



## RARE DISEASES

From discovery to diagnosis to treatment

It is estimated that about 25-30 million Americans suffer from a rare disease. In the United States, a rare disease is defined as affecting fewer than 200,000<sup>1</sup>. While each rare or ultra-rare disease affects only a small number of individuals, the impact on patients, families, and communities is enormous.

Rare diseases collectively affect a significant portion of the population. Most conditions are chronic, debilitating, and even life-threatening. Advances in genetics and genomics research have and will continue to benefit Americans living with rare diseases.

### Rare Diseases by the Numbers



Altogether, there are about **7,000** rare diseases in the U.S.



**1 in 10** Americans have a rare disease.



**1 of 2** rare disease patients are children



**1 in 4** rare diseases are lethal before age **5**



Over **90%** of rare diseases still lack an approved treatment.<sup>2</sup>



Between 2010 and 2015, the Office of Rare Diseases Research invested about **\$100 million in 180 research projects** to better understand and discover treatment options for individuals living with rare diseases.<sup>3</sup>

## NIH-funded Undiagnosed Diseases Network: Solving Medical Mysteries

Launched in 2014, the Undiagnosed Diseases Network (UDN) aims to solve the most challenging medical mysteries using advanced genomic and other technologies. Since its inception, over 300 patients with ultra-rare diseases have been diagnosed, and 31 new syndromes have been defined. The UDN provides a more financially feasible option for patients and their families: the average cost of a diagnosis through the UDN is \$18,903<sup>4</sup>; the average cost of care for an individual patient prior to enrolling in a UDN site totals over \$300,000.

### Real World Examples

#### Precision Medicine: A New Paradigm for Patient Care

Mila was six years old when she was diagnosed with an ultra-rare form of Batten disease—just 70 documented cases. Batten disease refers to a group of 10 neurodegenerative diseases with general symptoms including dementia, blindness, and epilepsy<sup>5</sup>. It is always fatal and there is no cure yet. NIH-funded researcher Timothy Yu, MD, PhD, designed a drug that would correct her rare mutation in the CLN7 gene. For months, Mila was experiencing about 30 seizures every day. The drug, fittingly named “milasen,” significantly reduced the number and intensity of the seizures.<sup>6</sup>

*“Mila’s treatment is allowing me the chance to ensure that her spark will ignite others, and one day soon there will be a bright light where there was once just darkness. Not just for Mila. Not just for Batten. But for the millions of children around the world with devastating genetic diseases.”<sup>7</sup>*

—Julia Vitarello, Mila’s Mom

#### A Significant Discovery

Seven children in America have been afflicted with a rare genetic disease that causes progressive neurodegeneration, marked by loss of motor function and language skills. Thanks to UDN scientists who closely studied the specific gene mutation in IRF2BPL and its function, families of the affected children were able to better understand the disease as a whole.<sup>8</sup>

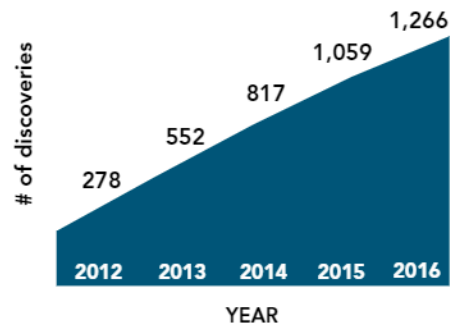
*“My family and I are forever indebted to these researchers for taking the time to patiently explain to us their findings on IRF2BPL mutation and how it was affecting Leo’s brain.”<sup>9</sup>*

—Satoshi Enoue, father of a patient with this rare disease

### How can Congress Support Research?

Researchers need robust, predictable, and sustained NIH funding. Congress can support the next genetics and genomics advances by continuing to fund biomedical research.

Cumulative Gene Disease Discovery



#### Additional Resources

Undiagnosed Diseases Network:  
[undiagnosed.hms.harvard.edu/](http://undiagnosed.hms.harvard.edu/)

Genetic and Rare Diseases  
Information Center:  
[rarediseases.info.nih.gov/diseases](http://rarediseases.info.nih.gov/diseases)

National Organization for  
Rare Disorders:  
[rarediseases.org](http://rarediseases.org)

References:  
[ashg.org/advocacy/fact-sheets/](http://ashg.org/advocacy/fact-sheets/)