UNDERSTANDING DIRECT TO CONSUMER GENETIC TESTING

What is Direct to Consumer (DTC) Genetic Testing?

DTC genetic testing is genetic testing which typically does not require an order from a medical provider. This type of genetic testing is different from clinical genetic testing which are typically ordered by a doctor or genetic counselor. Oftentimes these tests are advertised directly to the public through advertisements on social media or television. Examples of DTC companies include 23andMe, AncestryDNA, and Sequencingdotcom. The type of genetic test can vary amongst the DTC labs ranging from genetic ancestry information to analysis of an individual's whole genome (all of their DNA).

Benefits of Testing

Accessibility is often cited as a major benefit of DTC genetic testing. Depending on where an individual lives, there may be long wait times to see a genetics professional or a general lack of specialized care. The cost of DTC testing may also be less than clinical testing that goes through an individual's insurance. Finally, an individual does not have to deal with insurance guidelines regarding testing or appointment coverage. All these factors may make DTC the more compelling option for patients.

Drawbacks of Testing

Typically DTC genetic testing labs do not have to undergo the same oversight as clinical laboratories. For labs to be deemed reputable in the United States they are expected to be certified by the government through CLIA and the independent College of American Pathologists (CAP). It should be noted that certain companies may have FDA approval to clinically analyze specific genes. For example 23andMe is approved to analyze 44 different variants (there are over 4000 known disease causing variants) in the BRCA1 and BRCA2 genes. However, this is not a norm within the DTC industry. Due to a lack of governmental oversight, DTC laboratories are not required to adhere to the same data privacy rules as clinical laboratories. Since the 23andMe data breach in 2023, there has been increased concern regarding the safety and storage of clients' genetic information. Genetic information provided by DTC laboratories to law enforcement without client consent has also been frequently documented in the news cycle. The safety of an individual's private genetic information depends upon the policies and protections that each company has in place.

The methodologies utilized by DTC laboratories are often unknown or lacking in rigor. Unlike clinical genetic laboratories, DTC labs typically lack confirmatory methods. This means that there is a higher likelihood of a false positive on DTC genetic testing. DTC laboratories have also been noted to have decreased read depth when analyzing DNA. This means that DTC genetic tests do not read the DNA as closely for errors that may be

causing disease. In an attempt to cut costs, many DTC companies utilize AI interpretation to determine if a genetic variant may be disease causing. Interpretation of genetic variants is extremely complex and nuanced requiring years of training. Current AI technology is unable to properly evaluate genetic variants in the manner that a trained professional might.

The most concerning methodological issue with DTC genetic testing is the high rate of error. A study conducted in 2018 estimated that up to 40% of DTC genetic testing results are incorrect (Tandy-Connor et al., 2018). This means that 40% of individuals are receiving false negative or false positive results, leading to increased anxiety or false reassurance. Due to these methodological errors, individuals with a positive result on DTC testing are often asked by their healthcare providers to undergo clinical testing to confirm results. This can lead to a patient paying twice for testing.

What to Watch Out For

Clinical genetic testing is more likely to provide a patient with accurate genetic information, however DTC testing may be the best option for an individual due to existing barriers. When considering a DTC genetic testing laboratory, you should consider what their privacy policy is. Does the company have a mechanism for you to request that your data be deleted? What protections does it have in place to prevent a data breach? What is it potentially missing in its methodology and/or testing capabilities? Those seeking DTC testing should look out for concerning business practices, such as selling supplements that it is claimed will cure a person's symptoms. A notable red flag are companies that purport to offer genetic testing for hypermobile ehlers danlos syndrome (hEDS). This condition is known to be hereditary, however we have yet to identify which gene may be responsible for hEDS. Any company advertising its ability to genetically diagnose hEDS is misrepresenting our current scientific knowledge and seeking to profit off of chronically ill individuals. Evaluating for these red flags can help individuals identify which DTC companies have increased ethical standards.

How to Access Clinical Testing

For those interested in clinical genetic testing, a meeting with a genetic counselor is recommended. A referral can be placed by an individual's doctor or one can connect with a local genetic counselor via https://findageneticcounselor.nsgc.org.

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