



**Press release**  
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**GENOMICS | DIAGNOSTIC TESTS | GENETICS | R&D**

## **GENOMIC VISION and ROUEN University Hospital launch genomic biomarker identification trial to improve genetic counseling for Infantile Spinal Muscular Atrophy in France**

**Trial represents an important step in the partnership with Quest Diagnostics in the U.S.**

**Genomic Vision (FR0011799907 – GV)**, a French molecular diagnostics company specializing in the development of diagnostic tests for genetic diseases and cancers based on molecular combing technology, and the genetic laboratory of **Rouen University Hospital** announce that they will be collaborating in a Biomedical Research trial aimed at identifying the complex structural variations responsible for the infantile form of spinal muscular atrophy (SMA - Spinal Muscular Atrophy). These complex structural variations cannot be detected by current diagnostic techniques and are the cause of errors in screening couples at risk for conceiving a child with SMA.

The study will use Genomic Vision's proprietary molecular combing technology to examine the complex structural variations of the DNA of 360 subjects with an aim to make an accurate "map" of the SMN1 gene region in order to identify a genetic biomarker associated with these complex structural variations. The first of the trial's subjects are expected to enroll in October 2015. For more information on the study, please visit the website [www.ClinicalTrials.gov](http://www.ClinicalTrials.gov) (reference NCT02550691).

**Aaron Bensimon, Ph.D., Co-founder and CEO of Genomic Vision, remarked:** *"We are excited to be initiating this SMA study with Rouen University Hospital, as their clinicians have a great deal of clinical research experience. With this agreement, Genomic Vision is effectively launching the development of the SMA test while advancing its strategy to create partnerships with key opinion leaders, which is an important element to building our product portfolio of new diagnostic tests based on molecular combing. This is also an important phase in our collaboration with Quest Diagnostics in the United States, where there is considerable need for innovative diagnostic tools for this disease."*

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 or 1 million to 1.5 million healthy carriers in France. The high incidence of this disease, and its severity, are the reason why there is a strong demand for genetic counseling. Because of the complexity of these structural

variations, the approximately 8% of people in the general population who are healthy carriers of SMN1 gene anomalies go undetected by tests currently available.

*"This presents a real problem for genetic counseling of these hard-hit families. We need a new generation of tests which will allow detection of all the healthy carriers,"* **states Dr. Saugier-Weber, a geneticist and hospital practitioner at Rouen University Hospital.**

*"We believe it is essential to identify such a biomarker, as this should ultimately lead to the development of a simple diagnostic test that would increase reliability in detecting healthy carriers,"* **stresses Anne Jacquet, Director of Biomedical Research at Genomic Vision.**

Rouen University Hospital, as trial sponsor (Coordinator, Professor Frébourg; Scientific Supervisor, Dr. Saugier-Weber), has received authorization from the National Agency for Medicines and Health Products Safety (ANSM) and from the Ethics Committee.

### **ABOUT SPINAL MUSCULAR ATROPHY**

SMA is one of the two most frequent neuromuscular diseases of childhood, along with Duchenne muscular dystrophy. In France, its incidence is estimated to be on the order of 1 affected child for every 6,000 births or approximately 125 children with SMA per year. This disease is characterized by weakness (paralysis) and early-onset muscle wasting (atrophy) in the pelvis, shoulders, trunk, and extremities (arms and legs). Children with the most serious type of SMA (type I) have generalized muscle weakness, most often leading to death by respiratory failure before 2 years of age.

### **ABOUT ROUEN UNIVERSITY HOSPITAL**

The molecular genetics laboratory at Rouen University Hospital specializes in oncogenetics (colon cancer and Li-Fraumeni Syndrome) and neurogenetics (Infantile Spinal Muscular Atrophy and the various genetic causes for intellectual disability). Rouen University Hospital is currently sponsoring over one hundred biomedical research studies. The genetics trial, which is being conducted in partnership at Rouen University Hospital's Inserm Unit U1079, is one of the Hospital's research hallmarks.

### **ABOUT GENOMIC VISION**

Founded in 2004, Genomic Vision is a molecular diagnostics company specialized 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 notably target breast cancer and cancer of the colon. Since 2013, the Company has marketed the CombHeliX FSHD test for identifying a myopathy that is difficult to detect, Facio-scapulo-humeral dystrophy (FSHD), in the United States thanks to a strategic alliance with Quest Diagnostics, the American leader in diagnostic laboratory tests, and in France. Genomic Vision has been listed on Compartment C of Euronext Paris since April 2014.

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

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