Rare Disease

Pfizer is working to accelerate the development of effective therapies for patients affected by rare diseases, which are often debilitating conditions with exceptionally low prevalence.

We are positioned to use our global resources to help address the challenges of living with a rare disease and develop new medicines for those in need, through community partnerships, research and development, robust patient support and educational initiatives.

Accelerating our Gene Therapy Leadership

"The field of gene therapy research has made tremendous strides in recent years. We believe that gene therapy may hold the promise of bringing true disease modification for patients suffering from devastating diseases, and we hope to see this promise come to fruition – through new and existing in-house capabilities and potential partnership opportunities – in the years to come."

Mikael Dolsten
M.D., Ph.D., President of Pfizer Worldwide Research and Development

Gene therapy holds tremendous promise to potentially deliver highly specialized, transformative therapies to patients in areas of high unmet medical need, particularly in rare, monogenic diseases with loss of function.

Recognizing the promise of gene therapy, Pfizer has been making investments in this arena for the past several years, seeking to bring together the foremost expertise in recombinant Adeno-Associated Virus (rAAV) vector design and development through partnerships, deepening our existing in-house knowledge of disease biology, and expanding upon our strong expertise in complex biologic medicine manufacturing and analytical capabilities.

Pfizer amplified this commitment in 2016 by acquiring Bamboo Therapeutics, Inc., a biotechnology company based in Chapel Hill, N.C., focused on developing gene therapies for the potential treatment of patients with certain rare diseases related to neuromuscular conditions.

Bamboo's portfolio includes potential best-in-class rAAV-based gene therapies that will complement Pfizer's rare disease and gene therapy portfolios in two priority areas: neuromuscular, with a pre-clinical asset for Duchenne muscular dystrophy (DMD), and central nervous system, with pre-clinical assets for Friedreich's ataxia and Canavan disease, and a Phase 1 asset for giant axonal neuropathy.

Bamboo's approximately 11,000-square foot, fully staffed and operational manufacturing facility has experience producing Phase 1/2 materials using a superior suspension, cell-based production platform that increases scalability, efficiency and purity. This helps enable the DMD program and other projects requiring large amounts of rAAV. The facility, previously known as the University of North Carolina Vector Core facility, has served as a qualified supplier of rAAV vectors for several health care companies and academic institutions.

The acquisition of Bamboo has significantly progressed Pfizer's ability to develop and bring to market potentially life-changing treatments for patients in need. Learn more about how Pfizer are using the power of technology and innovative science to advance patient care.

Our collaboration with Spark Therapeutics, Inc. a fully integrated gene therapy company dedicated to challenging the inevitability of genetic disease, regarding investigational therapy SPK-9001, a potentially transformative treatment for hemophilia B that incorporates a bio-engineered rAAV vector, has seen encouraging progress in 2016. The companies have announced positive initial data from the first nine participants in a Phase 1/2 clinical trial evaluating SPK-9001. In July, SPK-9001 received breakthrough designation from the U.S. Food and Drug Administration (FDA), which is intended to expedite the development and FDA review of drugs to treat a serious or life-threatening disease or condition.