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## IMMUNE DEFICIENCY FOUNDATION

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*The National Patient Organization Dedicated to Advocacy, Education and Research for Primary Immunodeficiency Diseases*

### **Questions about Severe Combined Immunodeficiency Disease**

#### **Why have medical professionals called Severe Combined Immunodeficiency Disease (SCID) a pediatric emergency?**

SCID is a group of inborn disorders, all of which result in failure of affected infants to develop T-cells, a critical component of the immune system. As a result, the condition is fatal in infancy unless treated with bone marrow transplantation, or in some cases enzyme replacement or gene therapy. If diagnosed within the first 3 1/2 months of life before infections develop, the cost of this treatment can be as low as \$50,000 for each infant. By contrast, treatment costs can exceed \$1,000,000 for each infant diagnosed late, primarily due to costs of treating infections that can leave the child with long-term medical complications. Without a bone marrow transplant in those crucial 3 1/2 months, many of these infants die of infection even if a bone marrow transplant is attempted later. The diagnosis of SCID very early in life is a true pediatric emergency.

#### **How common is SCID?**

There is no central record of how many babies are diagnosed with SCID in the United States each year, but the best estimate is somewhere around 40 -100. SCID is a rare condition, but is as frequent as some conditions that newborns are currently tested for such as biotinidase deficiency or certain metabolic disorders. On the other hand, researchers have no clear idea of how many babies are not diagnosed and die of SCID-related infections each year. The actual number of cases is most likely higher.

#### **How is SCID diagnosed?**

Fewer than 20% of infants with SCID have a family member with the condition that allows doctors to suspect it and test at birth. Early diagnosis of SCID is rare because doctors do not routinely count each type of white blood cell in newborns. As a result, the average age at which babies are diagnosed with SCID is just over six months and some are diagnosed much later, usually because of recurrent infections and failure to thrive. Blood tests for SCID typically reveal significantly lower-than-normal levels of T cells and a lack of germ-fighting antibodies. Even if B cells are present in the blood of SCID patients, they do a poor job of producing antibodies. Low antibody levels and lack of specific antibodies after vaccination or a natural infection are characteristic features of SCID.

#### **Is there effective treatment for SCID?**

The most effective treatment for SCID is transplantation of blood-forming stem cells from the bone marrow of a healthy person. Bone marrow stem cells can live for a long time by renewing themselves as needed and also can produce a continuous supply of healthy immune cells. A bone marrow transplant from a tissue-matched sister or brother offers the greatest chance for curing SCID. However, most patients do not have a matched sibling donor, so transplants from a parent or unrelated matched donor are often performed. All transplants done in the first three months of life have the highest success rate.



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### **Can SCID be detected before birth (prenatally)?**

If the presence of SCID in the family's history is known, and the type of SCID has been identified, sequencing DNA from the fetus can test an at-risk pregnancy. SCID can be identified before the baby is born by removing and testing cells from the placenta (chorionic villus sampling or CVS), or by removing and testing a sample of the fluid surrounding the baby (amniocentesis). However, prenatal testing is only available when a previous family member with SCID has been recognized and had their gene mutation determined.

### **How Effective is Early Diagnosis?**

The sooner a child is diagnosed, the sooner treatment can begin and the more likely it is to be effective. Some babies develop fatal infections before their condition is recognized. Pediatricians are now encouraged to give infants as young as 6 weeks old the live rotavirus vaccine to prevent rotavirus infection; however, if this vaccine is given to an infant with SCID, the infant can contract serious diarrheal illness from the attenuated virus in the vaccine. The need for a way to recognize those infants who would be at risk if given an otherwise beneficial vaccine is crucial. Recent research shows that bone marrow transplants in the first three months of life work better than transplants at a later age. So it is critical to identify affected children immediately after birth. A survey of more than 150 patients commissioned by the Immune Deficiency Foundation found that SCID patients who were diagnosed early and treated by 3.5 months had a 91% survival rate; those treated after 3.5 months had a 76% survival rate.

### **Does Late Diagnosis Affect a Surviving SCID Patient's Quality of Life?**

If diagnosis is late, even a successful bone marrow transplant can still leave a patient with persistent health problems, such as the need for procedures as drastic as lung transplants because of the damage suffered before diagnosis. Most parents and physicians agree that ongoing health issues are not a result of the child having SCID, but because of the delay in diagnosis that leaves the infant critically ill with multiple infections. The poor quality of life in late-diagnosed SCID patients can eventually lead to disability or even death.

*The Immune Deficiency Foundation (IDF) applauds the Secretary of Health and Human Service's inclusion of SCID in the new national standards*

*IDF asks every state to include SCID Newborn Screening on their newborn screening panel immediately to save lives.*