

ABNORMAL NEWBORN HEMOGLOBIN SCREENING TEST: A GUIDE FOR PARENTS AND PRIMARY CARE PHYSICIANS

Introduction:

Babies are often referred by their primary care doctor to a pediatric hematologist because an “abnormal” hemoglobin has been identified by newborn screening. The Texas Department of State Health Services tests all newborn babies for sickle cell anemia and other potentially serious blood disorders involving hemoglobin. In the process, other hemoglobin abnormalities are sometimes discovered.

Hemoglobin is the substance that carries oxygen in the red blood cells. Normal newborn babies have two forms of hemoglobin, hemoglobin F (fetal hemoglobin) and hemoglobin A (adult hemoglobin), or a so-called “F, A” pattern. Most common abnormal hemoglobins (S, C, E, D, Barts, etc.) are easily diagnosed by the state’s laboratory and are not the subject of this information sheet.

On certain occasions, however, the newborn screening blood test uncovers an unknown abnormal hemoglobin in addition to the normal hemoglobin F and hemoglobin A. This pattern is called “F, A, Other”. It is reported to the pediatrician, with the suggestion that the abnormality be evaluated by a pediatric hematologist.

Your baby’s test shows such an “F, A, Other” pattern. The purpose of this information sheet is to describe why the abnormal hemoglobin is unlikely to be a problem for the baby and whether additional testing is necessary.

A baby inherits his or her hemoglobin genes from the parents. In your baby’s case either the mother or the father probably have the very same abnormal hemoglobin in addition to the normal hemoglobin A. Since most of these hemoglobin abnormalities are of no significance, the parent is generally unaware that they have it.

Although hemoglobin has hundreds of building blocks, many of them are not that important. However, it is common for an abnormality in one of them to occur and be picked up by the newborn screening test, yet cause no ill effects in the baby. This is called a “silent” mutation.

On rare occasions, however, the abnormal hemoglobin can cause problems, such as anemia (a reduction in the blood count, usually due to increased fragility of the red blood cells) or the hemoglobin “holding on” to oxygen either to tightly or to loosely. In rare circumstances, the abnormal hemoglobin causes the iron within it to be chemically altered so as to cause a blue or gray color in the baby. It must be stressed, however, that most of the time the abnormal hemoglobin really causes no problems at all.

Here’s What We Suggest That The Pediatrician Does to Assure That Nothing Serious is Happening:

Medical History & Physical Examination: The first step is to review the baby’s medical history regarding anemia, jaundice (a yellow color to the skin), cyanosis (a blue or gray color to the skin) and symptoms of anemia such as tiredness, poor feeding, and poor weight gain. Usually

there are no such problems. We also suggest inquiring about anemia or other problems with the blood in the family. Questions about the baby's ancestry are also important since certain hemoglobin problems occur in individuals from specific ethnic groups. It should also be assured that a physical examination on the baby shows no abnormalities that could be due to an abnormal hemoglobin.

Blood Tests on the Baby: The next step is to perform a blood count to check for anemia and other problems involving the blood. Most importantly, we suggest a hemoglobin electrophoresis test, ideally after the baby is 3 months of age. This can help determine the kind of hemoglobin abnormality the baby has. Sometimes a specific abnormal hemoglobin can be identified by the laboratory. They are often characterized by a letter of the alphabet or the city where the abnormal hemoglobin was first discovered. Knowing what the hemoglobin is and the results of the blood count will help determine whether the abnormal "other" hemoglobin has any significance (which is rare) or whether it is "silent" and of no importance whatsoever (the usual situation).

Quite often, it is not possible for the laboratory to determine the exact name of the abnormal hemoglobin. The primary goal, however, should simply to be certain that it will cause no problems for the baby. If the family is interested in knowing the name of the hemoglobin (sometimes it will be one never discovered before!), additional testing in special laboratories can be carried out. However, sometimes the family's insurance will not cover these expensive special tests, they are do not recommended routinely.

Testing Parents: Sometimes the parents might wish to be tested since one of them often also has the abnormal hemoglobin. It is frequently reassuring to know that the mother or father has it but is healthy.

The Bottom Line: Most babies with an "F, A, Other" newborn hemoglobin screening test do not have a blood disease, are healthy, and do not require follow-up by a hematologist. Regular blood counts, medicines, special diets or other treatments are unnecessary. If you have questions, please contact the Referral Coordinator at the Center for Cancer and Blood Disorders, and arrangements can be made for a formal hematology consultation.

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