NBS Hb Screen (non-S) by HPLC or IEF

Confirm by alternative method (IEF, HPLC, electrophoresis or DNA studies) and do CBC

[F] Hb β° (Thalassemia Major)

[FE] HbEE or HbΕβ°

[FEA] HbE/β+

[FC] HbCC or HbCβ°

[FC] HbC/β+

[FA] HbE/β+

[FA+Barts Hb]

[FAV]

Refer to specialist in hemoglobin disorders

CBC to differentiate

Refer to specialist in hemoglobin disorders

Refer to specialist in hemoglobin disorders

Quantitative HPLC for Barts hemoglobin

Do CBC

< 25% Hb Barts

> 25% Hb Barts

Anemia for age

Normal

Refer to specialist in hemoglobin disorders

No further testing required

No further testing required

Refer to specialist in hemoglobin disorders

Refer to specialist in hemoglobin disorders

No further testing required

No further testing required

Refer to specialist in hemoglobin disorders

No further testing required

Refer to specialist in hemoglobin disorders

Offer family genetic counseling

Abbreviations/ Key

CBC: Complete Blood Count

F, S, A, E, C, V, and Barts = The hemoglobins seen in neonatal screening.

‡ = Repeat testing at 6 months age is required if genotyping to confirm the newborn screening result is not done.

Actions are shown in shaded boxes; results are in the unshaded boxes.

Disclaimer: This guideline is designed primarily as an educational resource for clinicians to help them provide quality medical care. It should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. Adherence to this guideline does not necessarily ensure a successful medical outcome. In determining the propriety of any specific procedure or test, the clinician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this guideline. Clinicians are also advised to take notice of the date this guideline was adopted, and to consider other medical and scientific information that become available after that date.

© American College of Medical Genetics, 2009 (Funded in part through MCHB/HRSA/HHS grant #U22MC03957)