NEBRASKA
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DEPT. OF HEALTH AND HUMAN SERVICES

ACT SHEET FOR POSITIVE NEWBORN SCREENING RESULT
(FA + Barts, FA + other* + Barts)
Alpha Thalassemia

Disease Category: Hemoglobinopathy

Meaning of the Screening Result: Hemoglobin Bart's on a newborn screen is highly suggestive of Alpha thalassemia – any of 4 types. Alpha thalassemia 2 - silent carrier is a result of a single gene deletion. Alpha thalassemia trait results from loss of two genes. Hemoglobin H disease is a thalassemia resulting from the loss of 3 genes. Hydrops fetalis results from the 4-gene deletion which would be unlikely to be detected on a newborn screen since newborns do not survive more than a few hours.

Other* = S, C, E, D, or V

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).**
- Encourage parents to seek testing for thalassemia and hemoglobin variants followed by genetic counseling.
- Following initial confirmatory testing, **referral to pediatric hematologist** may be indicated for definitive diagnosis.
- **Report findings** to Nebraska Newborn Screening Program.

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

Condition Description and Clinical Expectations: The alpha thalassemias result from the loss of alpha globin genes. There are normally four genes for alpha globin production so that the loss of one to four genes is possible. A single gene deletion causes **alpha thalassemia 2 (silent carrier)** with no clinically detectable problems but may cause small amounts of hemoglobin Bart's to be present in newborn blood samples. **Alpha thalassemia trait (Alpha thalassemia 1)** a two gene deletion causes a mild microcytic anemia, which may resemble iron deficiency anemia. A three gene deletion causes **Hemoglobin H disease**, which is a moderately severe form of thalassemia. A four gene deletion causes **hydrops fetalis** and is usually fatal in utero.

Confirmation of Diagnosis: Presence of Bart's hemoglobin is confirmed by hemoglobin electrophoresis. In addition to the standard newborn hemoglobinopathy confirmation, we recommend that these babies have the following labs drawn at their 6 months well baby check: CBC with retic count, ferritin, and a hemoglobin electrophoresis. The patient with alpha thalassemia - silent carrier should have a normal CBC and electrophoresis. The hemoglobin in patients with alpha thalassemia trait may be slightly below normal to normal, with microcytic red blood cells and an elevated red blood cell number. The ferritin level reflects stored iron reserves. Many subtle hemoglobin defects like alpha thalassemia trait are not reliably diagnosed in the presence of iron deficiency. Documenting that the patient's iron level is normal improves the validity of the work up.

Genetic counseling: Parent studies may be indicated after confirmatory testing of the newborn, to detect the more serious forms of alpha thalassemia. Particularly in those children who have a Southeast Asian heritage, or in multiracial children, family studies may be useful as these ethnic groups have an increased risk for the more serious forms of alpha thalassemia. Documenting this risk status may be useful in genetic counseling or family planning.

If you have additional questions, please call Dr. James Harper of the pediatric hematology clinic at UNMC at (402) 559-7257. Other hematology specialists are available at (402) 955-3950 Dr. David Gnarra at Children's Hospital or (308) 762-2125 Dr. Howard Koch in Alliance.