Sickle Cell Disease and Trait

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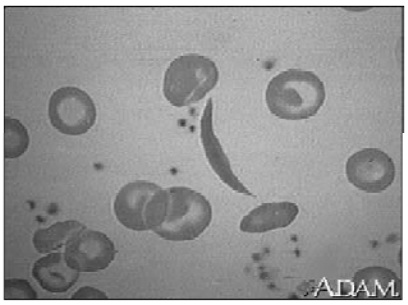
Sickle Cell Disease



What is Sickle Cell Disease?

- Sickle cell disease (SCD) is a group of inherited red blood cell disorders that affect hemoglobin-
  - The molecule in red blood cells that delivers oxygen to cells throughout the body

What is Sickle Cell Disease?

- People with this disorder have atypical hemoglobin molecules called hemoglobin S
  - Which can distort red blood cells into a sickle, or crescent, shape
- It is a condition in which the red blood cells can take on a curved shape, like a sickle

What is Sickle Cell Disease?

- Normal red blood cells are round, soft discs
  - Like doughnuts without holes
- 2.5 million Americans have the genetic trait for the disease
- 70,000 people in the United States have sickle cell disease
- About 1,000 children are born with sickle cell disease in this country each year

What is Sickle Cell Disease?

- A person with sickle cell disease has one different substance in the way it makes hemoglobin
- This substance is the amino acid valine
  - In one spot where there should be glutamic acid

What is Sickle Cell Disease?

- This one change causes the chemical to form long strings when it lets loose of its oxygen
- This change causes the red cell to become deformed into a "sickle" shape

Different Types of Sickle Cell Disease

- There are three common types of sickle cell disease in the United States
  - Hemoglobin SS (sickle cell anemia)
  - Hemoglobin SC disease
  - Hemoglobin sickle beta-thalassemia

Normal vs. Sickle Hemoglobin

- Normal

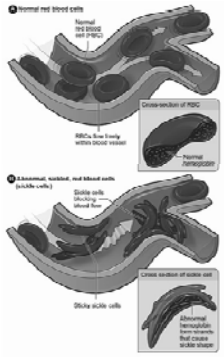
  - Disc-shaped
  - Soft (like a bag of jelly)
  - Easily flow through small blood vessels
  - Lives for 120 days

Sickle

- Sickle

  - Sickle-shaped
  - Hard (like a piece of wood)
  - Often get stuck in small blood vessels
  - Lives for 20 days or less

Cross Section



Cross Section

- Blockage of the small vessels by the sickled cells causes painful crises
  - Trouble breathing and damage to organs

Affected Populations

- Sickle Cell Disease is found in:
  - Africans
  - Turks
  - Greeks
  - Saudi Arabians
  - Egyptians
  - Iranians

Affected Populations

  - Italians
  - Latin Americans
  - Asiatic Indians

In The United States

- Sickle cell disease is present in 1 out of 400 African Americans in the United States
- It is the most common genetic disease in this country
- All new born babies should be tested at birth for sickle cell disease, so prevention can be started right away
- A simple blood test can be done from the baby's blood

Newborn Screening

- Most states now perform the sickle cell test when babies are born
- The simple blood test will detect sickle cell disease or sickle cell trait
- Other types of traits that may be discovered include:
  - Hemoglobin C trait
  - Hemoglobin E trait

Newborn Screening

  - Hemoglobin Barts
    - Which indicates an alpha thalassemia trait
  - Beta thalassemia trait

Medical Manifestations of Sickle Cell Disease

- Medical emergencies
  - Acute chest syndrome
  - Infection
  - Acute bone marrow necrosis

Stroke

- Disorders of the auditory system
  - Hearing loss
  - Auditory perceptual disorders
  - Impaired auditory comprehension
  - Disorders of speech and language
    - Language development at a slower rate

Stroke

- Receptive and expressive language disorders
- Word finding difficulty
- Disorders of literacy
  - Reading and writing difficulty
  - Decreased academic performance

Medical Manifestations of Sickle Cell Disease

- Splenic sequestration crisis
  - Children should be seen as speedily as possible in the emergency room
  - Circulatory collapse and death can occur in less than thirty minutes

Medical Manifestations of Sickle Cell Disease

- Aplastic crisis
- Hepatic sequestration crisis
- Priapism

Sickle Cell Trait

What is Sickle Cell Trait?

- Clinical syndromes associated with sickle cell trait
- Education and counseling
- Recommendations
- Case studies:

What is Sickle Cell Trait?

- Terrell Owens
- Devaughn Darling
- Korey Stinger
- Rashidi Wheeler
- Eraste Autin

What is Sickle Cell Trait?

- Sickle cell trait is a person who carries one sickle hemoglobin producing gene inherited from their parents and one normal hemoglobin gene
- Normal hemoglobin is called type A
- Sickle hemoglobin called S

What is Sickle Cell Trait?

- Sickle cell trait is the presence of hemoglobin AS on the hemoglobin electrophoresis
- This will NOT cause sickle cell disease
- Other hemoglobin traits common in the United States are AC and AE traits

Clinical Syndromes Associated with SCT

- Hyphema-vision abnormalities
  - Bleeding in the front of the eye
- Renal and urinary track infection
- Complications of strenuous exercise:
  - Risk factors for exercise-related death of young adults with SCT include:

Clinical Syndromes Associated with SCT

- Environmental heat stress during the preceding 24 hours
- SCT should not become overheated or dehydrated
- Frequent rest breaks and hydration are encouraged to prevent exercise-related deaths
- Exercise is encouraged with the preceding cautions in mind:

Clinical Syndromes Associated with SCT

- Splenic infarction
  - May occur with hypoxemia from systemic disease or from exercise at sea level or at high altitude
  - Associated with flights in unpressurized aircraft at 15,000 feet or more but may occur rarely at mountain altitudes higher than 6,000 feet above sea level

Clinical Syndromes Associated with SCT

- Individuals who have SCT do not have vaso-occlusive symptoms under physiologic conditions and have a normal life expectancy
- The inheritance of SCT should have no impact on career choices or lifestyle

Clinical Syndromes Associated with SCT

- SCT is found in 8 percent of African Americans
  - Also prevalent in persons of Mediterranean, Middle Eastern, Indian, Caribbean, and Central and South American descent

Clinical Syndromes Associated with SCT

- Nevertheless, numerous individuals with SCT have participated successfully in long-distance races in the Cameroon and in high-altitude sports, including the Olympics in Mexico City
- Caution should be taken for deep sea diving

Education and Genetic Counseling

- All persons with SCT should be educated about:
  - The inheritance of SCD
  - The availability of partner testing
  - Genetic counseling
  - Prenatal diagnosis

Summary of Risks

- Splenic infarction at high altitude, with exercise, or with hypoxemia
  - Deficient oxygen
- Isothenuria with loss of maximal renal concentrating ability
- Hematuria secondary to renal papillary necrosis
  - Presence of blood cells in the urine

Summary of Risks

- Fatal exertional heat illness with exercise
- Sudden idiopathic death with extreme exercise
- Glaucoma or recurrent hyphema following a first episode of hyphema
  - Vision problems
- Bacteruria in women
  - Bacteria in the urine

Summary of Risks

- Bacteruria or associated with pregnancy
- Renal medullary carcinoma in young people
  - Ages 11 to 39 years
- Early onset of end stage renal disease from autosomal dominant polycystic kidney disease

Kark's Factors Can Help Prevent Sudden Collapse:

- Start gradually
  - Gradual conditioning helps develop better oxygen-carrying abilities
- Avoid exercising in high temperatures
  - When the temperature is above 80 degrees, all exercise should stop

Kark's Factors Can Help Prevent Sudden Collapse:

- Hydrate
  - Water is essential
  - Trainers should monitor fluid
  - In no case should withholding water even be considered
- Avoid repeated high-temperature exposure
- Wear lighter clothing

Kark's Factors Can Help Prevent Sudden Collapse:

  - This helps dissipate body heat
- Do not drink stimulants such as "Red Bull" and do not use ephedrine

SCT and Athletes

- Some people with SCT have been shown to be more likely than those without SCT to experience heat stroke and muscle breakdown when doing intense exercise
  - Such as competitive sports or military training under unfavorable temperatures or conditions
    - Very high or low

SCT and Athletes

- Studies have shown that the chance of this problem can be reduced by avoiding dehydration and getting too hot during training
- People with SCT who participate in competitive or team sports should be careful when doing training or conditioning activities
  - Student athletes

SCT and Athletes

- To prevent illness it is important to:
  - Set your own pace and build your intensity slowly
  - Rest often in between repetitive sets and drills
  - Drink plenty of water before, during and after training and conditioning activities

SCT and Athletes

  - Keep the body temperature cool when exercising in hot and humid temperatures by misting the body with water or going to an air conditioned area during breaks or rest periods
  - Immediately seek medical care when feeling ill

SCT and Athletes

- But with proper precautions, virtually nobody should have to die a sudden death because they have SCT
  - That includes professional and collegiate athletes

SCT and Athletes

- U.S. Armed Forces, including the U.S. Marine Corp, have adopted the Kark Protocols (2001) recommended above
  - Training deaths in sickle cell trait recruits dropped to zero in one facility

Research Promise For The Future

- Bone marrow (stem cell) transplants that replace the patient's blood
- Making apparatus with donor cells from blood relatives have been successfully performed in a limited number of patients

Research Promise For The Future

- The first child to receive an unrelated stem-cell transplant in 1998 was found to be free of sickle cells in March of this year (2001)
- Gene therapy also appears promising for the future

Research Promise For The Future

- Sickle cells genes might be inactivated or inhibited while the gene for normal adult hemoglobin is increasingly expressed
- Until a cure for the disease is found, you have to know whether you carry the gene or not so you can be prepared

Case Studies

- Terrell Owens, NFL all-pro wide receiver
  - Talks about his 20 year old cousin's death from a sickle cell crises
  - His pal, Rohan Sutherland, was recently diagnosed with sickle cell disease

Football Players with SCT

- The death of Florida State University linebacker, Devaughn Darling, in a spring conditioning drill
- Darling had SCT, however ephedrine was found in his body
- He was training in high heat

Football Players with SCT

- The recent deaths of Minnesota Vikings lineman, Korey Stringer, Northwestern University defensive back, Rashidi Wheeler, and University of Florida freshman fullback, Eraste Autin, have posed additional questions
  - All three athletes were training in extremely hot weather

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