

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

**PAYMENT INFORMATION**

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

<p><input type="checkbox"/> <b>Patient Pay</b> (please complete section in its entirety)**</p> <p><input type="checkbox"/> <b>Check</b> (please attach to forms)*</p> <p><small>*Please make checks payable to Partners Personalized Medicine*</small></p> <p><input type="checkbox"/> <b>Credit card</b> (please fill out credit card information in its entirety)</p> <p><b>Card type:</b> <input type="checkbox"/> Mastercard <input type="checkbox"/> Visa <input type="checkbox"/> AMEX</p> <p><b>Name</b> (as it appears on card): _____</p> <p><b>Credit card number:</b> _____</p> <p><b>Expiration Date:</b> _____ <b>3 Digit Security Code:</b> _____</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><b>Patient Pay Billing Address:</b> _____</p> <p><b>City:</b> _____ <b>State:</b> _____ <b>Zip Code:</b> _____ <b>Country:</b> _____</p> <p><b>Home:</b> _____ <b>Cell/Work:</b> _____ <b>Email:</b> _____</p>	<p><input type="checkbox"/> <b>Referring Institution</b> (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><b>Bill to Name/Department:</b> _____</p> <p><b>Address:</b> _____</p> <p>_____</p> <p><b>City:</b> _____ <b>State:</b> _____</p> <p><b>Zip Code:</b> _____ <b>Country:</b> _____</p> <p><b>Phone:</b> _____</p> <p><b>Contact Person:</b> _____</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 (K<sub>2</sub>EDTA or K<sub>3</sub>EDTA) 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](http://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](http://www.partners.org/personalizedmedicine/Laboratory-For-Molecular-Medicine/Ordering/Policies)) for your convenience and DOES NOT need to be returned to the LMM.

# ECTODERMAL DYSPLASIA REQUISITION FORM

Patient Name: \_\_\_\_\_ Date of Birth: \_\_\_\_/\_\_\_\_/\_\_\_\_ (MM/DD/YYYY)

## TEST TO BE PERFORMED

Please check box(es) to order. If ordering reflex testing, please indicated order (i.e. 1, 2, 3). Testing can be conducted concurrently if desired.

### Hypohidrotic Ectodermal Dysplasia

- EDA Gene Sequencing \$700  
 EDA Deletion/Duplication (MLPA) - whole blood sample required \$600

### Hidrotic Ectodermal Dysplasia 2

- GJB6 (Connexin 30) Gene Sequencing Test \$500

### GJB2 Related-Ectodermal Dysplasias

- GJB2 (Connexin 26) Gene Sequencing Test \$500

### DSP Related-Ectodermal Dysplasias/Skin Fragility

- DSP Gene Sequencing Test \$1,700

### Familial Variant Testing

- Familial Variant(s)\*\* OR  Research Confirmation\*\* \$500

\*\*If proband testing was performed elsewhere, please attach a copy of the original result and send positive control sample, if available\*\*

Gene \_\_\_\_\_ Variant \_\_\_\_\_

Proband Name \_\_\_\_\_

Relationship to Patient \_\_\_\_\_ LMM Accession #: PM- \_\_\_\_\_

## CLINICAL INFORMATION

- Clinical status:**  Affected  Unknown  Unaffected  
**Purpose of study:**  Diagnostic  Carrier testing  Prenatal  Other \_\_\_\_\_

- Clinical diagnosis:**  Hypohidrotic Ectodermal Dysplasia  Hidrotic Ectodermal Dysplasia 2

**ICD-10 Code(s):** \_\_\_\_\_

### Clinical Features:

- Sparse hair  Scalp  Body  No Other features: \_\_\_\_\_  
Sweat level  Reduced  Absent  Normal \_\_\_\_\_  
Hypodontia  Yes → # of permanent teeth \_\_\_\_\_  No \_\_\_\_\_  
Conical crowns  Yes  No \_\_\_\_\_

Previous Genetic Testing:  Yes  No Gene(s)/Tests: \_\_\_\_\_

Result (if variants detected, please elaborate): \_\_\_\_\_

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

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

## FAMILY HISTORY

**Family History:**  Yes  No (Sketch below or attach pedigree if appropriate)

Please list affected family members: \_\_\_\_\_

Paternal Ancestry: \_\_\_\_\_

Maternal Ancestry: \_\_\_\_\_

Consanguinity:  Yes  No

○ = Female □ = Male ◇ = Gender Unspecified

● ■ ◆ = Affected Individual ⊙ = Carrier