Gene Therapies Continue to Emerge as Effective Treatments for Rare Diseases, But Complex Challenges Remain

To mark International Rare Disease Day, Genethon identifies the most urgent priorities in overcoming these obstacles

Editor's Note: Video featuring Genethon CEO Frederic Revah: Rare Disease Day 2022: Ensuring Patient Access to Treatments is our Priority

PARIS, FRANCE (February 28, 2022) – Genethon, a non-profit research organization focused on developing gene therapies for rare diseases, today recognizes International Rare Disease Day by reflecting on successes and highlighting challenges that remain in bringing hope to 300 million patients worldwide who suffer from more than 7,000 rare diseases.

"As a world leader in gene therapy, Genethon is making a unique contribution to finding treatments and cures for rare diseases," said Frederic Revah, Ph.D., Genethon CEO. "Products emerging from our research make up a global pipeline of treatments for patients who have no therapeutic alternatives. Today, thousands of patients around the world are already benefiting from our research."

Products resulting from Genethon's research include a gene therapy on the market for spinal muscular atrophy and 12 other treatments in clinical trials for rare diseases of the muscle, immune system, eye, and liver. Another seven products emerging from Genethon's research should enter clinic trials in the next few years.

However, Dr. Revah observes. "While therapeutic successes are multiplying, demonstrating the full relevance of this breakthrough technology, the development of gene therapy for rare diseases is coming up against scientific, technological, clinical, economic and financial obstacles, which jeopardize making it available to as many people as possible."

To overcome these obstacles Genethon has identified three priority initiatives. One of the most urgent is to invest massively in innovation to continue to improve vectors and bioproduction processes. The objective is to produce better, faster and in greater quantities, which will also have the effect of lowering costs.

Genethon's dedicated team of 35 experts and its privileged partnership with Yposkesi, Europe's largest contract development and manufacturing company, has all the assets to quickly obtain results.

Another priority involves have regulations evolving for clinical trials and product registration to optimize development costs for rare diseases. The third focuses on ultra-rare diseases, which account for 85% of all rare diseases and have no commercial model. To finance

development of therapies and offer these treatments to patients will require an innovative model of public-private cooperation between pharmaceutical companies, non-profit laboratories and public institutions.

“These are major challenges, and the solutions ultimately will have a positive impact well beyond rare diseases,” Dr. Revah said. “The expertise developed in creating gene therapies for rare diseases could benefit more frequent diseases, as it already has for certain cancers and, in the near future, for others such as neurodegenerative diseases."