

SPECIMEN INFORMATION

Specimen: Blood DNA Other _____ **Date Collected:** (mm/dd/yyyy) ____ / ____ / ____
 Cord Blood* CVS* Amnio*
 DNA* derived from: _____ Space for Lab Use Only
 (Choose One) Cord Blood CVS Amnio

**Maternal specimen is required to perform Maternal Cell Contamination testing. For MCC sample, complete only the first page of the requisition form to submit with specimen. Note an additional \$950 charge will be added for duplicate analysis, MCC studies, internal control tests and rush processing.*

PATIENT INFORMATION

First name: _____ **MI:** _____ **Institution:** _____
Last name: _____ **Medical Record Number:** _____
Date of Birth: (mm/dd/yyyy) ____ / ____ / ____ **Is the patient adopted?** No Yes
Gender: Male Female Unknown/Unspecified **Is the patient deceased?** No Yes, date: _____
Is patient pregnant? No Yes **EDD:** _____ **Race and Ethnicity: Please check ALL that apply**
Address: _____ White Ashkenazi Jewish Asian
City: _____ **State:** _____ **Zip Code:** _____ Hispanic Black/African American
Phone: _____ Native Hawaiian or other Pacific Islander
Email: _____ American Indian/Native Alaskan Other _____

REFERRING PROVIDER INFORMATION

| Referring Provider | Genetic Counselor / Additional Contacts |
|--------------------------------|--|
| Name (First, Last): _____ | Name (First, Last): _____ |
| Phone: _____ Fax: _____ | Phone: _____ Fax: _____ |
| Email: _____ | Email: _____ |
| Institution: _____ | Institution: <input type="checkbox"/> Same as Referring Provider <input type="checkbox"/> Provided below |
| Address: _____ | _____ |
| _____ | _____ |
| City: _____ State: _____ | Place facility sticker here |
| Zip Code: _____ Country: _____ | _____ |

PAYMENT INFORMATION

Please note: Payment information must be completed for testing to begin.

| | |
|---|--|
| <p><input type="checkbox"/> Patient Pay (please complete section in its entirety)**</p> <p><input type="checkbox"/> Check (please attach to forms)* <small>*Please make checks payable to Partners Personalized Medicine*</small></p> <p><input type="checkbox"/> Credit card (please fill out credit card information in its entirety)</p> <p>Card type: <input type="checkbox"/> Mastercard <input type="checkbox"/> Visa <input type="checkbox"/> AMEX</p> <p>Name (as it appears on card): _____</p> <p>Credit card number: _____</p> <p>Expiration Date: _____ 3 Digit Security Code: _____</p> <p><small>**For patient pay, please provide billing address and contact information. If same as above, please note section as such.**</small></p> <p>Patient Pay Billing Address: _____</p> <p>City: _____ State: _____ Zip Code: _____ Country: _____</p> <p>Home: _____ Cell/Work: _____ Email: _____</p> | <p><input type="checkbox"/> Referring Institution (please complete section in its entirety)</p> <p><small>*For new referring facilities, please complete and submit the New Institution Add Form*</small></p> <p>Bill to Name/Department: _____</p> <p>Address: _____</p> <p>_____</p> <p>City: _____ State: _____</p> <p>Zip Code: _____ Country: _____</p> <p>Phone: _____</p> <p>Contact Person: _____</p> |
|---|--|

HEARING LOSS REQUISITION FORM

Patient Name: _____ Date of Birth: ____/____/____ (MM/DD/YYYY)

TESTING TO BE PERFORMED

Check box(es) to order test(s). For reflex testing, indicate order of testing in space provided (i.e. 1, 2, 3).

Hearing Loss Tests

- ___ Connexin Test: *GJB2* Sequencing and *DFNB1 (GJB6)* Deletions
___ Connexin Test **REFLEX** to OtoGenome™ Test for Hearing Loss (110 Genes)*
___ OtoGenome™ Test for Hearing Loss and Related Syndromes (110 Genes)*

OtoGenome Test is performed via next-generation sequencing (NGS) and includes CNV analysis when NGS data meets necessary quality standards. Includes Usher, Pendred, Jervell and Lange-Nielsen, Branchio-Oto-Renal, Waardenburg, Alport, Alstrom, Muckle-Wells, Deafness Infertility syndromes.

Pendred Syndrome or Hearing Loss with EVA

- ___ *SLC26A4* (PDS) Gene Sequencing Test

Low Frequency Nonsyndromic Hearing Loss and Wolfram Syndrome

- ___ *WFS1* Gene Sequencing Test

X-linked Hearing Loss with Perilymphatic Gusher

- ___ *POU3F4* Gene Sequencing Test

SINGLE GENE TESTS

Please contact the lab for single gene tests at 617-768-8500 or Imm@partners.org.

Single Gene Test

- ___ _____ Gene Sequencing Test

FAMILIAL VARIANT TEST

Familial Variant Testing

(First 3 variants, \$500; each additional variant, \$50.)

- ___ Familial Variant(s) OR Research Confirmation
(If proband testing was performed elsewhere, please attach a copy of the original result and send positive control sample, if available.)

Gene #1 _____ Variant #1 _____

Gene #2 _____ Variant #2 _____

Gene #3 _____ Variant #3 _____

Gene #4 _____ Variant #4 _____

Proband Name _____ Relationship to Proband _____

LMM Accession #: PM- _____

PLEASE PROCEED TO PAGE 4 TO COMPLETE THE FAMILY HISTORY

HEARING LOSS REQUISITION FORM

Patient Name: _____ Date of Birth: ____/____/____ (MM/DD/YYYY)

CLINICAL INFORMATION

Clinical status: Affected Unknown (no screening/evaluation) Unaffected (all screening/evaluation(s) normal)

Purpose of study: Diagnostic Carrier testing Other: _____

Age at onset of hearing loss: _____ **ICD-10 Codes:** H90.5 (unspecified sensorineural hearing loss)
 Other _____

Type of hearing loss: Sensorineural Conductive Auditory neuropathy/dys-synchrony Mixed

Laterality: Bilateral Unilateral

Progression: Stable Progressive Fluctuating Unknown

Severity (PTA): *Please attach audiogram if available*

Left Ear: Mild (15-30dB) Moderate (31-50dB) Moderately-severe (51-70dB) Severe (71-90dB) Profound (>90db)

Right Ear: Mild (15-30dB) Moderate (31-50dB) Moderately-severe (51-70dB) Severe (71-90dB) Profound (>90db)

Audiogram shape/frequencies:

Left Ear: Flat (all frequencies) Sloping (high frequency) Saucer-shaped (mid frequency) Rising (low frequency)

Right Ear: Flat (all frequencies) Sloping (high frequency) Saucer-shaped (mid frequency) Rising (low frequency)

Exposure to aminoglycoside antibiotics (e.g gentamicin, neomycin, tobramycin, amikacin): Yes No Unknown

Auditory neuropathy/dys-synchrony:

- No
- Present OAEs
- Absent ABR w/ cochlear microphonic
- Unknown

Eye finding:

- None
- Unknown
- Retinitis pigmentosa - Age of onset: _____
- Other (explain): _____

Vestibular problems:

- None
- Delayed walking
- Dizziness/Vertigo
- Balance problems
- Unknown

BOR features:

- None
- Ear tags
- Ear abnormalities
- Branchial arch abnormality
- Renal abnormality
- Other (explain): _____

Temporal bone abnormalities on CT/MRI:

- None
- EVA (enlarged vestibular aqueducts)
- Mondini dysplasia
- Stapes fixation
- Perilymphatic gusher
- Unknown
- Other (explain): _____

Waardenburg features:

- None
- White forelock
- Heterochromia
- Hypoplastic/vivid blue irises
- Dystopia canthorum
- Musculoskeletal abnl.
- Hirschsprung

Electrocardiogram (ECG) finding:

- None
- Long QT
- Unknown
- Other (explain): _____

CAPS/Muckle Wells features:

- Urticaria-like rash
- Conjunctivitis
- Nephritis
- Amyloidosis

Alport features:

- Hematuria
- Proteinuria
- ESRD

Previous genetic testing: No Yes - Test/Results: _____

Other relevant medical problems: None Yes (explain): _____

Has another family member already had genetic testing for this disease? No Yes

If yes, please describe and attach a copy of the genetic test lab report and pedigree.

FAMILY HISTORY

Sibling with or other family history of similar hearing loss? Yes No

List affected individuals and the nature of their hearing loss (Sketch below or attach pedigree if appropriate): _____

Paternal Ancestry: _____

Maternal Ancestry: _____

Consanguinity: Yes No

○ = Female □ = Male ◇ = Gender Unspecified
● ■ ◆ = Affected Individual ⊕ = Carrier