REQUEST FOR X-INACTIVATION STUDIES

Please provide the following information. We cannot perform your test without ALL of this information. PLEASE PRINT ALL ANSWERS

PATIENT INFORMATION*

<table>
<thead>
<tr>
<th>FIRST NAME</th>
<th>MI</th>
<th>LAST NAME</th>
<th>BIRTH DATE (MM/DD/YYYY)</th>
<th>GENDER</th>
</tr>
</thead>
</table>

ANCESTRY
- Western/Northern European
- Central/Eastern European
- Latin American/Caribbean
- African
- Asian
- Jewish (Ashkenazi)
- American Indian
- Near East/Middle Eastern
- Native Hawaiian or Pacific Islander

Specify countries: ____________________________

Other: ____________________

CLINICAL INFORMATION

Patient is a known carrier of an X-linked disorder? ☐ No ☐ Yes

If yes, disease name: ____________________________________________

Mutation detected: _____________________________________________

Is the patient a known carrier AND symptomatic: ☐ No ☐ Yes

Additional clinical information:
_________________________________________________________________
_________________________________________________________________
_________________________________________________________________

ICD-10 CODE(S)* ___________________________________________________

TEST REQUESTED*

X-inactivation studies:

☐ Individual

☐ Individual and parents

* Required information

** For all prenatal requests, please call the laboratory prior to sending a prenatal sample. Please refer to the special requirements for prenatal samples on the Instructions for Sample Submission page.

9/28/2015
PATIENT REGISTRATION FORM

Please provide the following information. We cannot perform your test without ALL of this information. PLEASE PRINT ALL ANSWERS

PATIENT INFORMATION

FIRST NAME       MI       LAST NAME

BIRTH DATE       GENDER

STREET ADDRESS

CITY

STATE

ZIP

PHONE

PHYSICIAN INFORMATION*

REFERRING PHYSICIAN

PHONE

FAX

GENETIC COUNSELOR

PHONE

FAX

EMAIL ADDRESS FOR COUNSELOR

EMAIL ADDRESS FOR PHYSICIAN

INSTITUTION AND DEPARTMENT

STREET ADDRESS

CITY

STATE

ZIP

PAYMENT OPTIONS* (must choose one) [a receipt will be mailed to the patient for self-pay options]

☐ I have enclosed a check payable to the “Genetic Diagnostic Laboratory” for $____________________

☐ Please charge my credit card for the amount of $____________________

☐ VISA     ☐ Master Card     ☐ Discover     ☐ American Express

  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 my testing originated has agreed to pay all charges for the testing.

  INCLUDE Billing Address, Person Authorizing Payment, Telephone, and Fax below:

BILLING ADDRESS

BILLING ADDRESS

NAME OF INDIVIDUAL AUTHORIZING PAYMENT

PHONE

FAX
VERIFICATION OF CORRECTLY IDENTIFIED BLOOD TUBES

I am a participant in genetic DNA testing.

I have been shown the tubes containing my blood for this genetic testing and my name has been correctly placed on each one of these tubes.

I have signed a copy of the consent form regarding this genetic testing to be sent along with my blood samples. I have been given a copy of the consent form to keep.

Participant Name: _____________________________________________

Participant/Parent Signature: ______________________________________

Date: ___________________
INFORMED CONSENT FOR X-INACTIVATION STUDIES

Background: Evaluating for skewed patterns of X-inactivation can be useful in analysis and diagnosis of X-linked conditions. Skewed patterns of X chromosome inactivation can result in symptomatic female carriers of X-linked recessive conditions, as well as asymptomatic females in X-linked dominant conditions. X-inactivation studies can also be used to determine appropriate testing for relatives of carriers or affected individuals.

Purpose: I, or my child, will be tested for skewed patterns of X-inactivation as described above. I understand that the testing will take approximately 3-4 weeks to complete. The purpose of this genetic testing is to determine whether I, or my child, have skewed patterns of X-inactivation can be useful in analysis and diagnosis of X-linked conditions; the information may help establish appropriate medical management.

Results: I understand that there are three possible results to this testing:

POSITIVE: The analysis detects that I, or my child, have a clinically significant skewed pattern of X inactivation. My or my child’s healthcare provider may make medical management recommendations based on this information.

NEGATIVE: The analysis did not detect a skewed pattern of X inactivation. This result reduces the likelihood that I, or my child, have skewed X inactivation. Methods currently in use are unable to detect all skewing and therefore may still have skewed X inactivation that was not detected by the current technology.

INCONCLUSIVE: The laboratory could not determine whether there was or was not skewed X inactivation in the sample analyzed.

Disclosure Policy: Results will be reported to me only through the physician or genetic counselor who requested the testing due to the complexity of DNA based testing. The results of genetic testing are protected by the Health Insurance Portability and Accountability Act (HIPAA) of 1996 (Public Law 104-191). Release of test results is limited to authorized personnel as required by law. Additionally, results can be released to other medical professionals or other parties with my written consent.

Limitations: While genetic testing is highly accurate for detection of the majority of disease causing mutations, a small fraction of mutations 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. Occasionally, testing may reveal a variant of unknown significance that is unable to be definitively interpreted as positive or negative for disease-association based on our current knowledge of the variant. My signature below acknowledges my voluntary participation in this test, but in no way releases the laboratory and staff from their professional and ethical responsibility to me. Furthermore, the DNA analysis performed at the University of Pennsylvania Genetic Diagnostic Laboratory is specific only for X inactivation 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 (i.e. Genetic Information Nondiscrimination Act (GINA) of 2008 (Public Law 110-233). There are currently no laws specific to discrimination based on genetic information for life insurance, long term care, or disability insurance companies. Additional information about GINA can be found on the Genetics Public & Policy Center’s website at www.dnapolicy.org.

Use of Specimens: Any blood or tissue specimens obtained for the purposes of this genetic testing become the exclusive property of the Genetic Diagnostic Laboratory. After the specific tests requested have been completed and reported, the laboratory may dispose of, retain, or preserve these specimens for research or for validation in the development of future genetic tests. In all circumstance described previously, my identity will be protected and research results will not be provided to me or to any other party. If use of this genetic material results in a scientific publication, it will not contain any identifying information. Indicate consent or denial to the above sentence by initialing below. My refusal to consent to research will not affect the reporting of my genetic results.

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Initials _______
_____ I consent to the use of my DNA sample for future test validation and/or research purposes.

_____ I do not consent to the use of my DNA sample for future test validation or research purposes.

In the event that my sample is used for research purposes, the laboratory may wish to contact my physician/genetic counselor for additional information regarding my sample. This includes, but is not limited to, information on personal health and family history as it relates to the genetic testing. If there are new developments in the field, my physician/genetic counselor may be contacted by the Genetic Diagnostic Laboratory staff to offer me the opportunity to have additional clinical testing. Indicate consent or denial to the above sentence by initialing below. My refusal to consent to research will not affect the reporting of my genetic results.

_____ I consent to be contacted by the Laboratory in the future for research purposes.

_____ I do not consent to be contacted by the Laboratory in the future for research purposes.

**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. I will be given a copy of this consent form to keep.

**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, 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: ________________________________

Date: __________________

(or Parent/Guardian if patient is a minor)

Name and Relationship: ________________________________

(Parent/Guardian if patient is a minor)

9/28/2015