

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*

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*

☐ Point mutation analysis of c.617G>A in ACVR1

☐ Prenatal diagnosis: point mutation analysis of c.617G>A in ACVR1**

*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.

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 (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

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 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 (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) am/is a carrier of an altered ACVR1 gene related fibrodysplasia ossificans progressiva. I understand that the testing will take approximately 2-4 weeks to complete. 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 which is related to fibrodysplasia ossificans progressiva. 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. My or my child's healthcare provider may 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 your physician to help determine if the VUS can be further classified as to whether it is disease-causing for 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 mutation in the genes tested.

NEGATIVE: No clinically significant mutations was identified in the ACVR1 gene. This result reduces the likelihood that I, or my child, have a mutation in the gene tested. Methods currently in use are unable to detect all mutations and therefore may still carry a DNA mutation that was not detected by the current technology.

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 RB1 analysis 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.

Initials _____

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.

_____ 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 associate 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)