

Treating PIDD Kids – Post-Diagnosis



Most children with PIDD can lead relatively normal lives, but it's necessary for parents as caretakers to understand their child's treatments and psychological needs, as well as the financial considerations and environmental concerns, to best manage their child's disease.

By Amy Scanlin, MS

For parents, a child's diagnosis of a primary immune deficiency disease (PIDD) is a scary and, often, confusing time. What were frustrating repeated occurrences of ear infections, sinus infections, bronchitis or pneumonia now is a diagnosis that sounds far more frightening than was ever thought possible. And while the diagnosis can be a relief and bring apparent closure to the question: "What is going on

with my child?” — especially for those kids who also have health problems of the digestive tract, anemia or even autoimmune diseases as a result of immune system dysfunction — there also is the recognition that a whole new world of issues is arising.

Kids can take heart that with a prompt diagnosis and proper treatment, they very often will lead normal lives of play, sports and, yes, even school. Parents can take heart that while PIDD was once thought to be a rare disease, because only the most severe forms were more easily recognizable, milder forms of PIDD are now recognized as being more common than previously thought, and also more readily treatable.¹

PIDD Diagnosis and Treatment

Typically symptoms of a severe PIDD are noticed when a child is still in infancy through his or her first few years of life. In some cases, though, diagnosis does not occur until a child is a teen.

To make a diagnosis, typically a protein vaccine (tetanus and diphtheria toxoids) and a polysaccharide vaccine (pneumococcal) are administered to determine a child's ability to make specific antibodies in response to the vaccines' antigens. The pre-immunization response (blood taken before the immunization) is tested and compared with the post-immunization response (blood typically obtained about four weeks after the vaccination). This response allows doctors to better understand the severity of the disease and how best to treat it. In a child 6 years of age and younger, more than half of the vaccine components tested should demonstrate good antibody responses, and for a child older than 6 years of age, 75 percent of the vaccine components tested should result in good antibody responses.

Based on the results, a diagnosis of some form of antibody deficiency may be made, and subsequent treatment therapies can be determined. “We need to stratify the extent of the problem and make a decision as to how aggressive therapy needs to be based on the history, physical examination and laboratory test results,” says Dr. Terry Harville, medical director at the Departments of Pathology and Laboratory Sciences and Pediatrics at the University of Arkansas for Medical Sciences.

If recurrent respiratory infections are the main problem and no specific antibody deficiency can be ascertained or only a few of the vaccine components fail to respond adequately, then treatment with antibiotics may be appropriate. If there is an inadequate antibody response in the

testing, therapy with immunoglobulin (IG) replacement may be needed, and antibiotics may also be used. IG replacement therapy may also need to be started if there is a failure to achieve an antibody response, or if the doctor determines it necessary based on his or her expertise when considering the severity or types of infections. For an inadequate or nonexistent antibody response, there should not be a trial to see whether antibiotics alone can slow down or prevent illnesses.

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Sometimes, certain antibodies are missing, even though the majority of antibodies may be fine. This situation may be called a specific antibody deficiency. This is typically one of the most difficult to diagnose, since the testing may not demonstrate a severe deficiency in antibody production. And, it can be one of the most frustrating to treat because IG replacement therapy may be required, but a patient's insurance carrier may not want to pay for the therapy since a more severe antibody deficiency cannot be demonstrated.

As a group, antibody deficiencies represent the most common type of PIDD. Millions of antibodies, called immunoglobulins (IgGs), circulate in the blood, helping the immune system to fight all kinds of infections.² By definition, antibody deficiencies require replacement of the missing antibodies with IG infusions, either by the intravenous route (IVIG) or the subcutaneous route (SCIG), in order to prevent the complications associated with recurrent infections and, most importantly, to prevent death. IgGs are obtained by “batching” them from the plasma of thousands of donors. The plasma is processed to isolate the antibodies and to remove viruses or other infectious agents. When infused regularly (every three to four weeks for IVIG therapy, or up to several times per week for SCIG therapy), the infusions can help to bring IgG levels to normal or near-normal levels for a child's age to prevent infections.¹ It is now recognized that maintaining the serum IgG levels well into the normal range helps to prevent many complications associated with infections.

Whether treatment is administered in a hospital or at home, follow-up doctor visits help a child to be re-evaluated, and they allow the doctor to learn of any infections or illnesses a child may have had since his or her last visit. They also are a good opportunity for the doctor to determine whether a child's treatment dosage is still correct for his or her growing body. The frequency of follow-up visits is determined as part of the initial treatment plan, and it is continually adjusted as needed. A child also may need to be seen for other issues in addition to his or her PID. Asthma, allergies, arthritis, celiac disease or other things keeping him or her ill may need to be identified, evaluated and treated.

If breakthrough infections occur, antibiotic treatment also may be required. Frequently, the dose of antibiotics may need to be higher and the duration longer. For example, a five-day course would constitute undertreatment. Further, a 10-day course may need to be expanded to 14 days, and if sinusitis is present, one to three months of continuous antibiotics may be required. Despite being on adequate dosing of replacement IG, if frequent breakthrough infections are occurring or chronic sinus or chronic lung infections are present, then continuous dosing with antibiotics may be necessary. Although in the past, low-dose antibiotics may have been used to act as "prophylaxis" against infections, it is now recognized that patients do better with full treatment dosing on an ongoing basis.³

There are situations in which IG replacement therapy may not be sufficient. For instance, if there is a problem with the function of neutrophils, such as with chronic granulomatous disease, it may be necessary to use daily antibiotics, typically trimethoprim/sulfamethoxazole to help reduce or prevent bacterial infections, itraconazole or voriconazole to help reduce or prevent fungal infections, and gamma interferon injections to maintain appropriate health. When the issue is a more severe problem with T lymphocyte production or function that also affects the ability of B lymphocytes to function appropriately for making normal antibodies (which would typically be diagnosed in infancy), a child may require hematopoietic stem cell transplantation (HSCT). That child will initially receive any antibiotics needed and be started on IG



replacement therapy as soon as possible. "We also have to formulate a plan to protect these kids from viruses and other infections," says Harville. A child may need isolation, especially from other children who may be infected with viruses, until the immune system has been reconstituted via HSCT.

HSCT is used for reconstitution of immunity in the most severe immunodeficiencies, as well as for blood disorders and cancer therapy. Hematopoietic stem cells, when working properly, produce billions of new blood and immune cells daily. Bone marrow is historically the most common source for HSCTs. However, today, frequently the source of stem cells for transplantation is from the peripheral blood. Donors no longer are required to undergo anesthesia and have needles poked through their bone to obtain bone marrow. Now, donors sit for a few hours, typically reading, watching movies or sleeping, while their blood exits through a catheter from a vein into a machine that removes the stem cells, and then is returned to their bodies.⁴ This allows for minimal discomfort and maximal yields of stem cells, since testing can be performed to determine how much of the donors' blood needs to be processed in order to achieve the needed amount of stem cells.

Financial Considerations

"This is not a cheap disease," says Dr. Harville. IG replacement therapy can cost up to \$5,000 to \$10,000 a month, and still other costs can be considerable depending on the other issues affecting a patient's health and well-being. For example, missed work to take an ill child to be evaluated can place a strain on employment, and a child may be missing school frequently.

A financial assessment arranged through a social worker at the clinic or hospital can assist the family in paying for gas, accommodations and meal vouchers if the family must travel a long distance for treatment. Some clinics and hospitals have a "Medicaid van" that will pick up a patient who lives a distance from the clinic and return him or her at the end of the day, if reliable transportation is not otherwise available for that patient. There are many plans in place to help a family in need, and getting a social worker involved early is crucial.

For many, once the stress of the financial implications have been thought through and planned for, a family is better able to focus positively on the medical treatments ahead.

Environmental Concerns

Even if treatment for a PIDD has begun, a child's environment cannot be overlooked. "Parents must be careful of environmental issues that may produce adverse outcomes," says Dr. Harville. "All the therapy provided won't help a child if an underlying issue exacerbating the illness is still present in their environment."

Smoking and mold are two very common irritants that can promote more severe respiratory disease, despite IG replacement therapy and antibiotic usage. Mold behind walls, under sinks, in an attic or crawl space, places that may be otherwise unseen, can cause major problems for a child, even if others in the home have fewer respiratory symptoms. In addition, even being around a smoker who may not be smoking at the time can have an adverse impact. The residual smoke on the smoker's clothing and in his or her hair can be sufficient to create problems, especially for a baby being held by the smoker.

Parents should take notice of when a child experiences symptoms and flare-ups to help find clues as to what environmental agents may be causing them. As an example, if a child gets sick every time he or she is in the home or car of a smoker, the answer is that child should no longer be in those places. It's not good enough that the person isn't smoking at the time.

Psychological Healing

A positive relationship is widely recognized between mental health counseling and the presentation of chronic illness symptoms in adults, including improvements in immune function, as well as a decrease in the effects of stress symptoms. It is suggested that the same can be true for a child, particularly in the areas of relaxation and hypnosis. These interventions are already used successfully in situations such as pediatric anxiety.⁵

In Dr. Harville's experience, often a main stressor for a PIDD child is the parent under stress and the resultant child's perception of the parent's stress. "A child who picks up on the parent's worry doesn't do as well," he says. On their own, "kids don't tend to worry as much, because their situation may be normal for them." A child may get upset that he or she can't do all the things his or her friends can do on the playground, but for a child, that state of health is his or her "normalcy."

If a child starts sympathizing with the parent's worry, it should be determined whether he or she has acquiesced to the issue rather than coped with it. Some children are more intuitive than others and may, in turn, have more psychological problems, but more typically, a child will worry more about whether he or she is going to get that new computer game or the latest MP3 player than whether his or her lungs are deteriorating.

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Joining a local support group and organizations that provide learning and support resources such as the Immune Deficiency Foundation can be a critical part of the emotional journey toward healing. According to Dr. Harville: "All the assurance I can give during a visit isn't the same as another parent sharing their story." In the process of a support group, each parent also finds comfort in helping others. Volunteers are often able to sit with families while they wait, provide emotional support, a hand to hold and guidance.

Parents' Roles as Helper, Healer, Caretaker

When the diagnosis of a PIDD is made, parents immediately take on the roles of coordinator, observer and secretary, in addition to caregiver. When a child has a more complicated disease or other complications, the parents' task of having to potentially coordinate visits to multiple specialists — with each determining what he or she believes is the right course of treatment — while carefully observing how a child is responding to the treatments can be more than a full-time job. Yet, parents also must be careful note-takers and communicators to ensure that the multiple specialists are each informed of each other's care approaches to treatment, and how each of their parts in the overall care is impacting a child. This, then, can become essentially a second full-time job for many when more complications are present.

Play often helps parents to see what is on a child's mind.

Parents are encouraged to take notes, including times of day, location and symptoms, which can help doctors to know how a child is doing and what they can do better to help a child in his or her healing. As an added benefit, this also helps parents feel they play a productive and proactive part in a child's medical care.

As a child ages, understanding of his or her condition typically changes. A young child may view the many doctor visits as punishment because he or she doesn't understand the disease and the need for treatment. That child may only see that he or she is missing out on what other children can do. It is important for parents to never use the threat of "the doctor will give you a shot" as a means to try to control a child's behavior. This creates anxiety in a child who frequently has previously undergone "shots" and blood draws, and who will likely require many more in the future. While it is easy to say that parents must help a child to understand that these visits and sometimes treatments are not punishment, the question becomes how to do this when a child has such limited understanding of the situation and the parents cannot reward each doctor's visit.

Play often helps parents to see what is on a child's mind. Whether children are drawing or playing, parents are often able to gently guide them to talk about what they are thinking and feeling.⁶ It is a child's great ability for

imagination that makes him or her such a good candidate for relaxation and hypnosis interventions for stress management. Helping a child channel this imagination in a positive direction with the help of his or her doctor can result in less anxiety and better well-being.

"I tell kids and their parents that you can't pick your genetics," says Dr. Harville about the guilt parents feel over their child's condition. "It's not their fault; it is just the way things are, and together we'll figure out how to best manage the situation. There's no one at fault, and no one is the culprit."

Once a child becomes older, there is greater understanding about his or her specific health needs. Educational materials can help to explain how treatments are beneficial, and they can enhance an older child's curiosity and, in turn, ease his or her anxiety.⁶

The diagnosis of a PIDD is a scary and often frustrating time for parents. However, in most cases, management of the disease can be straightforward, and a child can lead a relatively normal childhood. In addition to medical treatments, environmental impacts may have a significant effect on the health of a child with PIDD. Making changes to a routine to severely limit or completely curtail exposure to irritants will pay off. Even when things appear down, keeping a positive attitude is important. It helps parents to better cope and can help prevent a child from "feeling" the parents' anxieties that in turn create anxieties in a child. Kids watch parents' cues to learn how they should feel. A child's team of medical experts is there to help and has many resources to make this potentially arduous journey more comfortable for all. ■

AMY SCANLIN, MS, is a freelance writer specializing in medical and fitness writing.

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