

GENOMICS | DIAGNOSTIC TESTS | GENETICS | R&amp;D

## Genomic Vision to demonstrate 'molecular combing' technology's ability to identify novel cancer-causing BRCA gene variants

Quest Diagnostics to aid analysis of study on up to 1,000 de-identified patient specimens

**Bagneux France (January 14, 2016)** – Genomic Vision (GV: EN Paris), a molecular diagnostics company specializing in the development of diagnostic tests for genetic diseases and cancers based on molecular combing technology, today announced its plans to initiate a patient study to demonstrate the value of Genomic Vision's FiberVision® molecular combing technology in detecting novel BRCA gene variants associated with a predisposition to breast and ovarian cancer.

Quest Diagnostics (NYSE: DGX), a strategic collaborator of Genomic Vision, plans to support the study, which will seek to determine the extent to which molecular combing can identify BRCA gene variants missed by other test methods. The analysis will be conducted at Quest's advanced clinical laboratory in San Juan Capistrano, Calif. and at Genomic Vision's laboratories in France. The study is expected to begin this month and be completed over the summer 2016. It is expected to involve testing on 500 to 1,000 de-identified specimens.

Loss of function variants of the BRCA1 and BRCA2 genes significantly increase a person's inherited risk of developing breast, ovarian and certain other cancers. Clinical laboratories use DNA sequencing combined with either microarray or multiplex ligation-dependent probe amplification (MLPA) to identify these variants. Scientists at Genomic Vision and Quest Diagnostics believe molecular combing is able to identify large genomic re-arrangements that conventional methods fail to identify.

*"Next generation sequencing is a hugely important innovation that has radically improved the detection of genetic variants involved in cancer. But it has its limitations, particularly for detecting large genomic rearrangements," states Dr. Charles (Buck) Strom, Medical Director of Quest Diagnostics Nichols Institute. "Our goal for this study is to understand the capabilities of molecular combing in detecting these types of uniquely complex genetic variants. The findings will help inform our understanding of the clinical value of molecular combing and the potential opportunities for a BRCA molecular combing lab-developed test for patients and physicians in the United States."*

**Dr. Camille Chypre, Genomic Vision's VP Research & Development, comments:** "We are very excited about this study that will assess the role of structural variations in the development of breast and ovarian cancer. Molecular combing has a potential to detect with accuracy the hereditary predisposition to this pathology in high risk patients."

**Aaron Bensimon, Ph.D., Genomic Vision's co-founder and Chairman, comments:** "The decision to run this clinical study came out from the common will of Quest Diagnostics and

*Genomic Vision to maximize the success of the launch of Quest's lab-developed test in the US market. Molecular combing is a uniquely advanced technology, and developing support for its clinical value in BRCA gene variant detection is an important element in our strategy."*

Genomic Vision and Quest Diagnostics have a strategic agreement under which Quest retains exclusive rights to develop, validate and market tests based on molecular combing in the fields of breast and ovarian cancer, such as BRCA gene mutation testing, as well as hereditary colon cancer (Lynch syndrome), spinal muscular atrophy (SMA) and facioscapulohumeral muscular dystrophy (FSHD), in the United States, India and Mexico. In 2014, Quest Diagnostics introduced a molecular combing FSHD lab-developed test to physicians in the United States.

Quest Diagnostics is a leader in genetic, cancer and women's health diagnostics. The company provides a suite of tests for identifying gene variants involved in germline (genetic) cancers and somatic tumors under the BRCAVantage™ and OncoVantage™ brand names.



## **ABOUT GENOMIC VISION**

Founded in 2004, Genomic Vision is a molecular diagnostics company that specializes in the development of diagnostic tests for genetic diseases and cancers based on molecular combing. Using this innovative technology that allows the direct visualization of individual DNA molecules, Genomic Vision detects quantitative and qualitative variations in the genome that are at the origin of numerous serious pathologies. The Company is developing a solid portfolio of tests that initially target breast and colon cancers. Since 2013, the Company has marketed the CombHelix FSHD test for identifying facioscapulohumeral dystrophy (FSHD), a myopathy that is difficult to detect. It is marketed in the United States through a strategic alliance with Quest Diagnostics, the American leader in diagnostic laboratory tests, and in France directly by the Company. Genomic Vision has been listed on Compartment C of Euronext Paris since April 2014.

## **ABOUT MOLECULAR COMBING**

DNA molecular combing technology significantly improves the structural and functional analysis of DNA molecules. DNA fibers are stretched over glass slides, as if "combed", and uniformly aligned over the entire surface. It is then possible to identify genetic anomalies by locating specific genes or sequences in the patient's genome using genetic markers, a technique developed by Genomic Vision and patented under the name Genomic Morse Code. This exploration of the entire genome at high resolution via a simple analysis enables the direct visualization of genetic anomalies that are undetectable by other technologies.

For further information, please go to: [www.genomicvision.com](http://www.genomicvision.com)

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