Suggested Follow-up for Hemoglobinpathies

Possible Causes: Autosomal recessive disorders that affect the red blood cells and result from genetically determined changes in the molecular structure of hemoglobin. Many hemoglobinopathy disorders with varying degrees of severity can be identified. Newborn screening can also identify genetic carriers of some hemoglobinopathy disorders.

Next Steps if Abnormal: Repeat hemoglobinopathy screening on filter paper and send to the DHEC laboratory. Refer to pediatric hematologist. Consider penicillin prophylaxis upon receipt of newborn screening report if the hemoglobin pattern is FS. Further evaluation/testing is necessary to establish diagnosis.

Neonatal Presentation: None.

Emergency Treatment: None.

Standard Treatment: Sickling disorders—Penicillin/antibiotic prophylaxis beginning in infancy and continuing through early childhood. Prompt evaluation/management of acute illness to lessen development of sickling crisis. Pain management strategies. Transfusion may be necessary. Medications to increase production of fetal hemoglobin and lower leukocyte counts may be used.

Advice for Family: Provide basic information about hemoglobinopathies. The handout, *When Baby Needs a Second Test for a Hemoglobinopathy*, may be used for this purpose.

Internet Resources:

http://sickle.bwh.harvard.edu/menu_sickle.html

http://www.scinfo.org/

http://www.genetests.org/query?dz=sickle

http://www.acmg.net/resources/policies/ACT/condition-analyte-links.htm