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Advances in Medical Ethics Journal

Commentary

An Overview on Genetic Testing

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DESCRIPTION

Genetic testing, often known as DNA testing, is used to detect changes in the sequence of DNA or the structure of chromosomes. Measurement of the results of genetic changes, such as RNA analysis as a gene expression output or biochemical analysis to assess specific protein output, is another type of genetic testing. Genetic testing can be used in a medical environment to diagnose or rule out suspected genetic illnesses, estimate risks for certain conditions, or acquire information that can be utilized to modify medical treatments based on an individual's genetic makeup. Genetic testing can also be used to determine biological relationships, such as a child's biological parentage (genetic mother and father) *via* DNA paternity testing, or to estimate a person's lineage in broad terms. Plant and animal genetic testing can be used for the same reasons as human genetic testing (e.g., to assess relatedness/ancestry or predict/diagnose hereditary disorders), to gather data for selective breeding, or to assist endangered species in increasing genetic diversity.

The number of genetic testing available has grown over time. Counting the number of chromosomes per cell was one of the first techniques of genetic testing, which was introduced in the 1950s. Certain genetic diseases, such as trisomy 21 (Down syndrome) or monosomy X, can be diagnosed if the number of chromosomes differs from the predicted number (46 in humans) (Turner syndrome). In the 1970s, chromosomal banding, a technique for staining specific areas of chromosomes, was developed, allowing for a more precise investigation of chromosome structure and the detection of diseases large structural rearrangements were involved in genetic diseases. Genetic testing has extended to include the fields of molecular genetics and genomics, which may detect changes at the level of individual genes, sections of genes, or even single nucleotide "letters" of DNA sequence, in addition to studying full chromosomes (cytogenetics). There are tests available for more than 2,000 genetic diseases, according to the National Institutes of Health, and one study claimed that there were more than 75,000 genetic tests on the market in 2017.

"The examination of chromosomes (DNA), proteins, and specific metabolites in order to detect heritable disease-related genotypes, mutations, phenotypes, or karyotypes for therapeutic purposes," were according to the National Institutes of Health. It can provide lifelong information on a person's genes and chromosomes.

Diagnostic testing

Cff DNA (cell-free foetal DNA) testing is a non-invasive (for the foetus) test. It is performed on a sample of the mother's venous blood and can reveal information about the foetus as early as the first trimester. It is the most sensitive and specific Down syndrome screening test as of 2015. Newborn screening is performed shortly after delivery to detect genetic problems that can be addressed early. A heel prick is used to get a blood sample from the infant 24–48 hours after birth, which is then sent to the lab for analysis. The newborn screening technique varies by state in the United States, however all states are required to screen for at least 21 diseases. The presence of aberrant results does not necessarily imply that the child has the disorder.

To confirm the condition, diagnostic tests must be performed after the initial screening. The most common application of genetic testing is the routine screening of neonates for certain diseases. In the United States, millions of infants are tested each year. Infants are now tested for phenylketonuria (a hereditary condition that can lead to mental illness if left untreated) and congenital hypothyroidism in all 50 states (a disorder of the thyroid gland).

Diagnostic testing is used to determine the presence or absence of a certain genetic or chromosomal problem. When a certain illness is suspected based on physical mutations and symptoms, genetic testing is frequently utilized to confirm the diagnosis. Diagnostic testing is available at any point in a person's life; however it is not available for all genes or genetic diseases. The results of a diagnostic test can have an impact on a person's health-care decisions and disease treatment. People with a family history of Polycystic Kidney Disease (PKD) who suffer abdominal discomfort or tenderness, blood in their urine, frequent urination, and pain in the sides, a urinary tract infection, or kidney stones may choose to have their genes checked, and the results may confirm the diagnosis of PKD. Despite the numerous implications of genetic testing in illnesses such as

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Received: 12-Apr-2022, Manuscript No. LDAME-22-16747; **Editor assigned:** 15-Apr-2022, PreQC No. LDAME-22-16747(PQ); **Reviewed:** 02-May-2022, QC No. LDAME-22-16747; **Revised:** 09-May-2022, Manuscript No. LDAME-22-16747(R); **Published:** 17-May-2022, DOI: 10.35248/2385-5495.22.8.11.

Citation: Simmon L (2022) An Overview on Genetic Testing. Adv Med Ethics J.8: 11.

epilepsy and neurodevelopmental disorders, many patients (particularly adults) lack access to these sophisticated diagnostic techniques, indicating a significant diagnostic gap.

Carrier testing identifies persons who have one copy of a gene mutation that causes a genetic condition when two copies are present. Individuals with a family history of a genetic problem, as well as people from ethnic groups with a higher risk of specific genetic conditions are eligible for this type of testing. If both parents are tested, the test can reveal a couple's chance of having a child with a genetic disorder such as cystic fibrosis.

Non-diagnostic testing is a type of non-diagnostic testing

- Forensic testing-this type of testing uses DNA sequences to identify a person for legal purposes. Unlike the tests outlined above, forensic testing does not look for disease-causing gene alterations. This type of testing can be used to identify crime or disaster victims, rule out or incriminate a suspect in a crime, or establish biological links between individuals (for example, paternity).
- Using unique DNA markers, paternity testing identifies the same or similar inheritance patterns between related individuals. Based on the fact that we all inherit half of our DNA from father and half from mother, DNA professionals assess individuals to find a match of DNA sequences at specific highly divergent indicators to determine relatedness.
- A genetic genealogy DNA test is performed to determine ancestry or ethnic heritage. Research testing entails the discovery of unknown genes, the study of gene function, and the advancement of genetic knowledge. Patients and their healthcare professionals rarely have access to the findings of testing conducted as part of a research study.