

Informed Consent for Genetic Testing

***Please check Treatment or Procedure: DNA Analysis (for specific gene, disease or genetic condition):**

- Filaggrin** Detection of at least one of five mutations (R501X, 2282del4, R2447X, S3247X and 3702delG) in the filaggrin gene.
- C2 Type 1** Detection of a 28 base pair deletion (rs9332736) within the C2 gene that leads to lack of Complement C2 protein production.
- CFTR** Detection of mutations and genetic variations in the Cystic Fibrosis Transmembrane Conductance regulator (CFTR) gene via full gene sequencing of all 27 exons, the polyT and TG repeat region of intron 9, and two intronic regions of the gene.
- STAT3** Detection of genomic mutations within the DNA binding and SH2 (SRC homology 2) domains of the signal transducer and activator of transcription 3 (STAT3) gene.
- Factor XII** Detection of a single nucleotide polymorphism in the Factor XII gene causing a Threonine to Lysine change at position 309 (T309K) or a Threonine to Arginine change at position 309 (T309R).

Other: _____

Before agreeing to have this treatment or procedure it is important that you read and understand this consent form. This consent describes the treatment or procedure and any risks it may involve. Please ask your doctor to explain any words or information you do not clearly understand.

What is involved in the procedure?

- A blood, saliva or tissue sample is collected for isolation and purification of DNA for molecular genetic testing. No more than 30 mL (about 1 ounce) of blood will be obtained.

What are the risks and discomforts of the treatment or procedure?

- There may be temporary pain and swelling at the site where the blood is drawn.
- DNA testing may cause emotional stress. You may have concerns about discrimination (insurance or work-related). National Jewish Health is committed to protecting your privacy. We treat all results with medical confidentiality. There are federal and state laws in place that protect you from discrimination by health insurance and employment.

What are the benefits of having this treatment or procedure?

- This test is designed to detect disease-causing mutations for the condition listed above. Whether or not mutations are present, this information may help diagnose and manage your condition. It will not detect all mutations within this gene, nor detect mutations in other genes. This test may have 3 possible outcomes:
 - Positive: The test shows that I have this condition or am at risk for developing this condition.
 - Negative: The test failed to find any significant abnormality in the gene. A negative result does not rule out a hereditary cause for this condition. There may be a small chance that I have this condition. Due to limitations in technology and incomplete knowledge of genes, some changes in DNA or protein products that cause disease may not be detected by the test.
 - Unknown Variant: The test revealed a change in one of the tested genes. It is unknown whether or not the change is disease-causing. As new information becomes available, it is possible that the variant may be re-classified as benign or disease-causing.
- This test may reveal unrecognized biological relationships (such as non-paternity). It may also reveal some other unknown familial genetic patterns.
- The results of this test may have implications for other family members. They may predict whether another family member has or is at risk for developing this condition, or is a carrier of this condition.
- Although genetic analysis often yields precise information, several sources of error are possible. These include, but are not limited to:

- Clinical misdiagnosis of the condition
- Sample misidentification

- Sample contamination
- Inaccurate information regarding family relationships

What may happen if you refuse this treatment or procedure?

- If you decide not to have this test, it may make the diagnosis and treatment difficult.

What are the alternatives to the treatment or procedure?

- You may choose not to undergo this procedure.

What are the benefits and risks of the alternatives?

- If you decide not to have this test, it may make the possibility of correct and/or appropriate diagnosis and treatment difficult.

No Guarantee:

- I understand that no guarantee or assurance has been made concerning the results of the test and that it may not diagnose my condition.
- National Jewish Health may contact me or my health provider if new information is learned that affects the interpretation of previously reported test results. A reasonable effort will be made to contact me through my doctor, or another person designated in writing. I may indicate my desire to opt out of being contacted by checking this box
- **DNA analysis is a fee-for-service test.** I understand that I am responsible for all charges for testing in the event my health plan does not reimburse for the test.
- My (or my child's or my unborn child's) sample may be used for test validation or education after personal identifiers are removed. Refusal to permit the use of my sample will not affect my test result. For such use, the sample may be stored indefinitely. I can withdraw my consent at any time by contacting the laboratory at (800) 550-6227 or by checking this box

Will my health information be kept confidential?

- Genetic testing is complex and there are important implications of test results. Results will be released only to:
 - the doctor ordering the test
 - an insurance provider requiring test results for reimbursement purposes
 - persons designated by me in writing
 - or as required by law

What if I have questions?

- If I have any questions about the treatment or procedure I can ask to speak with my doctor or a genetic counselor before signing this consent. I will not sign this consent unless I have read and understand the procedure.

My signature below indicates I have read, or had read to me, the above information and understand it. I have also read or had explained to me the specific disease(s) or condition(s) tested for, and the specific test(s) I am having performed. I have had the opportunity to discuss the purposes or possible risks of this testing with my doctor, a genetic counselor or someone my doctor has designated. The significance of a positive or negative test result based on my family history has been explained. I know that genetic counseling is available to me before and after the testing. I have all the information I want and all my questions have been answered.

I give my consent to have blood, saliva or a tissue sample sent to National Jewish Health Advanced Diagnostic Laboratory for DNA analysis for the above condition.

***Patient or Guardian Printed Name**

***Patient or Guardian Signature**

Date

Time

Provider Statement:

I have discussed the treatment/procedure, benefits, risks, consequences and alternatives, along with the benefits, risks and side effects related to the alternatives, and the risks related to not receiving the proposed care, treatment, and services with the patient or guardian. To the best of my knowledge, the patient or his/her guardian understands such consent to the proposed treatment/procedure.

***Health Provider Signature (who is reviewing this information with the patient)**

Date

Time