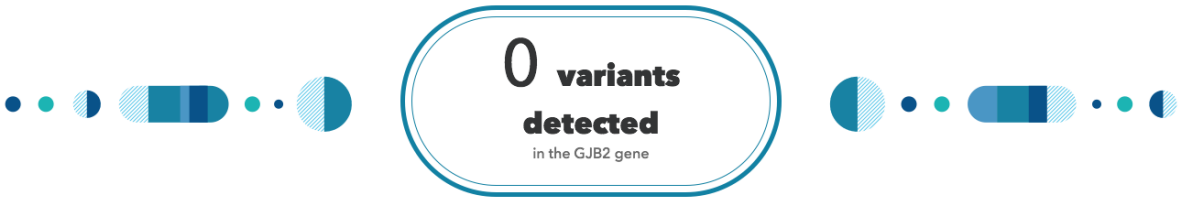
Nonsyndromic Hearing Loss and Deafness, DFNB1 (GJB2-Related)

DFNB1 is a type of inherited hearing loss that can be moderate to severe. Symptoms are typically noticed in newborns. A person must have two variants in the GJB2 gene in order to have GJB2-related DFNB1.

Erin, you **do not have the variants** 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

- Tests for **multiple variants** in the GJB2 gene.
- To identify carrier status for DFNB1.

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 test** for variants that can cause autosomal dominant or other forms of hearing loss.

Important Ethnicities

- This test is most relevant for people of **Ashkenazi Jewish** and **European** descent.
- This test does **not** include the majority of GJB2 variants that cause DFNB1 in people of East Asian descent.

You are likely not a carrier.

We ruled out the tested variants for DFNB1.

These variants are most common in people of **Ashkenazi Jewish or European** descent.

You still have a chance of being a carrier for DFNB1.

We cannot estimate your chances because sufficient data is not available.

About Nonsyndromic Hearing Loss and Deafness, DFNB1 (GJB2-Related)

Also known as: Connexin 26-Related Sensorineural Hearing Loss

When symptoms develop

Symptoms are typically present at birth.

How it's treated

There is currently no known cure. Treatment options include hearing aids, cochlear implants, and educational programs for people with hearing loss.

Typical signs and symptoms

- Moderate to profound hearing loss at birth

Ethnicities most affected

This condition affects people of all ethnicities, but is best studied in people of Ashkenazi Jewish, European, and East Asian 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.

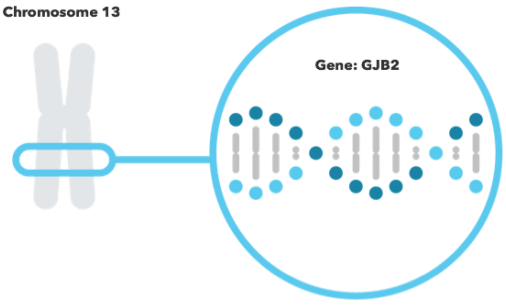
Learn more

DFNB1 is caused by variants in the GJB2 gene.

GJB2

The GJB2 gene contains instructions for making a protein called gap junction beta 2, also known as connexin 26. This protein helps transport potassium ions and other molecules between cells. Proper movement of potassium ions in the inner ear is needed for the brain to process sound. Certain variants in the GJB2 gene impair the function of this protein.

Read more at Genetics Home Reference



You have no variants detected by this test.

Variants Detected	View All Tested Markers
0	2

Marker Tested	Your Genotype*		Additional Information
35delG Gene: GJB2 Marker: i4000434	**C** Typical copy from one of your parents	**C** Typical copy from your other parent	> **Biological explanation** > **Typical vs. variant DNA sequence(s)** > **Percent of 23andMe customers with variant** > **References** [4 , 9 , 12 , 15] | ClinVar
167delT Gene: GJB2 Marker: i4000435	**A** Typical copy from one of your parents	**A** Typical copy from your other parent	> **Biological explanation** > **Typical vs. variant DNA sequence(s)** > **Percent of 23andMe customers with variant** > **References** [9 , 10 , 12] | 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

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, European, and East Asian descent only.

- For people with partial ethnicity from one or more groups mentioned above, post-test carrier risk depends on the exact mixture in the person's background.
- 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 63	[5 , 10]
European	1 in 150	[3]
East Asian	1 in 30	[2 , 6 , 14]

Test Details

Indications for Use

The 23andMe PGS Carrier Status Test for Nonsyndromic Hearing Loss and Deafness, DFNB1 (GJB2-Related) is indicated for the detection of two variants in the GJB2 gene. This test is intended to be used to determine carrier status for DFNB1 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 and European descent.

Special Considerations

- The severity of hearing loss can vary, but there are no other symptoms associated with this condition.
- 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.

Ashkenazi Jewish	76%	[10]
European	79%	[8]
East Asian		[2 , 6 , 14]

Analytical Performance

Accuracy was determined by comparing results from this test with results from sequencing for 103 samples with known variant status. 103 out of 103 genotype results were correct. About 1 in 13,000 samples may receive a **Not Determined** result for one or more variants included in this test. 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. Chan DK et al. (2014). "GJB2-associated hearing loss: systematic review of worldwide prevalence, genotype, and auditory phenotype." Laryngoscope. 124(2):E34-53.
2. Chen Y et al. (2015). "SNaPshot reveals high mutation and carrier frequencies of 15 common hearing loss mutants in a Chinese newborn cohort." Clin Genet. 87(5):467-72.
3. Cohn ES et al. (1999). "Clinical phenotype and mutations in connexin 26 (DFNB1/GJB2), the most common cause of childhood hearing loss." Am J Med Genet. 89(3):130-6.
4. D'Andrea P et al. (2002). "Hearing loss: frequency and functional studies of the most common connexin26 alleles." Biochem Biophys Res Commun. 296(3):685-91.
5. Dong J et al. (2001). "Nonradioactive detection of the common Connexin 26 167delT and 35delG mutations and frequencies among Ashkenazi Jews." Mol Genet Metab. 73(2):160-3.
6. Han SH et al. (2008). "Carrier frequency of GJB2 (connexin-26) mutations causing inherited deafness in the Korean population." J Hum Genet. 53(11-12):1022-8.
7. Hwa HL et al. (2003). "Mutation spectrum of the connexin 26 (GJB2) gene in Taiwanese patients with prelingual deafness." Genet Med. 5(3):161-5.
8. Kelley PM et al. (1998). "Novel mutations in the connexin 26 gene (GJB2) that cause autosomal recessive (DFNB1) hearing loss." Am J Hum Genet. 62(4):792-9.
9. Kenneson A et al. (2002). "GJB2 (connexin 26) variants and nonsyndromic sensorineural hearing loss: a HuGE review." Genet Med. 4(4):258-74.
10. Lerer I et al. (2000). "Contribution of connexin 26 mutations to nonsyndromic deafness in Ashkenazi patients and the variable phenotypic effect of the mutation 167delT." Am J Med Genet. 95(1):53-6.
11. Liu XZ et al. (2002). "The prevalence of connexin 26 (GJB2) mutations in the Chinese population." Hum Genet. 111(4-5):394-7.
12. Morell RJ et al. (1998). "Mutations in the connexin 26 gene (GJB2) among Ashkenazi Jews with nonsyndromic recessive deafness." N Engl J Med. 339(21):1500-5.
13. Smith RJH et al. (1993). "Nonsyndromic Hearing Loss and Deafness, DFNB1"
14. Taniguchi M et al. (2015). "Carrier frequency of the GJB2 mutations that cause hereditary hearing loss in the Japanese population." J Hum Genet.
15. Zelante L et al. (1997). "Connexin26 mutations associated with the most common form of non-syndromic neurosensory autosomal recessive deafness (DFNB1) in Mediterraneans." Hum Mol Genet. 6(9):1605-9.