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

ACT Sheet

Newborn Screening ACT Sheet

[FAV]

Hemoglobin Variant Carrier

Differential Diagnosis: Any hemoglobin variant (V) that does not migrate with hemoglobin S, E, C or is not specifically identified.

Condition Description: This group of hemoglobin variants are inherited types of red blood cell disorders characterized by abnormal hemoglobin production. They are due to genetic changes in either the alpha or the beta globin chain and refer to the presence of normal hemoglobin F and A along with any hemoglobin variants (V) other than those specifically identified (e.g. S, E, or C). Most variants are benign, although there are rare exceptions.

You Should Take the Following Actions:

- Inform family of the newborn screening result.
- Ascertain clinical status (most newborns are asymptomatic, however hypoxia, jaundice, scleral icterus, cherry-red or blue skin discoloration may occur).
- Evaluate the newborn (most newborns are asymptomatic; if there are signs of desaturation, jaundice, scleral icterus, or cherry-red or blue skin discoloration, consult with a pediatric hematologist immediately).
- If no clinical signs or symptoms, discuss with pediatric hematologist regarding guidelines on further care.
- Coordinate confirmatory diagnostic testing and management as recommended by specialist.
- Provide family with basic information about the specific identified hemoglobin variant.
- Refer for genetic counseling.

- Report final diagnostic outcome to newborn screening program.

Diagnostic Evaluation: The hemoglobins are listed in order of the amount of hemoglobin present on the newborn screen (F>A>V). Isoelectric focusing, high performance liquid chromatography (HPLC,) or capillary zone electrophoresis): may confirm the newborn screening result. Complete blood count: the CBC and reticulocyte count may or may not demonstrate anemia depending on the variant present. Molecular genetic testing, methemoglobin levels, oxygen dissociation or hemoglobin stability studies: may be required to confirm the diagnosis depending on the variant.

Clinical Considerations: Infants are usually asymptomatic at birth. Consultation with an expert in hemoglobin disorders should be performed if CBC or reticulocyte counts are abnormal. The type of variant identified will determine the prognosis. Carriers of certain Hb variants (e.g., Hb D-Los Angeles, Hb O-Arab) are at risk for having children affected by sickle cell disease. Rarely a variant may alter oxygen affinity or stability in a clinically significant manner. If concerns regarding oxygenation or jaundice, immediately refer to hematology with specific expertise in hemoglobin variant management.

Additional Information:

How to Communicate Newborn Screening Results
Clinicaltrials.gov

Referral (local, state, regional, and national:

Find A Hematologist (Filter by Pediatric Hematology-Oncology)
Find a Genetics Clinic Directory
Genetic Testing Registry

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Local Resources *(Insert Local Website Links)*

State Resource Site (Insert Website Information)

Name	
URL	
Comments	

Local Resource Site (Insert Website Information)

Name	
URL	
Comments	

Appendix *(Resources with Full URL Addresses)*

Additional Information

How to Communicate Newborn Screening Results

- https://bit.ly/NBSResultsHRSA

Clinicaltrials.gov

- https://clinicaltrials.gov/

Referral (local, state, regional and national)

Find A Hematologist (Filter by Pediatric Hematology-Oncology)

- https://www.hematology.org/education/patients/find-a-hematologist

Find a Genetics Clinic Directory

- https://clinics.acmg.net

Genetic Testing Registry

- https://www.ncbi.nlm.nih.gov/gtr/