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**Lab Dept:** Anatomic Pathology

**Test Name:** PENDRED SYNDROME (SLC26A4) KNOWN MUTATION

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***General Information***

**Lab Order Codes:** PENSK

**Synonyms:** Hearing Loss Genes; SLC26A4

**CPT Codes:** 81403 – Molecular Pathology, Level 4

**Test Includes:** Carrier testing for SLC26A4.

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***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:** 3 months

**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.

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***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

<b>Collection:</b>	Routine venipuncture
<b>Special Processing:</b>	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.
<b>Patient Preparation:</b>	None
<b>Sample Rejection:</b>	Mislabeled or unlabeled specimens

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### ***Interpretive***

<b>Reference Range:</b>	Benign for familial variants
<b>Methodology:</b>	PCR and Sanger sequencing assays of the appropriate exon for the 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.
<b>References:</b>	<a href="#">Harvard Medical School, Laboratory for Molecular Genetics, Partners Health Care Center for Personalized Genetic Medicine</a> Phone: 617-768-8500 Fax: 617-768-8513 Email: Imm@partners.org
<b>Updates:</b>	7/29/2010: CPT updates from University of Iowa. 5/20/13: Moved from U of IA to Harvard.