
Lab Dept: **Anatomic Pathology**

Test Name: **CONNEXIN 26 (GJB2) KNOWN MUTATION**

General Information

Lab Order Codes: CX26K

Synonyms: Hearing Loss Genes; Connexin 26; GJB2

CPT Codes: 81253 – GJB2 gene analysis, known familial variant

Test Includes: Carrier testing for Connexin 26 gene mutation.

Logistics

Test Indications: Testing of a relative for a specific known mutation (carrier testing).

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: 2-3 weeks

Special Instructions: Please send the completed [Harvard requisition and consent](#) with the patient specimen to the laboratory. The consent portion of the form is required.

**Incomplete or missing paperwork may delay the start of testing.

Specimen

Specimen Type: Whole blood

Container: Lavender top (EDTA) tube

Draw Volume: 7 mL (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 at least two unique identifiers (Name, Date of Birth, Medical Record Number). The laboratory accepts samples Monday through Friday from 9am – 5pm. Labeled samples should be sent overnight at room temperature and accompanied by a completed requisition form.

Patient Preparation: None

Sample Rejection: Mislabeled or unlabeled specimens

Interpretive

Reference Range: Benign for familial mutation

Critical Values: N/A

Limitations: N/A

Methodology: PCR and Sanger sequencing assay of the appropriate exon for known mutation. Only the single site of the mutation is reviewed and recorded for the absence or presence of the familial mutation. Confirmation of results is done by doing two unique DNA dilutions and two different set ups to rule out chance for error. High quality data must be observed in at least two unique set ups to complete the test.

References: [Harvard Medical School, Laboratory for Molecular Genetics, Partners HealthCare Center for Personalized Genetic Medicine](#) May 2013
(617)768-8500 Fax: (617)768-8513 Email: Imm@partners.org

Updates: 5/20/2013: Moved from University of Iowa to Harvard.