
Main Line HealthCare



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Main Line Primary Care

Genetic Testing - The Blueprints of our Lives

Genetic medicine begins with a genetic test. The test itself is nothing more than a mouth swab or blood draw that is sent to an appropriate laboratory. Such tests analyze specific sections of a patient's DNA, RNA, or metabolites found in the sample. The patient's profile is then correlated to the latest research that interprets the implications in terms of specific diseases or drug metabolism.

Currently genetic tests can detect susceptibility to more than 1,354 diseases. More are added every day. The tests are conducted in more than 610 laboratories throughout the United States, and that number is growing on a perpetual basis. (This data is provided by www.Genetests.org, a non-profit database supported by the NIH)

Genetic Testing for Diagnosis

Genetic testing can lead to more informed and more accurate diagnosis and treatment. When a patient comes to you with symptoms, you know that an immediate and certain diagnosis is often not possible. You gather the evidence by tests and data and prescribe various therapeutic measures. Genetic testing is a source of a new level of evidence, based on inherited tendencies and susceptibilities unique to each patient. Test results help you flag the most likely possibilities, reduce the guess work, and begin appropriate treatments. Genetic testing, although powerful, is an additional source of data for each patient. It rarely gives a definitive result by itself. It is data to be weighed with other evidence, such as physical and diagnostic examinations, biopsies, and more conventional tests.

Examples of Genetic Tests for Disease

Muscular Dystrophy: A test for the deletion in the dystrophin gene, the cause of Duchene's muscular dystrophy, can be used to identify women who are carriers of this condition.

Hemochromatosis: A patient with hereditary hemochromatosis can be spared cirrhosis by the early initiation of phlebotomy treatments. Gene tests check for a mutation in the HFE gene causing Iron overload.

Breast Cancer: Mutations in the BRCA1 and BRCA2 genes are associated with an increased risk of breast and ovarian cancer. Mutations in either of these genes confer a lifetime risk of breast cancer of between 60-85% and ovarian cancer between 15-40%.

Genetic Testing for Prediction

A patient may ask about an inherited disease that runs in the family. Genetic tests can confirm if the mutation associated with the disease is carried by that family member.

Genetic counselors are trained to evaluate such inheritance patterns within families and across generations. They can help a patient decide when a genetic test is recommended and then interpret the test results for risk of the disease in the context of the full family history.

An example is Hereditary Colon Cancer. A thorough family history is done first, identifying cancer of all types and family members with cancer, age of onset, and history of adenomas. If molecular genetic testing verifies the diagnosis of one family member, other relatives should be encouraged to get tested. The types of hereditary cancer are described below:

- 1) Familial adenomatous polyposis presents as multiple colonic polyps in the distal colon. It occurs at an early age and patients may have a genetic mutation within the APC germline.
- 2) Hereditary nonpolyposis colorectal cancer is the most common form of hereditary colorectal cancer. It is caused by a germ line mutation in the MLH1 and MLH2 genes.