Journey to Newborn Screening

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The following is a summary of remarks delivered by Jennifer Puck, M.D. at the 2017 Plasma Protein Forum in Washington, D.C.

Severe Combined Immunodeficiency (SCID) is a primary immunodeficiency disease caused by a combination of T-cell and humoral deficiencies, resulting in the inability to produce antibodies. Babies born with SCID look healthy, but they soon develop recurrent infections that cannot be cured with standard therapies. Bone marrow transplants are an effective treatment, however unless SCID is diagnosed immediately, a transplant becomes difficult due to the multiple infections.

The first successful bone marrow transplant for a SCID patient occurred nearly 50 years ago in 1968. Since then, there have been numerous advancements in both diagnosis and treatment. Early testing began with families that had previous babies affected with SCID. Research by Dr. Rebecca Buckley revealed babies' survival was better in those families in which prior history allowed early diagnosis, with a 96 percent survival rate versus a 66 percent survival rate in patients without a family history. Many babies without a family history of SCID were diagnosed later due to a delayed onset of symptoms, likely caused by dwindling levels of maternal immune globulins that protects babies in early infancy. In the case of David Vetter, known as the "Bubble Boy" of Houston, doctors knew to protect David with a sterile bubble at birth because he had an older brother affected with SCID who did not survive.

Because of David and Dr. Buckley's work, it became evident that babies affected with SCID need to be diagnosed as early as possible, and before any infections occurred in order to have optimal treatment and the best chance of survival. To pursue early diagnosis, the National Institutes of Health's Office of Rare Diseases and the National Institute of Allergy and Infectious Diseases founded the Primary Immunodeficiency Consortium whose research focused on improving newborn screening techniques.

Research began by testing dried blood spot samples collected by hospital nurseries and sent to state health departments for screening. Initial attempts to detect SCID using plasma or cellular proteins were unsuccessful. However, a breakthrough came when Dr. Jennifer Puck and her research team thought to use a polymerase chain reaction test on DNA circles that are created when T-cells and B-cells reproduce their DNA. Dr. Puck and her team discovered it was possible to isolate these DNA from dried blood spots and accurately diagnose babies affected with SCID.

The test was first piloted in Wisconsin and is now included in most state newborn screenings. Each state performs and funds newborn screenings independently, therefore the inclusion of SCID must be raised locally to convince policymakers of the test's value. In 2010, the U.S. Secretary of Health and Human Services announced the addition of SCID to the Recommended Uniform Screening Panel, due to the efforts of the Jeffrey Model Foundation and the Immune Deficiency Foundation. Today screening is almost universal in the United States, with only two states (Indiana and Louisiana) not screening for SCID.

The future work of the Consortium is focused on developing best practices for treating SCID and, ultimately, curing babies. After more than 50 years of medical and scientific research, SCID has evolved from a fatal diagnosis to a severe but treatable condition, but early diagnosis is imperative to start treatment before a life-threatening infection could cause irreversible damage. •

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