

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

[Comprehensive Sickle Cell Center Directory](#)

[Sickle Cell Information Center](#)

[Find Genetic Services](#)

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.

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LOCAL RESOURCES: Insert State newborn screening program web site links

State Resource site (insert state newborn screening program website information)

Name	<input type="text"/>
URL	<input type="text"/>
Comments	<input type="text"/>

Local Resource Site (insert local and regional newborn screening website information)

Name	<input type="text"/>
URL	<input type="text"/>
Comments	<input type="text"/>

APPENDIX: Resources with Full URL Addresses

Additional Information:

Grady Comprehensive Sickle Cell Center

<http://www.scinfo.org/>

Management and Therapy of Sickle Cell Disease

<http://www.nhlbi.nih.gov/health/prof/blood/sickle/index.htm>

Sickle Cell Disease in Children and Adolescents: Diagnosis, Guidelines for Comprehensive Care, and Care Paths and Protocols for Management of Acute and Chronic Complications.

<http://www.dshs.state.tx.us/newborn/pdf/sedona02.pdf>

American Academy of Pediatrics

<http://pediatrics.aappublications.org/cgi/content/full/109/3/526>

Sickle Cell Disease Association of America

<http://www.sicklecelldisease.org/>

Referral (local, state, regional and national):

Testing

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>

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