Investment in research saves lives and money

Sickie Cell Disease

Sickle cell disease (SCD) is a life-threatening hereditary condition that occurs when an individual inherits two sickle cell genes or other change in hemoglobin from each parent. Complications of SCD include pain, anemia, multiple organ damage, and susceptibility to infections. Sickle cell trait (SCT) occurs when an individual only has one copy of the gene. Individuals with SCT are asymptomatic carriers of the sickle cell gene.

Today:

- An estimated 100,000 Americans are currently living with sickle cell disease (SCD).*
- Up to 1.5% of individuals born in the U.S., or one in 66, have sickle cell trait (SCT). If two individuals with SCT have a child there is a 25% chance they will have SCD and a 50% chance they will have SCT. †
- Through newborn screening via a blood test, nearly 40,000 infants have been identified and diagnosed with SCD within a 20 year period. *
- The sickle cell gene is especially common among African-Americans; it is estimated 1 in 12 African-Americans have SCT and 1 in 500 have SCD. *
- The only known cure for SCD is a bone marrow or stem cell transplant, both of which are very limited by the availability of a genetically similar donor. *

How Research Saves Lives:

- In 1973, the average lifespan for an individual with SCD was 14-years-old. With improved screening techniques, penicillin prophylaxis, stroke prevention and hydroxyurea therapy, the life expectancy of an individual with SCD is now between 40 and 60 years. **
- Despite increased susceptibility to infections, it is estimated that fewer than 50% of individuals with SCD receive the flu vaccine. In a National Institutes of Health (NIH) funded study, a pediatric SCD clinic at Boston University implemented several evidence-based approaches to increase vaccine compliance, including enhanced electronic health records, utilization of a SCD patient registry, and parent education. Through these techniques, the clinic was able to raise their vaccination rate to 90% in just two flu seasons. *
- Children with SCD are more likely to develop, and subsequently die from, invasive pneumococcal disease (IPD), compared to children without SCD. Due to the introduction of the pneumococcal conjugate vaccine in 2000, IPD rates for children with SCD have dropped by 53%. Additionally the vaccine is thought to be a major contributor to the 42% decline in the mortality rate of young African-American children with SCD between 1999 and 2002. *

How Research Saves Money:

- NIH-funded research at Johns Hopkins University 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 cost for treating SCD- a savings of approximately $3,000 per treated child. **
- Preliminary studies have found that among SCD patients experiencing a vaso-occlusion episode (VOE), also referred to as a ‘pain crisis,’ those with dental infections were 72% more likely to be admitted into the hospital. It is estimated that the prevention of dental infections in SCD patients could save $2.5 million per year in avoided medical costs. [1]
- Researchers funded by NIH established specialized Sickle Cell Infusion Centers, which are dedicated to treating VOE and are incredibly cost effective. These centers produced a 7.6% healthcare cost savings by reducing hospitalizations by 52% and emergency department visits by 8%. [2]

The Cost:

- The total cost of direct medical care for individuals with SCD is $1.1 billion annually.*
- Up to 43% of children with SCD will be admitted into a hospital over the course of a year. Each admission will cost up to $10,013 in direct medical costs.a
- On average, Medicaid spends $1,049 per year per child with SCD for prescription medications.*

Melissa Creary was diagnosed with Sickle Cell Disease (SCD) at the age of 3. Newborn screening had not yet been implemented and a family member recommended her parents see a hematologist. Despite her diagnosis at a young age, it wasn’t until she was in high school that she had her first hospitalized pain crisis. It was around this time she learned the mechanisms and causes of SCD while taking an Advanced Placement (AP) Biology course, and this knowledge sparked an interest that has led her to pursue a career studying SCD.

After receiving degrees in biology and public health from Emory University, Melissa helped to establish SCD research protocols in the Department of Biochemistry at Morehouse School of Medicine in Atlanta, GA, and later came to the Division of Blood Disorders at the Centers for Disease Control and Prevention (CDC). There she worked with CDC leadership to create a national public health data collection system and increase awareness of SCD among the general public. “The work being done at the CDC was instrumental to a public health approach for SCD. CDC will always be in need for more funds to do work on SCD.”

As a Ph.D. candidate at Emory, Melissa speaks regularly with people who have SCD and compiles their narratives, to not only provide more information about the nature of the disease, but to humanize the condition and provide researchers with vital perspectives to direct their work. Melissa understands the importance of integrating the patient voice into research, “…it’s about individuals […] when people share their stories, their names, their images, it validates their experiences living with SCD.”

“If you think research is expensive, try disease.” - Mary Lasker 1901-1994
Hope for the Future:

- The immune system of many individuals with SCD develops resistance to the foreign cells in their blood transfusion treatments, requiring more transfusions for the same result. Using the CRISPR technique, NIH-funded researchers at Johns Hopkins University were able to correct the mutation that causes SCD in red blood cells collected from affected individuals. This may well lead to an alternative, more sustainable, and safer treatment.*

- Treatment of vaso-occlusion episode (VOE), a common symptom of SCD, has remained relatively unchanged over the past 80 years, consisting of pain management during the episode. An investigational drug, rivipansel, is showing encouraging results addressing VOE. Those on the experimental medication used less additional pain management medications.*

- Preliminary research conducted at Albert Einstein College of Medicine has uncovered the first possible treatment for the prevention of organ failure in individuals with SCD. The results suggest that antibiotics used to deplete an individual’s microbiome would reduce these common long-term complications. Additionally, the treatment was also shown to reduce VOE. Clinical trials are expected to begin soon.*

- SCD is the most common cause of strokes in children. Multiple studies have suggested the combination of Transcranial Doppler Ultrasonography (TCD) screening, blood transfusions, and hydroxyurea reduces the likelihood of strokes in high-risk children with SCD.†‡

The Bottom Line:

SCD is a chronic, painful, and life-threatening hereditary condition with no standard treatment or universal cure.

As knowledge continues to grow around the possibilities of gene editing, stem cells and bone marrow transplants, researchers are closer than ever to developing a universal cure for individuals with SCD. For this goal to become a reality, researchers must have access to the necessary resources they need to continue their lifesaving and life-improving work.

Incidence of Sickle Cell Trait in 2010, by state

Incidence per 1,000 newborns screened at birth

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

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