Lab Dept:	**Anatomic Pathology**
Test Name:	**BRANCHIO-OTO-RENAL SYNDROME (EYA1) KNOWN MUTATION**

General Information

Lab Order Codes:	BRORK
Synonyms:	Hearing Loss Genes; EYA1; Oto-facio-cervical syndrome; branchio-otoic syndrome
CPT Codes:	81403 – Molecular Pathology, Level 4
Test Includes:	Carrier testing for EYA1.

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 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 (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 familial variant
Methodology:	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.
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: lmm@partners.org
Updates:	7/29/2010: CPT updates 1/30/2013: CPT update 5/20/2013: Moved from U of IA to Harvard.