Rare Diseases

In the United States, a disease is considered rare if it affects fewer than 200,000 people. It is estimated that 1 in 10 Americans, and 350 to 400 million people worldwide, are living with a rare disease. For the majority of these individuals, no treatment options are available.\(^1\), \(^2\), \(^3\)

### TODAY

- **95%** of rare diseases lack any FDA-approved treatment.\(^4\)
- **50%** of those with a rare disease are children.\(^1\)
- **Approximately 7,000** rare diseases and disorders have been identified to date.\(^1\)
- It takes **18%** longer to develop an orphan* drug compared to medicines for more common conditions.\(^4\)
- Nearly **1/3** of children with a rare disease die before the age of 5.\(^1\)

*The FDA defines “orphan” drugs as medications for conditions affecting fewer than 200,000 people in the United States, or that will not be profitable within seven years of FDA approval.

### Research Delivers Solutions

The international research effort to sequence all human genes, known as the **Human Genome Project**, led to the identification of the precise genetic cause of many rare diseases. This knowledge has led to **breakthroughs in treatments, symptom management, and even cures**. For example, a 2016 study used genetic sequencing to provide a clinical diagnosis for **42%** of patients with persistent abnormalities in brain matter, whose conditions had previously gone undiagnosed using standard methodologies.\(^7\), \(^8\)

A rare disease known as **homozygous familial hypercholesterolemia (HoFH)** is a **life-threatening** condition that prevents the body from removing ‘bad cholesterol.’ Individuals with untreated HoFH often die before the age of 30. A new treatment option, **Evolocumab**, has been shown to reduce levels of ‘bad cholesterol’ among these patients by **60%** on average, greatly decreasing mortality risk and improving overall health.\(^9\)

**Severe combined immunodeficiency (SCID)** is a rare and fatal immune disorder that causes death before an infant’s second birthday. As a result of **research-based newborn screening**, children with SCID can be diagnosed and treated with a bone marrow transplant within 3 months of birth, a period during which they are still protected by their mother’s immune cells. Cost-benefit research estimates that every dollar invested in newborn screening for SCID produces **$5** in economic and societal benefits.\(^10\)

### Majority Favor Doubling Funding for Medical Research Over the Next Five Years

Do you favor or oppose doubling federal spending on medical research over the next five years?

- **41%** Strongly favor
- **29%** Somewhat favor
- **15%** Somewhat oppose
- **10%** Strongly oppose
- **5%** Don’t know

Source: A Research!America poll of U.S. adults conducted in partnership with Zogby Analytics in January 2019
Rare Diseases

Then. Now. Imagine.

THEN
In the early 1980s, 20-25 million people in the U.S. were affected by approximately 5,000 rare diseases, and there were only 10 drugs available for treatment.1, 2

NOW
The Orphan Drug Act was passed in 1983, creating the orphan drug designation and providing needed incentives for researchers and manufacturers to develop therapies for rare diseases. Since then, the FDA has approved over 500 orphan drugs. In 2018 alone, 90 rare disease indications were approved and 34 novel treatments for rare diseases were approved – 58% of all 2018 FDA drug approvals.3

IMAGINE
A cure for all rare diseases.

Number of Conditions Tested As Part of Newborn Screening, 2018

Newborn Screening

Newborn screening allows for the detection of numerous rare diseases, such as SCID, phenylketonuria, and cystic fibrosis. Early diagnosis, preventive treatment, and care lead to much better health outcomes for these infants.

Quest for Diagnosis

5.6 to 7.6:
That’s the average number of years it takes to correctly diagnose a rare disease patient in the U.S.4

25% of patients with the most “common” rare diseases wait between 5 and 30 years to receive a correct diagnosis, with 40% receiving an incorrect initial diagnosis.5

3. “RARE Disease Patients Have Cost the NSF.” Business Intelligence News, 2018
4. “Orphan drug development brings unique challenges.” PhRMA.
6. “Rare Disease Caregiving in America.” National Alliance for Caregiving, 2018
10. “Newborn screening for severe combined immune deficiency (SCID).” CDC.
15. Gainotti et al. “Meeting Patients’ Right to the Correct Diagnosis.” 2018

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