"Raw" DNA Data

Genetic Information Fact Sheet

This handout is intended to provide you with information about raw DNA data and what you can do with this data. As with any genetic testing, there are important things to consider before you decide what actions you will take with your raw DNA data.

What is DNA?

Deoxyribonucleic acid, or DNA, is the main component of our chromosomes found in almost every cell of our body. DNA is made up of four building blocks called nucleotides: adenine (A), thymine (T), guanine (G), and cytosine (C). The order of these four nucleotides is your "genetic code". A gene is a section of DNA that tells the body how to make a certain protein. Each gene is like an instruction that the body reads and then interprets to build a protein. Proteins play critical roles in the body and are required for the structure, function, and regulation of the body's tissues and organs. Currently, it is thought that every human has about 20,000 protein coding genes in most cells of their body. The complete set of genetic information within the cell is called the genome. Greater than 99% of a human genome is identical while about 0.1% is unique to the individual.

What is DNA sequencing and genotyping?

DNA sequencing is the process of determining the exact order of nucleotides within a person's DNA. Sometimes the entire genome is sequenced (whole genome sequencing) while other times only protein-encoding genes (whole exome sequencing) or desired genes (single-gene sequencing) are sequenced. DNA genotyping is the process of determining what nucleotides are present at specific places within the DNA. Therefore, genotyping is like reading a few highlighted words on a page while sequencing is like reading whole sentences or paragraphs.

What can sequencing or genotyping tell us?

Once the nucleotide information is available, the laboratory can determine if there are any changes (also called variants) within the DNA. An example of a variant would be the replacement of an adenine (A) for a guanine (G). Most variants are benign, meaning they are not harmful and have no effect on the gene they are located in. These variants are what simply make us unique. Other variants may predispose you to disease or cause disease. Last, there are many variants that we do not know if they are harmful or not (variants of uncertain significance).

What is raw DNA data?

Raw DNA data is a computer file that provides the order of the nucleotides within the regions sequenced or provides a list of the nucleotides at the specific locations tested during genotyping. Raw DNA data is not interpreted; meaning it does not tell you what variants may be present in your DNA sequence. Raw DNA data is also not validated, meaning it may not be accurate as it has not undergone the checks that are performed on clinical test results (see below).

What do people do with raw DNA data?

Some individuals choose to upload their raw DNA data to a third-party website to receive an "interpretation" of their information. However, several things should be considered before deciding to do so:

Validity: There are many different platforms and resources third-party sites use to interpret raw DNA data. Therefore, an individual may receive different results from different companies. In addition, a study found a percentage of variants, reported by third-party sites as disease causing variants, were actually benign when tested by a clinical laboratory.

Utility: Given the questionable validity of third-party site interpretations, individuals and medical providers are advised to not make medical decisions or changes in clinical care based on results of raw DNA data only. In addition, results may not be clinically meaningful. In other words, they may not change your medical care.

Informed consent: An informed consent provides the known risks and benefits of using a third-party interpretation service. One study looking at various third-party interpretation sites found that several companies had insufficient informed consent documents, which can lead to consumer harm if benefits and risks are not fully disclosed.

Food and Drug Administration (FDA): Currently, the FDA does not regulate third-party interpretation services. Therefore, the efficacy and safety of the information obtained from these sites has not been well studied and proven.

Are there risks in uploading my raw DNA data to a third-party website?

Yes, there are some risks to be considered before pursuing interpretation of raw data:

Privacy: The information obtained from direct-to-consumer testing, including raw DNA data and interpretation services, is not held to the same privacy standards as medical records, specifically those outlined by the Health Insurance Portability and Accountability Act (HIPAA). While many companies claim to not share or use your genetic information, there are not current practices to ensure this will not happen. In addition, a study looking into third-party interpretation services found that several companies were not clear on the risks associated with a privacy breach.

Psychological impact: Some individuals may learn they carry a variant in a gene that increases their risk for potentially serious conditions (cancer or heart disease, for example). One could also learn assumed family relationships are incorrect such as learning a family member is adopted or that paternity is incorrect. This information could have a negative impact on an individual for which they were not prepared.

Misinterpretation: While third-party sites will attempt to interpret the raw DNA data, there remains ambiguity in what the results actually mean. Studies have found that misunderstanding of the information obtained from third-party interpretation services is common in consumers. This can lead to unnecessary anxiety/worry and medical care costs.

What should I do with the information I learn from a third-party website?

If you choose to upload your raw data DNA to a third-party interpretation site, we highly recommend researching the company first and reading their informed consent carefully, keeping in mind the information discussed in this handout. It is also critical to have any results from your raw DNA data confirmed by a clinical laboratory that has been CLIA-approved (see above regarding validity). This process is best done through a medical provider or genetic counselor who is familiar with genetic test results. Once the variant has been confirmed as pathogenic (disease-causing), appropriate changes in clinical care may be recommended.

If you have any further questions regarding raw DNA data, we recommend contacting a genetic counselor in your area (www.findageneticcounselor.com).

References

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