

ACT SHEET FOR POSITIVE NEWBORN SCREENING RESULT (FAC) HEMOGLOBIN C TRAIT (Hb AC)

Disease Category: Hemoglobinopathy

Meaning of the Screening Result: Hemoglobin FAC pattern on newborn screen is highly suggestive of Hemoglobin C trait. However, IEF (the test used for screening) does not quantitate results. Without quantitation those newborns with A and C hemoglobins where the C hemoglobin is nearly as much as the A hemoglobin, could be at risk of a thalassemia.

YOU SHOULD TAKE THE FOLLOWING ACTIONS:

- Contact the family to inform them of the screening result and to offer education and counseling.
- Reassure the family that infants do not have clinical problems related to the carrier state for Hemoglobin C.
- Order confirmatory testing (hemoglobin electrophoresis).
- Encourage parents to seek genetic counseling and testing as indicated.
- Report findings to Nebraska Newborn Screening Program.

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

Condition Description: Individuals with Hemoglobin C trait are carriers of the gene for Hemoglobin C.

Clinical Expectations: Prognosis is for a normal life expectancy. Target cells may be seen in the peripheral blood smear. Mild microcytosis may also be noted. Rarely patients may have a borderline low hemoglobin, though the vast majority of the individuals with Hemoglobin C trait have normal levels of hemoglobin. Carriers are at risk for having children affected with Hemoglobin C disease, Hemoglobin S-C disease or Hemoglobin C Beta thalassemia.

Confirmation of diagnosis: Diagnosis is confirmed by hemoglobin electrophoresis and parental or DNA studies as indicated. (However, if on confirmation a thalassemia or co-existent thalassemia is identified, it is important to refer these patients to pediatric hematology for further work-up, to determine clinical significance.)