Severe Combined Immunodeficiency (SCID) Fact Sheet

The California Newborn Screening (NBS) Program has begun a pilot program to expand the newborn screen to include the identification of Severe Combined Immunodeficiency (SCID) and related disorders. A baby in your care has had either a positive result or inconclusive results for this screening test. An inconclusive result means the DNA from the baby's newborn screening sample did not amplify, so we were unable to complete the testing. Therefore, more testing needs to be done. Since a newborn is protected by maternal antibodies in the first 2 – 4 months of life, a baby with SCID or a related disorder may not have any symptoms in the first few months.

What is SCID?

Severe combined immunodeficiency (SCID) represents a group of rare, sometimes fatal, congenital disorders characterized by little or no immune response. The defining feature of SCID, commonly known as "bubble boy" disease, is a defect in the specialized white blood cells (T and B lymphocytes) that defend us from infection by viruses, bacteria and fungi. Without a functional immune system, SCID patients are susceptible to recurrent infections such as pneumonia, meningitis and chickenpox, and can die in their first year of life. Restoring a functional immune system, usually by bone marrow transplantation, can cure SCID if the diagnosis is made before devastating infections occur.

What immune disorders are picked up by the SCID screening test?

The test measures T cell receptor excision circles, or TRECs, that are byproducts of normal T lymphocyte development. Any of a spectrum of disorders with poor T lymphocyte production (as in DiGeorge syndrome) or increased loss of T lymphocytes may lead to a positive test. While not as severe as SCID, these conditions need to be identified promptly and may require treatment. HIV or AIDS are not identified by TREC screening.

Why is SCID being added to the California NBS Program now?

Testing methodology has recently become available that can accurately and efficiently identify babies who may have SCID. This new methodology allows for efficient testing of large numbers of samples, making it possible to screen all newborns in the State. When identified early, treatment is more effective. In February, 2010, the American College of Medical Genetics (ACMG) added SCID to its panel of disorders recommended for newborn screening, and in May 2010, the U.S. Secretary of Health and Human Services endorsed this recommendation. As a result, the NBS Program is currently in the pilot phase of adding SCID testing to the California NBS panel.

What do I do now?

Two issues need to be addressed immediately. First, ***do not*** vaccinate a baby suspected of having SCID with a live vaccine, including the rotavirus vaccine. (For babies that may seek care in Mexico, ask parents to tell the pediatric care provider that live vaccines are contraindicated, including the BCG vaccine against tuberculosis.) Second, a Newborn Screening Coordinator will, with your consent, arrange for follow-up testing. If the first test result is inconclusive, a second heelstick filter paper specimen will be collected and the test will be run again. If the test is deemed positive, or if the baby has had two heel stick tests with inconclusive results, a CBC with differential and blood lymphocyte evaluation by flow cytometry will be performed.

Blood lymphocyte evaluation by flow cytometry will determine if the baby has SCID or a related disorder. If the results of the follow-up test are abnormal, you will be contacted by Dr. Jennifer Puck at UCSF in Northern California or Dr. Sean McGhee at UCLA in Southern California to facilitate

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getting babies into care with a pediatric immunologist. If the results are normal, you will be notified by NBS staff and no further follow-up for SCID is necessary.

What if this baby needs a transfusion?
Make sure any blood ordered for this baby is irradiated, leukoreduced and CMV negative. While this may be standard practice for many facilities, it's best to make sure to specify this in the order.

Remember:
Due to biological variability of newborns, the Newborn Screening Program will not identify all newborns with SCID or a related condition. While a positive screening result identifies newborns at an increased risk to justify a diagnostic work-up, a negative screening result does not rule out the possibility of any immune system defect. Health care providers should remain watchful for excess infections in their patients. A newborn screening result should not be considered diagnostic, and cannot replace the individualized evaluation and diagnosis of an infant by a well-trained, knowledgeable health care provider.

If a baby exhibits symptoms consistent with an immune disorder, has had multiple infections, or you suspect SCID even though the NBS result was negative, contact Drs. Puck or McGhee to consult about further testing.

For more information about SCID, we recommend the following:

Websites

The Jeffrey Modell Foundation: www.jmfworld.org

The Immune Deficiency Foundation: www.primaryimmune.org

The National Human Genome Research Institute of the NIH: www.genome.gov/13014325

Articles

Routes JM, Grossman WJ, Verbsky J, et al. Statewide newborn screening for severe T-cell lymphopenia. *JAMA*. 2009;302(22):2465-2470.

Addition of severe combined immunodeficiency as a contraindication for administration of rotavirus vaccine, *MMWR Morbidity Mortality Weekly Report*. 2010 Jun 11;59(22):687-8

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