IDeaS for Change

National Center for Advancing Translational Sciences

More than 7,000 known rare diseases affect an estimated **30 million people** in the United States, most of whom are children. Taken together, **nearly 10%** of the U.S. population have a rare disease. That matches the number of people with common conditions such as diabetes, heart failure and Alzheimer's disease.

Innovative Study Reveals the High Medical Cost of Rare Diseases

Rare diseases can take a profound physical and emotional toll on patients and their families. But the true economic cost of rare diseases to people, health care providers, insurers and society is difficult to calculate.

The IDeaS (Impact of Rare Diseases on Patients and Healthcare Systems) study, led by NCATS, takes a major step toward quantifying the tremendous financial burden of rare diseases. IDeaS researchers searched medical and insurance records in four health care system's databases to measure the prevalence of a sample of 14 rare diseases. They estimated those diseases' direct medical costs and mapped the lengthy journey some people with rare diseases travel to an accurate diagnosis.

The IDeaS findings revealed:

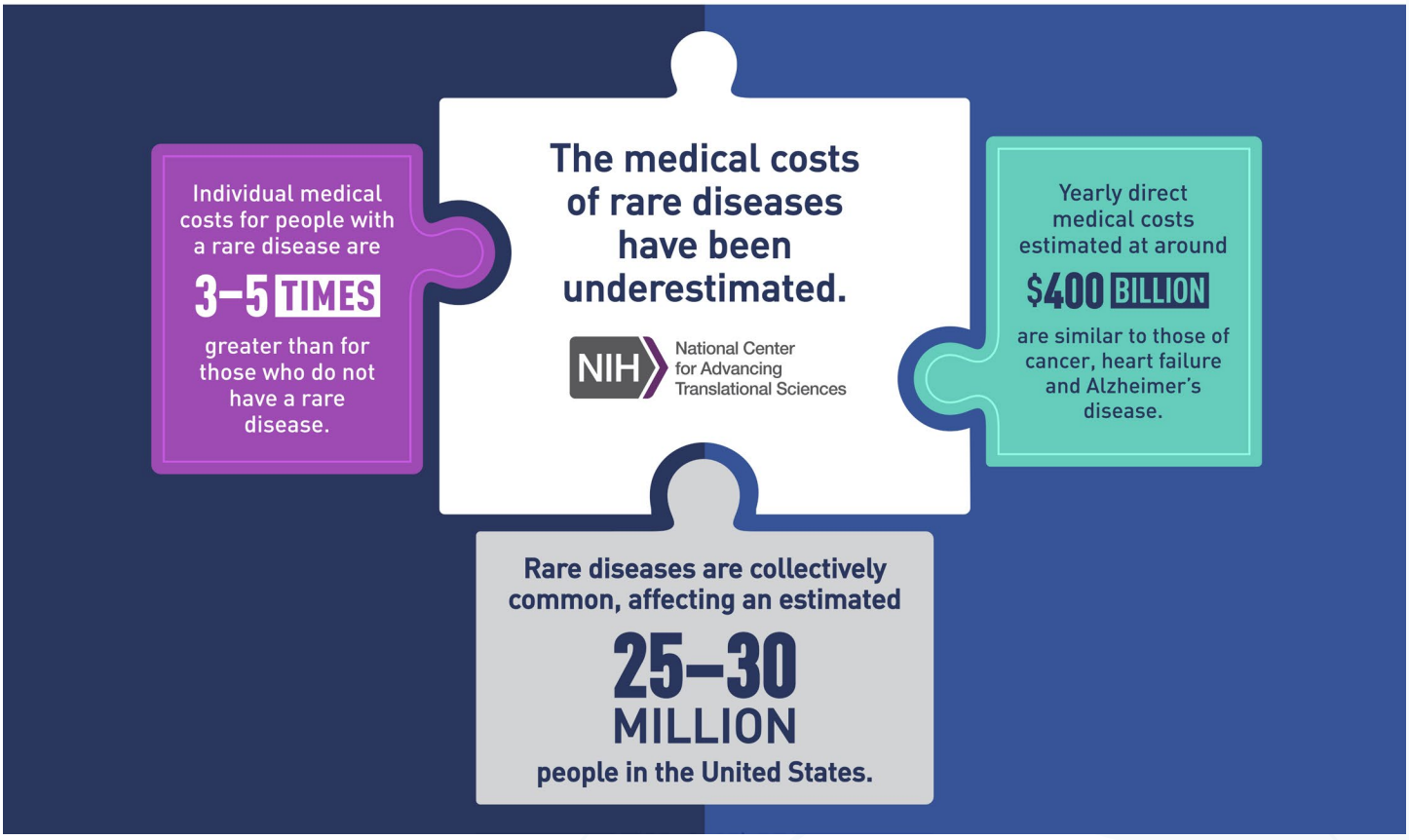
- Health care costs for people with rare diseases are significantly underestimated. The costs for those with rare diseases may be **three to five times greater** than the costs for people without a rare disease.
- The average annual cost of caring for people without rare diseases was **$5,862**, based on insurance data from the Eversana Life Sciences database. For those with rare diseases, costs ranged from $8,812 to **$140,044**.
- Medicare and Medicaid data revealed that average annual care costs for people without rare diseases was **$2,211**. For those with rare diseases, annual costs ranged from $4,859 to **$18,994**.
- Based on the Eversana cost data, the total yearly direct medical costs for people in the United States with rare diseases may reach **$400 billion per year**. That rivals the annual direct medical costs for cancer, heart failure and Alzheimer's disease.
- The diagnostic odyssey for those with rare diseases is real and prolonged. Delayed diagnosis lengthens people's journey to effective treatment. Long diagnostic odysseys result in irreversible disease progression and ongoing high medical costs.
- Disease coding and classification obstacles make it difficult for health care databases to accurately quantify the true scope and cost of rare diseases. Even when accurately diagnosed, only 20% of rare diseases have their own code in the International Classification of Diseases.

- Advances in genetic analysis and machine learning tools could speed disease identification, reshape the clinical course of a disease and reduce costs of care.
- Making more disease-modifying treatments available could slow or reverse the devastating progression of disease and preserve the quality of life and productivity of millions of people with rare diseases.

The IDeaS findings underscore rare diseases' profound costs to people and the nation's health care system. They also highlight the urgent need for more research to speed diagnosis, rethink treatment and reduce the physical and financial burden of rare diseases.

IDeaS is a collaboration among NCATS, Eversana Life Sciences, Oregon Health & Science University, Sanford Health and a health insurer in Australia. View the complete IDeaS study in the *Orphanet Journal of Rare Diseases*. Learn how NCATS and its partners work to reshape the lives of those with rare diseases at ncats.nih.gov/rare-diseases.

RARE DISEASES: Individually Rare, Collectively Common



Source: The IDeaS Intiative: Pilot Study to Assess the Impact of Rare Diseases on Patients and Healthcare Systems.