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Information Bulletin

Genetic Testing

What is genetic testing?

Genetic testing examines DNA or proteins to identify anomalies linked to particular diseases, or to determine ancestry, paternity or ethnicity.

How is the testing done?

Genetic testing usually involves taking a biological sample which can be blood, cells or other body fluids and tissues. Typically a genetic test looks for changes in the DNA sequence which mean that a protein may not be produced in the right amount, or produced in a form that will not function, or is not produced at all.

Genomic testing looks at the whole genome (all genes and regions in between), exome sequencing and panels where more than five genes are evaluated simultaneously.

Targeted genetic testing looks for variations in specific genes. This can be done in a variety of ways including gene testing by genotyping, exome sequencing or microarray analysis.

Genetic screening includes genetic tests that are recommended to all members of a particular group (e.g., newborns, members of particular ethnic groups).

Why consider a genetic test?

Genetic testing can serve many purposes, but the results are not always easy to understand. Geneticists and genetic counsellors can help to interpret results and support decision making. Different types of genetic testing are carried out for different purposes.

Pre-symptomatic or predictive genetic testing is used to assess the risk of developing a specific disease. This information can help to inform decisions around lifestyle and healthcare.

Diagnostic testing determines what is making a person ill and, more than understanding the cause of a disease, diagnostic testing reveals the molecular characteristics of the disease to help better understand and classify it. In some cases, but not all, this can help physicians determine the most appropriate treatment for each patient and help better manage a particular disease and manage health issues.

Prognostic testing can help to predict the course of a disease by assessing the level of expression of certain genes. This information can be used to optimize treatment and decision making.

Phamacogenomic testing assesses how certain drugs are processed by an individual's body, which can help physicians determine which drugs will be most effective.

Carrier testing is useful to couples considering pregnancy and looks for genetic changes that could be passed on to offspring and cause serious disorders. Some of the more common disorders screened for include cystic fibrosis, sickle cell disease, thalassemia, and Tay-Sachs disease, but there are more than 100 others that can be tested for.

Prenatal genetic screening is used to estimate the risk of certain DNA abnormalities before babies are born. These include Down syndrome, Edward syndrome, Patau syndrome and several other uncommon syndromes.

Newborn screening is used to test for inherited diseases that can be treated early in life and have dire consequences if left untreated.

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Limitations of genetic testing

Genetic testing provides a limited amount of information about diseases with a genetic component. In the case of predictive genetic testing, tests often assess the probability of developing a particular disease and cannot determine when the disease will manifest or how severe the symptoms will be. Most genetic diseases result from the effects of several different genes or from interactions between genes and the environment. In these cases, genetic testing is not very informative. Despite advances in DNA analysis, identifying and understanding the meaning of genetic changes is not always easy.

Ethical Issues

There are many reasons to utilize genetic testing. Individual reasons for accessing or abstaining from genetic testing are complex and private. At the moment, Canada does not have any laws in place to prevent individuals from being compelled to submit to genetic testing or to share the results of genetic tests, although BILL S-201, An Act to prohibit and prevent genetic discrimination has been introduced before Parliament.

http://www.parl.gc.ca/HousePublications/Publication.aspx?DocId=6257111

More Information and Resources

http://healthycanadians.gc.ca/healthy-living-vie-saine/pregnancy-grossesse/fertility-fertilite/gen-test-eng.php

https://ghr.nlm.nih.gov/handbook/testing?show=all

http://www.huffingtonpost.ca/2013/04/17/genetic-non-discrimination-act-bill_n_3102527.html

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