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 history, 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 or 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
<table>
<thead>
<tr>
<th><strong>Processed Volume:</strong></th>
<th>Same as Draw Volume</th>
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<tbody>
<tr>
<td><strong>Collection:</strong></td>
<td>Routine blood collection</td>
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<tr>
<td><strong>Special Processing:</strong></td>
<td>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. <strong>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.</strong></td>
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<td><strong>Patient Preparation:</strong></td>
<td>None</td>
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<tr>
<td><strong>Sample Rejection:</strong></td>
<td>Mislabeled or unlabeled specimens</td>
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**Interpretive**

<table>
<thead>
<tr>
<th><strong>Reference Range:</strong></th>
<th>Benign for variants</th>
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<tbody>
<tr>
<td><strong>Methodology:</strong></td>
<td>Sanger sequencing PCR</td>
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<tr>
<td><strong>References:</strong></td>
<td>Harvard Medical School, Laboratory for Molecular Genetics, Partners Health Care Center for Personalized Genetic Medicine March 2018 Phone: 617-768-8500 Fax: 617-768-8513 Email: <a href="mailto:IMM@partners.org">IMM@partners.org</a></td>
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