Henry Ford Health System
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Fax: 313.916.2385
www.henryford.com/pathology

HENRY FORD CENTER FOR PRECISION DIAGNOSTICS

Informed Consent Requirements

Genetic counseling and informed consent is recommended prior to all genetic testing and is required prior to predictive testing.

Due to the complexities of genetic testing, a frank discussion about the risks and benefits, and possible outcomes, is recommended prior to placing a test request for any type of genetic testing. Counseling should be performed by a genetic counselor or qualified individual.

The ordering clinician is responsible for explaining the issues with genetic testing as relevant to the specific test requested within the clinical context presented.

1. The patient has the right to refuse testing
2. Limitations of testing
    a. Different methodologies detect different types of changes (sequencing, deletion/duplication analysis, microarray, chromosome analysis)
    b. Disorders can be associated with changes in one of multiple genes
    c. A positive result may not indicate severity of the condition
3. Benefits of testing
    a. Confirmation of a diagnosis provides definitive answers for symptoms and may indicate precise clinical follow up and treatment
    b. A result indicating increased risk for developing a disease provides benefit by improving compliance with screening, early detection and early intervention.
    c. A negative result, in some cases, can rule out a specific disorder and thereby improve diagnosis of the true cause of the presenting symptoms
    d. Genetic testing can provide critical information for other family members
    e. A confirmed diagnosis may allow for enrollment in research studies which could lead to new treatments
4. Risks associated with testing
    a. The physical risk from a blood draw is minimal. Some genetic tests require alternate sample types that can be associated with a slightly increased risk (amniocentesis, muscle biopsy) due to the procedure.
    b. Reactions to receiving results of genetic testing vary greatly between individuals and can be stressful. Psychological and social risks may occur based upon individual perceptions of the impact on quality of life.
    c. Genetic results can impact extended family and testing of additional family members may be recommended. Providing guidance to patients for structuring these conversations may help reduce stress.
    d. Results of genetic testing can indicate the genetic background of family members and may reveal unexpected results such as non-paternity.
    e. Some genetic testing may result in a diagnosis of a different disease that is outside the scope of testing. When possible, testing should be as limited as possible to confirm or rule out the disorder. If a familial mutation is known, it is recommended to test for only that mutation.

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5. Possible result outcomes
    a. Negative or "normal" indicates no change in the region tested (refer to limitations). A negative result does not necessarily rule out a disease. Some diseases are caused by more than one gene, or by genetic changes not detected by the test methodology used. A referral to genetic counseling may be indicated to assess for residual risk or alternative testing methods.
    b. Positive result indicated that a disease-associated change was detected. A positive result may indicate carrier status (for recessive disorders), which may have an increased risk for having an affected child. A positive result may indicate a a change that is known to cause disease, yet may not reliably predict severity. A positive result for predictive testing (hereditary cancer syndromes) indicates an increased risk for disease, but does not mean that the disease will occur.
    c. Inconclusive (VUS-variant of unknown significance) results indicate that a change was detected, yet that change is not clearly associated with disease, nor has it been shown to occur within unaffected population. An inconclusive result can be frustrating or stressful for the patient and they should be informed of this possibility prior to initiating testing. Inconclusive results are monitored and maybe updated to benign (normal) or disease-causing (pathogenic). Patients with inconclusive results are encourage to maintain contact with their clinician so they can be contacted if the status of the finding is updated in the future.
6. Secondary or unexpected findings
    a. Occasionally genetics testing may identify a change resulting in a diagnosis that is outside of the scope of the intended testing.
    b. The risk of secondary findings is increased by testing of a larger portion of the genome including large sequencing panels and microarray whole genome testing.
    c. Reducing the risk of secondary findings is accomplished by ordering the most specific, targeted testing for the patient presentation and indication.
    d. Referral to formal genetic counseling or contact the center for precision diagnostics is indicated for questions or concerns.
7. For additional detailed information on informed consent, visit the State of Michigan's link listed below:
    Informed Consent for Genetic Testing Handbook
    http://www.michigan.gov/documents/InformedConsent_69182_7.pdf

Definitions

Diagnostic – testing can confirm a suspected diagnosis (Down Syndrome)

Predictive – a positive result indicates an increased risk for developing a disease, yet is not certain (Hereditary Cancer Predisposition Syndrome)

Presymptomatic – a positive result indicates the individual will eventually develop the disorder (Huntington Disease)

Variant of Unknown significance (VUS) – a change that is not clearly benign nor pathogenic

Secondary finding – unintended result that is outside the scope of testing