

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)*</p> <p><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>
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Patient Name: _____ Date of Birth: ____/____/____ (MM/DD/YYYY)

SPECIMEN & SHIPPING REQUIREMENTS

The preferred blood specimen is a 7 ml blood sample (3-5ml for infants) collected in a lavender top (K₂EDTA or K₃EDTA) blood tube. Smaller blood samples or other tissue specimens may also be acceptable for certain tests. All samples must have two patient identifiers, preferably the patient's name and date of birth. Please contact the laboratory for more details.

Each sample must be accompanied by a requisition form. The ordering provider must sign the declaration below.

The blood sample (with forms) should be shipped overnight at room temperature to:

Laboratory for Molecular Medicine
65 Landsdowne Street
Cambridge, MA 02139

For more detailed information about shipping requirements and procedures, see our website www.partners.org/personalizedmedicine/lmm.

LABORATORY FOR MOLECULAR MEDICINE POLICIES

By requesting testing from the Laboratory for Molecular Medicine (LMM), the ordering provider indicates that he/she understands AND accepts the policies of the LMM, as noted below, and has communicated these policies to the patient.

1. Our testing process includes highly skilled technicians and advanced technology. As in any laboratory, there is a small possibility that the test will not work properly, or an error may occur.
2. Listed turn around times (TATs) represent the typical TAT for a test, but are not guaranteed.
3. If the requisition form is incomplete, and the healthcare provider cannot provide the required information, lab staff may need to contact patients directly to obtain or verify the information needed to complete the form.
4. Test results, as well as any updates to those results, may become part of a patient's permanent medical record (electronically or otherwise) or be made available (electronically or otherwise) to the ordering healthcare institution and its healthcare team.
5. Results will only be released to the ordering provider and other providers listed on the requisition form. The ordering provider assumes the responsibility to disclose the test results and direct care as appropriate.
6. Test results and submitted clinical information may be shared with other clinical laboratories for the purpose of improving our understanding of the relationship between genetic changes and clinical symptoms. Sharing data in this manner may enable us to provide better interpretations of your genetic findings as well as assist other patients with similar results. We will protect your privacy/confidentiality by removing your name and other direct identifiers, such as SSN or medical record number, from data shared with other laboratories.

RESEARCH POLICIES & OPPORTUNITIES

Blood or other samples sent to the LMM may be used by Partners Healthcare System (PHS), by medical organizations connected to PHS, or by educational or business organizations approved by PHS, for research, education and other activities that support PHS's mission, without your/the patient's specific consent. Other types of research performed in association with the Laboratory for Molecular Medicine require that we obtain consent from the patient (see below).

PATIENTS - Please check off and initial below whether we can contact you to let you know about research studies in which you/your child may be able to participate. These research studies may include:

- A request for additional clinical records about your condition
- Studies to find new causes for your condition
- Studies to evaluate newly developed treatments for your condition

Please check one option: _____ Yes, you can contact me _____ (patient initials)
If yes, please provide your contact information on the first page

_____ No, please do not contact me _____ (patient initials)

ORDERING PROVIDER SIGNATURE

I, _____ (print name), as ordering provider, certify that the patient being tested and/or their legal guardian have been informed of the risks, benefits, and limitations of the testing ordered, as well as the policies of the LMM listed above. I have obtained informed consent, as required by my own state and/or federal laws. In addition, I assume responsibility for returning the results of genetic testing to my patient and/or their legal guardian and for ensuring that my patient receives appropriate genetic counseling to understand the implications of their test results.

Signature (Ordering Provider) _____ Date _____

Please Note: A patient consent form is available on our website (www.partners.org/personalizedmedicine/Laboratory-For-Molecular-Medicine/Ordering/Policies) for your convenience and DOES NOT need to be returned to the LMM.

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 (109 Genes)*

___ OtoGenome™ Test for Hearing Loss and Related Syndromes (109 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.

___ Mitochondrial Gene Panel: 12SrRNA and tRNAs_r (UCN)

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

(Sequencing - 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