
Genetic Services Policy Project

Sickle Cell Disease

What is Sickle Cell Disease (SCD)?

- SCD is an autosomal recessive inherited blood disorder that affects red blood cells.
- SCD presents in varying degrees of severity with multi-system manifestations.
- Sickle cell crisis can cause both acute (5-7 days) and chronic (weeks to months) pain syndromes.
- Other symptoms include: painful swelling of the hands and feet, pallor, jaundice, severe anemia with splenic enlargement, respiratory symptoms, acute chest syndrome, and neurological changes.
- With disease management, most individuals with SCD live beyond 40 years of age.

What causes SCD?

- SCD results when a person has two copies of a gene for hemoglobin S (Hb SS), which causes red blood cells to contain an abnormal type of hemoglobin.
- SCD also encompasses disorders combining Hb S with another abnormal hemoglobin: hemoglobin C (Hb SC), sickle β -thalassemia (Hb S β ⁺-thalassemia and Hb S β ⁰-thalassemia), D-Punjab, and O-Arab.

What does it mean to be a carrier of sickle cell?

- Sickle cell carriers have one copy of Hb S, and are almost always asymptomatic.
- Life expectancy for carriers is normal.

What is the carrier frequency for the Hb SS gene?

- Carrier frequency (Hb S) varies by ethnicity:
 - 1:14 in African Americans
 - 1:176 in Native Americans
 - 1:183 in Hispanics
 - 1:360 in Middle Eastern groups
 - 1:625 in Caucasians not of Middle Eastern origin
 - 1:1336 in Asians

How is SCD detected?

- Newborn screening for hemoglobinopathies typically identifies individuals with SCD.
- Historically some confusion between carrier status and the presence of SCD.

What are standard treatments and therapies for SCD?

- Life-long comprehensive care is required to minimize morbidity and reduce early mortality.
- Some individuals require extensive therapies and hospitalization for the specific symptoms of sickle cell disease.
- Sickle cell crises are often managed with drug therapy.
- Coping mechanisms, pain management, and cohesive family units help to prevent psychological instability and the development of a chronic pain syndrome.

- Newer treatments, like hydroxyurea, help reduce the frequency of severe pain, acute chest syndrome and the need for blood transfusions in adults.

What are costs associated with SCD?

- Charges for chronic transfusion for stroke prevention range from \$9,828 to \$50,852 per patient per year.
- Estimated average direct costs per hospitalization were \$6,300 for the years 1989 through 1993, or \$475.2 M for 75,000 hospitalizations.
- In 2004, there were 20,271 hospital discharges for children with sickle cell disease and vaso-occlusive crises (VOC) with an average length of stay of 4.4 days.
- Nearly two-thirds of SCD patients are covered by Medicare or Medicaid.

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