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Surviving Life in a Bubble and Other Immunodeficiencies

SCID Specialty Care Center Helps Children Live a Normal Life

What many people know about severe combined immunodeficiency (SCID) doesn't go beyond what they watched in the 1976 Emmy-nominated "The Boy in the Plastic Bubble," where the star, played by John Travolta, must live out his life in incubator-like conditions. While the film occurred almost three decades ago, SCID still does exist. And it's an illness that is much more complex and debilitating than any Hollywood screenplay could depict.

For children born with this rare genetic disorder, NewYork-Presbyterian/Morgan Stanley Children's Hospital has one of only several New York State-designated SCID Specialty Care Centers. Patients cared for at the facility are born with few or no T cells (the white blood cells in that combat infections), making them susceptible to contagions that can not only be reoccurring, but also life-threatening. As a health care requirement, all infants born in New York State are screened for SCID.

Yesim Yilmaz Demirdag, MD, Director of the SCID Specialty Care Center, and her team of highly qualified allergists and immunologists, genetic counselors, hematologists with expertise in bone marrow transplantation, nurses, social workers, and other support staff, evaluate newborns with abnormal screening for SCID, the most severe form of primary immunodeficiencies. The doctors, who have specialty training in immunodeficiency syndromes, also diagnose and treat newborns with abnormal SCID screening who do not have SCID but other forms primary immunodeficiencies, such as Di George Syndrome."Our initial testing includes complete blood count and differential of white blood cells and flow cytometric analysis to assess lymphocyte subsets," says Dr. Demirdag. "If these are abnormal, we perform further testing such as lymphocyte function test and genetic testing."

Newborns with abnormal SCID screening are evaluated further by an allergist/immunologist who performs comprehensive diagnostic testing, such as blood tests to confirm or rule out an SCID diagnosis. If the SCID diagnosis is made, the newborn is referred for bone marrow transplantation or treated with enzyme replacement, depending on the type of the SCID.

Also known as stem cell transplantation, bone marrow transplantation is the most effective treatment for SCID. "The survival rate is 96 percent if the infant receives bone marrow transplantation before 3.5 months of age and before he/she develops severe infections," says Dr. Demirdag. "The survival rate is only 66 percent if the transplant is delayed."

The ideal donor is a perfectly HLA-type matched sibling who has a normal immune system. But if a matched sibling donor is not available, success has been had with matched unrelated donors and even half-matched related donors, such as parents.

Through comprehensive screening tests for SCID infants may be diagnosed with other conditions in which T lymphocytes are deficient—these are often milder forms of primary immunodeficiencies. The team at the SCID Specialty Care Center cares for these infants with treatments including prophylactic antibiotics and immunoglobulin replacement treatments.

Testing for SCID, including genetic testing, is available onsite at the SCID Specialty Care Center, and patients are seen within 24 hours of referral. With initial testing, many results are available within 24 hours.

And the quick response is a relief for parents waiting on the results. "As one of the top children's hospitals in the nation, our hospital is fully equipped to offer the best diagnostic and therapeutic services to these patients," says Dr. Demirdag.

To refer a patient for the evaluation of SCID or other immunological disorders, call (212) 305-2300. — ***Cecilia Martinez***