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 ß-thalassemia (Hb Sß±-thalassemia and Hb Sß°-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.

References

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