Carrier Screening ACT Sheet
Sickle Cell Carrier/Trait

Carrier Screening: Carriers are primarily identified through population screening programs.

Condition Description: Carriers are heterozygous for the sickle cell (Hb S) mutation and are usually asymptomatic.

YOU SHOULD TAKE THE FOLLOWING ACTIONS:

- Confirm screening result by hemoglobin separation by electrophoresis, isoelectric focusing or high performance liquid chromatography (HPLC). DNA studies may be used to confirm the genotype.
- Inform individual of carrier screening result. Provide counseling and education.
- Consult with a genetic counselor or geneticist for additional testing recommendations for the reproductive partner.
- Perform urinalysis annually. Ask about gross hematuria at every visit.

Clinical Considerations: Carriers do not have anemia. For the large majority of carriers, no restrictions of physical activity are indicated. In hypoxic conditions (strenuous exercise, dehydration, high altitude), sickling may occur. Rarely, this may lead to splenic infarction, hematuria secondary to renal papillary necrosis, or exertional heat illness (rhabdomyolysis, heat stroke, or renal failure). High-performance athletes should be made aware of non-specific early warning signs of heat related illness.

Sickle cell trait has also been reported in association with renal medullary carcinoma in young people (ages 11 to 39 years) and early onset of end stage renal disease in patients with autosomal dominant polycystic kidney disease.

Reproductive Implications: When an individual is found to be a carrier of the sickle cell mutation, the reproductive partner should be offered carrier screening for mutations in the same gene. If both parents are carriers of the sickle cell mutation (i.e. have sickle cell trait), or if one parent has sickle cell (Hb S) trait and the other parent is a carrier of another sickling mutation (e.g. Hb C trait, beta thalassemia trait or another type of trait), there is a 1 in 4, or 25%, chance in each pregnancy that the offspring will have sickle cell disease.

Additional Information:

Sickle Cell Information Center
Sickle Cell Disease Association of America

Referral (local, state, regional and national):

Testing
Clinical Services
Find Genetic Services

ACMG
American College of Medical Genetics and Genomics
Translating Genes Into Health®

APPENDIX: Resources with Full URL Addresses

Additional Information:

Sickle Cell Information Center
http://www.scinfo.org/

Sickle Cell Disease Association of America
http://www.sicklecelldisease.org/index.cfm?page=chapters

Referral (local, state, regional and national):

Testing
http://www.ncbi.nlm.nih.gov/sites/GeneTests/lab/clinical_disease_id/2775?db=genetests&country=United%20States

www.rbclab.com

Clinical Services
http://www.genetests.org/

Find Genetic Services
http://www.acmg.net/gis

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