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DTC-Genetics and Individualized Genomics: Emerging Challenges of Bringing Forensic Science and Clinical Tests for the General Population

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Abstract

The popularization of direct to consumer genetic (DTCG) services such as genetic ancestry, biogeography and genetic susceptibility to diseases have been increasing in the last years and it is estimated that the market will continue to grow in the next decades. However, the public availability of the databases generated from the companies providing these services have raised some concerns about the privacy of customers and unanticipated future applications of their data and samples stored by the companies. Here, we briefly discuss these issues and present some potential alternatives to address the situation, including keeping a well informed customer services, the use medical support and more regulation to access the databases.

Keywords: Genetics; Diseases; Population; Biogeography; People; Direct-to-consumer genetic tests

Abbreviations: STRs: Short Tandem Repeats; SNPs: Single Nucleotide Polymorphisms; DTCGT: Direct-To-Consumer Genetic Tests; FDA: Food and Drug Administration; WGS: Whole Genome Sequencing; WES: Whole Exome Sequencing; GWAS: Genome Wide Association Studies; INDELs: Insertion-Deletion Markers; CODIS: Combined DNA Index System; DTCG: Direct to Consumer Genetic.

Introduction

The use of genetic markers has proven to be efficient and reliable in forensic research, criminal caseworks and paternity testing [1]. Among the most widely used markers are short tandem repeats (STRs) [2], insertion-deletion markers (INDELs) and single nucleotide polymorphisms (SNPs) [3-5]. These markers have also been successfully employed in anthropological research to determine the structure of human populations, gene ancestry, biogeography and genetic association studies of complex diseases [6-9]. Traditionally, forensic genetics and genomic data have been generated and used in academic research, Government's agencies and clinical settings with specific purposes and scopes. In recent

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years, however, the advances in DNA sequencing and genotyping technologies as well as the decreasing costs and time needed for analysis have promoted the spread of Internet based companies offering direct-to-consumer genetic tests (DTCGT) services to the general population. Here we briefly discuss some of the current findings and challenges that need to be clarified for companies, customers and Government's regulators for a better utilization of these services and customer's data protection.

Genetic Ancestry, Biogeography and Family Connections

Among the services offered by DTCGT companies are genetic ancestry, biogeography, individual gene admixture and more recently genetic predisposition to diseases.

These services have become so popular among the population that altogether the genetic information accumulated by these companies' databases is now estimated by ranging a few millions of individuals tested and the market is expected to increase in the next years [10,11]. Most people requesting the services are interested in searching their ancestral genealogy roots, finding unknown relatives, or simply "recreational" curiosity [12]. Bringing the power of genetic tools to the non-professional population is celebrated by some who think that this might bring science closer to people. However, it is also criticized by others who think that these services require clear statement of roles and responsibility of all parties in the use and privacy of the information generated which compete to companies, the costumers and the government's regulator represented by the Food and Drug Administration (FDA) [10]. This is especially important because the occurrence of "incidental findings" is expected in some of these tests. When genetic tests are performed under clinical, academic and justice settings, individuals receive health, psychological and privacy support in any given situation before and after tests results. However, these companies operate outside the traditional health system and the individuals might not receive the health support needed to overcome the stress generated by some potential negative outcomes from their genetic data.

For example, it has been reported that individuals requesting DTCGT services incidentally (unexpectedly) have discovered that one of his/her parents or supposed family members are not their relatives [13]. There might be very serious situations that might uncover familial infidelity issues or untold children adoptions. How the customers will manage psychologically and emotionally these potential incidental findings? Another question is that since companies retain the customers' DNA samples and data, what if third parties access the DNA samples or genetic data without the customers' consent and somehow try to profit from the data? Moreover, some of these DTCGT companies have opened biomedical research branches and have filled up for patents [14]. Under clinical research settings, there is also possibility of getting patents but most academic institutions are non-profit organizations and the incomes are invested in more research. Additionally, patients have a rigorous informed consent and their genetic data is protected. However, in DTCGT companies, customer's genetic information is public; being shared with other customers in the database and recently, the data was even used by the police to catch killers. In particular, a high profile case, a suspect linked to 50 rapes and 12 murders between1976-1986 was recently arrested in California because the police found the suspect using the databases from a DTCGT company [13].

The police had a DNA profile from the crime scene belonging to the suspect, but they didn't have the match identifier. So, the police created a profile for the suspect and uploaded the suspect's DNA to the DTCGT Company's database to compare the suspect DNA information to trace the perpetrator directly or at least some genetically distant relatives. They found a perpetrator's family member, which eventually led to an arrest and further investigation. In this case, the suspect's relatives submitted their DNA for the specific purpose of ancestry/genealogy, but without their consent they also helped the police to find their relative. Discussions from the genealogy's users have divided point of views between justifying the police due to the gravity of the crimes and those alarmed who just now know that that from here and the future they may help the police to trace a relative. The access of genetic information in the clinical and academic settings, as well as in the FBI throughout the Combined DNA Index System (CODIS) is private and tightly regulated [15].

Access to these data is restricted, and requires court's authorization under very special situations. However, it just happened that there is no clear boundary of information privacy in DTCGT companies' databases, which involves national and worldwide customers. Moreover, the further use of DNA samples bank from customers is not well stated and it might be used for other purposes non-intended by the customers. For instance, in recent years tremendous research efforts have been invested to study the use DNA information to generate computer generated images from suspects, a process called forensic DNA phenotyping. It is currently possible to get information with high accuracy about sex, ethnicity, and eye and hair color from a murderer's DNA sample

found in the crime scene. However, long-term research is aimed at identifying genes involved in craniofacial morphometric, age, and height. Estimates indicate that with the improvement of genomic data analysis and artificial intelligence it would be possible in the coming decades to predict human faces and overall body shapes from forensic DNA phenotyping data could be used to solve crimes and archeological/anthropological research [16]. The use of DNA samples from DTCGT costumers could be potentially used in DNA phenotyping and other non-intended applications. Altogether, these cases and examples uncover some unanticipated situations and findings that open a new debate on challenging ethical issues that need to be addressed for a better regulatory legal frame and protection for customers.

Genetic Risk Analysis for Diseases

Although DTCGT services are dominated by ancestry/biogeography and similar tests, another debate regarding with new services related with detection of genetic predisposition to diseases, personal micro biome, whole genome sequencing (WGS) and whole exome sequencing (WES) have emerged [10,17]. Extensive genetic epidemiology and genome wide association studies (GWAS) have point out considerable genetic disparities related to African, European, Asian, Hispanics and Native-American ancestry to certain diseases and some health-related phenotypes. These diseases are highly inheritable and major risk factors for other complex diseases such as cardiovascular disease, diabetes, strokes, Alzheimer disease, and cancers [18-22].

Based on this knowledge, some DTCGT companies now provide services aimed at identifying genetic variants related to susceptibility of diseases and prediction of personal risks on individuals and their implementation was recently approved by the FDA. The tests would help customers to verify if they are carriers of a familial or unknown recessive mutations related with common hereditary diseases. The impact of the services would help for example, on family planning, susceptibility to some cancers, chronic and late-onset degenerative diseases. These services for risk to genetic diseases have a better control involving medical supervision. In different cancers, for example, some specific genetic mutations have been identified of high risk. In particular, familiar history of breast and ovarian cancer shave been highly associated with mutations in the BRCA1/2 gene. In some cases some women have undergone double mastectomy surgery doe to the discovery of having the BRCA1 mutation associated with familiar breast cancer. In a DTCGT situation, for instance, if a woman requesting these services discovers that she has this mutation, she will need to follow close attention with medical supervision, since this surgery procedure would be too radical if truly needed.

Concerns about these services have pointed to the potential negative psychological effects on stress and anxiety that incidental findings from genetic or genomic risk susceptibility data could have on individuals taking the test. Incidental findings resulted from these tests have the potential to cause great distress and it is suggested that companies should make counseling available, before and after people take tests [23]. Moreover, most incidental findings might be beyond our current knowledge but some confer high risk to serious diseases and reproductive decisions that could be mitigated by timely medical intervention [24]. With the advances of biomedical research, it is inevitable that more DTCGT for risk to diseases will be available and the impact of incidental findings will also increase in the future as more genetic mutations related with diseases are discovered. The discovery of more genetic risk factors for diseases will also help to gather information useful in forensic research to gather data from criminals and anthropological/archeological applications.

Conclusions and Recommendations

There is no easy answer for some of the questions stated above and we cannot anticipate all potential uses that might be possible in the future for DTCGT and DNA databases. However, customers need to know what they are giving away when they request services such as ancestry and make them clear about other possibilities. Conversely, customers should have the option of requesting the ancestry analysis and give them the option of not being part of the databases. Companies may also have the option of using genetic data from “neutral” reference populations for the genetic estimations. If the police want to use the DTCGT companies’ databases to solve crimes they would need a warrant with an open debate and legislation; so more people might be willing to participate and share their data. Information sharing has always been crucial for successful law enforcement, which could solve crimes and ultimately save lives. The effectiveness of DNA databases continues to grow as the databases get larger [15].

On the other hand, there seems to be a consensus among the scientific community that it is just a matter of time for the massification and use of genomic data is not only inevitable but also beneficial for biomedical research and personalized medicine [17]. However, customers should be well informed about the limitations of the genetic data and what are the real predictive conclusions we can make

of them. For example, a recent report showed that DTCGT for risk to diseases could give up to 40% of false positive results; and some gene variants were designated as "high risk" under DTCGT services whereas they were classified as benign by a third party interpretation [11]. Therefore, it is imperative to know if the services provided correspond to raw genotype data with low accuracy and require more clinical confirmations. Efforts are ongoing to identify molecular mechanism and common risk alleles for cancer and other diseases; however, we are currently unable to anticipate the full range of uses and consequences. The impact of individual genetic variation and the effect of non-genetic factors should also be considered for better interpretation. Moreover, at this early stage of translational medicine involving personalized genomic data, it is often unclear whether data generated from these services will have clinical significance outside academic research [25]. It is absolutely necessary the inclusion of a better genetic and medical counseling for best control of these services and customers' protection and benefit [23,25].

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