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Genomic Vision Presents the Initial Results of the Pilot Study Undertaken with Quest Diagnostics in Spinal Muscular Atrophy (SMA) at the ASHG 2016 Annual Meeting in Vancouver

Molecular combing allows a more accurate mapping of the SMN locus in the African-American population than genetic sequencing

Bagneux (France) - Genomic Vision (FR0011799907 – GV), DNA molecular combing specialist that develops tests for the diagnostics market and tools for the life sciences research market, today announces that it has presented, at the Annual Meeting of the American Society of Human Genetics (ASHG 2016, October 18 - 22, 2016, Vancouver, Canada), the initial results of the pilot study undertaken with Quest Diagnostics that aims to identify a biomarker to improve genetic counseling for Spinal Muscular Atrophy (SMA) in the African-American population.

SMA is a hereditary genetic disease caused by a defect of the SMN1 gene in both of the patients' copies of chromosome 5. The disease is transmitted in an autosomal recessive manner, which means that the healthy parents of an affected child carry the SMN1 gene defect, although they are completely asymptomatic. The frequency of healthy carriers of this disease in the general population is 1/40 to 1/60. The high incidence of this disease, and its severity, are the reasons why there is a strong demand for genetic counseling.

Due to the particularly complex genomic organization of the SMN locus, around 8% of individuals globally and 30% of healthy carriers in the African-American population cannot be detected efficiently using traditional molecular biology techniques. Detecting these healthy carriers is therefore crucial to improving genetic counseling among this population.

The poster presented during the annual meeting (entitled *Molecular Combing reveals structural variations in the Spinal Muscular Atrophy locus in African-American population*, session: *Molecular and Cytogenetic Diagnostics*) concerned the accurate mapping of the SMN locus in the African-American population using molecular combing technology.

Anne Jacquet, Director of Biomedical Research at Genomic Vision, comments: "*The SMN locus is a very complex region that no technology has yet been able to accurately characterize. The initial results obtained*

with molecular combing using a specific Genomic Morse Code for the studied SMN region has revealed a more complex and variable genomic organization than that described in human genome sequencing databases. We have notably identified, at various points along the SMN locus, variable numbers of copies of the gene within a same individual and from one individual to another. This more accurate mapping of the SMN locus should provide us with crucial information for developing our screening test for healthy carriers, notably for the African-American population within which 30% of healthy carriers are currently not efficiently detected that is thus a real problem for providing families with genetic counseling."

ABOUT GENOMIC VISION

Founded in 2004, Genomic Vision is a DNA molecular combing specialist that develops tests for the diagnostics market and tools for the life sciences research market. Using its 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. Utilizing this technology DNA fibers are stretched over glass slides, as if "combed," and are 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 visit www.genomicvision.com

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