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. The majority of children affected by the most severe form, SMA type I, do not survive past two years of age.

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.

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. Known as the “first molecular disease”, SCD was only curable with a bone marrow transplant—a treatment option not available for most patients.

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.

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/