

## **Hemoglobin Bart's- For Physicians**

As part of routine newborn screening all babies are tested for sickle cell disease and other hemoglobinopathies. Screening of all specimens is done by isoelectric focusing (IEF). Results are then confirmed by IEF and citrate agar electrophoresis.

Hemoglobin Bart's was present in your patient. Hemoglobin Bart's is a common hemoglobin variant that is only detectable during the newborn period. Its presence indicates that one or more of the infant's four alpha globin genes are dysfunctional, which results in alpha thalassemia. The severity of the alpha thalassemia increases as the number of alpha genes that are dysfunctional increases.

### **Possible Alpha Thalassemia Variations:**

#### *One dysfunctional alpha gene: Silent carrier*

No clinical implications

#### *Two dysfunctional alpha genes: Alpha thalassemia trait*

Patient has mild anemia and microcytosis, but condition is benign and requires no treatment

#### *Three dysfunctional alpha genes: Hemoglobin H disease*

This generally results in moderate hemolytic anemia and some degree of splenomegaly. Patients with hemoglobin H disease also have chronic hemolysis and are susceptible to accelerated hemolysis when exposed to certain medications.

#### *Four dysfunctional alpha genes: Fetal Hydrops syndrome*

Severe anemia begins in utero and typically results in fetal demise in third trimester. This disorder is lethal.

### **Follow Up Recommendations:**

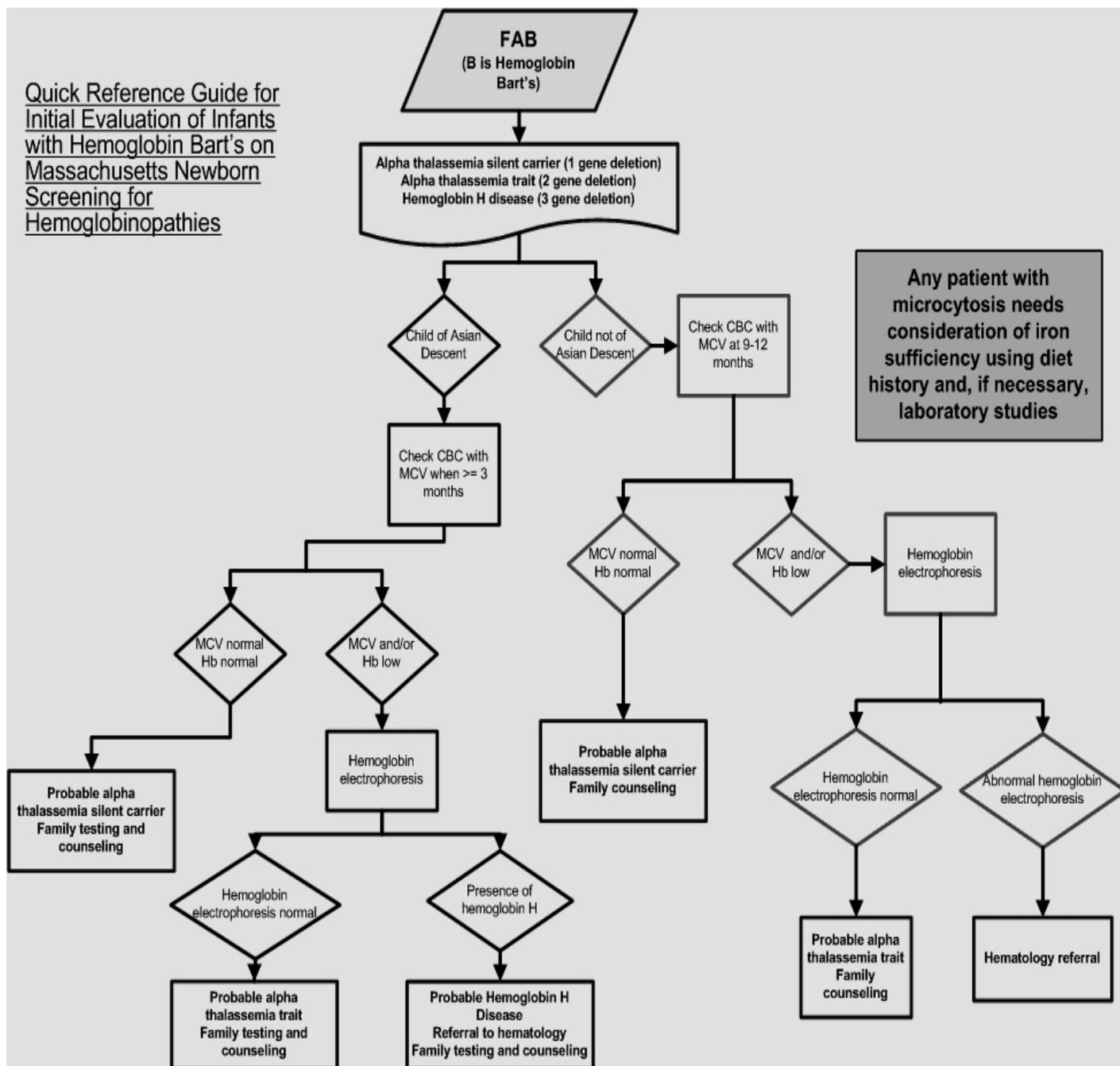
**\*\*Newborn screening is unable to determine the clinical severity. Further studies are necessary. Please see attached flow chart for evaluation recommendations.**

After further evaluations and have been completed and the child's alpha thalassemia status has been determined, the family should be offered genetic counseling for parental testing and to discuss the inheritance of hemoglobinopathies.

If you have any further questions, please contact the New Hampshire Newborn Screening Program at (603) 271-4225.

# New Hampshire Newborn Screening Program

Quick Reference Guide for Initial Evaluation of Infants with Hemoglobin Bart's on Massachusetts Newborn Screening for Hemoglobinopathies



Taken from the “Quick Reference Guide to Results from Massachusetts Newborn Screening Guide for Hemoglobinopathies”