GENETIC DIAGNOSTIC LABORATORY
UNIVERSITY OF PENNSYLVANIA SCHOOL OF MEDICINE
DEPARTMENT OF GENETICS
560 Clinical Research Building • 415 Curie Boulevard • Philadelphia, PA 19104
Tel: (215) 573-9161 • Fax: (215) 573-5940 • Email: gdllab@pennmedicine.upenn.edu
CLIA ID: 39D0893887

REQUEST FOR FIBRODYSPLASIA OSSIFICANS PROGRESSIVE (FOP) TESTING

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

PATIENT INFORMATION* Sample Collection Date & Time: _____

FIRST NAME _____ MI _____ LAST NAME _____ BIRTH DATE (MM/DD/YYYY) _____ GENDER _____

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

ICD-10 CODE(S):* ☐ M61.10 Progressive myositis ossificans
☐ Q66.89 Other deformities of foot
☐ Q66.89 Other anomalies of toes: congenital
☐ Other: _____

Does the patient have extra-skeletal bone formation? ☐ No ☐ Yes

If yes, list locations and age of appearance: _____

Does the patient have any additional skeletal abnormalities? ☐ No ☐ Yes

If yes, please describe: _____

Does the patient have malformed great toes? ☐ Hallux Valgus ☐ Malformed 1st metatarsal
☐ Monophalangism ☐ None

Does the patient have tibial osteochondromas? ☐ No ☐ Yes

Additional clinical observations: _____

_____

_____

TEST REQUESTED*

☐ Site specific analysis of ACVR1 gene (c.617G>A)

☐ Prenatal site specific analysis of ACVR1 gene (c.617G>A) **

*Required information
** 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.

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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 (MM/DD/YYYY) 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

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INFORMED CONSENT FOR GENETIC TESTING FOR A SPECIFIC MUTATION IN FIBRODYSPLASIA OSSIFICANS PROGRESSIVA (FOP) (ACVR1 c.617G>A)

Background: Fibrodysplasia ossificans progressiva (FOP) is a disorder of heterotopic (extra-skeletal) ossification in which muscles and connective tissue such as tendons and ligaments are replaced by bone over time. This bone formation generally begins in early childhood and typically initiates in the neck and shoulders. Malformation of the great toe is another characteristic clinical feature.

The incidence of FOP is ~1 in 2 million. Most cases are spontaneous (new) mutations with no previous family history, although autosomal dominant inheritance is also observed. Heterozygous mutations in the ACVR1 gene at c.617G>A (p.Arg206His) have been identified in patients with classic features of FOP (progressive heterotopic ossification and malformed great toes) in 100% of cases. It is possible that patients with heterotopic ossification and atypical features of FOP (FOP variants) may not carry the ACVR1 c.617G>A mutation.

Purpose: The diagnostic samples will be used for the purpose of attempting to determine if I (or my child/fetus) am/is a carrier of an altered *ACVR1* gene related fibrodysplasia ossificans progressiva. This information may help establish appropriate medical management.

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

PATHOGENIC VARIANT: A clinically significant variant is detected in the *ACVR1* gene. This may explain my personal or family history of FOP. 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 *ACVR1* gene which is the likely deleterious. This may explain my personal or family history of FOP. My or my child's healthcare provider will make medical management recommendations based on this information.

VARIANT OF UNCERTAIN SIGNIFICANCE: The laboratory may detect an alteration in the *ACVR1* gene 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 FOP.

LIKELY BENIGN VARIANT: A variant is detected the *ACVR1* gene which is not likely to be clinically significant. This result reduces the likelihood that I, or my child, have a clinically significant variant in the gene(s) tested.

NEGATIVE: No clinically significant variants were identified in the *ACVR1* gene. 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 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 the current knowledge of the variant. The DNA analysis performed at the University of Pennsylvania Genetic Diagnostic Laboratory is specific only for the 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.

Initials _____

Use of Specimens After Clinical Test Performed: I understand my blood or tissue specimen 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. Samples from New York residents will be disposed of 60 days after clinical testing is complete.

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: http://www.med.upenn.edu/genetics/gdl/.

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: http://www.med.upenn.edu/genetics/gdl/.

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, 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: _____ Date: _____
(*or Parent/Guardian if patient is a minor*)

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

Initials _____