Nonsyndromic Hearing Loss and Deafness, DFN1 (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 DFN1.

Erin, you **do not have the variants** we tested.

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

0 **variants** detected
in the GJB2 gene

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**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 DFN1.

**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 DFN1 in people of East Asian descent.

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You are likely not a carrier.

We ruled out the tested variants for DFN1.

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

You still have a chance of being a carrier for DFN1.

We cannot estimate your chances because sufficient data is not available.
About Nonsyndromic Hearing Loss and Deafness, DFN1 (GJB2-Related)

Also known as: Connexin 26-Related Sensorineural Hearing Loss

<table>
<thead>
<tr>
<th>When symptoms develop</th>
<th>Typical signs and symptoms</th>
<th>Ethnicities most affected</th>
</tr>
</thead>
<tbody>
<tr>
<td>Symptoms are typically present at birth.</td>
<td>Moderate to profound hearing loss at birth</td>
<td>This condition affects people of all ethnicities, but is best studied in people of Ashkenazi Jewish, European, and East Asian descent.</td>
</tr>
</tbody>
</table>

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.

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.

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.

<table>
<thead>
<tr>
<th>Marker Tested</th>
<th>Your Genotype*</th>
<th>Additional Information</th>
</tr>
</thead>
<tbody>
<tr>
<td>35delG</td>
<td>C</td>
<td>Biological explanation</td>
</tr>
<tr>
<td>Gene: GJB2</td>
<td>Typical copy from your parent</td>
<td>Typical vs. variant DNA sequence(s)</td>
</tr>
<tr>
<td>Marker: i4000434</td>
<td></td>
<td>Percent of 23andMe customers with variant</td>
</tr>
<tr>
<td></td>
<td></td>
<td>References [4, 9, 12, 15]</td>
</tr>
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</table>

*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

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

<table>
<thead>
<tr>
<th>Ethnicity</th>
<th>Carrier Risk</th>
<th>References</th>
</tr>
</thead>
<tbody>
<tr>
<td>Ashkenazi Jewish</td>
<td>1 in 63</td>
<td>[5, 10]</td>
</tr>
<tr>
<td>European</td>
<td>1 in 150</td>
<td>[3]</td>
</tr>
<tr>
<td>East Asian</td>
<td>1 in 30</td>
<td>[2, 6, 14]</td>
</tr>
</tbody>
</table>
Indications for Use

The 23andMe PGx 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.

<table>
<thead>
<tr>
<th>Ethnicity</th>
<th>Carrier Detection Rate</th>
</tr>
</thead>
<tbody>
<tr>
<td>Ashkenazi Jewish</td>
<td>76% [10]</td>
</tr>
<tr>
<td>European</td>
<td>79% [8]</td>
</tr>
<tr>
<td>East Asian</td>
<td>[2, 6, 14]</td>
</tr>
</tbody>
</table>

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.
References


