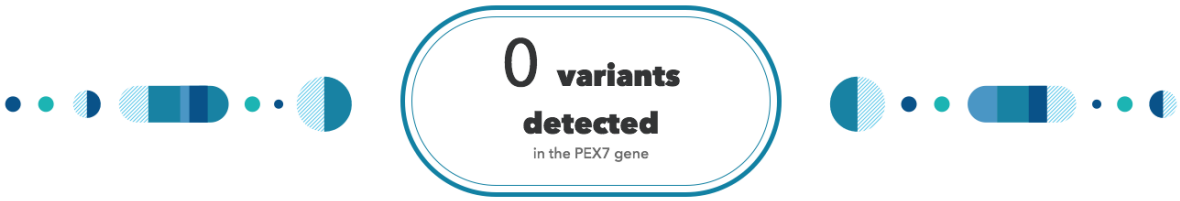
Rhizomelic Chondrodysplasia Punctata Type 1

RCDP1 is a rare genetic disorder. It is characterized by bone abnormalities, cataracts, and intellectual disability. A person must have two variants in the PEX7 gene in order to have this condition.

Erin, 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 L292X variant in the PEX7 gene.
- To identify carrier status for RCDP1.

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 subtypes of RCDP.

Important Ethnicities

- This test does **not** include a large fraction of PEX7 variants that cause RCDP1 in any ethnicity.

You are likely not a carrier.

This result may be less relevant for you because the variants that cause RCDP1 are rarely found in people of your ethnicity.

We ruled out the tested variant for RCDP1.

This variant is very rare in all ethnicities.

You still have a chance of being a carrier for RCDP1.

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

About Rhizomelic Chondrodysplasia Punctata Type 1

Also known as: A type of Peroxisome Biogenesis Disorder, Chondrodystrophia Calcificans Punctata

When symptoms develop

Symptoms are typically present at birth or develop during infancy.

How it's treated

There is currently no known cure. Treatment focuses on managing symptoms and providing supportive care through physical therapy. Treatment may include cataract removal.

Typical signs and symptoms

- Skeletal problems
- Childhood cataracts
- Intellectual disability
- Frequent lung infections

Ethnicities most affected

This condition is most common in people of European descent.

Read more at

Genetics Home Reference

GeneReviews

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.

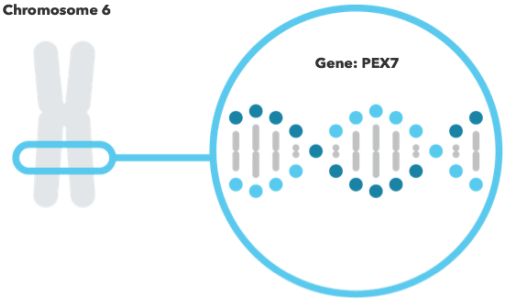
Learn more

RCDP1 is caused by variants in the PEX7 gene.

PEX7

The PEX7 gene contains instructions for making a protein called peroxisome biogenesis factor 7, also known as PEX7. This protein helps peroxisomes (compartments within cells that make and break down fats and other substances) work properly. Certain variants in PEX7 disrupt peroxisome function by preventing the production of fats that are required for normal development.

Read more at Genetics Home Reference



You have no variants detected by this test.

Variants Detected	View All Tested Markers
0	1

Marker Tested	Your Genotype*		Additional Information
L292X Gene: PEX7 Marker: i3002517	**T** Typical copy from one of your parents	**T** Typical copy from your other parent	> **Biological explanation** > **Typical vs. variant DNA sequence(s)** > **Percent of 23andMe customers with variant** > **References** [1 , 2 , 4 , 5 , 6] | ClinVar

*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

Post-test carrier risk for RCDP1 is the chance of still being a carrier for the condition if you do not have the variant tested. This chance depends on how common it is to be a carrier for RCDP1 and whether the variants we tested tend to be found in people of your ethnicity.

Because you do not have the variant we tested, your chances of still being a carrier are lower than for someone who has not been tested. However, we cannot provide an exact estimate because the information needed to calculate post-test carrier risk is not available for your ethnicity.

Test Details

Indications for Use

The 23andMe PGS Carrier Status Test for Rhizomelic Chondrodysplasia Punctata Type 1 is indicated for the detection of the L292X variant in the PEX7 gene. This test is intended to be used to determine carrier status for RCDP1 in adults, but cannot determine if a person has two copies of a tested variant.

Special Considerations

- This test does not include a large fraction of PEX7 variants that cause RCDP1 in any ethnicity.
- There are currently no professional guidelines in the U.S. for carrier testing for this condition.

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.

European	about 50%	[2 , 3]

Analytical Performance

Accuracy was determined by comparing results from this test with results from sequencing for 49 samples with known variant status. 49 out of 49 genotype results were correct. Fewer than 1 in 100,000 samples may receive a **Not Determined** result. This can be caused by random test error or unexpected DNA sequences that interfere with the test. It can also be caused by having two copies of a variant tested.

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

1. Braverman N et al. (1997). "Human PEX7 encodes the peroxisomal PTS2 receptor and is responsible for rhizomelic chondrodysplasia punctata." Nat Genet. 15(4):369-76.
2. Braverman N et al. (2002). "Mutation analysis of PEX7 in 60 probands with rhizomelic chondrodysplasia punctata and functional correlations of genotype with phenotype." Hum Mutat. 20(4):284-97.
3. Braverman NE et al. (1993). "Rhizomelic Chondrodysplasia Punctata Type 1"
4. Brites P et al. (1998). "Molecular basis of rhizomelic chondrodysplasia punctata type I: high frequency of the Leu-292 stop mutation in 38 patients." J Inherit Metab Dis. 21(3):306-8.
5. Motley AM et al. (2002). "Mutational spectrum in the PEX7 gene and functional analysis of mutant alleles in 78 patients with rhizomelic chondrodysplasia punctata type 1." Am J Hum Genet. 70(3):612-24.
6. Seaver LH et al. (2009). "ACMG practice guideline: genetic evaluation of short stature." Genet Med. 11(6):465-70.