

Patient Information Sheet

Informed Consent for Genetic Testing

1. These tests look for changes in DNA, chromosomes, genes or gene products that are known to be associated with risk of specific diseases. The purpose of these tests is to help your doctor more accurately diagnose your current condition and/or future risk of disease.
2. In addition to assisting in making a diagnosis, these tests may reveal a genetic pre-disposition for one or more of these diseases. You may wish to get genetic counseling before consenting to this test. If a positive result is obtained, additional testing and/or genetic counseling follow-up may be advised.
3. Your doctor has ordered one or more of the following tests:

FACTOR V LEIDEN - This is a test for a variation in another gene that affects blood clotting. If this variant is present, it may indicate a higher than average risk for developing blood clots.

PROTHROMBIN GENE MUTATION - This is a test for a variation in another gene that affects blood clotting. If this variant is present, it may indicate a higher than average risk for developing blood clots.

HEMOCHROMATOSIS MUTATIONS - This is a test for a variation in a gene that controls iron storage in the body, and helps to diagnose hereditary hemochromatosis, a disease of iron overload.

4. The results of these genetic tests assist in diagnosis, but do not by themselves permit diagnosis of a disease condition. Genetic testing is ordinarily highly accurate; however, in some cases results may not be obtained or may be inconclusive. Only certain variations in these genes are known and available for testing, so a "normal" result in these genetic tests cannot guarantee that you do not have or will not develop a particular disease. Some genetic tests are only done by a few laboratories in the world, and may need to be sent to out of state laboratories that are not certified by New York State Health Department. In some instances, the clinical utility of the test may not be established.
5. Because interpretation of genetic test results is complex, the test results will be provided to your physician who will inform you of the results. To the extent permitted by law, all of the records and results of this testing are confidential and will not be released to anyone other than you, your referring doctors, and Strong Memorial Hospital Medical Records without your consent.
6. Besides providing excellent medical care, one of the missions of the University of Rochester and Strong Memorial Hospital is advancing medical science. Our doctors learn about better ways to care for patients and improve the health of people. No tests other than those authorized will be performed on your sample, and your sample will be destroyed after testing or not more than sixty days after the sample is taken, unless you consent to allow us to use the remaining sample for research. With your consent, our researchers will be able to use the remaining part of blood samples not needed for the specific tests above for approved development of new or improved laboratory tests. Names and other identifying information are kept strictly confidential. Samples with consent for research are stored indefinitely. We sometimes conduct follow-up studies based on new medical information. If you are eligible, someone will contact you personally. Participation in such studies is voluntary.