



GENOMICS | GENETICS | R&D | DIAGNOSTIC TESTS

GENOMIC VISION: OXFORD UNIVERSITY SELECTS THE MOLECULAR COMBING TECHNOLOGY TO STUDY DNA REPLICATION IN THE CONTEXT OF A RARE DISEASE

- The FiberVision® platform will be used to understand genetic mutations in Ruijs-Aalfs Syndrome (RJALS), an autosomal recessive disease characterized by chromosomal instability, premature aging, and early onset of hepatocellular carcinoma in children

Bagneux (France), February 8, 2018 – 7:45 am CET- Genomic Vision (FR0011799907 – GV), company specialized in the development of diagnostic tests for the early detection of cancers and hereditary diseases, and applications for life sciences research, announced today the adoption of its molecular combing technology with the use of the FiberVision® platform by the University of Oxford, UK, to further understand the ubiquitin-proteasome system and its role in the Ruijs-Aalfs Syndrome (RJALS).

RJALS, also known as SPARTAN syndrome, is a human autosomal recessive disease characterized by chromosomal instability, premature aging, and early onset of hepatocellular carcinoma in children.

Kristijan Ramadan, Senior Group Leader and Associate Professor at the Cancer Research UK / Medical Research Council Oxford Institute for Radiation Oncology and Department of Oncology, declared: *"We aim to understand how the components of the ubiquitin-proteasome system affects DNA Replication during its initiation, elongation and termination. We have recently identified a human syndrome (Ruijs-Aalfs Syndrome - RJALS), caused by monogenic and biallelic mutations in SPRTN protease gene/protein. We found that the main cause of RJALS is DNA Replication stress phenotype detected by DNA fibre technique. Now, we aim to use the technology of Genomic Vision coupled DNA combing to better understand how the SPRTN mutations detected in RJALS affect DNA firing, the velocity of DNA Replication fork, fork staling and fork termination. We believe that the FiberVision® platform will help us to better and more thoroughly investigate the role of SPRTN protease in DNA Replication and ultimately understand how SPRTN protease preserves DNA replication and thus protect us from DNA Replication stress and consequently accelerated ageing and cancer."*

Aaron Bensimon, co-founder and CEO of Genomic Vision, added: *"The acquisition of our FiberVision® platform by the University of Oxford, one of the leading academic team supporting excellent science, and training the very best scientists, is a new recognition of the advantages of our technology. The team of Prof. Ramadan is a perfect illustration of the use of molecular combing to understand genetic mutations in severe and rare complex disease. RJALS is a critical disease that causes extremely serious symptoms to patients. I strongly believe that molecular combing is an added value to the studies on SPRTN. The FiberVision® platform*

is the perfect tool to uncover the molecular mechanisms of SPRTN and its role in the disease onset. The platform facilitates the test performance with more reliable results. In addition, this fantastic project by Prof. Ramadan validates our business model of allowing academic research teams to find new applications where an unmet medical need can be addressed using the GV technology."

ABOUT GENOMIC VISION

GENOMIC VISION is a company specialized in the development of diagnostic solutions for the early detection of cancers and serious genetic diseases and tools for life sciences research. Through the DNA Molecular Combing, a strong proprietary technology allowing to identify genetic abnormalities, GENOMIC VISION stimulates the R&D productivity of the pharmaceutical companies, the leaders of the diagnostic industry and the research labs. The Company develops a robust portfolio of diagnostic tests (breast, ovarian and colorectal cancers, myopathies) and analysis tools (DNA replication, biomarkers discovery, gene editing quality control). Based near Paris, in Bagneux, the Company has approximately 50 employees. GENOMIC VISION is a public listed company listed in compartment C of Euronext's regulated market in Paris (Euronext: GV - ISIN: FR0011799907). For further information, please visit www.genomicvision.com

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