

Barbara L. Ballard

Advisory Committee on Heritable Disorders in Newborns and Children

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I would like to thank the committee for this opportunity to represent the families of children with Severe Combined Immune Deficiency. My name is Barbara Ballard and I am the mother of a boy with X-linked SCID. I am also the administrator for group of SCID families dedicated to supporting one another in this journey we call SCID row.

It was 25 years ago this week that David Vetter, the "Texas Bubble Boy" died. Despite being diagnosed as a newborn it took doctors years to offer any treatment other than a plastic bubble because transplants were only an option if you had a matched donor. There were no donor registries, PEG-ADA had not yet been developed, and half matched transplants would not be available for more than another decade. Today bone marrow transplants are the standard of care for the majority of SCID patients. An effective method to diagnose SCID from a simple blood spot now exists. SCID families passionate to improve the rate of diagnosis have given their children's very blood toward improving the rate of diagnosis.

Let me talk a moment about Quality of Life for those children who are lucky enough to be survivors of this disease. I specifically want to talk about those children who were not diagnosed as newborns, those children who had to be so sick that they came to the brink of death before doctors could diagnosis the problem. My son Ray is one such child. Ray is now 15 years old. Born seemingly normal, he thrived for several months until he caught his first cold. Within days of first entering my pediatrician's office with a child that I thought might have a virus, he was in the PICU and on a ventilator with PCP pneumonia. He spent 4 ½ months on a ventilator, had 13 chest tubes, and was trached. He received his first bone marrow transplant at a year old while on the ventilator in the Duke PICU. An enteral virus ultimately caused severe GI damage. His GI tract never fully recovered and he remains fed by enteral and parenteral means. Infection and Graft vs. Host disease caused his graft to fail and he has required 2 additional booster transplants. He managed to come off the ventilator and his trach was eventually removed, but he has severe lung damage and scarring which significantly limits his ability to participate in normal childhood activities.

All the infections had to be countered with multiple antibiotics, antivirals and antifungals. Ultimately, we learned that one of the antibiotics used to save his life, had also left him deaf.

My son's medical costs maxed out a \$2million insurance policy by the time he was 5 years old. Though Ray survived when many have not, his life will continue to have many costly challenges which could have been prevented. Benefits of early diagnosis for him would have been a life without these ongoing costs and challenges.

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My son is not alone in having long term medical complications resulting from a delay in diagnosis. As the administrator for a support group of SCID families, I can tell you many similar stories. It is for all the SCID children, surviving and lost, that I speak today.

Modern viruses are becoming more of a risk even to the general population. The best way to battle many of them has been the development of live virus vaccines. It is now considered “safe” to give a live rotavirus vaccine to an infant who is only 6 weeks old. How is a pediatrician to know that a 6 week old infant has SCID, unless there is a mandatory screening test for newborns? The manufacturers’ own literature tells you to “check with your doctor if your baby’s immune system is weakened”. The vials in which the vaccine is delivered must be disposed of in a biohazard waste container. It is unconscionable that the administration of a live vaccine to children as young as 6 weeks has been approved without first providing a method to identify those children for whom this vaccine could be devastating. The responsibility to protect those children who are most at risk of injury from these vaccines now lies with you, the members of this committee. As more live virus vaccines are developed to protect the general population it compounds the risk to our undiagnosed SCID babies and compounds your obligation to protect them.

There are those who would argue that a false positive test for SCID would be too dire for the family involved. I disagree. When I asked the SCID families their perspective on this argument; those families were overwhelmingly shocked to learn that there was more concern for a family with a healthy baby who might be asked to repeat a test, then for a family with an undiagnosed SCID baby who might not learn of that diagnosis until after they buried their child.

SCID is a disease which cannot be seen or identified at birth without a blood test. Children with SCID are born looking and acting seemingly normal. A simple and reliable test for SCID now exists. We can easily identify affected children before they contract their first cold. Without this early diagnosis and the now standard medical treatments, the damage caused by infections allowed to ravage the bodies of these children, will cause irreversible damage and very often death. How many children must suffer with a diminished quality of life, how many children must die before you say that it’s too many? We have the technology, we have the science, we now need the prudence to step up to the plate and make this test a standard of care.

