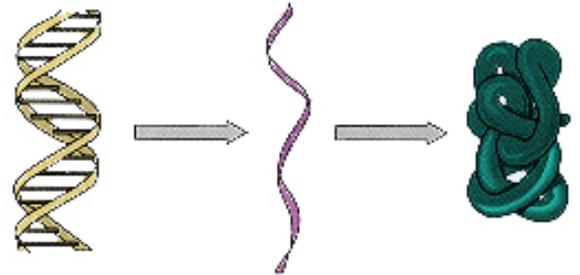


Understanding Genetics:

A Guide for Patients on Whole Genome and Whole Exome Sequencing



What is Whole Genome Sequencing?

Your “genome” can be found in every cell of your body and is your complete set of DNA. It is the unique blueprint for your body. Your genome is made of individual units called genes, and each gene has a different purpose. The part of your genes that code for proteins is called the “exome.”

- Whole genome sequencing (WGS) is a complete readout of all the information that makes up all of your DNA.
- Whole exome sequencing (WES) is a subset of the whole genome, and is a readout of the parts of your DNA that code for proteins.
- Currently, WGS is not widely offered. However, as technology and scientific knowledge advances this may change. WES is available if a doctor orders the testing.

What Can You Learn From WES/WGS?

The amount of information that we can learn from these tests is constantly changing due to our expanding understanding of genetic sequences. Both tests can provide information about your family, your current health, and future health concerns. You can learn about:

- Specific, single gene disorders that you may have.
- Certain ‘multi-factorial’ gene disorders. These are disorders due to the effects of multiple genes, sometimes in combination with environmental factors.
- Pharmacogenomic profiles:
 - What medications may be beneficial to you specifically.
 - What medications may be ineffective or harmful to you.
- Genetic diseases you may have a predisposition to:
 - Alzheimer’s disease (and other forms of dementia), breast cancer, and others.
- Family relationship information:
 - Degrees of relatedness can be discovered between family members who have had WES/WGS done

Who Gets These Tests?

These tests are not recommended for the general public.

- A limited subset of patients receive them:
 - Individuals who have had other types of genetic tests, which did not result in a diagnosis.
 - Certain people involved in clinical trials or research studies.
- Anyone considering these tests must consent and understand the potential benefits and limitations of the testing.

What You Can’t Learn from Sequencing

Seventy-five percent of people who undergo sequencing do not receive an official diagnosis.

- Some diseases are not caused by genetic conditions.
- Some conditions are genetic, but scientists have not identified the gene that causes them.
- The technology of WES/WGS is not perfect, and some genetic information is currently not obtainable.

Identifying the disease gene does not mean that there is a treatment or a cure, or even a name for that disease.

The Impacts of WGS

It is important to understand that sequencing can result in a number of secondary and potentially unwanted findings. Learning of these items can cause emotional distress that could be avoided by not being sequenced. Some examples include:

- Detection of variants of unknown significance (VUS). These are genetic changes that are identified in your genetic code, but the medical significance is not known.
- Identification of risk factors for secondary diseases that you were not expecting such as Alzheimer's. These diseases may not have a cure and/or may not affect you for many years.
- Identification of non-paternity.
- Identification of other at-risk family members. Your results mean other family members may get information about their genes and learn they may be at increased risk for a disease as well. Not everyone in your family may want to know this information.

Questions to Consider Before Testing

- Would another, simpler or targeted DNA test be better?
- What will my results mean for my family members who may share a similar genetic makeup?
- Is there any information about disease risk that I would not want to know?
- Do I want to know about something that may not affect me in the next 10 or more years?
- Do I want to know about a genetic disease for which there is no treatment?
- How do I feel about receiving uncertain results, or results that tell me 'maybe' rather than 'yes' or 'no'?
- Will my insurance company pay for testing? Some insurance companies cover WES, others do not. It is important to talk with your insurance company about their policy prior to testing.

References and Other Information

Visit the sites below for information on testing, coverage of testing, and to find a genetic counselor near you

Genes In Life: Answering Your Questions about Health & Genetics- Genesinlife.org

Genetic Information Nondiscrimination Act- GINAhelp.org

Find a Genetic Counselor Directory- NSGC.org

References:

<http://www.genome.gov/19516567>

<http://genesinlife.org/testing-services/testing-genetic-conditions/whole-genome-sequencing>

http://www.ashg.org/education/genetic_testing.shtml

<http://www.cincinnatichildrens.org/service/d/diagnostic-labs/molecular-genetics/whole-exome-sequencing/families/default/>

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