Genomic Vision Technology Used to Genetically Characterise Patients Affected by Familial Adult Myoclonic Epilepsy (FAME)

- Identification and characterization by GV’s technology of a new genetic cause for FAME
- Extensive somatic variability in expansion length and structure highlighted
- Discovery of a correlation between the age at seizure onset and the length of a specific part of the expansion, opening the path to the development of a new diagnostic assay

Bagneux (France) - GENOMIC VISION (FR0011799907 – GV), a biotechnology company developing molecular and Artificial Intelligence tools to control quality and safety of genetically modified genome, is pleased to announce that the Institute of Human Genetics at the University of Duisburg in Essen (Germany), has developed a new assay to identify and characterise patients with FAME (Familial Adult Myoclonic Epilepsy) using Genomic Vision molecular combing proprietary technology and support.

FAME is a rare, very slowly progressive genetically heterogeneous disorder characterised by cortical tremor and seizures. The recent publication in the prestigious journal Nature Communications “Unstable TTTTA/TTTCA expansions in MARCH6 are associated with Familial Adult Myoclonic Epilepsy type 3” describes how molecular combing has been implemented to characterise nucleotide expansions (TTTTA/TTTCA) in a specific gene for one form of this disorder (FAME3).

Genomic Vision Molecular Combing technique proves to be a robust and accurate technique to precisely measure the variability in length and structure of these repeats and to identify for the first time micro-rearrangements occurring near or within this expansion site. Professor Christel Depienne, Ph.D. and professor at the University hospital Essen explains: “We are very pleased that molecular combing technique allowed us to characterise the expansions in FAME loci whereas methods such as next generation sequencing could not provide results with enough coverage and accuracy. We really believe that this technique has a lot of potential to characterise all forms of FAME and will lead to the development of an excellent diagnostic assay which will allow to improve the management and the quality of life of patients affected by this condition”.

The fine characterization of FAME3 patients with molecular combing has revealed an inverse correlation between the age at seizure onset and the length of the expansion, highlighting the needs to better characterize expansions in FAME patients. Aaron Bensimon, co-founder and CEO of Genomic Vision adds: “We know that Molecular Combing is the appropriate approach to assess all DNA rearrangements in genetic diseases, and in particular to size length variations of repeated regions and are confident that it will become the standard. These variations are the causes of several neurological diseases that we are currently investigating to develop new diagnostic assays. We are delighted that the Institute of Human Genetics, at the University of Duisburg-Essen uses GV’s technology to characterise the FAME condition”.

ABOUT GENOMIC VISION

Genomic Vision is a biotechnology company developing molecular and Artificial Intelligence tools to control quality and safety of genetically modified genome in particular in genome editing technologies and biomanufacturing processes.
Genomic Vision proprietary molecular tools provide robust quantitative measurements that are needed to enable high confidence characterization of DNA alteration in the genome. These tools are currently used for monitoring DNA replication in cancerous cells, for early cancer detection and the diagnosis of genetic diseases.

Based near Paris, in Bagneux, the Company has approximately 30 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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