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Department of Health Services



ARNOLD SCHWARZENEGGER
Governor

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Dear Newborn Screening Providers/Stakeholders:

This letter is to inform you of a change in protocol related to follow-up of newborns who received a red blood cell transfusion prior to collection of an adequate newborn screening specimen. Beginning July 1, 2007, **post-transfusion testing for hemoglobinopathies of babies with hemoglobin patterns of A, AF, or FA will no longer be routinely done by the NBS Program.** Instead it will be up to the babies' physicians to request that the testing be performed through the state-funded hemoglobin confirmatory laboratory at Children's Hospital and Research Center, Oakland (CHRCO). As before, there will be no charge for this service when done through the NBS Program.

Background

Hemoglobin screening results utilizing the standard newborn screening technology are invalid for three months if a newborn has been transfused with whole blood or packed red blood cells prior to specimen collection. In July 2005 the Newborn Screening Program initiated mandatory follow-up hemoglobin DNA testing (which could be performed at any time post-transfusion rather than after three months) for all newborns without an adequate newborn screening test prior to transfusion. Prior to that time, transfused babies with a hemoglobin pattern containing S,C, D, E, or V were routinely tested by the Program to rule out a hemoglobinopathy, but for patterns of A, AF or FA, the Program recommended that the babies be tested but did not require it. For reasons described below, this practice will be resumed.

Prior to 2005, physicians were urged by Newborn Screening follow-up coordinators to have transfused babies (with the aforementioned hemoglobin patterns of A, AF, or FA) tested by electrophoresis for hemoglobinopathies three months after the last transfusion. However, because very few of these babies were tested, testing was made mandatory to assure that hemoglobinopathies would be picked up in transfused babies. This meant that NBS Area Service Center (ASC) staffs would contact physicians/NICUs and make arrangements for specimen collection and testing. However, despite the efforts of the ASCs, of the 1,635 babies screened only after a transfusion in 22 months, only 51% of them had DNA testing. Of those babies tested, there have been 19 hemoglobin traits identified and no hemoglobinopathies confirmed. Although these figures are not surprising given the known prevalence rates of traits and hemoglobinopathies in the California population, it was determined that in view of the significant added workload of the Newborn Screening Area Service Centers and CHRCO, the

cost of mandatory follow-up outweighed the benefit. Furthermore, transfused babies who have a hemoglobinopathy most likely would have had a hemoglobin pattern (post-transfusion) of S, C, D, E, or V, all of which get followed up with DNA testing.

The Newborn Screening Program still recommends that the transfused babies in question be tested for hemoglobinopathies. The difference is that the babies' doctors will have the option of contacting the ASC to request follow-up rather than the ASCs initiating the process. The Newborn Screening results mailers will alert the physicians to the fact that the hemoglobin result of the post-transfusion screen is uninterpretable due to a transfusion and that they may contact the ASC to arrange follow-up.

Transfusions also render galactosemia results uninterpretable. For galactosemia, the mailer advises physicians to have the parents tested if there is a family history of the disease or if the baby is symptomatic. The test for biotinidase deficiency, to be added to California's screening panel this summer, is also thought to be affected by transfusions. The state's metabolic specialists are in the process of developing recommendations for biotinidase deficiency testing in the event that a baby was transfused before specimen collection.

The fact that there are multiple newborn screening tests that are invalidated by transfusions underscores the importance of NICU staff collecting the NBS specimen prior to a baby getting transfused, no matter how young the baby is. If a baby is under 12 hours of age when the specimen is collected, then a second specimen needs to be collected at least 24 hours after a transfusion is completed. Together the two specimens will provide a complete screening panel, thereby eliminating the possibility of missing a disorder or the need for more complicated and inconvenient follow-up testing.

We appreciate your cooperation in assuring that all newborns get screened correctly and completely. Please share this information with your staff. Questions or concerns should be directed to your designated ASC (see attachment).

Sincerely,



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cc: Heidi Lerner, RN, MSN, MPH, Nurse Consultant
Newborn Screening Area Service Center Project Directors