Detection of primary immune deficiency disorders

Thomas Rand, MD, PhD

Primary immune deficiency disorders (PIDD) are a group of diseases in which the immune system does not handle infections normally. PIDD are largely genetic diseases that are lifelong. Like other genetic diseases, another relative may be similarly affected. In some PIDD, the immune system may appear normal for some time but develop abnormality over time, particularly for common variable immune deficiency, one of the more common diagnoses within PIDD.

Because frequent infections and difficulty recovering from infections are common, whereas PIDD are rare, medical diagnosis is like finding a needle in a haystack.

Within the last three years, most states including Oregon, Washington, and Alaska, added severe combined immune deficiency (SCID) to newborn screening. SCID includes a number of PIDD that are fatal without treatment, typically by transplant bone marrow from a matched donor.

Clinicians must have a low threshold for ordering screening tests, because many normal results must be tested to find the rare PIDD. Complete blood count (CBC) and immunoglobulins IgG IgM IgA are the most basic screening tests ordered to evaluate difficulties with infection. CBC findings in PIDD may be nonspecific such as low lymphocyte number, but follow up testing will lead to the proper diagnosis. Immunoglobulins IgG IgA IgM are antibody proteins and are the first step in diagnosis of PIDD that have deficiency of antibody. Since Human Immunodeficiency Virus (HIV) infection is more common than PIDD, then HIV testing is usually done when considering immune deficiency.

Beyond purely the numbers of infections, some types of problems trigger more urgent consideration of PIDD. “10 Warning Signs of PICC” (figure) has been promoted by The Jeffrey Modell Foundation and The Immune Deficiency Foundation.

With appropriate medical management, the lives of people with PIDD may be transformed for the better. For example, gamma globulin replacement therapy is indicated for sufficiently severe deficiency of antibody in PIDD. Having seen first-hand astounding results of treatment, clinicians experienced with PIDD are big advocates for screening tests such as CBC, immunoglobulins IgG IgM IgA and newborn screening for SCID.

Thomas Rand, MD, PhD
Pediatrics
PeaceHealth Medical Group
4545 Cordata Pkwy, Suite 1E
Bellingham, WA 98226
360-738-2200
trand@peacehealth.org