M.Sc Mazin Eidan Hadi
mazin.eidan@uomus.edu.iq

What is the Genetic Testing?

Genetic testing is "the analysis of chromosomes (DNA), proteins, and certain metabolites in order to detect heritable disease-related genotypes, mutations, phenotypes, or karyotypes for clinical purposes." It can provide information about a person's genes and chromosomes throughout life. **Genetic tests can be used for many different purposes.**

Testing include:

1-Newborn screening: Newborn screening is used just after birth to identify genetic disorders that can be treated early in life. A blood sample is collected with a heel prick from the newborn 24–48 hours after birth and sent to the lab for analysis.

2-Diagnostic testing: Diagnostic testing is used to diagnose or rule out a specific genetic or chromosomal condition. In many cases, genetic testing is used to confirm a diagnosis when a particular condition is suspected based on physical mutations and symptoms.

3-Carrier testing: Carrier testing is used to identify people who carry one copy of a gene mutation that, when present in two copies, causes a genetic disorder.

4-Preimplantation genetic diagnosis: Genetic testing procedures that are performed on human embryos prior to the implantation as part of an in vitro fertilization procedure. Pre-implantation testing is used when individuals try to conceive a child through in vitro fertilization. Eggs from the woman and sperm from the man are removed and fertilized outside the body to create multiple embryos. The embryos are individually screened for abnormalities, and the ones without abnormalities are implanted in the uterus.

5-Prenatal diagnosis: Used to detect changes in a fetus's genes or chromosomes before birth. This type of testing is offered to couples with an increased risk of having a baby with a genetic or chromosomal disorder.

6- Predictive and PR symptomatic testing: Predictive and PR symptomatic types of testing are used to detect gene mutations associated with disorders that appearafter birth, often later in life. These tests can be helpful to people who have a family member with a genetic disorder Predictive testing can identify mutations that increase a person's chances of developing disorders with a genetic basis, such as certain types of cancer.

7-Pharmacogenomics: type of genetic testing that determines the influence of genetic variation on drug response. When a person has a disease or health condition, this type of genetic testing can be used for cancer patients undergoing chemotherapy, A sample of the cancer tissue can be sent in for genetic analysis by a specialized lab. After analysis, information retrieved can identify mutations in the tumor which can be used to determine the best treatment option.

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Non-diagnostic testing includes:

1- Forensic testing: Forensic testing uses DNA sequences to identify an individual for legal purposes. Unlike the tests described above, forensic testing is not used to detect gene mutations associated with disease. This type of testing can identify crime or catastrophe victims, rule out or implicate a crime suspect, or establish biological relationships between people (for example, paternity).

2- Paternity testing: This type of genetic test uses special DNA markers to identify the same or similar inheritance patterns between related individuals. Based on the fact that we all inherit half of our DNA from the father, and half from the mother, DNA scientists test individuals to find the match of DNA sequences at some highly differential markers to draw the conclusion of relatedness.

3- Genealogical DNA test: To determine ancestry or ethnic heritage for genetic genealogy

4- Research testing: Research testing includes finding unknown genes, learning how genes work and advancing our understanding of genetic conditions. The results of testing done as part of a research study are usually not available to patients or their healthcare providers.

Types of Genetic Testing

Several different methods are currently used in genetic testing laboratories. The type of test will depend on the type of abnormality that is being measured. In general, three major types of genetic testing are available—cytogenetic, biochemical, and molecular testing to detect abnormalities in chromosome structure, protein function, or DNA sequence, respectively.

1- Cytogenetic Testing

Cytogenetics involves the examination of whole chromosomes for abnormalities. Chromosomes of a dividing human cell can be clearly analyzed under a microscope. White blood cells, specifically T lymphocytes, are the most readily accessible cells for cytogenetic analysis since they are easily collected from blood and are capable of rapid division in cell culture. Cells from other tissues such as bone marrow (for leukemia), amniotic fluid (prenatal diagnosis), and other tissue biopsies can also be cultured for cytogenetic analysis.

Following several days of cell culture, chromosomes are fixed, spread on microscope slides and then stained. The staining methods for routine analysis allow each of the

chromosomes to be individually identified. The distinct bands of each chromosome revealed by staining allow for analysis of chromosome structure.

2- Biochemical Testing

The enormous numbers of biochemical reactions that routinely occur in cells require different types of proteins. Several classes of proteins exist to fulfill the multiple functions, such as enzymes, transporters, structural proteins, regulatory proteins, receptors, and hormones. A mutation in any type of protein can result in disease if the mutation ultimately results in failure of the protein to correctly function.