

## REQUEST FOR LYMPHEDEMA GENETIC TESTING PANEL

*Please provide all of the following information. We may not be able to perform the test without complete information. PRINT ALL FIELDS.  
Note that there is a 48-hour window for test cancellation. Cancellation requests after this period will be accommodated at the discretion of the lab.*

### PATIENT INFORMATION\*

FIRST NAME \_\_\_\_\_ M \_\_\_\_\_ LAST NAME \_\_\_\_\_ ASSIGNED SEX \_\_\_\_\_ BIRTH DATE (mm/dd/yyyy) \_\_\_\_\_

### CLINICAL INFORMATION\*

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

- I89.0 Lymphedema     J94.0 Chylothorax     I89.9 Noninfective disorder of lymphatic vessels and lymph nodes

Additional clinical features, please specify: \_\_\_\_\_  
\_\_\_\_\_  
\_\_\_\_\_  
\_\_\_\_\_

### SAMPLE INFORMATION\*

UNAFFECTED TISSUE SAMPLE	AFFECTED TISSUE SAMPLE #1 [Include pathology report]	AFFECTED TISSUE SAMPLE #2 (OPTIONAL) [Include pathology report]
<input type="checkbox"/> Blood <input type="checkbox"/> Skin punch biopsy from _____ <input type="checkbox"/> Cultured cells from _____ <input type="checkbox"/> Frozen tissue from _____ <input type="checkbox"/> Isolated DNA from _____ <input type="checkbox"/> Other _____	<input type="checkbox"/> Skin punch biopsy from _____ <input type="checkbox"/> Cultured cells from _____ <input type="checkbox"/> Frozen tissue from _____ <input type="checkbox"/> Isolated DNA from _____ <input type="checkbox"/> Other _____	<input type="checkbox"/> Skin punch biopsy from _____ <input type="checkbox"/> Cultured cells from _____ <input type="checkbox"/> Frozen tissue from _____ <input type="checkbox"/> Isolated DNA from _____ <input type="checkbox"/> Other _____
Collection Date/Time: _____	Collection Date/Time: _____	Collection Date/Time: _____
<input type="checkbox"/> Included with test request form <input type="checkbox"/> Will be submitted at a later date	<input type="checkbox"/> Included with test request form <input type="checkbox"/> Will be submitted at a later date	<input type="checkbox"/> Included with test request form <input type="checkbox"/> Will be submitted at a later date

### TEST REQUESTED\*

**STAT REQUEST** (Check this box in addition to the test below if you intend to order STAT results in half (50%) of the listed TAT and a total cost of +20% of the base cost listed for the ordered test, this option excludes *Expedited* and *Prenatal* test requests.)

- Lymphedema Panel [43 genes]  
 *PRENATAL* \*\* Lymphedema Panel [43 genes]

\*Required Information

\*\*Please call the laboratory prior to sending a prenatal or STAT sample. Please refer to the test catalog for prenatal sample requirements at [www.med.upenn.edu/genetics/gdl](http://www.med.upenn.edu/genetics/gdl)

## PATIENT REGISTRATION FORM

Please provide all of the following information. We may not be able to perform the test without complete information.  
PRINT ALL FIELDS.

### PATIENT INFORMATION

FIRST NAME	M	LAST NAME	ASSIGNED SEX	BIRTH DATE (mm/dd/yyyy)
STREET ADDRESS			PHONE	
CITY	STATE	ZIP		
<b>ANCESTRY</b>				
<input type="checkbox"/> European (non-Finnish)	<input type="checkbox"/> African	<input type="checkbox"/> European (Finnish)	<input type="checkbox"/> East Asian	
<input type="checkbox"/> Amish	<input type="checkbox"/> South Asian	<input type="checkbox"/> Jewish (Ashkenazi)	<input type="checkbox"/> Admixed American	
<input type="checkbox"/> Middle Eastern	<input type="checkbox"/> OTHER (specify countries):			

### ORDERING PHYSICIAN INFORMATION

ORDERING PHYSICIAN	Physician Contact (email address or phone number)	FAX
GENETIC COUNSELOR	GC Contact (email address or phone number)	FAX
INSTITUTION	DEPARTMENT	
STREET ADDRESS		
CITY	STATE	ZIP

### BILLING INFORMATION [MUST SELECT ONE]

I have enclosed a check payable to the "Genetic Diagnostic Laboratory" for \$ \_\_\_\_\_

Please charge my credit card for the amount of \$ \_\_\_\_\_

VISA       MasterCard       Discover       AmEx

Card Number: \_\_\_\_\_ Exp Date: \_\_\_\_\_

Name of cardholder as it appears on card: \_\_\_\_\_

I have Pennsylvania Medicaid. A copy of my Medicaid card is attached.

INSTITUTIONAL BILLING. The institution where testing is ordered has agreed to pay all charges for testing.  
INCLUDE: Billing Address, Person Authorizing Payment, Telephone Number, and Fax Number below.

\_\_\_\_\_

Billing Address

\_\_\_\_\_

Billing Address

\_\_\_\_\_

Name of Individual Authorizing Payment      PHONE      FAX

### SEND FINAL TEST RESULTS TO: [IF NONE SELECTED, WILL GO TO DR &/or GC LISTED ABOVE]

<input type="checkbox"/> Ordering Physician	<input type="checkbox"/> Genetic Counselor
<input type="checkbox"/> Central Lab Services/ Send Out Department	<input type="checkbox"/> Other: _____
Fax: _____	Fax: _____

\*Required Information

\*\*Please call the laboratory prior to sending a prenatal or STAT sample. Please refer to the test catalog for prenatal sample requirements at [www.med.upenn.edu/genetics/gdl](http://www.med.upenn.edu/genetics/gdl)

06/2025

Genetic Diagnostic Laboratory  
Department of Genetics

## INFORMED CONSENT: GENETIC TESTING FOR LYMPHEDEMA PANEL

**Background:** This test provides analysis of 43 genes associated with lymphatic malformations, which encompass a variety of disorders and can occur as an isolated feature or as part of a syndrome. Genetic diagnosis can confirm a clinical diagnosis and can be used to guide follow-up care and treatment strategies. Identifying the genetic cause also allows for risk assessment among family members.

**Purpose:** The diagnostic samples will be used for the purpose of attempting to determine if there is an alteration in the following genes related to somatic overgrowth: *ABCC9, ADAMTS3, ALG8, ARAF, BRAF, CBL, CCBE1, CCDC88A, CDC42, CELSR1, EPHB4, FAT4, FLT4, FOXC2, FZD6, GATA2, GJA1, GJC2, HGF, HRAS, ITGA9, KIF11, KRAS, MAP2K1, MAP2K2, MET, MPI, NAGA, NRAS, PIEZO1, PIK3CA, PTEN, PTPN11, PTPN14, RAF1, RASA1, RIT1, SHOC2, SOS1, SOX18, THSD1, VEGFC,* and *ZNHIT3*. 14 SNPs are also analyzed to detect 45,X. This information may help establish appropriate medical management.

**Results:** I understand that there are four possible results with this testing:

**PATHOGENIC VARIANT:** A clinically significant variant is detected in gene(s) analyzed. This may explain my personal or family history of overgrowth. My or my child's healthcare provider will make medical management recommendations based on this information.

**LIKELY PATHOGENIC VARIANT:** A variant is detected in the gene(s) analyzed which is the likely deleterious. This may explain my personal or family history of overgrowth. My or my child's healthcare provider will make medical management recommendations based on this information.

**VARIANT OF UNKNOWN SIGNIFICANCE:** The laboratory may detect an alteration in the gene(s) analyzed which is currently of unknown significance, called a "variant of unknown significance (VUS)". The laboratory will work with my physician to help determine if the VUS can be further classified as to whether it is associated with overgrowth.

**NEGATIVE:** No clinically significant variants were identified in the gene(s) analyzed. This result reduces the likelihood that I, or my child, have a clinically significant variant in the gene(s) tested. Methods currently in use are unable to detect all variants and therefore I may still carry a variant that was not detected by the current technology.

**Disclosure Policy:** The Genetic Diagnostic Laboratory will release my test results to the ordering healthcare provider &/or genetic counselor, and otherwise only as permitted by law. The results will be kept confidential to the extent allowed by law. If I provide separate written consent, the lab will release my test results to other medical professionals or third persons I want to receive my results.

**Limitations:** While genetic testing is highly accurate for detection of the majority of disease causing variants, a small fraction of variants may be missed by the current technology. Due to the nature of the testing, there is a small possibility that the test will not work properly or that an error will occur. Testing may reveal a variant of unknown significance that is unable to be definitively interpreted as positive or negative for disease-association based on the current knowledge of the variant. The DNA analysis performed at the University of Pennsylvania Genetic Diagnostic Laboratory is specific only for gene(s) analyzed and in no way guarantees my health.

There are federal laws in place that prohibit health insurers and employers from discriminating based on genetic information, such as test results. There currently are no federal laws prohibiting discrimination based on genetic information by life insurance, long term care, or disability insurance companies but state laws may restrict this. I understand I can ask my ordering provider or genetic counselor for more information about how insurers might use genetic information.

Genetic testing may identify information about biological family relationships that may be unexpected. Genetic testing may identify a medically relevant finding that is not related to the indication for testing. If genetic testing suggests that biological relationships between family members are not as reported to the Genetic Diagnostic Laboratory and/or another incidentally identified genetic result, I understand that these findings may be disclosed to the healthcare provider(s) who ordered the genetic testing.

Initials: \_\_\_\_\_

**Use of Specimens After Clinical Test Performed:** I understand my blood &/or tissue specimen(s) will not be returned to me or the ordering healthcare provider, and becomes the property of the lab upon receipt. The laboratory is not a DNA banking facility; therefore this is no guarantee that samples will be available or usable for additional or future testing.

After the laboratory completes the ordered clinical test, the lab may retain and preserve the specimen to validate the development of future genetic tests or for future research or education purposes. The laboratory is committed to continuous improvement and therefore I understand my coded sample may be used to validate a new assay. If testing reveals a clinically significant result during the validation process of a new assay related to the original indication for testing, my health provider may be contacted. If the lab uses the specimen for future research or education purposes, the specimen will be de-identified by removing my personally identifying information. My name, address and other personal identifying information will not be linked to the samples, or the results of the research, and I will not be identified in any research results or publications. I will not receive a copy of the research results. I can decline for my sample to be retained at the lab by filling out "Research Opt Out" form found on the following website: <https://genetics.med.upenn.edu/genetic-diagnostic-laboratory>

I understand the lab may wish to contact me, or my ordering healthcare provider, for additional information. The additional information may include, but would not be limited to, information about health and family history that might be relevant to the research. I understand I can decline future contact from the lab by filling out "Research Opt Out" form found on the following website: <https://genetics.med.upenn.edu/genetic-diagnostic-laboratory>

**Genetic Counseling** provided by a qualified specialist (i.e. genetic counselor/ medical geneticist) is a recommendation for individuals proceeding with genetic testing. This service is available before and after genetic testing. Additionally, other testing or further physician consults may be warranted.

The Genetic Diagnostic Laboratory is also an available resource to ask more questions about this testing. The laboratory genetic counselor can be reached at 215-573-9161 and Arupa Ganguly, PhD, FACMG can be reached at 215-898-3122.

**HEALTHCARE PROVIDER STATEMENT:**

I have explained to \_\_\_\_\_ the purpose of this genetic testing, the procedures required and the possible risks and benefits to the best of my ability.

\_\_\_\_\_  
Printed Name of Professional Obtaining Consent

\_\_\_\_\_  
Signature of Professional Obtaining Consent

\_\_\_\_\_  
Date

**CONSENT OF PATIENT:**

I have read and received a copy of this consent form. I agree to have genetic testing performed for myself, child or my fetus, and accept the risks. I understand the information provided in this document and I have had the opportunity to ask questions I have about the testing, the procedure, the associated risks and the alternatives.

Patient's Printed Name: \_\_\_\_\_

DOB: \_\_\_\_\_

Patient's Signature: \_\_\_\_\_  
(or Parent/Guardian if patient is a minor)

Date: \_\_\_\_\_

Name and Relationship: \_\_\_\_\_  
(Parent/Guardian if patient is a minor)