American College of Medical Genetics **ACT SHEET**

Newborn Screening ACT Sheet **[FAV]**

Hemoglobin Variant Carrier

Differential Diagnosis: This refers to the presence of any beta hemoglobin variant (V) other than hemoglobin S. This result is different from FAS which is consistent with sickle carrier.

Condition Description: Generally benign genetic carrier state (trait) characterized by the presence of fetal hemoglobin (F), and hemoglobin A and V. The hemoglobins are listed in order of the amount of hemoglobin present (F>A>V).

YOU SHOULD TAKE THE FOLLOWING ACTIONS:

- Contact family to inform them of the screening result and to reassure them that these individuals usually
 do not have clinical problems.
- Do complete blood count (CBC) and reticulocyte count and repeat at 6 months.
- Order hemoglobin profile analysis (usually performed by electrophoresis).
- Offer family members referral for genetic evaluation and counseling.
- Report findings to newborn screening program.

Diagnostic Evaluation: CBC and reticulocyte count. Hemoglobin separation by electrophoresis, isoelectric focusing, or high performance liquid chromatography (HPLC), shows FAV. DNA studies may be used to confirm genotype.

Clinical Considerations: Infants are usually normal at birth. Carriers of certain Hb variants (e.g. HbD-Los Angeles, Hb O-Arab) are at risk for having children affected by sickle cell disease. Counseling at reproductive age should be considered. Consultation with an expert in hemoglobin disorders should be considered if CBC or reticulocyte count are abnormal.

Additional Information:

Grady Comprehensive Sickle Cell Center

Management of Sickle Cell Disease

Sickle Cell Disease in Children and Adolescents: Diagnosis, Guidelines for Comprehensive Care, and

Care Paths and Protocols for Management of Acute and Chronic Complications

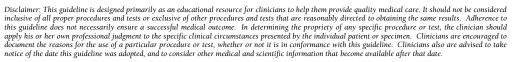
American Academy of Pediatrics

Sickle Cell Disease Association of America

Referral (local, state, regional and national):

Testing
Clinical Services

<u>Comprehensive Sickle Cell Center Directory</u>
<u>Sickle Cell Information Center</u>
<u>Find Genetic Services</u>





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State Resource si Name	te (insert state newborn screening program website information)
URL	
Comments	
Comments	
Local Resource S	ite (insert local and regional newborn screening website information)
Name	
URL	
Comments	
APPENDIX: Resourc	es with Full URL Addresses
Additional Information Grady Comprehon http://www.scinf	ensive Sickle Cell Center
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	l Therapy of Sickle Cell Disease .nih.gov/health/prof/blood/sickle/index.htm
Protocols for Ma	ase in Children and Adolescents: Diagnosis, Guidelines for Comprehensive Care, and Care Paths and nagement of Acute and Chronic Complications. state.tx.us/newborn/pdf/sedona02.pdf
American Acade http://pediatrics.a	my of Pediatrics appublications.org/cgi/content/full/109/3/526
Sickle Cell Disea http://www.sickle	ase Association of America ecelldisease.org/
Defensed (least state	regional and national).
Testing	regional and national):

Clinical Services

Comprehensive Sickle Cell Center Directory

http://www.scinfo.org/index.php?option=com_content&view=article&id=197&Itemid=34

Sickle Cell Information Center

http://www.scinfo.org/

Find Genetic Services

http://www.acmg.net/GIS/Disclaimer.aspx

Disclaimer: This guideline is designed primarily as an educational resource for clinicians to help them provide quality medical care. It should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. Adherence to this guideline does not necessarily ensure a successful medical outcome. In determining the propriety of any specific procedure or test, the clinician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this guideline. Clinicians also are advised to take notice of the date this guideline was adopted, and to consider other medical and scientific information that become available after that date.

