RARE DISEASE

Kristin Chenoweth
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Emerging Breakthroughs in Personalized Medicine Improves Care for Rare Disease Patients

New tests and therapies may help doctors diagnose rare disease patients a lot faster and lead to more effective treatments.

Although scientists have long understood that most rare diseases are caused by harmful genetic mutations, it often takes several years for doctors to identify which gene is causing a patient's specific symptoms. These years take a toll on patients and their families, who watch loved ones suffer despite dozens of costly tests and visits to various doctors and specialists. Even after the disease is diagnosed, existing daily maintenance medications can sometimes prove woefully inadequate.

Fortunately, new tests and therapies emerging in an era of personalized medicine are helping physicians use diagnostics to determine which medical treatments will work best for each patient.

Genetic testing
A new technology called next-generation sequencing (NGS) can test for thousands of genetic mutations at once. Scientists hope NGS tests may someday replace the battery of single-gene tests that doctors often use to understand the potential causes of a patient's symptoms. The use of NGS tests may help shorten the diagnostic odysseys that many rare disease patients must endure before receiving an accurate diagnosis, thereby reducing associated expenses and improving patients' lives.

Personalized medicine
Meanwhile, to improve the prospects for patients with rare diseases after they are diagnosed, the biopharmaceutical industry is developing an emerging group of personalized medicines known as gene therapies. Gene therapies promise to deliver lasting benefits by reversing the genetic causes of diseases. The U.S. Food and Drug Administration approved the first gene therapy, called Luxturna (voretigene neparvovec), in 2017. By revising a harmful genetic mutation, Luxturna can restore vision to patients with Leber congenital amaurosis, a rare genetic retinal disease.

Researchers studying the benefits of NGS tests, gene therapies and other personalized treatments hope that their work will help advance a new era in healthcare that quickly targets more effective treatments to patients who will benefit from them.

Edward Abrahams, President, Personalized Medicine Coalition

America's Most Successful Public Health Program at Risk

A life-saving bill is set to expire on September 30, 2019. The Senate must act. Twelve-thousand babies and their families are depending on it.

George Fox loved growing up in Florida. When he and his high school sweetheart got married and settled down in the Florida Keys, they looked forward to a dream life. After their son, Phoenix, was born, George beamed when the doctor remarked on how strong his baby appeared.

Within weeks, however, little Phoenix began showing symptoms of a life-threatening heart abnormality. After months of tests, he was diagnosed with Pompe disease, an incurable muscle-wasting condition that can cause heart weakness and, if left untreated, can result in death. One in 28,000 Americans suffer from Pompe.

Screening programs save lives

Of the 4 million babies born in the United States each year, 1 in 300 are found to have a potentially devastating condition through screening. Newborn screening is the most successful public health program in the history of our country.

A bill currently before Congress, the Newborn Screening Saves Lives Reauthorization Act, or S. 2158, would continue to provide assistance to states in order to improve and expand newborn screening programs.

Time is running out

If S. 2158 does not pass, other states may not be able to implement testing for Pompe and other newly-detectable diseases. Please go to RareScreening.org to contact your Senators and ask them to co-sponsor and support S.2158.

Mark Dant, Chairman of the Board of Directors, The EveryLife Foundation for Rare Diseases

"Building Bridges of Hope to a Cure"

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