



UW Medicine
Department of Laboratory Medicine

Expansion of Hereditary Hearing Loss Testing

[Laboratory Medicine Home Page](#)

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*For additional ordering
information and instructions,
contact the Community
Services office at (206) 598-
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We are pleased to announce two additional tests for Hereditary Hearing Loss. Last month we introduced the Hereditary Hearing Loss Screen (Test order code: HHL), which utilized DNA sequencing to screen for mutations in connexin 26 (GJB2 gene) in addition to screening for the large deletion that involves connexin 30 (GJB6 gene). These tests detect >99% of cases with autosomal recessive non-syndromic neurosensory hearing loss.

Hereditary Hearing Loss, Connexin 30 DNA

This test is most useful for patients in whom previous testing detected only a single mutation in the GJB2 gene and their diagnosis remained questionable. Detection of this deletion would confirm their diagnosis and the etiology of their hearing loss. This test detects the large 342 kb deletion (GJB6_D13S1830del342kb) involving a portion of the GJB6 gene, which in addition to a GJB2 mutation can cause hearing loss.

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. Contact the lab prior to sending a sample for prenatal testing.
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:	HHL30
CPT Codes:	83891, 83898, 83894, 83912

Hereditary Hearing Loss, Relative

This test is designed to test individuals who have a relative with a known mutation in the GJB2 gene. It will detect the presence or absence of a specific GJB2 (connexin 26) mutation(s) known to be present in that particular family. The region of the GJB2 gene carrying the mutation is amplified and sequenced directly.

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. Contact the lab prior to sending a sample for prenatal testing.
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:	HHLREL
CPT Codes:	83891, 83898, 83894, 83904 x4, 83912

