Most of us, when we hear hoof beats, think of horses. It's so easy to look at a picture and see what you most expect. As ambulatory nurses, we need to look for the unexpected—a zebra instead of a horse. The Immune Deficiency Foundation (IDF) would like us to “Think Zebra.”

Primary immune deficiency disorders (PIDD) were once believed to occur rarely in the general population. Historically they have been considered pediatric disorders because they were thought to be diagnosed most often during childhood. According to the National Institute of Child Health & Human Development (NICHD), “PI diseases were once thought to be rare, mostly because only the more severe forms were recognized. Today physicians realize that PIDs are not uncommon. They are sometimes relatively mild, and they can occur in teenagers and adults as often as in infants and children” (NICHD, 2011).

The 2007 National Patient Survey results published by the IDF conclude that diagnoses of PIDD are far more common than suggested in the literature. “The exact number of persons with PI is not known. It is estimated that each year about 400 children are born in the United States with a serious PI. The number of Americans now living with a primary immunodeficiency is estimated to be between 25,000 and 50,000” (NICHD, 2011). Survey findings indicate the prevalence of PIDD in the general population to be 1 in 1,200 persons (Boyle & Buckley, 2007). Findings also confirm that primary immune deficiency disorders are no longer strictly a pediatric condition in the United States (IDF, 2007). More than half of new patients surveyed were not diagnosed with PIDD until they were age 30 or older. The average length of time from onset of symptoms to diagnosis is 12.4 years (IDF, 2007). Few of the patients participating in the survey reported a family history of PIDD; however, 9 out of 10 patients reported they experienced repeated, serious, or
unusual infections prior to diagnosis (IDF, 2007). The goal of this article is to raise awareness regarding PIDD in the ambulatory nursing community.

Primary immune deficiency disorders are a group of conditions in which the patient has an intrinsic immune system defect in his or her immune system. There are currently more than 150 different diagnoses recognized by the World Health Organization as PIDD (Boyle & Buckley, 2007). Making a PIDD diagnosis is challenging because there aren’t any clearly recognizable or unique symptoms. The patient frequently presents with symptoms of infections, which can be persistent and/or chronic. It is not unusual in our practice to hear from a new patient and family with a child who has been sick for a long period of time – sometimes years. Often several different physicians and specialists saw the child before there was a correct diagnosis and appropriate referral for treatment. If the patient, family, or health care provider does not look beyond the individual episode requiring treatment, the underlying cause may go undetected.

The Jeffrey Modell Foundation (2012) created the 10 Warning Signs of PIDD to help raise awareness of these disorders. The list for children is as follows (variations for adults are noted):

1. Four or more new ear infections within one year (two or more for adults)
2. Two or more serious sinus infections within one year (in absence of allergy for adults)
3. Two or more months on antibiotics with little effect
4. Two or more pneumonias within one year (one or more for adults)
5. Failure of an infant to gain weight or grow normally
6. Recurrent deep abscesses of the skin or internal organs
7. Recurrent need for intravenous antibiotics to clear infections
8. Persistent thrush or fungal infection
9. Two or more deep-seated infections including sepsis (not noted in adult criteria)
10. A family history of PIDD

Also included in diagnosing an adult is an infection with normally harmless tuberculosis-like bacteria and chronic diarrhea with weight loss (Jeffrey Modell Foundation, 2012). This information is available on the Foundation Web site (www.info4pi.org).

Evaluation may include a detailed medical history, thorough physical examination, blood tests, and vaccines to test the immune response. A routine CBC with differential that shows a low lymphocyte count (especially if it persists in follow-up testing) may warrant closer examination by an immunologist. Other lab tests frequently used in diagnosis are IgG, IgA, IgM, and IgE levels, delayed-type hypersensitivity skin tests, mitogen and antigen studies that look at T cell response to stimuli, and CD19 and CD20 for B cell lymphocytes present in the circulation. Frequently, patients are given vaccines (not live virus) followed by titers to check for the body’s response to those vaccines. Patients with undiagnosed and untreated PIDD are unable to produce antibodies sufficient to fight infection. Administration of live viruses to a patient with a compromised immune system can result in serious illness or death. In addition, siblings and other household contacts should not receive live virus vaccines due to the potential for disease transmission (Winkelstein, 1996).

“PIDD vary in prevalence from relatively common to very rare” (Burton, Murphy, & Riley, 2009, p. 6). Examples of some of the better-known disorders are noted in Table 1 (see page 10). This table presents only basic information regarding PIDD. The wide range of symptoms and variability among disorders makes it impossible to include details for any one specific disorder in this format. It is worth special note that severe combined immune deficiency (SCID) usually manifests in infancy, and without early diagnosis, has poor outcomes for the patient. SCID is sometimes known as “bubble boy disease.” When diagnosed at birth, these patients receive a bone marrow transplant within the first ten days of life. “In January of 2010, the Advisory Committee on Heritable Disorders in Newborns and Children voted to add screening for SCID to the core panel of universal screening of newborns in the United States” (Gill, 2010). To date, it is the only form of PIDD for which routine screening has been established.

For many patients with PIDD, treatment includes IgG replacement therapy given intravenously or subcutaneously. Intravenous infusions of immune globulin (IVIG) are usually given every 3-4 weeks and can be given in an infusion center or at home through a home care agency. Home IVIG infusions should be provided through a home care agency with a nurse present during infusion. Subcutaneous infusions are usually given weekly at home and administered by the patient or a family member following initial training by a health care provider (Skoda-Smith, Torgerson, & Ochs, 2010). Having options for the method of administration of IgG gives the patient some control over his or her treatment options and lifestyle.

Stem cell transplants have been used for some patients with PIDD with positive outcomes. “The primary immunodeficiency diseases for which HSCT (hemopoietic stem cell transplant) is most commonly performed include those diseases that are characterized by deficient T-lymphocytes or combined deficiencies of T-lymphocytes and B-lymphocytes. HSCT is most often used to treat SCID. HSCT has also been used in some patients to treat other primary immunodeficiency diseases such as the Wiskott-Aldrich syndrome, hyper-IgM syndromes, and chronic granulomatous disease” (IDF, 2007). Clinical trials with gene therapy are also being done with these same patient populations with mixed results. Stem cell transplants and gene therapy show tremendous potential for the treatment of T-lymphocyte disorders and combined T-lymphocyte and B-lymphocyte disorders, but more research is needed.

“Throughout adult life, patients remain stable in both the management of their condition and the condition itself by adhering to the chronic care management (CCM) model” (Burton et al., 2009, p. 8). This model includes disease prevention, evidence-based interventions, along with
several other tenets of traditional care supported by research and education. Burton and colleagues (2009) further discussed the new patient-centered PIDD CCM based on the Expert Patient Program started in the United Kingdom in 2002. The five core proficiencies identified as necessary to becoming an expert patient are as follows:

- Problem-solving
- Decision-making
- Resource maximization
- Developing effective partnerships with health care providers
- Appropriate interventions

“The focus of this new CCM for PIDD is enabling complete patient self-management from the patient’s perspective” (Burton et al., 2009, p. 8).

Assisting patients to be experts in their own care would help to establish them as effective partners with their health care providers. This type of health care consumer/provider relationship is consistent with the National Prevention Strategy (U.S. Department of Health and Human Services, 2012) and would support the goals of Health Care Reform. For patients with PIDD, being educated about their disorder and having the ability to advocate for their health care needs can make a significant difference in their ability to reach their optimal health potential. “Effective management of chronic disease is the key to both reducing health care expenses and improving patients’ quality of life” (Burton et al., 2009, p. 5).

Resources for health care providers, patients, and families are available through the Internet or in print. The Immune Deficiency Foundation (www.primaryimmune.org) and the Jeffrey Modell Foundation (www.info4pi.org) are both reliable sources of information. The IDF mission includes PIDD research and patient advocacy among its many offerings. Their “Think Zebra” campaign was established to promote awareness of PIDD. They have also published a handbook for patients and families as well as a nurses’ guide to IVIG. The handbook contains basic information as well as a section dedicated to each of the most common diagnoses. The IDF also sponsors an online continuing education course for nurses that includes detailed information about PIDD. The course is free, provides 5 contact hour credits, and can be found on their Web site. IG Living magazine (www.igliving.com) is a useful resource for patients and families receiving immune globulin therapy. It has been in publication for several years and is sponsored by several manufacturers of IVIG products. Subscriptions are free.

All of us, as ambulatory care nurses, are in a position to listen to our patients’ stories, hear what they are saying, and collaborate with them to look at the big picture. Early diagnosis and treatment leads to the best possible outcomes for PIDD patients. So remember: The next time you hear hoof beats, “Think Zebra!”

References


Suggested Reading


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