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The Right of Genetic Testing in Healthcare

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Genetic testing is a type of medical examination that identifies changes in genes, chromosomes, or proteins. Genetic test results can either confirm or rule out a suspected genetic predisposition or help determine a person's potential for developing or transmitting a genetic predisposition. Genetic testing involves looking at changes in

Genetics: Gene examines DNA sequences to detect mutations (mutations) in genes that can cause or increase the risk of genetic disorders. Genetic testing can be either small or large in size, analyzing DNA building block (nucleotide), one or more genes, or the entire human DNA (known as the genome).

Chromosomes: Chromosomal genetic testing analyzes complete chromosomes or long DNA lengths to determine whether there are major genetic mutations, such as an extra copy of a chromosome, which causes genetic predisposition. Genetic testing is voluntary. Because testing has advantages and disadvantages and risks, the decision about whether to test is personal and complex.

The Different types of test undertaken for the Genetics testing: Diagnostic tests are usually ordered when the clinician has reason to think - because the patient has the appropriate symptoms, known as a 'clinical presentation' - that the patient may have a specific genetic condition. The doctor may use the test to confirm their initial diagnosis, or alternatively to rule out a possible cause of the symptoms. Tests can also help distinguish between two or more cases with similar symptoms.

Hypothetical tests are performed to determine if someone is at risk or not before showing any symptoms. More and more people are ordering randomized controlled trials, but in a medical setting predictable screening is usually ordered when a genetic condition is found in a family member, and doctors want to determine who in the family is affected. In this case, the underlying genetic variation is often already identified, so more targeted tests are used.

Pharmacogenomics is the study of genomic drug response responses, and testing allows clinicians to obtain information about how the human genome affects their response to medication. This information may include whether a particular drug will work or not, and how likely it is that the drug will cause some side effects. This information can be taken into account by physicians and other factors such as patient age and weight, and any other medical conditions a patient has, when making decisions about the best treatment option and individual dosage.

Comparing the genes of a patient's tumor cells with those of a patient's healthy cells can also help to identify genes that work in other ways. For example, it may help to identify the target of new research as genes that appear to 'drive' cancer to develop and spread. It is expected that the wealth of information generated by the new sequencing methods will be useful especially when it comes to studying the progression of cancer and selecting patients for clinical trials. Indeed about 50% of the results retrieved through the 100,000 Genomes Project cancer program show the potential for treatment and / or clinical testing.

Other types of hypothetical tests also exist, although these currently have fewer clinical applications and are therefore often seen in research or in specific cases targeted at the consumer. There are consumer studies available that look at single genetic variations associated with an increased (or decreased) risk of certain diseases. There are also genetic studies that each have a small effect on a particular factor, and then seek to combine these results into a 'polygenic score' - a summary of a person's risk of infection, compared to a risk factor. To have the same attitude among people. Although interesting, those polygen points are the subject of ongoing debate and provide only possible information. They are rarely used in the clinic.

Conflict of Interest

The authors declared no potential conflicts of interest for the research, authorship, and/or publication of this article.

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