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BREAKING NEW GROUND THROUGH HIGH-QUALITY GENETIC TESTING

A conversation with **AARON ELLIOTT**, CEO of Ambry Genetics

Ambry Genetics drives innovation in diagnostics by launching cutting-edge genetic tests and technologies. Its broad collaborations, novel research projects, and networks for data sharing continually push the industry forward. The recent acquisition of Ambry Genetics by Konica Minolta merges genomics, protein, and imaging analyses to develop an even more complete perspective on human disease. Aaron Elliott, CEO of Ambry Genetics, discusses the company's approach to genetic testing and its unparalleled commitment to quality.

What is Ambry Genetics' approach to genetic testing?
Ambry was founded in 1999 and has always been focused on innovation. We've been first-to-market with a wide variety of tests and technologies, including clinical exome and hereditary cancer panels. We strive to develop the most accurate, highest quality tests possible. Our motto has always been to develop, design, and run every test as though the results are for one of your family members.

What are some of Ambry's major accomplishments?
In 2010, we were the first to commercially launch a next-generation sequencing assay, which led to a paradigm shift in the whole industry from low- to high-throughput technology. We were also the first company to launch a hereditary cancer panel that included genes besides *BRCA*1 and 2. Now, these have become the panels that everyone offers. In 2011, we were the first company to launch clinical exome sequencing, which is a test targeted for patients with conditions that are difficult to diagnose. Our comprehensive assay can diagnose these patients more than 40% of the time, which is the highest in the industry. The other big step that changed the genetic testing field was our lawsuit with Myriad Genetics in 2013, which drove the nail in the coffin for gene patents, and opened up *BRCA* testing to the entire industry.

What are the benefits of the increased accuracy of Ambry's tests?
Within the commercial clinical testing industry, low-cost providers probably have about a 90% accuracy rate. The remaining ~10% comes from Ambry's additional technical and variant assessment experience and capabilities that allow us to identify more patients. Any increased ability to accurately detect variants is very important when the positive rates in general are low. For 85% of high-risk patients, we can't definitively pinpoint why they have cancer at such a young age or have such a strong family history of cancer. We're in the business of rare variant detection — detecting very uncommon, complex mutations. That's our specialty. That additional accuracy and quality is why Ambry is in business and remains a leader in this space.

What is the scope of your clients and collaborations?
We invest a lot in research collaborations with cancer institutions and others to advance the field of genetics. We have more than 200 clinical research projects ongoing at any one time as well as various internal research projects. Many of these focus on trying to increase the diagnostic rate or better understand the clinical utility of hereditary testing, with the goal of providing answers to our patients about why they are

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at a higher risk. We have helped nearly 1.5 million people, which is a pretty impressive statistic. We're not testing the general population; we're testing people that are very sick or carry a very high risk of being impacted by these various diseases.

How has integration with Konica Minolta enhanced your approach to genetic testing?
Konica Minolta's precision medicine initiative is aimed at capturing the whole diagnostic workflow from imaging to molecular profiling. Konica Minolta is heavily entrenched in medical imaging. It already developed a proprietary protein labeling technology, bought Ambry for the genomics piece and also acquired Invicro, the gold standard of medical imaging for clinical trials. By integrating the genomics, protein, and imaging, we're going to get a much more holistic view of a patient's disease. This integration provides a whole new way of thinking about finding genetic associations and the future of biomarker development.

What makes Ambry stand out?
The most unique feature of Ambry is the academic feeling of the company and the strong connections with our clients and institutions. When you talk to people that use Ambry Genetics, they refer to us as collaborators, not as a service provider. Another feature that makes us scientifically unique is our effort to resolve variants of unknown significance as either causative or not causative of a patient's symptoms or cancer. In addition to having an army of variant scientists we also operate the Ambry Translational Genomics lab, where we do different functional studies to determine whether a variant of unknown significance has an impact or not, which is something unique to Ambry and sets us apart from other labs. Once we have the answer, we share the data with the world.

What's next?
We're about to change the game again in hereditary cancer. In January 2019, we're going to launch the world's most accurate, sensitive test for hereditary cancer. It will be a completely different type of test than anyone has seen before and will once again demonstrate our commitment to finding answers.

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