NEBRASKA
Good Life. Great Mission.
DEPT. OF HEALTH AND HUMAN SERVICES

ACT SHEET FOR POSITIVE NEWBORN SCREENING RESULT (FAV, FVA, FV) HEMOGLOBIN VARIANT (unknown variant on screen)

Meaning of the Screening Result: Newborn Screening results of FAV, FVA, or FV indicate a variant hemoglobin present on screen which the screening test cannot specify. Most variants are of a benign nature, however if a co-existent thalassemia is present, clinical significance must be determined.

YOU SHOULD TAKE THE FOLLOWING ACTIONS:

- **Contact the family** to inform them of the screening result and offer education and counseling.
- **Order confirmatory testing (hemoglobin electrophoresis by IEF and HPLC).**
- Reassure the family that infants typically do not have clinical problems related to the carrier state for most variant hemoglobin types.
- Encourage parents to seek testing for thalassemia and hemoglobin variants followed by genetic counseling.
- **Consider contacting a specialist in hemoglobinopathies for consultation or referral** on diagnostic evaluation and management as needed.
- **Report findings** to Nebraska Newborn Screening Program.

Condition Description: There are hundreds of hemoglobinopathies of a benign nature that typically show as FAV, FVA or FV on the newborn screen.

Clinical Expectations: Variable, depending on specific variant, and co-existent hemoglobins. For most of these, the patient will have no untoward symptoms. However, if a coexistent thalassemia that is clinically significant is present, patients can have variable symptoms from microcytic anemia to severe complications.

Confirmation of Diagnosis: Order a newborn confirmation **hemoglobin electrophoresis by IEF (Isoelectric Focusing) and HPLC (High Performance Liquid Chromatography).** Depending on these results, further testing may be necessary and family studies may be required to definitively diagnose the baby.

Pediatric specialists in hemoglobinopathies are available at Children's Hospital (402)955-3950 & UNMC/Nebraska Med Center (402)559-7257.