

Testimony of Ashley C. Gould, General Counsel, 23andMe, Inc.
“Direct-To-Consumer Genetic Testing and Consequences to the Public Health”
Committee on Energy and Commerce
Subcommittee on Oversight and Investigations, U.S. House of Representatives
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Chairman Stupak, Ranking Member Burgess, and Members of the Subcommittee:
Good morning, and thank you for inviting me to discuss this important issue.

My name is Ashley Gould, and I am the General Counsel of 23andMe, Inc., a leading personal genetics company dedicated to research and helping individuals understand their own genetic information through DNA analysis technologies and web-based interactive tools. I ask that the full written testimony be submitted for the record.

I. Overview

Direct-to-consumer (DTC) genetic testing leverages the findings from the Human Genome Project. Genetic testing has the potential to allow individuals to access their genetic information – the building blocks of their genome. Customers empowered with this information have made lifestyle changes aimed at reducing their risks of developing disease and have provided information to their doctors to aid in diagnosis and treatment. These actions have improved and even saved lives. Consumers can only be helped if such services operate at the highest levels of both science and ethics. We thus greatly appreciate the Subcommittee’s interest in this increasingly important area of personal health, and we support sensible U.S. Food and Drug Administration (FDA) regulation of genetic and other forms of direct-to-consumer testing.

Before explaining our DTC genetic testing services more specifically, we would like to explain that 23andMe operates pursuant to the following core beliefs and facts:

- A. Consumers have a fundamental right to access their personal genetic information;
- B. 23andMe takes a holistic approach in providing genetic information to customers. Customers are provided with as much scientifically-sound information as is available so that they can learn as much as possible;
- C. 23andMe provides a platform for customers to participate in the research process, so that we can all learn more about genetics and diseases;
- D. Genomics has reached the stage where 23andMe can provide personal genetic information to consumers in a cost-effective manner. 23andMe relies on experts, both internally and on an outside board of prominent science advisors, to provide its services based firmly on peer-reviewed science;

- E. 23andMe provides genetic testing results to consumers in a secure, confidential, and privacy-protected environment;
- F. 23andMe encourages our customers to consult with medical professionals before taking any medical action based on DTC results; and
- G. 23andMe supports federal standard-setting or other sensible regulatory activities to protect consumers and increase transparency.

II. Background

23andMe met with the FDA even before we launched our service, explained the full scope of our proposed services, and were encouraged to continue with our service by the then-FDA commissioner. Even while we have not been actively regulated by the FDA, we have embraced the scientific standards normally applied in a regulated environment. Based on FDA and Clinical Laboratory Improvement Amendments (CLIA) standards, we have created a sophisticated, high-quality testing process.

To process and analyze our customers' saliva samples, 23andMe contracts with the National Genetics Institute (NGI) – a wholly owned subsidiary of the Laboratory Corporation of America. NGI is a CLIA Program-certified laboratory that provides advanced clinical genetics testing services for blood screening, medical testing, and clinical research. Licensed as a clinical laboratory provider by both state and federal agencies, NGI participates in a number of approved quality control programs, and holds active Biologics Licenses from the FDA for screening of plasma for blood-borne infectious agents. It also provides advanced genetic testing services to physicians, hospitals, and clinics, and has supported numerous pharmaceutical and biotechnology companies in the clinical development of new infectious disease and oncology therapies. (See Attachment A). We believe that NGI complies with the highest professional, regulatory, and corporate quality-assurance standards.

NGI processes customer saliva samples using a chip that analyzes nearly 600,000 data points – the building blocks of the genome that I referred to above. After NGI completes the analysis, the collected data for each sample is encrypted using leading technologies to protect privacy, and sent electronically to 23andMe, which then uploads the data to the customer's individual account. 23andMe then notifies its customers that their data are loaded and ready for viewing.

Throughout this process, 23andMe provides strong safeguards for our customers' privacy and confidentiality. We have invested in a sophisticated technical environment for the storage and security of customer data and have had independent security audits to verify that security is state-of-the-art. As a part of our Terms of Service, we educate customers about our service and what will be provided – including how to use their information, how their information is protected, and the scientific limitations. During account registration, each customer is required to focus his or her attention on relevant highlighted sections of our Terms of Service and agree to them. In addition, we have

consulted on privacy issues with leading experts in the field, including the Electronic Frontier Foundation, to adopt the highest standards and best practices for storing and safeguarding our customer data.

Equally important, 23andMe grounds its service in five core elements: (A) informed consent; (B) educating customers about personal considerations they should take into account when using our service; (C) educating customers about the science and methodologies behind our service; (D) updating customers with new information as the science advances; and (E) emphasizing that customers should consult with health care professionals before taking any medically-related actions based on our reports.

A. Informed Consent and Privacy

We require informed consent from customers before they use our services so that they understand our permitted uses of their data and what our reports indicate – both the information and its limitations. We take extra precautions with certain reports. For example, we ask customers to be certain they want to know potentially significant information, and insert interstitial pages that must be clicked through before viewing our Parkinson’s and BRCA reports. This way, a customer has the chance to reflect before making an affirmative choice to view his or her data.

No individual customer information is ever disclosed to third-parties without explicit consent (unless required by law). In addition, we recently introduced an institutional review board-approved consent document, which allows customers to decide if they would like to participate in research intended for peer-reviewed publication. In such instances, customer information will only ever be provided on an aggregate basis. As a constant reminder to our customers, our consent document, terms of service, and privacy statement are all available for review via links on every page of our website.

B. Education About Customers’ Personal Considerations

Our customers come to our service because they are motivated to learn about their genes. Customers are not compelled to learn any information, and we designed the web site to allow customers to choose to learn the information that interests them. Our Terms of Service page clearly states that information customers learn could cause distress. Specifically, we disclose at length the following risks and considerations:

- “Once you obtain your Genetic Information, the knowledge is irrevocable;”
- “You may learn information about yourself that you do not anticipate;”
- “The laboratory may not be able to process your sample, and the laboratory process may result in errors;”
- “You should not change your health behaviors solely on the basis of information from 23andMe;”
- “Genetic research is not comprehensive;”

- “Genetic Information you share with others could be used against your interests;” and
- “23andMe Services are for research, informational, and educational use only. We do not provide medical advice.”

For a complete and fully described list of disclosed risks and considerations, please see our Terms of Service. (See Attachment B). In addition, our public website contains a section addressing considerations before using our service or engaging in genetic testing. (See Attachment C).

C. Education and Transparency About the Science and Our Methodologies

We provide extensive information to our customers so they understand that the data we provide can change as new scientific studies are completed. We also explain the technology, algorithms, and methodology used to process and analyze their DNA. We have white papers readily accessible on our website that detail our inclusion criteria procedures and algorithms. In addition, for each trait, condition, or disease association we test, we explain whether the result is based on established research – meaning that the topic meets 23andMe’s criteria for findings that are very likely to reflect real effects. The scientific community has largely reached consensus on these topics. We also explain whether the result is based on preliminary research – meaning that these studies still need to be confirmed by the scientific community. It also includes topics where there may be contradictory evidence. The results of these studies are not conclusive. We report associations based on preliminary research so that our customers know the most current information about what their genome says, but we provide the customers with contextual information so they know the research is preliminary.

When a customer receives his or her results related to a disease association, the customer receives the risk prediction in context of the average person’s risk versus the risk for a customer with his or her genetic variations. We also educate our customers about the importance of the environment and other factors that also may influence their risk of disease. Disease is not determined by the genome alone. We provide extensive information about the condition and next steps, including encouraging customers to discuss their results with their physicians and to consult genetic counselors.

D. Updates on Changing Data

We are committed to keeping our customers educated on new scientific advances. After customers receive their initial results, we update them on new scientific findings. As new scientific studies uncover new information that changes our understanding of the genome and its meaning, we tell our customers how this new information changes their results. We believe customers have a right to be updated when advances in science change our understanding of their genomes, so we do not cut off our customers after one look – or a year of looks – at their information.

E. Emphasis on Physician and Genetic Counseling Support If Customers Have Questions

As stated, if customers have questions, we recommend that they should coordinate the receipt of their information with physicians or a genetic counselor. We do not offer genetic counseling services directly and in fact believe it is important for counselors to be independent of the company. We have provided links to such services previously and since early June 2010, we have offered our customers a referral to Informed Medical Decisions, Inc. (InformedDNA), an independent provider of genetic counseling services staffed by certified genetic counselors. These genetic counselors are trained professionals, who have additional training about the services and results that 23andMe provides.

To be clear, there is no financial relationship between 23andMe and InformedDNA. However, we do provide InformedDNA with background information about 23andMe's test offerings so that InformedDNA's genetic counselors will be prepared to discuss the tests we offer our customers. This service is optional for 23andMe customers, and customers who choose InformedDNA for genetic counseling services pay InformedDNA directly.

23andMe partnered with InformedDNA because it is the largest independent network of genetic counselors and is the only national provider whose services are a covered benefit for most individuals with commercial health insurance. (See Attachment D).

III. Concerns About DTC Genetic Testing

We recognize that there are concerns about customers having direct access to genetic testing, including the risk that individuals might make poor choices based on their results, and consistency of results among the different companies in this sector. We take those concerns extremely seriously.

A. Genetic Concerns

The best way to avoid poor choices is to have educated customers. Here are some of the things we are doing to educate customers: We have created educational videos that are on our public website (these have over 140,000 views on Youtube and are used by schools for genetic education). Our health reports have extensive information including description of the underlying scientific literature upon which reports are based, citations to these studies, the interplay between genes and the environment, and technical information and additional resources for those who are interested. We also educate the broader community. We have offered free genotyping to hundreds of physicians so they can learn about their data and understand their patients' needs, we are involved in medical school programs to further genetics understanding, and we are involved in numerous third-party research studies – some of which are described below.

With respect to concerns about people making poor choices, there is excellent scientific research being conducted about the impact and utility of DTC genetic testing. For example, the National Human Genome Research Institute and the National Cancer Institute of NIH, along with the Group Health Cooperative in Seattle and the Henry Ford Health System in Detroit, launched the “Multiplex Initiative” in May 2007 to study 2,000 people in Detroit who were offered a multiplex genetic test for eight common conditions. This study has shown that consumers understand that both genes and environment contribute to disease risk. As part of this research, researchers found that “[i]ndividuals who present to health care providers with online genetics information may be among the most motivated to take steps toward healthier lifestyles. These motives might be leveraged by health care providers to promote positive health outcomes.” There is more to come, and it will educate all of us about what the actual issues might be and how to address them.

We want to better understand these issues ourselves, and to that end, 23andMe has worked with the Genetics and Public Policy Center at Johns Hopkins University to conduct an independent study of how 23andMe customers understand and use their results. We expect these results to be released later this summer, and we will keep you informed about them. We are also working on a related study with Stanford University.

23andMe has over 50,000 customers, and through monitoring community feedback, we are confident that DTC genetic testing has a positive impact on customers’ lives.

B. The June 2010 Sample Mix-Up

Regarding concerns about accuracy and reliability, let me briefly address the reported June 2010 incident in which customers received results belonging to other people. A single human error by a certified technician at our contracted laboratory caused this incident. The technician accidentally and wrongly inverted a plate by 180 degrees, causing valid results for plate samples to be matched with the wrong person’s name. This mismatching error at the laboratory led to 23andMe’s receipt of mismatched results that were then transmitted to customers, affecting eighty-seven customers in all.

23andMe responded to the problem as soon as we learned of it from our community by notifying customers and removing the mismatched results in less than 24 hours. We were then able to give the customers their correct results within six days. While 23andMe regrets the error and takes the incident seriously, the company rapidly resolved it. Since the incident, both 23andMe and our laboratory have further strengthened our quality control systems to prevent such problems. The laboratory error that occurred here could have happened in any laboratory for any genetic tests coming from any hospital, doctor’s office, or other laboratory client – it was not a problem uniquely related to 23andMe samples or DTC services to any extent. (See Attachment E).

C. Standards Setting

We fully appreciate the concerns that different companies can return different risk predictions, which can happen even when the tests are accurate. There are several scientific reasons for differences: companies employ slightly different criteria for the inclusion of disease-associated markers in their reports; new associations between genetic markers and diseases are being discovered at a rate faster than companies' development cycles; and companies test for an imperfectly overlapping set of genetic variants for reasons including the ability of different genotyping technologies to assay certain variants. We have asked NIH and FDA to help on this point. In addition, the industry is already looking to other reputable, independent entities that could take up this challenge, such as NIST, an independent standard setting entity.

IV. **How DTC Genetic Testing and 23andMe Helps Consumers Improve Their Lives**

Over the past decade, scientists have discovered that an increasing number of diseases can be linked to variations in an individual's DNA. These discoveries have launched the revolution in personal health, empowering individuals as never before to manage their personal health before they get sick. Thanks to such advances, we now understand that each of us is born with genetic predispositions for developing diseases.

In some cases, this information can truly save a life. A striking example is that of Kirk Citron. Kirk found out from 23andMe that he had about three times the normal risk for venous thromboembolism – something he never would have suspected. Knowing this information, Kirk did a little digging and found out his father's father had a blood clotting condition that had him on blood thinners for the last 20 years of his life. A few weeks later, Kirk tore his ACL and was told he needed to have knee surgery (which is one of the risk factors for blood clots).

Kirk went to his primary doctor and his orthopedist and told them he was worried about the risk, given his family history and his DNA test. Both doctors were somewhat dismissive of Kirk's concerns, but Kirk was insistent enough that they agreed to change the post-surgery protocol to give Kirk five days of blood thinner.

Kirk had the surgery and had the five days of blood thinner. Five days later, he suddenly felt short of breath. Kirk saw his doctor and was immediately admitted to the hospital – he had experienced a pulmonary embolism – a blood clot that traveled to his lung.

In discussing it afterwards, Kirk's doctor said two things: first, the fact that Kirk had been on the blood thinner for five days almost certainly meant that the attack was much less severe than it might have been (it could have been fatal). Second, having the information ahead of time allowed Kirk's doctor to make the diagnosis much faster than otherwise would have been the case. Once Kirk was in the hospital, and on a new round of blood thinners, he was out of danger, and now is home and doing just fine.

Our tests also detect genetic variations that have been linked with heart attacks, Crohn's Disease, macular degeneration, Parkinson's disease, Type 2 diabetes, and many others. As scientists continue to make breakthroughs, the number of diseases we can test for will only increase. Because new, reliable studies are being published weekly, even daily, we are regularly adding traits, diseases, and conditions to our testing. As we do so, we educate our customers on these latest scientific discoveries as they happen, but we only link them to our customers' personal data after we determine that they meet our standards of reliability.

23andMe's DTC service is consistent with the FDA's long history of approving at-home, over-the-counter tests for HIV, hepatitis, and fecal blood (which might be caused by colon cancer). In addition, FDA already permits customers to have direct access to tests for potentially less worrisome conditions, such as high cholesterol and pregnancy. As the FDA explains on its website, these tests help consumers "detect possible health conditions when [they] have no symptoms, so [they] can get early treatment and lower [their] chance of developing later complications ... [and] detect specific conditions when there are no signs so that [they] can take immediate action." (See Attachment F). Although the results of the FDA-approved, over-the-counter tests may lead to customers receiving potentially distressing information, the FDA has permitted consumers to have direct access to these tests. Our tests are even less worrisome, as they do not diagnose a disease but rather only provide more limited data.

V. How DTC Genetic Testing and 23andMe Help Develop and Accelerate Medical Research

Besides the direct consequences of genetic testing on healthcare, 23andMe is also strongly rooted in the development of innovative solutions for accelerating medical research. One of the unique features of 23andMe's DTC genotyping service is the company's focus on 23andWe, a community-centered research effort in which consumers are encouraged to contribute to medical science by answering surveys.

To date, through 23andWe surveys, 23andMe has amassed one of the largest databases of genetic and health information in existence. This database includes over 50,000 customers, of whom more than half have participated in at least one survey, and of whom roughly 10% have participated in at least 20 surveys in total. Our customers have answered over 10 million research questions. Last month, we published our first paper describing the results of a number of replications and several novel findings based on our customer data in the open access, peer-reviewed journal PLoS Genetics, and many more publications covering novel associations with more serious medical conditions are on the way.

We believe that this model of research has the potential to transform research in this field in two specific ways. First, unlike most research studies to date where the primary source of funding has been a government agency such as the NIH, our research model has been primarily driven by consumer interest. Second, our active cohort of

engaged customers has enabled us to pursue research into over 600 health conditions simultaneously, in contrast to traditional approach of funding a genotyping study for a single health condition at a time. This allows research to proceed at a much faster pace than possible in more traditional settings. For example, in March 2009, we began a project to study the genetics of Parkinson's disease. In less than a year, we were able to assemble a database of nearly 4,000 genotyped patients with Parkinson's disease, which enabled us to replicate previous findings as well as to discover a number of novel associations that had not previously been reported in the scientific literature. These novel associations are not yet reported to our customers because they have not yet been published in a peer-reviewed journal, which is one of our standards for inclusion.

Because we have such a large research database and engaged customers, we are able to assess the clinical significance of genome associations. We are in a unique position to tell our customers, and the community, how clinically significant this information is.

VI. 23andMe Supports a Strengthened Regulatory Framework for DTC Genetic Testing

23andMe has been working with federal authorities – and looks forward to continuing to work with them – on improving the regulatory framework for all diagnostic testing, including genetic testing. These are complicated issues, as evidenced by the fact that FDA just held a two-day meeting this week and heard from numerous medical, laboratory, academic, and industry groups about how to change the regulation of laboratory testing. We have met with the FDA several times and have had discussions with them on how they might regulate this new industry that does not fit squarely into any existing regulatory paradigm. We have also engaged with other officials at the U.S. Department of Health and Human Services and NIH.

We have been working on a proposed framework that we will present to the FDA tomorrow. This framework takes into account rapid technological innovation, and will ensure the analytical and clinical validity of all laboratory tests. We hope that the framework will lead to a scalable regulatory system for the FDA to adopt.

VII. Conclusion

Thank you again, Chairman Stupak, Ranking Member Burgess, and Members of the Subcommittee for giving me the opportunity to speak with you today and for your work on these issues.