



GENOMICS | GENETICS | R&D | DIAGNOSTIC TESTS

## GENOMIC VISION: THE PERFORMANCES OF THE FSHD DIAGNOSTIC ASSAY PRESENTED AT THE 2018 INTERNATIONAL FSHD RESEARCH CONFERENCE (IRC) IN LAS VEGAS

- The team of Prof. Frank Baas (Leiden University Medical Centre) presented the results of a study conducted on 80 patients
  - The results demonstrate an improvement of the FSHD detection thanks to molecular combing compared to the common Southern Blot technology

**Bagneux (France), June 11, 2018 – 7.45 am (CEST) - Genomic Vision (FR0011799907 – GV)**, a company specializing in the development of diagnostic tests for the early detection of cancers and hereditary diseases, and applications for life sciences research, today announces that Prof. Frank Baas (Leiden University Medical Centre), a worldwide recognized key opinion leader on FSHD (Facioscapulohumeral Muscular Dystrophy), has presented the results of an efficacy study of the FSHD diagnostic assay based on the FiberVision® platform in comparison to the traditional technology of Southern blot. This presentation was held last weekend in Las Vegas at the 2018 FSHD International Research Conference (IRC).

During this event which gathered the most prestigious key opinion leaders on FSHD, the team of Prof. Baas presented the results of a comparison study led in Leiden with two groups of 40 individuals. Entitled « Evaluation of FSHD1 testing in diagnostics using FiberVision® platform based on Molecular Combing technology », the study demonstrated that the molecular combing technology allowed a 100 % correct classification of individuals not affected by the disease. The FSHD diagnostic assay of Genomic Vision also allowed to precisely quantify the D4Z4 repeated sequence involved in the pathology in 95% of the samples of affected patients whose complete analysis was conducted.

**Aaron Bensimon, cofounder and CEO of Genomic Vision, declared:** « *While our FSHD diagnostic array is already used by several reference hospital centers, such as La Timone in Marseille, we are extremely proud that its performances are highlighted. Presented during the prestigious IRC, the performance study comparing our technology to Southern blot demonstrates the sharp precision of molecular combing and positions it as a new standard for the FSHD detection while deepening the knowledge and the understanding of the genetic events of the pathology.* »

Facioscapulohumeral muscular dystrophy (FSHD) is the third most prevalent muscular hereditary myopathy worldwide. This genetic disease is characterized by atrophy and weakness of the face, shoulders, and ambulatory muscles. There is great variability in the clinical severity, from a severe infantile form to individuals who remain asymptomatic throughout most of their life. It is estimated that this autosomal dominant disease affects 1 in 10,000 to 1 in 20,000 people. Today there are 870,000 individuals worldwide affected by FSH, a number that could be

significantly higher given the undiagnosed cases. About 70% of FSHD patients inherit the disease from a parent, while 30% of the cases are sporadic and associated with *de novo* mutations.

## 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](http://www.genomicvision.com)

## CONTACTS

### Genomic Vision

Aaron Bensimon  
Co-founder, Chairman & CEO  
Tel.: +33 1 49 08 07 50  
[investisseurs@genomicvision.com](mailto:investisseurs@genomicvision.com)

### Ulysse Communication

**Press Relations**  
Bruno Arabian  
Tel.: +33 1 42 68 29 70  
[barabian@ulyse-communication.com](mailto:barabian@ulyse-communication.com)

### NewCap

**Investor Relations**  
Dušan Orešanský / Emmanuel Huynh  
Tel.: +33 1 44 71 94 92  
[gv@newcap.eu](mailto:gv@newcap.eu)

## FORWARD LOOKING STATEMENT

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