screening in individuals of Ashkenazi Jewish descent." *Genet Med.* 10(1):54-6. \*
- Schiffmann R et al. (2005). "Mucopolidosis IV." [Updated 2015 Jul 30]. \*

## Change Log

Your report may occasionally be updated based on new information. This Change Log describes updates and revisions to this report.

Date	Change
Oct. 12, 2016	Mucopolidosis Type IV report created.



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