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

DFNB1 is an inherited condition characterized by mild to profound hearing loss that is typically present from birth. People with GJB2-related DFNB1 most often have two variants or two copies of a variant in the GJB2 gene.

Jamie , you have **two copies of a variant** we tested.

People with this result may have DFNB1-related hearing loss, which is typically present from birth and does not usually progress over time. Your result may also be relevant if you're considering having children.

Variant detected

in the GJB2 gene

This test **does not diagnose** DFNB1. If this result is unexpected, please discuss this report with a healthcare professional.

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

See Frequently Asked Questions

Intended Uses

- Tests for **multiple variants** in the GJB2 gene.
- To identify carrier status for DFNB1.
- Informs individuals with certain combinations of variants in the GJB2 gene that people with their result may have hearing loss related to DFNB1.

Limitations

- Does **not test** for all possible variants for the condition.
- Does **not report** if someone has two copies of the W24X or R143W variant.
- Does **not test** for variants that can cause autosomal dominant or other forms of hearing loss.

Important Ethnicities

- This test is relevant for people of many ethnicities, including people of **Ashkenazi Jewish**, **East/Southeast Asian**, **European**, and **Ghanaian** descent. It may also be relevant for people of **Hispanic/Latino**, **Northern African/Middle Eastern**, and **South Asian** descent.

People with your genetic result may have DFNB1-related hearing loss.

Your result may also be relevant if you're considering having children.

Please talk to a healthcare professional if you have concerns.

People with your result may have DFNB1-related hearing loss, which is typically present from birth and does not usually progress over time. However, not everyone with two copies of a variant in the GJB2 gene will experience noticeable hearing loss. A healthcare professional can answer questions you may have about your results. See Frequently Asked Questions for more information.

Your results may be relevant for your family.

Because you have two copies of a variant, you will pass a variant on to each of your children. **If your partner is a carrier for DFNB1**, each child may have a **50% chance** of having this condition. Your relatives may also wish to consider testing if they plan to have children.

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.

Typical signs and symptoms

- Mild to profound hearing loss typically present from birth
- Unlike some other forms of hearing loss, DFNB1 does not impact balance or movement.

Ethnicities most affected

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

How it's managed

Although DFNB1-related hearing loss is not reversible, support options include hearing aids, cochlear implants, and educational programs. An audiologist can help assess hearing and discuss support options.

Read more at: MedlinePlus GeneReviews

It is important to talk to a healthcare professional if you are concerned about your results.

People with your result may have DFNB1-related hearing loss. It is important to consult with a healthcare professional if you have any concerns about your result.

Print report

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

Find a genetic counselor

See our Frequently Asked Questions for more information.

FAQs

Learn more about this condition and connect with support groups.

Learn more

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DFNB1 is an inherited condition characterized by mild to profound hearing loss that is typically present from birth. People with GJB2-related DFNB1 most often have two variants or two copies of a variant in the GJB2 gene.

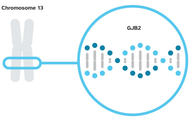
Overview | Scientific Details | Frequently Asked Questions

DFNB1 is most often 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 MedlinePlus*



You have two copies of a variant we tested.

Variants Detected | View All Tested Markers

Marker Tested	Your Genotype*			Additional Information
c.35delG **Gene:** GJB2 **Marker:** **i4000434**	(--) Variant copy from one of your parents		(--) Variant copy from your other parent	Biological explanation Typical vs. variant DNA sequence(s) Percent of 23andMe customers with variant References [23 , 26 , 34 , 41 , 6] | ClinVar*
W24X **Gene:** GJB2 **Marker:** **rs104894396**	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 [2 , 21 , 28 , 34] | ClinVar*
M34T **Gene:** GJB2 **Marker:** **rs35687622**	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 [17 , 20 , 24 , 31 , 34 , 7] | ClinVar*
V37I **Gene:** GJB2 **Marker:** **rs72474224**	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 [15 , 16 , 3 , 31 , 34 , 36 , 7] | ClinVar*
c.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 [19 , 26 , 30 , 34] | ClinVar*
c.235delC **Gene:** GJB2 **Marker:** **rs80338943**	G Typical copy from one of your parents		G Typical copy from your other parent	Biological explanation Typical vs. variant DNA sequence(s) Percent of 23andMe customers with variant References [11 , 27 , 3 , 32 , 36 , 42] | ClinVar*
S139N **Gene:** GJB2 **Marker:** **rs76434661**	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 [10 , 22 , 29 , 34 , 35 , 39 , 40 , 8] | ClinVar*
R143W **Gene:** GJB2 **Marker:** **rs80338948**	G Typical copy from one of your parents		G Typical copy from your other parent	Biological explanation Typical vs. variant DNA sequence(s) Percent of 23andMe customers with variant References [14 , 25 , 32 , 34 , 36] | 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 and Clinical Performance

Carrier frequency and carrier detection rate are *most* relevant for people without a variant detected.

For people who do not have the variant(s) tested, it may be possible to calculate an estimate of post-test carrier risk (the chances of still being a carrier) using information in this table. **View technical article on estimating post-test carrier risk.**

For people with one or more variants that could not be determined, their remaining chances of being a carrier may be similar to or less than the carrier frequency in people of their ethnicity.

Carrier frequency and carrier detection rate

This report provides two pieces of information to help interpret certain genetic results.

- **Carrier frequency** is the average chance of being a carrier for this condition. For example, a carrier frequency of 1 in 59 means that 1 out of every 59 people is expected to be a carrier for this condition.
- **Carrier detection rate** is an estimate of the percentage of carriers for this condition that would be identified by this test. For example, if the carrier detection rate is 86%, then our test is able to detect 86% of carriers for this condition. In cases where ranges are provided, the estimated carrier detection rate may depend on the region or country of ancestry.

Carrier frequency and carrier detection rate vary by ethnicity and are provided only where sufficient data is available.

Ethnicity	Carrier frequency	Carrier detection rate	References
Ashkenazi Jewish	1 in 20	93%	[19 , 9]
East/Southeast Asian	Up to 1 in 4	75-99%	[16 , 37 , 38 , 5]
European	1 in 20	75-98%	[12 , 13 , 37]
Ghanaian	Not available	91%	[37]
Hispanic/Latino	Not available	51-99%	[37]
Northern African/Middle Eastern	Not available	74-99%	[37]
South Asian	Not available	48-92%	[37]

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 eight variants in the GJB2 gene. This test is intended to be used to determine carrier status for DFNB1 in adults. This report also describes if a result is associated with personal risk of having hearing loss related to DFNB1, but it does not describe a person's overall risk of having DFNB1-related hearing loss. This test is relevant for people of many ethnicities.

Special Considerations

- The degree of hearing loss can vary, but there are no other symptoms associated with this condition.
- Some of the variants included in this report — including M34T and V37I — tend to be associated with milder hearing loss. In addition, some people with two GJB2 variants do not have any noticeable hearing loss.
- Most people with DFNB1 have two variants in the GJB2 gene. However, some people with the condition have one variant in the GJB2 gene and a second variant not tested (a deletion) in the GJB6 gene.
- There are currently no professional guidelines in the U.S. for carrier testing for this condition.

Test Performance Summary

Clinical Performance

This test is expected to detect the majority of DFNB1 carriers of many ethnicities, including Ashkenazi Jewish, East/Southeast Asian, European, Ghanaian, Hispanic/Latino, Northern African/Middle Eastern, and South Asian. See the Test Interpretation and Clinical Performance section above for additional details about carrier detection rates.

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

1. **Adadey SM et al. (2020). "Connexin Genes Variants Associated with Non-Syndromic Hearing Impairment: A Systematic Review of the Global Burden." Life (Basel). 10(11).** *
2. **Alvarez A et al. (2005). "High prevalence of the W24X mutation in the gene encoding connexin-26 (GJB2) in Spanish Romani (gypsies) with autosomal recessive non-syndromic hearing loss." Am J Med Genet A. 137A(3):255-8.** *
3. **Bruzzone R et al. (2003). "Loss-of-function and residual channel activity of connexin26 mutations associated with non-syndromic deafness." FEBS Lett. 533(1-3):79-88.** *
4. **Chan DK et al. (2014). "GJB2-associated hearing loss: systematic review of worldwide prevalence, genotype, and auditory phenotype." Laryngoscope. 124(2):E34-53.** *
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6. **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.** *
7. **Dahl HH et al. (2013). "Etiology and audiological outcomes at 3 years for 364 children in Australia." PLoS One. 8(3):e59624.** *
8. **Dodson KM et al. (2011). "Vestibular dysfunction in DFNB1 deafness." Am J Med Genet A. 155A(5):993-1000.** *
9. **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):150-3.** *
10. **Pleishman SJ et al. (2006). "The structural context of disease-causing mutations in gap junctions." J Biol Chem. 281(39):28958-63.** *

See all references

Change Log

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

Date	Change
May 24, 2022	Six variants were added to the report: W24X (rs104894396), M34T (rs35687622), V37I (rs72474224), c.235delC (rs80338943), S139N (rs76434661), and R143W (rs80338948). Customers who have these variants will see them detected in their result, and see updated content in their report. Customers with certain combinations of variants will now receive information on whether people with their result may have DFNB1-related hearing loss. In addition, the test can now report if a customer has two copies of some tested variants. Carrier detection rates and carrier frequencies were added or updated for several ethnicities, and post-test carrier risk information removed. Customized language based on customers' self-reported ethnicity was removed. Ethnicity-specific information can be found on the Scientific Details page.
Dec. 9, 2019	Information specific to people of South Asian descent was added. Customers may now see carrier detection rate information specific to that ancestry.
March 2, 2018	The carrier frequency and carrier detection rate were updated for customers who self-report having Ashkenazi Jewish ancestry. The chances of still being a carrier were also updated for customers with no variants detected who self-report having Ashkenazi Jewish ancestry. The carrier detection rate was updated for customers who self-report having European ancestry. The chances of still being a carrier were also updated for customers with no variants detected who self-report having European ancestry.
Feb. 16, 2016	Due to improvements in data analysis, some customers who previously received a "Not Determined" result for i4000435 may see a genotype at this marker. This may also update the overall report result for these customers.
Oct. 21, 2015	Nonsyndromic Hearing Loss and Deafness, DFNB1 (GJB2-Related) report created.

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

What does this test do?

This test looks for eight variants in the GJB2 gene linked to DFNB1.

People with one GJB2 variant can pass the variant on to their children.

People with two GJB2 variants or two copies of a variant may have DFNB1-related hearing loss, which is typically present from birth. They are also expected to pass a variant on to each of their children.

This test does not include all possible genetic variants linked to DFNB1.

Is this answer helpful? Yes No

What does this test **not** do?

This test does not diagnose DFNB1. Only a healthcare professional can do that.

This test does not include all possible variants linked to DFNB1.

Is this answer helpful? Yes No

My report says **two copies of a variant** were detected. What does this mean?

This means you have two copies of a genetic variant we tested. You inherited one variant from each of your parents.

People with this result may have hearing loss related to DFNB1, which is typically present from birth and does not usually progress over time. However, not everyone with two copies of a variant in the GJB2 gene will experience noticeable hearing loss. It's important to talk with a healthcare professional if you are concerned about your result.

This result also means you will pass a variant on to each of your children.

Is this answer helpful? Yes No

My report says people with my result **may have DFNB1-related hearing loss**. What does this mean?

DFNB1-related hearing loss is characterized by mild to profound hearing loss that is typically present from birth and does not usually progress over time. However, not everyone with two copies of a variant in the GJB2 gene will experience noticeable hearing loss. In people who do experience DFNB1-related hearing loss, the degree of hearing loss can vary, depending on which variants are causing the condition and other factors. For example, the V37I and M34T variants included in this report tend to be associated with milder hearing loss, and some people with these variants do not have any hearing loss. There are no other symptoms associated with this condition.

A healthcare professional can answer any questions you may have about your results.

Is this answer helpful? Yes No

The report says the test is relevant for people of many ethnicities. What if I'm not one of those ethnicities?

The genetic variants we tested are common in people of many ethnicities, including people of Ashkenazi Jewish, East/Southeast Asian, European, and Ghanaian descent. These variants are also frequently found in people of Hispanic/Latino, Northern African/Middle Eastern, and South Asian descent. However, these variants can be found in people of other ethnicities, too.

People with your genetic result — regardless of their ethnicity — may have hearing loss related to DFNB1. In addition, you will pass a variant on to each of your children.

Is this answer helpful? Yes No

My report says I have **two copies of a variant** linked to DFNB1. What are some things I could do?

People with your genetic result may have hearing loss related to DFNB1. It is important to talk to a healthcare professional if you are concerned about your result.

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

You may also want to share your results with other family members. Because you share DNA with your relatives, your genetic result could also be relevant for them.

Is this answer helpful? Yes No

How could my result affect my children?

Because you have two copies of a variant, you will pass a variant on to each of your children. If your partner is a carrier for DFNB1, each of your children may have a 50% chance of having this condition.

A genetic counselor can help you and your partner understand if additional testing might be appropriate. **Learn more about genetic counseling.**

Is this answer helpful? Yes No

Have more questions? **Check out our Customer Care Help Center.**

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