About Sickle Cell Disease: What's in YOUR GENES?

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What is Sickle Cell Disease?

SCD is a blood disorder

Sickle Cell Disease (SCD) is an **inherited blood disorder** that affects red blood cells. Normal red blood cells are round and flexible, which lets them travel through small blood vessels to

Causing misshapen blood cells

SCD causes red blood cells to **form into a crescent shape,** like a sickle.

Creating painful complications

The sickle-shaped red blood cells break apart easily, clump together, and stick to the walls of blood vessels, **blocking the flow of blood,** which can cause a range of serious health issues.

Image Source: Sickle Cell Disease Coalition (SCDC)

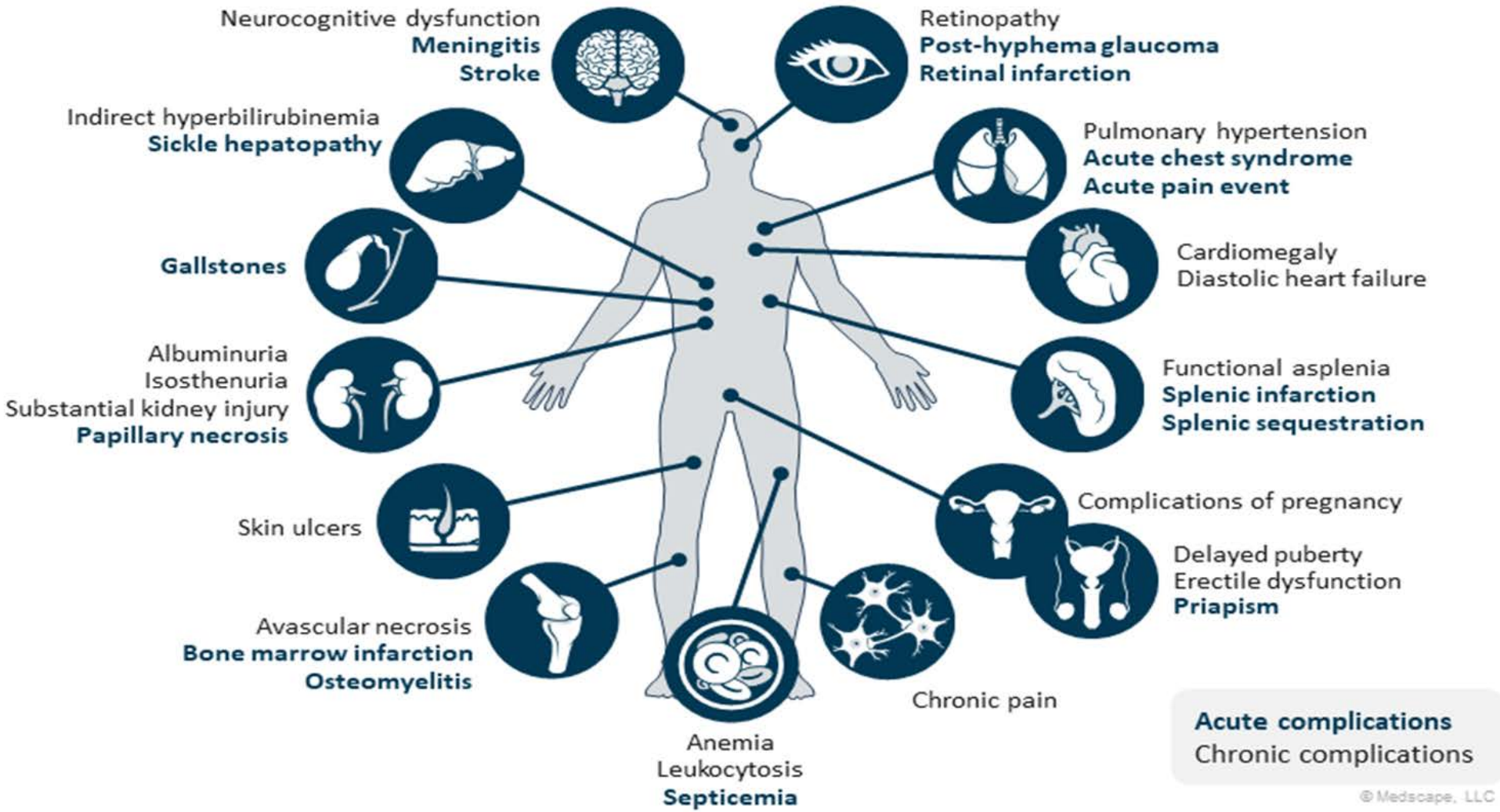
Sickle Cell Disease Complications

- People living with sickle cell disease (SCD) can start to have signs of the disease during the first year of life, as early as 5 months of age.

- **Common complications from SCD can include:**

  - Pain
  - Stroke
  - Organ damage
  - Anemia
  - Infections
  - Anxiety/Depression

SCD Is a Multi-System Disease



Kato GJ, et al. *Nat Rev Dis Primers.* 2018;4:18010.

History of Sickle Cell Disease

Hemoglobin Gene Genetic Mutation

- Sickle cell disease (SCD) results from a **genetic mutation** that occurs in the hemoglobin gene.
  - hemoglobin is the oxygen carrying component of red blood cells

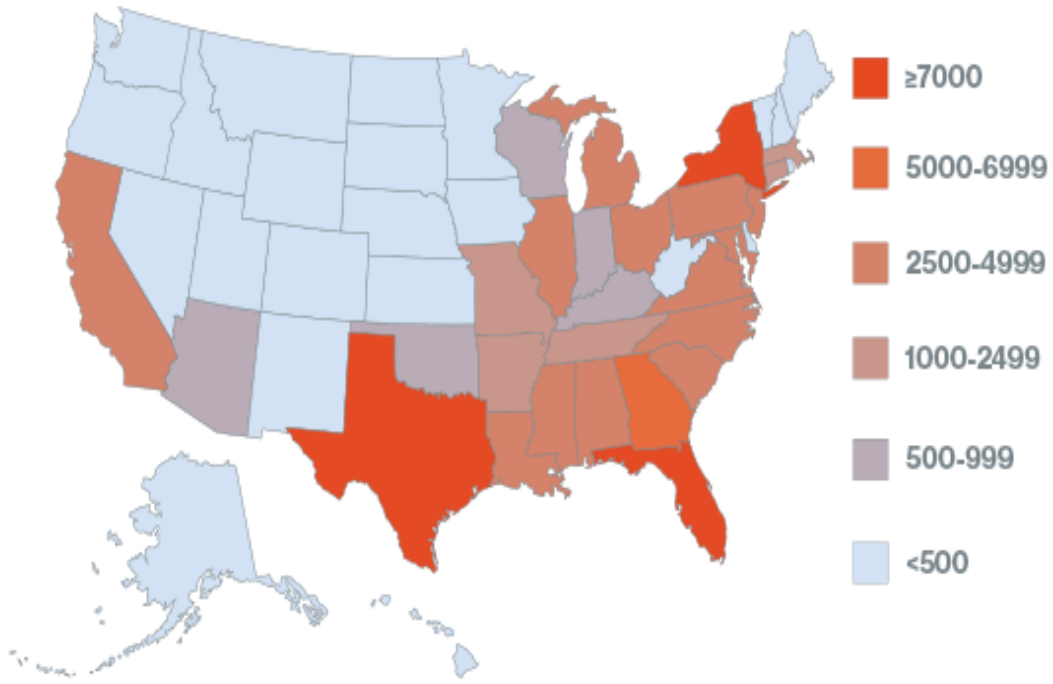
- There are several types of SCD that can result depending on the location of the mutation in the hemoglobin gene
  - sickle cell anemia (HbSS)
  - sickle hemoglobin-C disease (HbSC)
  - sickle beta thalassemia (HbSβ+, HbSβ0)

- Hemoglobin genetic mutation is common in people with ancestry from parts of Africa, South America, the Mediterranean basin, the Middle East, and India.

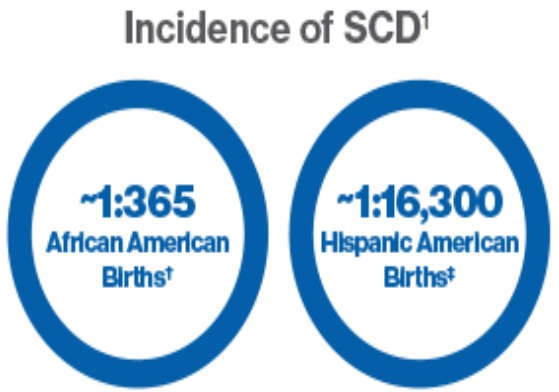
- Malaria was very common in these areas

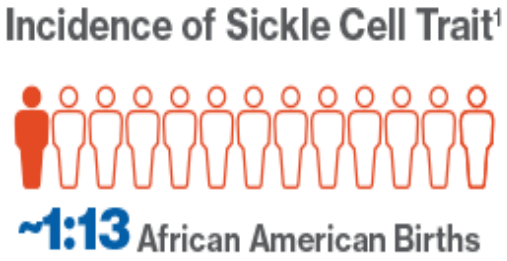
  People who inherit one mutated hemoglobin gene have sickle cell trait (SCT).
  - Parents can pass sickle cell trait to their children
  - When two persons with hemoglobin s gene, (sickle cell trait carriers) reproduce, there is a chance the child can be born with SCD.

~100,000 People in the United States Have SCD[1]



MORE THAN **55%** OF PEOPLE WITH SCD RESIDE IN JUST 10 STATES[2*]



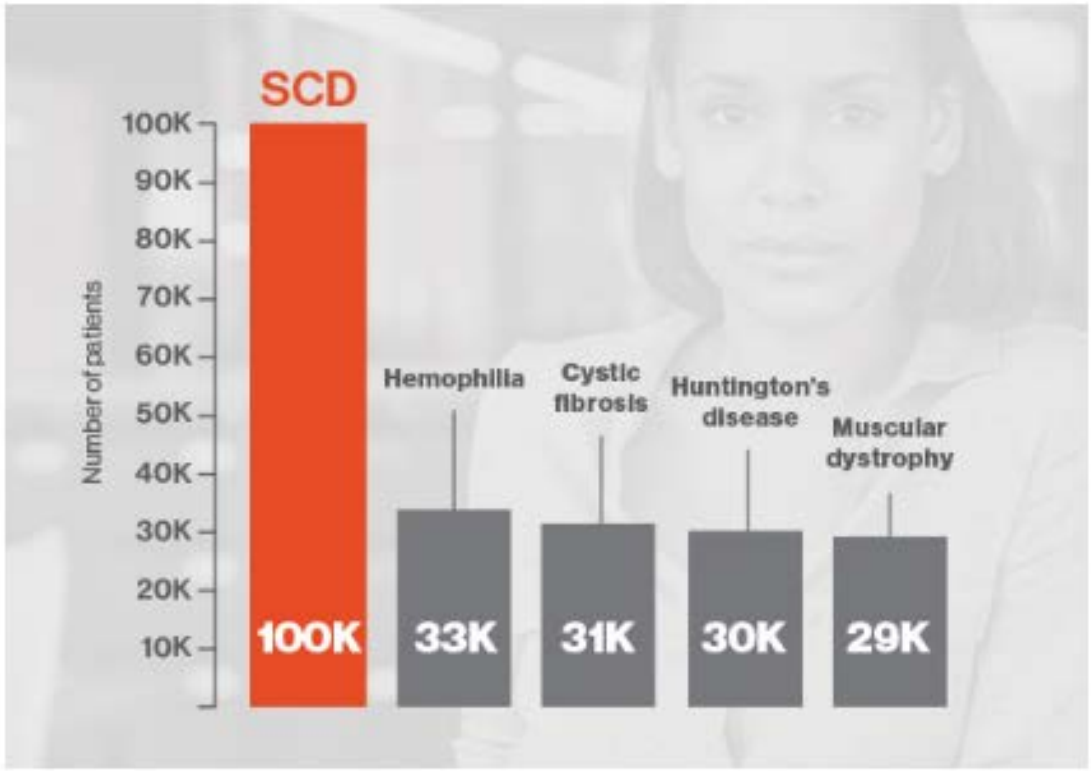


What's Your SCT Status?

https://www.rethinkscd.com/sickle-cell-disease/incidence-prevalence-prognosis/

SCD Is >3 Times More Prevalent Than Other Rare Inherited Disorders

Prevalence of Some Inherited Disorders in the United States[1,4-6]



Despite its higher prevalence, SCD awareness and funding are lower than those of other genetic diseases.[7]

https://www.rethinkscd.com/sickle-cell-disease/incidence-prevalence-prognosis/

The life span of those with the most severe form of SCD is on average 30 years shorter than for the general US population.

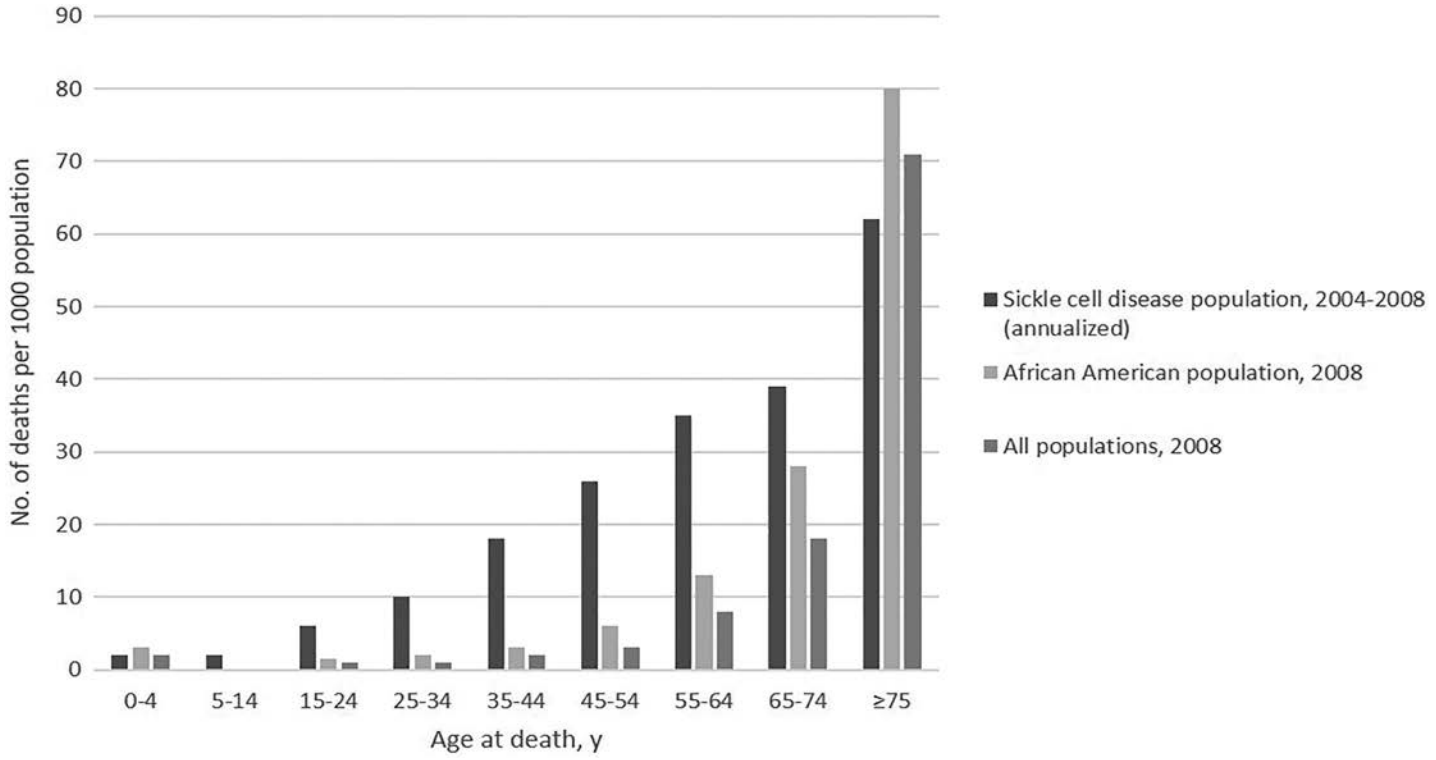
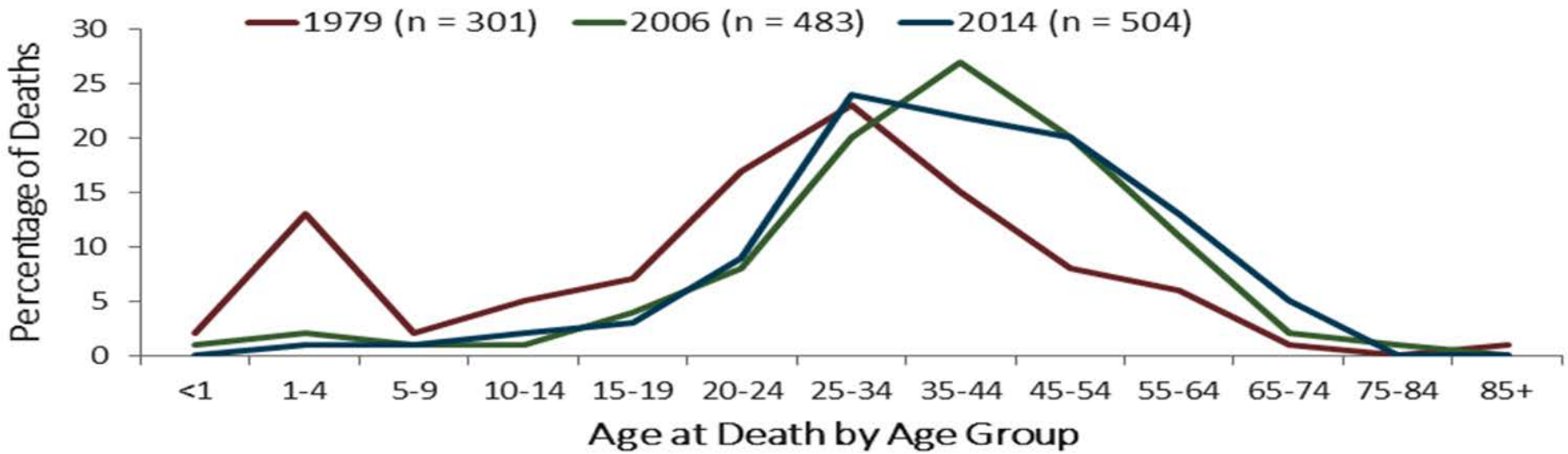


Figure 2. All-cause mortality rates for sickle cell disease identified through population-based surveillance (2004-2008), the African American population (2008), and all populations (2008), in California and Georgia. Source: Paulukonis et al.6

Reducing Health Care Disparities in Sickle Cell Disease: A Review. Le et al. Public Health Reports 2019, Vol. 134(6) 599-607

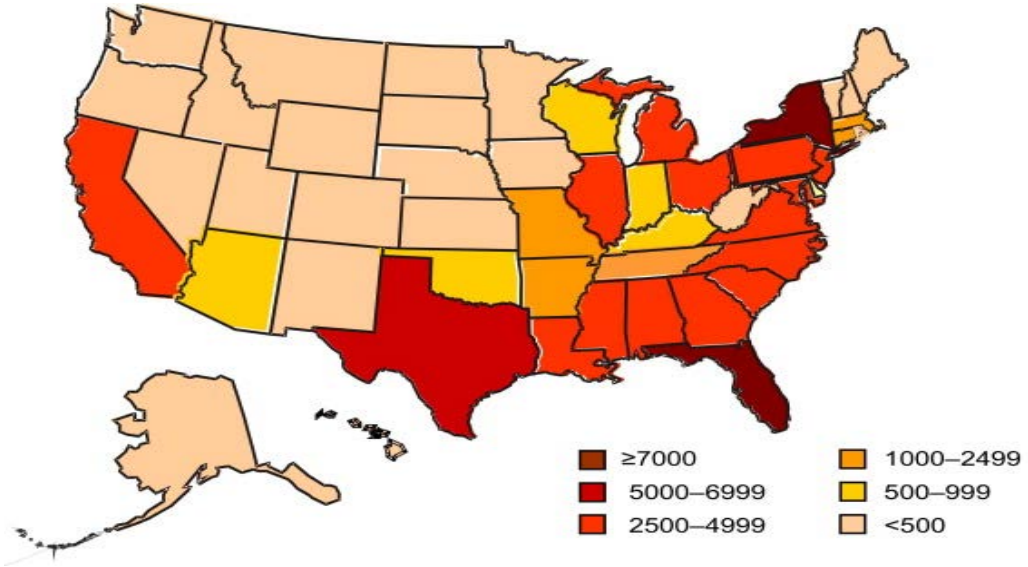
SCD and Mortality in the United States

- Childhood survival is 96% to 98% for all genotypes
- In 2014, most deaths (66%) occurred at ages 25 to 54 years
- More recent surveillance data from Georgia and California showed mean age at death was 43 years for women, 41 years for men

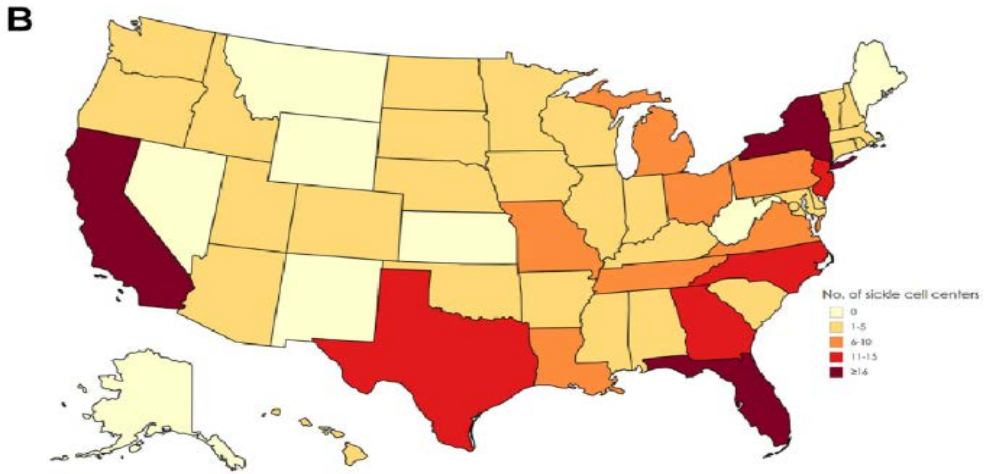


Quinn CT, et al. *Blood*. 2010;115:3447.
Paulukonis ST, et al. *Public Health Reports*. 2016;131:367-375.
Hassell KL, et al. *Am J Prev Med*. 2010;38:S512-S521.

Disparities in Access to Care for SCD



Population Estimates of Sickle Cell Disease in the U.S.



Number of sickle cell disease treatment centers per state, 2017

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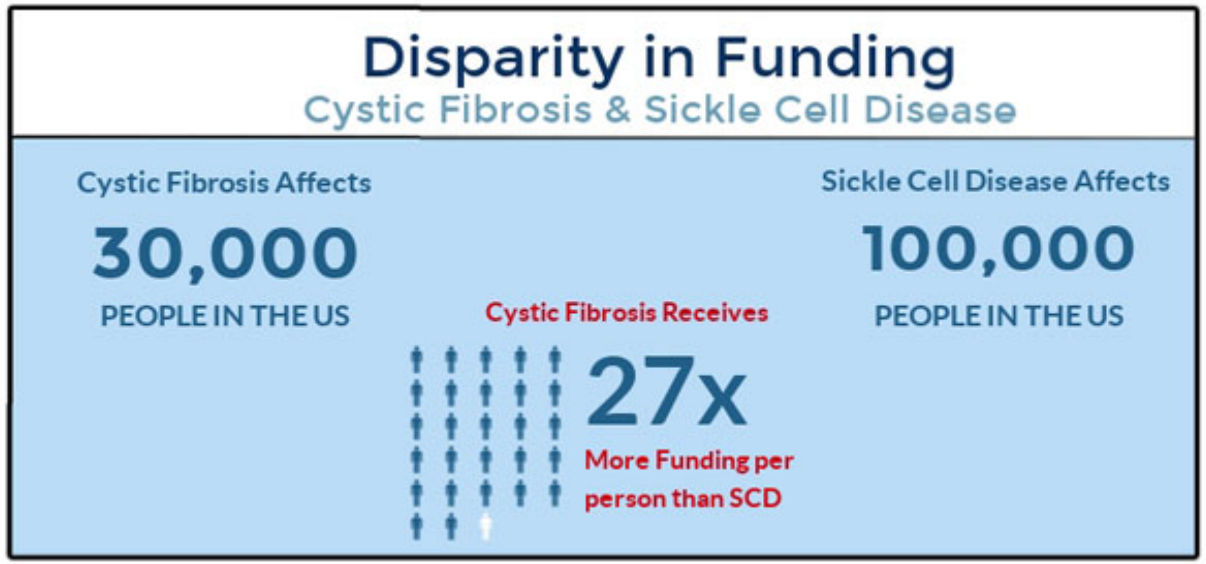
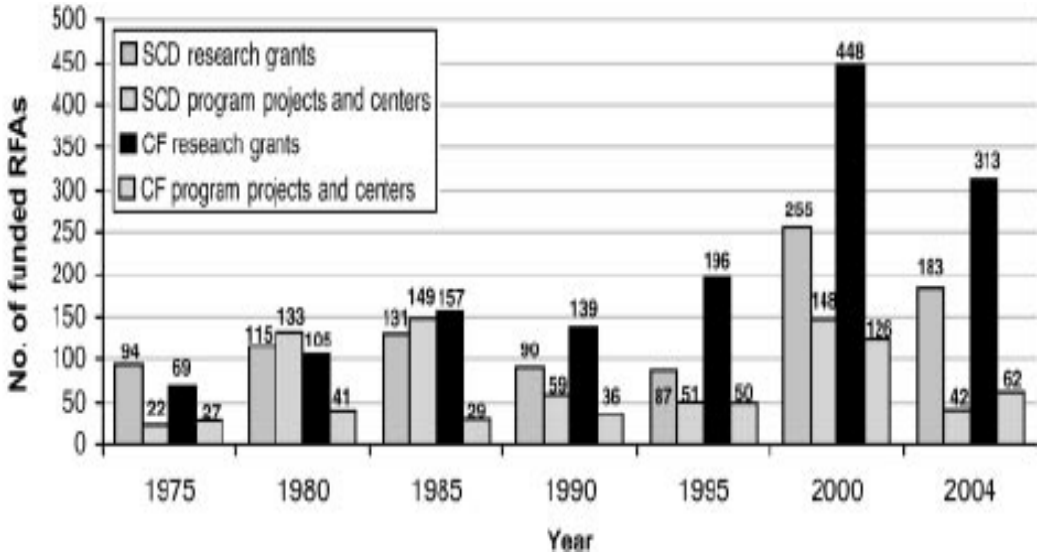
CURE
SICKLE
CELL.
The time has come.
Together we can cure
sickle cell disease.
#CuringSCD
NIH
National Heart, Lung,
and Blood Institute

Lack of Funding For SCD research





Number of federal government requests for applications funded for sickle cell disease and cystic fibrosis: 1975–2004.

Clinical Ways to Mitigate Sickle Cell Disease

- Immunizations
- Medicine to take to prevent infections and other diseases
  - Penicillin – antibiotic prophylaxis
  - Folic Acid – help make red blood cells
- Blood transfusion
- Limited therapeutics
  - Hydroxyurea
  - Crizanlizumab
  - Voxelotor
  - L-Glutamine
- Bone marrow transplantation
- Research Area: Gene Therapy

Blood Transfusion

- Red Blood Cell transfusion occurs from one person to another.
- Transfusing normal red blood cells can help a person living with sickle cell disease (SCD) deliver oxygen to the body and unblock vessels.
- Blood donations are crucial for keeping a strong blood supply for persons with SCD.
- Blood transfusions are needed in SCD for stroke prevention, during the perioperative period to prevent complications during/after surgery
- Pain management when accompanied by major organ damage or significant decline in hemoglobin below patient's baseline

Critical Blood Shortage

- Blood banks across the country are facing critical shortages, and at hospitals, type O blood is often in short supply and high demand.
- O positive is the most common blood type
- O negative is considered the universal blood donor—it's what doctors reach for in an emergency when a patient's blood type is unknown

Fifty-one percent of African Americans and 57% of Hispanics have type O blood, in comparison to 45% of whites.

AFRICAN-AMERICAN BLOOD DONATIONS ARE IMPORTANT

Blood is blood, right?

No.

Blood from an African-American donor to an African-American recipient is more likely to match.

Some African Americans have rare blood types that are unique to the black community like U-negative and Duffy-negative; this only emphasizes the urgent need for more black donors.

This can mean fewer health problems for the recipient. Many recipients, such as those with sickle cell, struggle to find matches that are genetically similar and have to wait until a suitable donor can be found.

Blood Donations Needed Among **African Americans**

African American individuals make up 13% of the U.S. population, but **less than 3%** of blood donors.

Bone Marrow Transplant

- Bone marrow transplantation (BMT) is currently the only known approved cure for sickle cell disease (SCD), but it is not for everyone.

- Benefits of BMT are that it:
  - replaces the abnormal stem cells residing in bone marrow with healthy cells.
  - Currently the only approved cure for SCD.

- While most individuals with SCD do well after BMT, there are possible life-threatening risks and considerations.
  - BMT requires a healthy brother or sister of the individual receiving the transplant to serve as a bone marrow donor.
  - Transplants may not be always effective, and more research is being done to support these efforts.

Sickle Cell Disease and Stigma

Stigma is a mark of disgrace or shame placed on an individual due to a circumstance or quality.

Lack of Understanding of SCD leads to Stigma

- Unfortunately, due to people not understanding sickle cell disease (SCD), stigmatizing beliefs can grow around the disease.
- Patients are viewed as drug seekers, often denied appropriate, timely medical care
    - Leads to the perpetuation of health care disparities and inequities

- People with SCD have painful episodes that are hard to describe, and they often appear to look “normal” thus creating disbelief of the severity of their condition

- People with SCD suffer from fatigue that other people may believe is just laziness.
- These stigmatizing beliefs can lead to bullying, marriage rejection, and exclusion from the community for individuals living with SCD.

- Nevertheless, there are ways to help.

Sickle Cell and the Community

- Family
  - Learn about the disease to help family members manage SCD (hydration, nutritional diet, temperature regulation, emotional support/reduce stress, educational support)

- School

Children with sickle cell disease (SCD) may:

- Miss school due to complications from SCD
- Need to drink water throughout the day
- Be tired in class and need occasional rests
- Have difficulties doing activities due to fatigue or pain
- May need to limit rigorous play

It is important for all teachers and school administrators to provide accomodations to learning and school activities for children with SCD.

Religious Communities

- Religion has been identified by individuals with sickle cell disease (SCD) as an important factor in coping with stress and supporting their quality of life.
- Religious leaders can play a role in raising awareness on SCD.
- Religious leaders can show support to those living with SCD and ensure stigmatizing beliefs are confronted in congregations.

- While vital sources of spiritual strength, religious leaders should not prescribe care for individuals with SCD.
- All treatment should be under the guidance of health care professionals.

- Couples looking to marry should be counseled on genetic testing and potential options depending on sickle cell status – by religious leaders and medical professionals.

Community Organizations

- Communities, towns, cities, and countries have a part to play in supporting persons living with sickle cell disease (SCD).

- Some examples include:
- Raising awareness about SCD
- Reducing the stigma around SCD by preventing the spread of misinformation
- Supporting families of persons living with SCD
- Advocating for support from governments for increased access to testing, including universal newborn screening, and strengthened access to healthcare for those living with SCD.

What Can You Do To Help Raise Sickle Cell Awareness ?

- Partner with local SCD community-based organizations

- Donate Blood
- Donate Bone Marrow-Register with Be The Match
- Get Sickle Cell Trait Tested

American Red Cross

BTSCC
Breaking The Sickle Cell Cycle Foundation Inc.

BLOOD ASSURANCE

What's In Your GENES?

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www.btsscycle.org

2nd Annual Sickle Cell Warrior Walk/Run 5K

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Sickle Cell Math Is Brutally Simple, but Not Widely Taught

An inexpensive blood test can warn couples if they face one in four odds of having a baby with the disease. No one ever told Lametra Scott and Rickey Buggs about it.

Dr. Scott encourages her son, Rickey, who has sickle cell disease, to take each day as it comes. Morgan Hornsby for The New York Times