ASHG
American Society of Human Genetics

Success Stories
in Human Genetics and Genomics Research

HOW HUMAN GENETICS DELIVERS HEALTH ADVANCES

Advances in federally funded genetics and genomics research are leading to new discoveries in preventing, diagnosing, and treating diseases—with much more to come. In this way, human genetics and genomics is providing hope for patients, families, and communities.

Rare Diseases

Then: Spinal Muscular Atrophy

Spinal muscular atrophy (SMA) is a rare childhood disease characterized by progressive muscle weakness.[1] The majority of children affected by the most severe form, SMA type I, do not survive past two years of age.[2]

Now

Investments in research to understand the cause of SMA have led to better and faster diagnosis and new treatment options. In 2016, the U.S. Food and Drug Administration (FDA) approved the first drug to treat SMA in both adults and children. Three years later, the FDA approved the first gene therapy for children with the most severe form of SMA.[3]

"We found out that he had spinal muscular atrophy so they told us take him home and love him. He's probably not gonna make it to two years. A week later we went into a clinic at Nationwide Children's and she told us about gene therapy…and they had one remaining spot in their trial….All these things they said your child will never do, Donovan's doing all of them and it's because of gene therapy."

—Mike and Laura Weisgarber,

Then: Sickle Cell Disease

In the U.S., sickle cell disease (SCD) is the most common inherited blood disorder, with many patients suffering from the disease being African American. Patients with SCD have abnormally shaped red blood cells leading to anemia and other complications.[4] Known as the "first molecular disease", SCD was only curable with a bone marrow transplant —a treatment option not available for most patients.[5]

Now

Clinical trials at the National Institutes of Health (NIH) and elsewhere are currently investigating new gene therapies and gene editing technologies to cure SCD. Early results from the trials are promising with patients living pain free and no longer needing frequent blood transfusions.

"The other day I was going into work, and I ran out of gas. This is the most horrible thing that can happen to me today, I'm ok."

—Jenelle Stephenson who received treatment as part of an NIH sickle cell disease clinical trial

Imagine

Finding a cure for all rare diseases for all patients. Genetic-based therapies have the potential to transform treatment of rare diseases. There are currently numerous genetic therapies in clinical trials for many diseases including inherited childhood blindness, Huntington's disease, and familial amyotrophic lateral sclerosis (ALS).

References:
ashg.org/advocacy/fact-sheets/

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