ASCLS Mission:

The mission of ASCLS is to make a positive impact in health care through leadership that will assure excellence in the practice of laboratory medicine.

Your Safety and Service Experience Are Important To Us!

Laboratory Patient Safety Tips:

Genetic Testing

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What is a genetic test?

It is a medical laboratory test that examines DNA – specifically, components of DNA such as chromosomes and genes, or proteins that are produced as a result of genetic programming.

- **DNA** (deoxyribonucleic acid) is the molecule within the nucleus of cells that provides the instructions/blueprint/code for how cells grow, develop, function and reproduce.
- In the nucleus of each cell, the DNA molecule is packaged into thread-like structures called **chromosomes**. Each **chromosome** is made up of DNA tightly coiled many times around proteins that support its structure.
- **Genes** are found on chromosomes at specific places and in a specific order. A gene is the basic physical and functional unit of heredity. **Genes**, which are made up of DNA, act as the instructions for making proteins.
- **Proteins** are the molecules in the body that are responsible for the growth and repair of cells.
- A **mutation** is a permanent change in the **DNA** structure that makes up a **gene**. This means that the instructions could be deleted, altered, rearranged, or in some cases turned off. If there is a mutation the cell may not function normally.

How can gene mutations affect my health and development?

To function correctly, each cell depends on thousands of proteins to work correctly. Sometimes, gene mutations prevent the proteins from working properly. When a mutation alters a protein that plays a critical role in the body, it can prevent normal development or cause a medical condition. A condition caused by mutations in one or more genes is called a genetic disorder.

Do all gene mutations affect health and development?

No; only a small percentage of mutations cause genetic disorders—most have no impact on health or development.

Why would my doctor recommend that I have a genetic test?

Genetic tests are used in conjunction with other medical information (such as physical examination, patient history, family history, and other test results) to identify genetic conditions, birth defects, chromosomal disorders, and other conditions, including some cancers.

What will the results of a genetic test tell my physician?

Together with the patient's medical history and family medical history, the results of a genetic test may:

- Confirm a diagnosis
- Guide treatment and management of an existing disease
- Determine if the individual is a carrier of a specific mutation, which could potentially affect their children
- Detect increased risk for developing a disease or condition in the future

For some situations, a genetic counselor may be consulted to provide guidance on the specific test to be ordered and to interpret the test results with the physician and the patient.

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Are there benefits, risks, limitations and costs to having a genetic test?

There are risks and benefits to undergoing any medical procedure. Benefits and risks may vary and must be determined for each individual's situation; however, risks of collecting a specimen for genetic testing are minimal and may include either collecting a blood specimen or cheek cells (using a swab).

Undergoing genetic testing may cause additional worry or anxiety, if it shows that the individual has a gene that may cause a genetic disorder for which there is no treatment.

Genetic tests are consistently more expensive than other laboratory tests. Genetic testing may not be covered by health insurance.

If your physician has ordered a genetic test for you, it is important to ask: What are the benefits and what are the risks of having this genetic test?

What questions should I be asking if I am told that I need a genetic test?

As a patient, you have the right to ask your physician questions about testing that will be performed to determine your diagnosis, treatment and prognosis. In addition, it is appropriate to ask your health insurance company specific questions on whether the test will be covered by your plan or if you will be responsible for the cost of the test.

Questions to ask your care provider:

- What is the name of the test?
- What type of information will it provide to me and my physician for my situation?
- Who will interpret the test information?
- Will I be able to talk with a genetic counselor?
- How much will it cost?
- Will my health insurance cover the cost?
- Do I need to get the testing approved by my insurance before I have the test?
- How much will my cost be, if my insurance does not pay for the test?
- If insurance will pay for the test, what is the co-pay and/or deductible that I will be charged if I have this specific test?
- Are there any possible risks associated with genetic testing results being in my medical record and the medical care or insurance coverage that I might need in the future?

Where can I find reliable resources on genetic testing?

NIH U.S. National Library of Medicine Genetics Home Reference: https://ghr.nlm.nih.gov