

PREPARED STATEMENT OF
DAVID BECKER Ph.D.
CHIEF SCIENTIFIC OFFICER, PATHWAY GENOMICS

BEFORE THE
U.S. HOUSE OF REPRESENTATIVES
COMMITTEE ON ENERGY AND COMMERCE
July 22, 2010

Mr. Chairman and Committee members,

Thank you for the opportunity to testify to you today. My name is Dr. David Becker, I am the Chief Scientific Officer at Pathway Genomics, a San Diego based genetic testing company.

Pathway Genomics utilizes advanced genetic testing technology and validated genetic information to give individuals and physicians the best available information about their genetics, allowing them to make informed health decisions. It should be noted that although Pathway provided genetic tests directly to consumers like many of our competitors, we voluntarily suspended that practice as we work with the U.S. Food and Drug Administration (FDA) to ensure that our tests are offered in a manner consistent with regulatory requirements.

Pathway operates its own on-site laboratory that is certified by the U.S. Centers for Medicare and Medicaid Services (CMS) under the Clinical Laboratory Improvement Act (CLIA) and licensed by the state of California. The company employs 70 geneticists, scientists, bioinformaticists, physicians, and genetic counselors, as well as software engineers, customer support representatives, and others to ensure that we offer our customers accurate information that is properly understood.

Benefits of Genetic Testing

Since the completion of the Human Genome Project in 2003, scientists, physicians, policy makers and consumers have eagerly anticipated the era of “Personalized Medicine,” which means providing targeted preventive care and therapeutic treatment based on an individual’s genetic makeup.

Today, billions of dollars in research are being dedicated to building on our current knowledge of the human genome. Almost daily, new and exciting discoveries about the relationships between genetics and our health are transforming traditional medicine.

Empowering consumers with information about their genes holds tremendous promise to promote health and improve lives. We at Pathway can attest to the positive value that genetic information has provided to the lives of our customers. Moreover, Dr. Francis

Collins, MD, PhD, the current Director of the National Institutes of Health (NIH), cited several instances in his book *The Language of Life* where individuals have learned from genetic reports and proactively made changes to prevent or manage disease. As a physician, Dr. Collins knew he needed to incorporate a healthy diet and regular exercise in his life, and his genetic testing finally motivated him to make these changes.

Some have argued that consumers should not be provided with information about their genes, based on a fear that the information would not be used appropriately, or that genetic testing might create unnecessary anxiety amongst patients who feel that they are destined to get certain diseases based on their genes. However, we believe that consumers are capable of understanding both the benefits of having information about their genes, as well as the fact that genetics are only one piece of the puzzle of living a healthy life.

Moreover, in 2009, Boston University School of Medicine's Risk Evaluation and Education for Alzheimer's disease (REVEAL) study found that asymptomatic first-degree relatives of Alzheimer's patients who received genetic testing results regarding a possible propensity for Alzheimer's disease (presence of an APOE ϵ 4 allele) did not have a change in the levels of anxiety, depression or overall general distress. While this study certainly highlights the need for responsible delivery of this information, it also indicates that patients are able to process genetic information effectively and use it to make better health decisions, even with respect to diseases like Alzheimer's.

Companies like Pathway are allowing individuals and their physicians to accurately determine their personal genetic makeup. As new genetic findings are made and validated, Pathway updates individuals about these discoveries and in consultation with their physician, individuals can use this information to improve the quality of their health care.

Pathway Genomics' Testing Services

Pathway currently offers three different health-related reports, as well as a genetic ancestry report. The three health related reports are:

- Health Conditions – This report provides a customer with an assessment of their genetic risk for as many as 25 different health conditions. The number of conditions will vary by the reported ethnicity of the individual, as a result of the quantity and quality of genetic research available.
- Pre-Pregnancy Planning (Carrier Status) – This report indicates the presence of genetic markers associated with 37 recessive genetic diseases. This information can be used to detect conditions that are unlikely to affect the tested individual, but can be passed to their children.
- Drug Response – This report suggests the likely response to as many as 10 different medications. This information may facilitate optimal medication

decisions, avoiding drugs that may be harmful or dangerous or assisting in establishing proper dosing.

Similar to our competitors, genetic risk is determined by applying the odds ratios associated with an individual's genetic markers against a population average to calculate a genetically adjusted risk of disease. However, as Dr. Francis Collins highlights in his book, quantifying the risk of contracting specific diseases may not incorporate other factors like family history, lifestyle and environment that for some diseases can be more significant factors than genetics. In consideration of these concerns, we want to highlight some significant differences in the way that Pathway reports risk information. After extensive internal discussions among our scientific and medical staff, we felt that a specific percentage based calculation implies a degree of precision that is not justified by the current state of genetic information. Therefore, we instead elected to categorize the genetic results into categories meant to motivate positive health behaviors appropriate to current genetic knowledge. From lowest to highest level of genetic risk, these categories are named, "Maintain a Healthy Lifestyle," "Learn More," "Be Proactive," and "Take Action."

Further, every customer is encouraged to fill out a health survey that collects relevant data on lifestyle, environment, and family history, and we use that to calculate a lifestyle risk score using a similar categorization system. This allows us to inform an overweight customer who smokes a pack of cigarettes a day that, even if they do not have genetic risks for lung cancer and hypertension, they may be at significant risk based on their lifestyle. We feel that this approach provides a more comprehensive assessment and makes it easier for our customers to understand how to properly use the information provided to improve their health.

Physician Oversight and Genetic Counseling Services

In addition to providing understandable genetic reports, Pathway felt that it was important to provide consumers with access to professionals trained in genetics to ensure that our customers and their physicians understand what their genetic reports say, and just as importantly the limitations of genetic information.

All of Pathway's physicians and genetic counselors are Board-certified or Board-eligible with the American Board of Genetic Counseling and have significant training and expertise supporting patients with understanding of genetic information. Our genetic counselors and physicians are available for guidance and support both before any services are ordered and post-testing at no additional charge to the customer.

These qualified experts review all health report results prior to delivery, looking for indications that warrant a separate communication encouraging the customer to speak with one of our genetic counselors or physicians. Performed at no additional charge, these personal consultations explain the results and appropriate next steps, including, as appropriate, consulting with their physician.

Validated Testing Platforms

Pathway has consistently strived to provide our customers with information that is accurate, understandable, and consistent with professional medical standards. Pathway has validated its laboratory-developed genetic tests according to CLIA standards and guidelines to assure accuracy of the data produced in our laboratory. This includes using reference samples provided by Coriell and ParagonDx, operating multiple genetic testing platforms to cross-validate results in-house and being engaged in a proficiency testing program with outside labs.

Further, Pathway maintains a competency program that ensures all laboratory employees are properly trained and capable of accurately performing our tests.

Accurate Genetic Reports That Reflect Current Knowledge

Pathway recognizes that it is equally important to ensure that its testing accurately represent the best scientific literature. Our reports are constructed using evidence-based standards, and only markers that have achieved high quality standards of clinical research are incorporated. To be used in our analysis, a marker must have been assessed in a research study containing a minimum of 1,000 cases and 1,000 controls, match the reported ethnicity of the tested individual, and demonstrate a statistically significant p-value and odds ratio. To reach our higher standard of a *validated* marker, there must be at least one additional study for the same ethnicity with a minimum of 500 cases and 500 controls, and the same risk allele must have demonstrated statistically significant outcomes in the same direction. Additionally, only one marker is selected per gene or region to avoid potential errors associated with signal duplication.

To stay on top of the constant flow of new information about the relationship between genetics and health outcomes, we have a team of seven full-time-equivalent curators, all with Ph.D. degrees, who are responsible for reviewing the latest research and ensuring that our reports reference the best available information. This information is used to update existing reports as well to expand those reports with new conditions as new discoveries are made.

“Direct-to-consumer” (DTC) genetic testing has garnered a lot of attention recently, and Pathway believes that genetic testing can be offered directly to customers in ways that help them improve their health without creating unacceptable risks or inconvenience. Pathway has commonly been labeled a DTC genetic testing company, though we feel that this is a misnomer because expert clinicians have always been intimately involved in the review, interpretation and presentation of results and assuring that the customer has a proper understanding of his or her report. However, as I mentioned previously, we have suspended web and retail access to our services, and the growth of our business to support these services, while we work with FDA.

Existing Regulation

Laboratories are already under significant regulatory oversight. This regulation includes CLIA, state regulations, and, in some cases, regulation by the FDA. Additionally, there are voluntary certifications like College of American Pathology (CAP), which certify laboratories with high standards of validation, proficiency, and quality assurance. Pathway Genomics operates its own on-site, CLIA-certified and California state-licensed laboratory, and is pursuing CAP Certification. These programs require us to follow very specific rules to ensure the quality and accuracy of our services, and affect not just our laboratory operations, but also all of our business functions.

Pathway also understands the critical importance of data privacy and data security. Consumers who have entrusted us with their personal information can be assured that Pathway utilizes administrative, technical, and physical controls to secure their genetic information. One element of these controls is our maintenance of a closed, secure system -- all of our testing takes place in-house in our own on-site laboratory mitigating many potential privacy and security risks. Pathway has a Chief Privacy Officer who administers the company's comprehensive privacy policy and security program. Each consumer receives a consent agreement as well as a user-friendly shortened summary of the privacy policy in addition to the full privacy policy.

Regulatory Changes

Pathway feels that bona fide genetic testing companies such as ours would benefit from a stable regulatory environment. Earlier this week, FDA held a public meeting to allow industry experts and laboratory providers, including Pathway, to discuss ways that FDA might regulate laboratory developed tests to ensure that high standards of accuracy and quality are maintained, while not creating an undue burden on innovation and advancing medical care.

While FDA's efforts to clarify the regulatory requirements for laboratory developed testing are still in progress, here are some areas that we believe could use additional guidance or clarification:

- Risk-based classification system for genetic tests
- Minimum scientific study criteria
- Standardized definitions for population risk
- List of the strongest effect marker for each condition
- Standardized labeling and website disclosures
- Genetic test registry with required disclosures
- Mandatory CLIA and CAP certifications
- Publicized and standardized compliance procedures, regarding incident management and reporting, customer complaints, etc.

While it is critical to ensure public safety, it is also important that new regulations do not stifle innovation or restrict an individual's access to information that impacts their health. With a stable and supportive regulatory structure, the field of genetics can not just improve the delivery of health care, but become a driver of high paying green jobs, investment, and advanced education. Failure to do so will cede America's leadership position in this emerging field of science to other countries.

Conclusion

Pathway is committed to improving the health and well-being of our customers. We are also committed to ensuring that everything we do is based on rigorous scientific standards and complies with all applicable laws and regulations. Genetic testing holds great promise to help people live more healthy and productive lives. Allowing consumers, in consultation with health care professionals, to access their genetic information empowers healthy and proactive choices. We are excited about the future of genetic testing, and look forward to working with FDA to ensure that the services we offer are of the highest quality and value to the American public.

Pathway Genomics appreciates the opportunity to testify at this hearing, and appreciates the Committee's consideration of this important issue.