Issues of direct-to-consumer (dtc) genetic testing

Part 1

Genetic testing is a type of medical test that reveals information about a person’s genetic composition. Oftentimes, a physician guides the patient through the process and explains the results to them. However, in recent years, direct-to-consumer (DTC) genetic testing has exploded in popularity. These tests differ in that customers can buy them online without the need of a healthcare provider. The most popular tests claim to provide insight into someone’s ancestry or health risks. For instance, someone who uses the popular DTC genetic testing company, 23andme, may find out that they have a half-sister whom they never knew about, or that they are more susceptible to lung cancer than the average American. Since there is the potential of a customer to receive life-changing news, it is important to understand the process and implications of DTC genetic testing.

From a customer’s point of view, the process is straightforward: spit in a tube, send it to a lab, and wait for results. However, the steps taken to go from a little bit of spit to a full list of genetic information is less simple. First, someone orders a genetic testing kit online without the need for a physician’s prescription. Costs of DTC genetic tests range from $100 to a few thousand dollars. Next, they spit their saliva into a vial at home and then send it to a laboratory for testing. Saliva is collected instead of blood because the DNA in saliva has the quality and purity needed for genotyping. Also, saliva is less costly for sampling materials, less invasive to customers, and safer for operators. [1] At 23andme, when the samples arrive, technicians first scan the barcode of each sample in order to identify each customer’s saliva. The first test is a visual inspection to see if enough saliva is present. If

there is not a sufficient amount of liquid, a replacement kit will be shipped to the customer to recollect enough saliva. DNA is then extracted from the saliva cells and is amplified by PCR so that there is enough DNA to be genotyped. 23andme uses a unique genotyping chip manufactured by Illumina designed to capture key features about one's health and ancestry. [2] The chip is a small glass slide with millions of microscopic beads on its surface. Each bead is very tiny, about the size of a bacterial cell and is covered with tiny DNA fragments that have been synthesized to correspond to a particular SNP (single nucleotide polymorphism.) A SNP is a variation in the genetic code that is caused by a single base pair substitution in the DNA. Scientists have identified about 10 million SNPs out of the possible 3 billion base pairs. According to 23andme, not all of the SNPs' functions have been identified yet, but they have picked 600, 000 SNPs to measure that have been proved to have contributions to health and ancestry to include in their analysis. The amplified DNA is then cut into smaller pieces and is labeled with a fluorescent dye. The DNA fragment used for testing is one strand of DNA that extends up to the SNP region, but does not include the SNP. [3] Afterwards, it undergoes hybridization, where the DNA strands are placed on the chip and incubated so that the DNA denatures and each separated strand forms a bond with the corresponding sequences on the beads. [4] Next, according to Laframboise, the bead is incubated with a mixture of nucleotides that have been tagged with a fluorescent dye. DNA polymerase moves across the bead and at each position, it elongates the DNA fragment by bonding the corresponding fluorescent nucleotide. For instance, if the SNP genotype of an individual was AA, it would be matched with red fluorescently

labeled T’s and would glow red. Similarly, when another SNP location is GG, it will be matched with green fluorescently labeled C’s and would glow green. If another SNP location is AG, then it will be matched with both red T’s and green C’s and will glow yellow. When the chip is scanned by a high energy light source, the color that each bead fluoresces is used to note which allele is present at every position that the beads are testing. [5] Since the genetic sequence of each bead and the function of the gene they encode are also already known, the intensity of the light emitted from the beads is examined to determine which genes are most active. This SNP genotyping technology is accurate 99. 9% of the time. [6] The next step is to understand how the SNPs can be interpreted.

A haplotype is the inheritance of a cluster of SNPs. A haplogroup is a group of similar haplotypes that are closely linked and that tend to be inherited together. By examining haplotypes, patterns of genetic variation that associate with health factors can be identified. For instance, if a haplotype is associated with a certain disorder, then the stretches of DNA near the SNP cluster can be examined to try to identify the genes responsible for causing the disorder. Additionally, the U. S. Library of Medicine claims that certain haplotypes refer to mitochondrial DNA or Y chromosome DNA. Since the Y chromosome is only passed from father to son, information is discovered about the paternal lines of ancestry. On the other hand, mitochondria have their own DNA that is passed down from mothers to sons and daughters. It provides information about the direct female ancestral line. [7] Together, they make up less than 1% of the total DNA and represent only a small fraction of someone’s ancestry. [8] For the remaining 99% of DNA, the haplotypes are

compared with those of others who have taken the tests to provide an estimate of a person’s ethnic background. For example, the pattern of SNPs might indicate that a person’s ancestry is approximately 50% Chinese, 25% Japanese, and 20% Indian. Genealogists use this type of test because Y chromosome and mitochondrial DNA results represent only single ancestral lines and do not capture the overall ethnic background of an individual. All of this genetic information has been discovered through a single vial of saliva.

Part 2

There are various reasons that compel people to utilize direct-to-consumer (DTC) genetic testing. These tests provide information on ancestry, paternity, and ethnicity which can help someone discover their identity. Others may choose to test to learn information about disease risk quickly and without the need for a health care provider. This knowledge may then motivate people to make healthy lifestyle changes to minimize the chances of developing a disease. However, according to an online survey, the most common reason that people seek DTC genetic testing is out of curiosity. [9] As DTC genetic testing is becoming more affordable, more people are purchasing these kits. As a Thanksgiving special, 23andme offered their ancestry kit for as low as $49 and their ancestry and health service for $199. [10] These lower prices allow curious cheaper access to this technology.

The societal and economic value DTC genetic testing is still unclear. The cost of health care may be reduced if people adopt better lifestyles through the knowledge obtained from the genetic tests. According to Hogarth, Javitt, and Melzer, these tests may also increase the amount of early detection and

intervention of diseases. Also, the cost and effectiveness of pharmacogenomics may be lowered through better targeting of drugs. [11] On the other hand, DTC genetic testing could raise the overall health care costs if more people get tested and then seek additional genetic counseling. An internet-based survey by McGuire found that 78% of those individuals who considered using genetic testing services would ask their physician for help interpreting test results. [12] This shows that patients will likely bring their results to their physicians, thus increasing pressure on the health care system.

Another downside of DTC genetic testing is that a negative result could give someone a false sense of security and cause them to miss regular checkups or continue to lead an unhealthy lifestyle that may lead to disease. There is also the possibility that DTC genetic testing will generate undue stress for individuals who misinterpret their results or receive false positives. A study that analyzed the results of 49 patient samples found that 40% of variants in a variety of genes reported in DTC raw data were false positives. [13] The possible inaccuracy of DTC genetic testing should be considered before drawing conclusions from results. Additionally, it is important to note that genetic risk is only part of the equation that results in health conditions. Environmental risk factors and lifestyle choices play a large role in health and therefore the results from DTC genetic testing is not conclusive.

When performing a genetic test, it is important that the results are brought to a genetic counselor who is skilled in risk assessment of the diseases as well as in the interpretation of these results in the context of relevant

medical and family history. [14] Consumers may make vital lifestyle changes for disease prevention based on misunderstood results. The genetic counselors will put the test to perspective and help prepare clients for potential findings, thereby reducing unnecessary stress. Lastly, a potential downfall is in the Genetic Information Nondiscrimination Act (GINA). GINA makes it illegal to discriminate against workers because of genetic information. However, there are loopholes where discrimination can occur. It only covers health insurance. It does not cover life insurance, long-term care insurance, or disability insurance. [15] Although genetic testing is an exciting technology, it is wise to consider the pros and cons before buying a kit. For those who have decided to use this service, it is imperative to seek an interpretation from a medical professional before acting on results.

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