



UW Medicine
Department of Laboratory Medicine

Hereditary Hearing Loss

[Laboratory Medicine Home Page](#)

For Technical & Clinical Information:

Karen Stephens, PhD
Director of Genetics
(206) 543-8285

**University of
Washington**

**Department of
Laboratory Medicine**

*For additional ordering
information and instructions,
contact the Community
Services office at (206) 598-
6066 or (800) 713.5198*

On August 1st, 2005, the Department of Laboratory Medicine will replace the current Deafness DNA Screen with the following test.

This test has a detection rate of 98% and involves DNA sequencing of the entire coding region and associated intron junctions of the coding region of the GJB2 gene that encodes the connexin 26 protein. Additional reflex testing will be performed in about 3-5% of referrals, which will bring the detection rate to near 100%. These will include cases referred for diagnostic testing in which only one GJB2 mutation is detected. In this scenario it is important to determine if there is a second rare mutation or if the individual is a coincidental carrier and their deafness has another etiology. Reflex testing will also be performed for cases referred for carrier testing where a specific mutation has not been identified previously in another family member. With reflex testing, the total test price is \$540.00 and the complete CPT codes would be 83891, 83894x3, 83898x3, 83904x5, 83912.

Specimen collection:	Adult: 5 mL lavender top tube. Child: 2 mL lavender top tube Cultured amniocytes in T25 or T75 flask. Chorionic villi: tissue in sterile tube or culture media.
Specimen transport:	Entire tube at 2-8°C. Amniocytes/CV transported at room temp.
Performed:	Weekly, results within 2-3 weeks. Prenatal samples are expedited.
Test Order Code:	HHL
CPT Codes:	83891, 83898, 83894, 83904 x4, 83912