

# Investment in research saves lives and money



facts about:

## Rare Diseases

*"If you think research is expensive, try disease."*

- Mary Lasker 1901-1994

### Today:

- ❑ A disease is considered rare if it affects less than 200,000 people in the U.S. Approximately 7,000 rare diseases have been identified to date.\*
- ❑ Today, 95% of rare diseases lack any FDA-approved treatment.\*
- ❑ It is estimated that 1 in 10 Americans -- or 30 million people -- and 350 million worldwide are living with a rare disease.\*
- ❑ Of those with a rare disease, 50% are children. Nearly one-third of children with a rare disease will die before the age of 5.\*

\* GLOBAL GENES <GLOBALGENES.ORG>  
+ PHARMA <WWW.PHARMA.ORG>

### The Cost:

- ❑ In 2000, less than \$20 billion was spent on drugs to treat rare diseases worldwide. By 2020, that amount is projected to increase to \$176 billion, around 19% of the world's total drug expense.^
- ❑ Individuals with a rare disease typically visit 8 physicians, and receive 2 to 3 misdiagnoses before they are correctly diagnosed. It typically takes more than 7 years after the onset of symptoms for this costly and resource-extensive process to produce results.‡
- ❑ On average, a treatment for a rare disease costs \$137,000 per person per year.^
- ❑ Over 50% of individuals with a rare disease report having to use their personal savings to cover the cost of medical care.‡

^ THE BOSTON GLOBE <WWW.BOSTONGLOBE.COM>  
‡ RARE DISEASE IMPACT REPORT, 2013. SHIRE.

SAVING LIVES  
SAVING MONEY

### HOW RESEARCH SAVES LIVES:

- ❑ Homozygous familial hypercholesterolemia (HoFH) is a rare, life-threatening condition that inhibits the body from removing 'bad cholesterol.' Individuals with untreated HoFH often die before the age of 30. In 2012 and 2013, two new treatment options were made available that, on average, reduced these individual's levels of 'bad cholesterol' by 50%, greatly decreasing their mortality risk and improving their overall health.\*
- ❑ Approximately 30,000 Americans are living with cystic fibrosis (CF), a potentially fatal disease that affects an individual's lungs and digestive system. In the 1960s, children with CF were not expected to live past 10 years old. With advances in treatment, the life expectancy for patients with CF has more than tripled to 37 for women and 40 for men.\*

### HOW RESEARCH SAVES MONEY:

- ❑ Sickle cell disease (SCD) is a hereditary, potentially life-threatening disease that affects the red blood cells. Researchers at Johns Hopkins Children's Center have found that young children with SCD who receive a daily dose of the medication, hydroxyurea (HU), experience less pain, require fewer blood transfusions, and are 30% less likely to be hospitalized compared to children with SCD who did not receive HU. The treatment was also associated with a 31% reduction in hospitalization costs, and a 21% net decrease in annual direct medical costs for treating SCD- a savings of approximately \$3,000 per treated child.‡
- ❑ Severe combined immunodeficiency (SCID) is a fatal immune disorder that causes death before an infant's second birthday. With research-based newborn screening available, children with SCID can be diagnosed and cured through a bone marrow transplant within 3 months of birth, while they are still protected by their mother's residual immune cells. Cost-benefit research estimates that for every dollar invested in newborn screening for SCID, there is nearly \$5 in economic and societal benefits.‡

^ AMERICAN ACADEMY OF NEUROLOGY <PATIENTS.AAN.COM>  
\* PHARMA <WWW.PHARMA.ORG>  
‡ WANG, W. C. ET. AL. LANCET, 2011. 377(9778):1663-1672.  
● WANG, W. C. ET. AL. PEDIATRICS, 2013. 132(4).  
+ THOMPSON, J. D. AND MIKE GLASS, WASHINGTON STATE DEPARTMENT, 2012.

## survivor:

NAME: Max Hasenauer  
AGE: 18  
CONDITION: x-linked agammaglobulinemia



When he was just 22-months-old, Max Hasenauer was diagnosed with X-linked agammaglobulinemia (XLA) after a scratch on the playground became life threatening. XLA is a rare genetic condition where the body does not produce the antibodies necessary to maintain a functioning immune system. Max's body is unable to fight off infections, making common illnesses fatal. In order to survive, he takes antibiotics daily and receives infusions of new antibodies every three weeks. In the days leading up to his infusion, he feels fatigued and ill as the old antibodies wear off and his body struggles to fight off infections.

Despite the treatments Max receives, his XLA can quickly become life-threatening, and it has impacted Max's life in many different ways. Unable to receive the flu vaccine due to his compromised immune system, Max has had to miss school during flu season. Now a sophomore in college, Max returns home every three weeks to receive his infusions. To his disappointment, he is unable to participate in his school's semester abroad program because of limitations in healthcare coverage abroad and the rarity of the condition.

While new treatments are certainly needed to improve Max's quality of life, he would not be alive today without the research that made his infusions possible. The National Institutes of Health (NIH) has launched the Therapeutics for Rare and Neglected Diseases program, a collaboration between NIH researchers, nonprofit organizations and pharmaceutical and biotechnology companies, to accelerate the development of new treatments for many rare diseases.

As Max's younger sister Amanda shared, "I support medical progress because I wouldn't have my brother without it!"

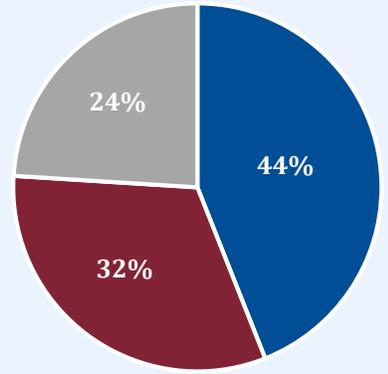
# facts about: } Rare Diseases

## Hope for the Future:

- Between 2010 and 2015, one-third of the drugs approved by the FDA were for rare diseases. Since the passage of the Orphan Drug Act, nearly 500 FDA-approved drugs have entered the market to treat rare diseases. Currently, there are more than 450 rare disease drug candidates in the pipeline.\*
- The Human Genome Project led to the identification of the precise genetic cause of more than 4,500 rare diseases. This knowledge has led to countless breakthroughs in treatments, symptom management, and even cures. As researchers continue to explore the human genome, more novel approaches will be developed, addressing the unmet needs of thousands of Americans with rare diseases.^
- For many genetic conditions (which include 80% of rare diseases), gene therapy may be the answer to a vast amount of medical conditions. For example, researchers at the University of Pennsylvania's Orphan Disease Center have been exploring gene therapy for the treatment of mucopolysaccharidosis type I (MPS I), a rare disease that causes stunted growth, deafness, and cognitive decline. They have seen promising results in animal models and are hoping to begin phase I clinical trials shortly.\*\*

### National Poll: Plurality Willing to Pay More in Taxes to Fund Research

*Would you be willing to pay \$1 per week more in taxes if you were certain that all of the money would be spent on additional medical research?*



A RESEARCH!AMERICA POLL OF U.S. ADULTS CONDUCTED IN PARTNERSHIP WITH ZOGBY ANALYTICS IN JANUARY 2015

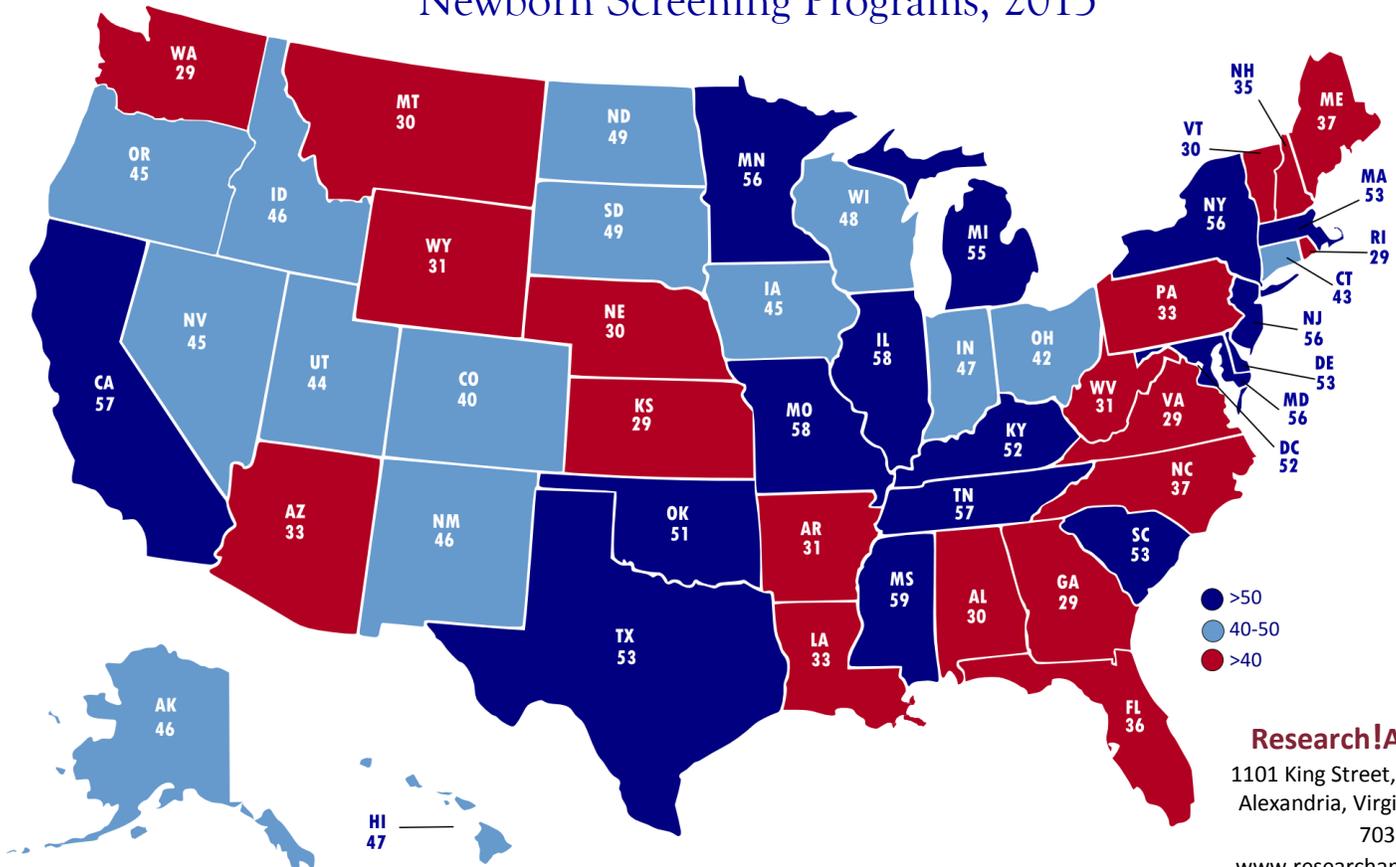
■ Yes ■ No ■ Not sure

\* PHARMA <WWW.PHARMA.ORG>  
^ NATIONAL INSTITUTES OF HEALTH <WWW.NIH.GOV>  
‡ PERELMAN SCHOOL OF MEDICINE, PENN MEDICINE <WWW.MED.UPENN.EDU>

### The Bottom Line:

*Combined, rare diseases affect one-tenth of the U.S. population. Yet the majority of these individuals do not have a single treatment option available. Both public and private sector support is critically needed to incentivize and propel drug candidates through the pipeline to the patients who desperately need them.*

## Number of Conditions Tested in State Newborn Screening Programs, 2015



### Research!America

1101 King Street, Suite 520  
Alexandria, Virginia 22314  
703.739.2577  
www.researchamerica.org  
info@researchamerica.org

SOURCE: BABY'S FIRST TEST

*The Albert and Mary Lasker Foundation is a founding partner in this series of fact sheets. www.laskerfoundation.org*