

## Nonsyndromic Hearing Loss and Deafness, DFNB1 testing via *Connexin 26/GJB2* Gene Sequencing

About 1/1000 (0.1%) children are born with profound hearing loss. Approximately 60% of these cases are caused by abnormalities in deafness genes. Of all deafness genes, mutations in the *Connexin 26/GJB2* gene are by far the most common. Up to 50% of mutations responsible for congenital deafness are found within this gene. The carrier rate in the general population for a recessive deafness-causing *GJB2* mutation is roughly 3%. *GJB2* mutations are responsible for recessive Nonsyndromic Hearing Loss and Deafness DFNB1.

The entire coding region of the *GJB2* gene is contained within one long exon (exon 2). Our test involves the complete sequencing in both directions of the coding region plus additional flanking DNA. Sequence analysis of the coding region can detect both mutations in greater than 95% of individuals with DFNB1. Turn around time for our test is generally less than 2 weeks.

### Specimen Requirements

- Collect 2-5 ml of whole blood in EDTA (purple top tube) or ACD (yellow top tube). 5 ml is the preferred volume.
- Only one blood tube is required for multiple tests.
- Ship whole blood specimens at room temperature.
- Do not freeze blood.
- During hot weather, include a frozen ice pack in the shipping container. Do not allow the ice pack to come in direct contact with the specimen tube.
- In cold weather, include an unfrozen ice pack to help moderate extremes in temperature. The DNA in whole blood is stable for at least 48 hours at 21°C, 5-7 days at 4°C.

<b>Sequence analysis of Connexin 26 (<i>GJB2</i>) gene</b>				<b>\$290.00</b>
<b>Molec Diag, Ascertainment</b>	<b>83890</b>	<b>x1</b>	<b>\$30.00</b>	
<b>Molec Diag, Isolation</b>	<b>83891</b>	<b>x1</b>	<b>\$30.00</b>	
<b>Molecular Diag, Amplif</b>	<b>83898</b>	<b>x 2 @\$50</b>	<b>\$100.00</b>	
<b>Mutat Id By Seq, Seg</b>	<b>83904</b>	<b>x 2 @\$30</b>	<b>\$60.00</b>	
<b>Molecular Diag, Separation</b>	<b>83894</b>	<b>x1</b>	<b>\$40.00</b>	
<b>Interpretation And Report</b>	<b>83912</b>	<b>x1</b>	<b>\$30.00</b>	<b>\$290.00</b>

**Accreditation Info. CLIA ID #: 52D1027685** (expires 1/18/07) (CAP#: 7185561, AU ID: 1407125 expires 12/20/06)

### **Ship to:**

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