
Lab Dept: **Anatomic Pathology**

Test Name: **CONNEXIN 26/30 MUTATION**

General Information

Lab Order Codes: CONI

Synonyms: Hearing Loss Genes; Connexin 26; Connexin 30; GJB2; GJB6

CPT Codes: 81252 x1 – GJB2 gene analysis; full gene sequence
81254 x1 – GJB6 gene analysis; common variants

Test Includes: GJB2 - gap junction protein, beta 2 (Connexin 26), locus 13q12 and
GJB6 – gap junction protein, beta 6 (Connexin 30), lambda-crystal, locus
13q12

Logistics

Test Indications: All persons with congenital/early childhood mild-to-profound sensorial
hearing loss and a negative family histor, should have Connexin screening.

Lab Testing Section: Anatomic Pathology - Sendouts

Referred to: Harvard Medical School Laboratory for Molecular Genetics

Phone Numbers: MIN Lab: 612-813-6280

STP Lab: 651-220-6550

Test Availability: Daily, 24 hours

Turnaround Time: Approximately 3 weeks

Special Instructions: Please send completed [Harvard requisition and consent](#) with the patient or
specimen to the laboratory. The consent portion of the form is required.
Please note that incomplete of missing paperwork may delay the start of
testing.

Specimen

Specimen Type: Whole blood

Container: Lavender top (EDTA) tube

Draw Volume: 7 mL (Pediatric minimum: 3-5 mL for an infant) blood

Processed Volume:	Same as Draw Volume
Collection:	Routine venipuncture
Special Processing:	Lab Staff: Send lavender tubes labeled with a minimum of Name, Date of Birth, and Age. The reference laboratory accepts samples Monday through Friday from 9am to 5pm. Labeled specimens and requisitions/consents should be sent overnight at room temperature. **Notice: Samples are NOT accepted on Saturdays, Sundays or holidays. Please keep blood samples collected on these days at refrigerated temperatures until the next business day.
Patient Preparation:	None
Sample Rejection:	Mislabeled or unlabeled specimens

Interpretive

Reference Range:	Benign for variants
Methodology:	First screen for 35delG variant. If negative or heterozygous then bidirectional DNA sequencing of exon 1 and coding region of exon 2 is performed. PCR is performed to detect the presence or absence of the deletion.
References:	Harvard Medical School, Laboratory for Molecular Genetics, Partners Health Care Center for Personalized Genetic Medicine Phone: 617-768-8500 Fax: 617-768-8513 Email: IMM@partners.org
Updates:	4/1/2006: CPT coding change previously listed as 83890 only 1/22/2008: CPT coding update 7/29/2010: CPT updates from University of Iowa 2/5/2013: CPT update 5/20/2013: Moved from U of IA to Harvard 1/21/2014: CPT update