Calling for Sickle-Cell Trait Research, Surveillance, and Public Education and Awareness (H Res 539, 115th Congress)

Brief Authors
Hannah Brown, PhD Candidate

Brief Editors
Allison Roder, PhD, Andrew Pericak, MEM

Sponsor / Author
Representative Barbara Lee (D-CA-13)

Co-Sponsors
Multiple

Last Action
Referred to House Subcommittee

Date of Last Action
Sep 29 2017

Congressional Session
115th Congress

Date Introduced
09/26/2017

Publication Date
Aug 16 2018

Originating Entity
House of Representatives
The Policy

What it does

Encourages increased awareness and research support for sickle-cell disease and promotes standardized strategies for sharing education, screening results, and counseling.

Synopsis

Calling for Sickle-Cell Trait Research, Surveillance, and Public Education and Awareness (H Res 539 [12]) aims to improve the overall awareness of sickle-cell disease (SCD). The nonbinding resolution recommends implementation of standardized protocols both for raising awareness of SCD and for dissemination of information, including screening results and counseling, to those carrying or affected by the disease.

This resolution specifically:

- Recognizes the challenges of treating health outcomes associated with SCD;
- Encourages the medical community to work with states’ newborn screening programs [13] to standardize the dissemination of screening results, education, and counseling regarding SCD;
- Urges the Department of Human and Health Services (HHS) to develop a public awareness campaign that promotes the importance of knowing one’s sickle-cell trait status no matter one’s racial or ethnic identity;
- Calls on HHS to expand access for screening and counseling for individuals with the sickle-cell trait; and
- Supports research concerning outcomes and implications SCD.

Context

All newborns in the US are currently required to be tested for SCD as part of the newborn screening program [13] within each state, district, or territory. This test is done through a heel prick that collects a small blood sample on a special paper that can then be sent to a lab to detect the type of hemoglobin present in the sample. If a baby is positive for SCD, health providers are instructed [14] to follow up with the family to make sure the parents are informed of the results. The child is then retested to confirm the diagnosis. These tests also inform the parents as to whether or not the child carries the SCT trait. Health providers are further encouraged to recommend counseling to these families and children.

It is also possible to diagnose SCD and SCT prenatally [15] (i.e., before birth.) This diagnosis is done by taking a sample from the amniotic fluid (the liquid in the sac carrying the growing embryo) as early as eight to ten weeks into pregnancy. This diagnosis could also be done by taking tissue from the placenta, the organ that connects the umbilical cord to the womb.

The Science

Science Synopsis

According to the National Institutes of Health’s National Heart, Lung, and Blood Institute [16], sickle-cell disease (SCD) encompasses a group of genetic disorders affecting red blood cells (RBCs) in which the
individual has abnormal or sickle-shaped RBCs that cannot efficiently transport oxygen throughout the body.

All tissues in the body need a steady supply of oxygen to function correctly. In order to reach all parts of the body, oxygen-carrying RBCs need to squeeze in and out of tight blood vessels. Hemoglobin [17], the protein in RBCs that carries and delivers oxygen, typically unloads its oxygen in various body tissues and then travels back into the RBCs to get reloaded with oxygen in the lungs.

Red blood cells normally have a flexible donut shape which allows them to travel through small blood vessels. But sickled hemoglobin form stiff rods inside these cells resulting in their deformation. Thus, passage through the circulatory system is restricted, resulting in reduction of oxygen delivery throughout the body.

**The Mayo Clinic** [18] outlines the following common symptoms of SCD: painful swelling of hands and feet, frequent infections due to damage to the spleen (the organ that fights infection), yellow tint to the skin and eyes (jaundice), fever, and abdominal swelling. Later in life, those affected by SCD are at risk for stroke, hypertension, blindness, ulcers, and gallstones.

Sickle cells also tend to burst and die more frequently than normal cells, leading to a reduction in RBC count, indicative of a condition called hemolytic anemia [19]. Symptoms of hemolytic anemia include severe fatigue, stress on many organs (such as the lungs, heart and kidneys,) and other severe complications involving the lungs, spleen, eyes, joints, and skin.

Sickle-cell disease is not contagious, meaning it does not spread laterally from person to person as does the common cold. Rather, SCD is inherited, meaning the genes causing it are passed from parent to offspring. Sickle-cell genes are **autosomal recessive** [20], meaning that you must inherit a sickle-cell allele from each parent in order to develop the most severe form of SCD, sickle cell anemia. However, inheritance of one sickle cell allele [21] (from only one parent) results in carrier status, meaning there is a 50% chance of the SCD trait being passed to the next generation.

Additionally, one copy of the SCD allele is sufficient to cause the body to produce some abnormal hemoglobin along with normal hemoglobin. This is referred to as **sickle-cell trait** [22] (SCT). People with SCT usually do not present symptoms of a disease, but may be at increased risk for complications in a variety of extreme environmental conditions such as high atmospheric pressure, low oxygen levels, and high altitude. Athletes with SCT are especially at risk when undergoing training and conditioning exercises under severe conditions due to an increased risk for heat stroke and muscle breakdown.

Sickle-cell disease and sickle-cell trait can both be diagnosed by a blood test. According to the **Centers for Disease Control and Prevention** [22] and the **Sickle Cell Disease Association of America** [23], approximately 70,000 – 100,000 people are affected by SCD in the United States, most commonly African American and Hispanic American individuals. Approximately 7% of African American babies have SCT.

Sickle-cell disease affects people throughout their lives; the only current cure is **hematopoietic stem cell** [24] transplantation, a costly treatment that depends on the availability of donors who are a close enough
genetic match to a given patient. However, if the disease is caught early enough, there are a number of
treatments available that will extend life expectancy and improve quality of life. The implementation of
these treatments has increased life expectancy [25] from fourteen years in 1973 to approximately 50
years today.

Recent research has looked at the possibility of a vaccine that protects against pneumococcal disease to
decrease mortality rates among children. One study [26], published in 2009, found that the introduction of
the vaccine led to a 42% decrease in mortalities among African American babies under four years old born
with SCD. Another study [27] found similar results in regards to children under four, but did not find a
significant decrease in mortality rates for children between the ages of four and fourteen. The researchers
of this latter study concluded that more research needs to be done in order to develop more effective
treatment approaches, such as hydroxyurea [28], for older children to significantly decrease mortality
rates in this age demographic.

Relevant Experts

Marilyn Jo Telen, MD [29] is a Professor of Medicine, an Associate Professor of Pathology, and a member
of the Duke Cancer Institute at Duke University. Her research and practice to date has been focused on the
molecular genetics and biochemistry of blood antigens and SCD. Dr. Telen is also the Director of Duke’s
Comprehensive Sickle Cell Center. Her work and publications focus on the composition of red blood cell
membranes and she is also involved in a large study looking for genetic predisposition to development of
SCD.

Relevant publications:

- Kanter, Julie, Marilyn J. Telen, Carolyn Hoppe, Christopher L. Roberts, Jason S. Kim, and Xiaoxi Yang.
  https://doi.org/10.1186/s12916-015-0473-6 [30].
- Elmariah, Hany, Melanie E. Garrett, Laura M. De Castro, Jude C. Jonassaint, Kenneth I. Ataga, James R. Eckman,
  Allison E. Ashley-Koch, and Marilyn J. Telen. 2014. “Factors Associated with Survival in a Contemporary Adult
  https://doi.org/10.1002/ajh.23683 [31].

John J. Strouse, MD [32] is an instructor in the Department of Medicine at Duke University. Dr. Strouse’s
work is centered around pediatric hematology and developing new strategies and therapies for treating
people with SCD.

Relevant publications:

- Stewart, Rosalyn W., Lauren N. Whiteman, John J. Strouse, C. Patrick Carroll, and Sophie Lanzkron.
  2016. "Improving Inpatient Care for Individuals with Sickle Cell Disease Using the Project ECHO
- Noronha, Suzie A., S. Christy Sadreameli, and John J. Strouse. 2016. "Management of Sickle Cell Disease in
Nirmish Ramesh Shah, MD [37] is an Assistant Professor in the Department of Medicine and Pediatrics at Duke University. Dr. Shah’s work focuses on understanding the biology of SCD as well as pain management, symptom analysis, and hospitalization strategies for those affected by this disease.

The diagnosis of sickle trait is greatly under-appreciated for its implications towards future generations. Due to its implications, a concerted effort is needed to assure the diagnosis of sickle cell trait follows patients as they grow up and move from pediatrics to adult medical care.

Relevant publications:


Jennifer Rothman, MD [41] is an associate professor of Pediatrics and an Assistant Professor in Medicine at Duke University. Dr. Rothman’s research and work centers around the development of SCD, bone marrow failure syndromes, and general hematology.

Relevant publications:


### The Debate

#### Endorsements & Opposition

At present, there have not been any publicly reported endorsements of or opposition to this resolution.

#### Status

H Res 539 was introduced in the House on September 26, 2017 and referred to the House Committee on Energy and Commerce [49]. On September 29, 2017, it was referred to the Subcommittee on Health [50].

#### Recommended Citation


#### License Info

[Creative Commons Attribution-ShareAlike 4.0 International License](https://creativecommons.org/licenses/by-sa/4.0/) [52]. Please distribute widely but give credit to [Duke SciPol](https://scipol.duke.edu), linking back to this page if possible.

#### Related Tags

sickle cell disease, [53] Newborn Screening, [54] newborn, [55] sickle cell trait [56]


**Links**

[1] [https://scipol.duke.edu/taxonomy/term/9](https://scipol.duke.edu/taxonomy/term/9)
[3] [https://scipol.duke.edu/engagements/9341](https://scipol.duke.edu/engagements/9341)
[5] [https://scipol.duke.edu/content-authors/hannah-brown-phd-candidate](https://scipol.duke.edu/content-authors/hannah-brown-phd-candidate)
[6] [https://scipol.duke.edu/content-authors/allison-roder-phd-0](https://scipol.duke.edu/content-authors/allison-roder-phd-0)
[7] [https://scipol.duke.edu/content-authors/andrew-pericak-mem](https://scipol.duke.edu/content-authors/andrew-pericak-mem)
[8] [https://scipol.duke.edu/scitalk/science-settled](https://scipol.duke.edu/scitalk/science-settled)