

Familial Mediterranean Fever GenotypR™ INFORMED CONSENT

1. What causes Familial Mediterranean Fever (FMF)? Familial Mediterranean Fever is an inherited condition characterized by recurrent episodes of painful inflammation in the abdomen, chest, or joints. These episodes are often accompanied by fever and sometimes a rash. The first episode usually occurs by the age of 20, but in some cases, the initial attack occurs much later in life. Typically, episodes last 12 to 72 hours and can vary in severity and length of time between attacks. A buildup of protein deposits (amyloidosis) occurs in some cases of FMF and can lead to kidney failure if left untreated. The disease primarily affects populations originating in the Mediterranean region, particularly people of Armenian, Arabic, Turkish, and Jewish ancestry. The frequency of affected individuals in these populations ranges from 1 in 250 people to 1 in 1,000 people.

2. What is the purpose of this test and what are its limitations? This test detects the presence of twelve mutations in the FMF gene: E148Q, P369S, F479L, M680I (G/C), M680I (G/A), I692del, M694V, M694I, K695R, V726A, A744S, R761H. This assay detects more than 74% of the mutations associated with FMF in the most commonly affected ethnic populations (Armenian, Arabs, Jews Turks). Everyone has two copies of the FMF gene; an individual may have two normal copies (unaffected non-carrier), two abnormal copies (affected), or one normal and one abnormal (carrier). If these mutations are not found by the testing procedure, it does not mean that the risk of carrying or developing FMF is not present. It simply means that these specific mutations have not been found, although other mutations may be present.

3. What is required to perform this test? You will be asked to provide 5 mL of blood, which is equal to about one tablespoon. DNA will be extracted from this blood sample and tested. The only discomfort that you may feel is the stick of the needle in your arm. You may also experience a small bruise at the site of the needle puncture. You will also be asked to provide information regarding your medical history, which is necessary for proper interpretation of your test result. In the unlikely event that you should be injured in the course of being tested, your physician will provide any necessary medical care. However, you would be expected to bear the cost of such medical care. Compensation will not be provided in the event of any injury.

4. Is there a cost for this test? This is a routine clinical laboratory test and the results may aid in your diagnosis; thus, you or your health insurer will be billed for this procedure.

5. What will happen to the DNA once the test is complete? The original blood sample will be discarded at the end of the testing process or stored not more than 60 days. The DNA will be retained for a minimum of 6 months. In some circumstances, a patient's DNA may be used anonymously as a negative or positive control sample in future testing, but, in this circumstance, all identifiers will be removed prior to re-testing and the DNA sample and results obtained will remain anonymous.

I understand and agree that my DNA remaining after testing may be stored for up to 6 months should additional testing be required. *Please initial.*

6. How will I obtain results from this test? DNA testing and interpretation of results are complex. The information from this test will be provided in the form of a written report to your physician who will inform you of the results. The laboratory will not provide results directly to patients. Your physician may suggest genetic counseling prior to performing this test or if your results are abnormal. To the extent permitted by law, all of your laboratory records and results are confidential and shall not be disclosed without your written authorization.

Patient Attestation of Informed Consent:

My signature below indicates that I have received information about this test, **FMF GenotypR™**, and that I have read and understood the material in this document. I have been given a full opportunity to ask questions that I may have about the testing procedure and related issues. I agree to undergo this testing.

Patient Signature

Date

Signature of Parent/Guardian if Patient is a minor

Print Name of Parent/Guardian

For the Physician:

As the referring physician, I understand the benefits and limitations of this study and have requested that the above-named patient be tested. I attest to the fact that I have provided the patient with the information contained above and fully answered any questions. I believe that the patient understands the information and is voluntarily signing this informed consent.

Signature of Physician/Health Care Professional

Print Name of Physician/Health Care Professional