



Funding Research to Find Cures/Treatments

Scientific research to identify, diagnose and treat PIDD and autoimmune diseases continues to increase with the combined efforts of doctors, organizations and individuals.

By Matthew David Hansen, DPT, MPT, BSPTS

The hopes and prayers of people living with a primary immune deficiency disease (PIDD) and/or autoimmune disease are to someday be cured of the condition or, at the very least, to find a way to stay healthy and improve their quality of life. Although the prospect of an individual miracle cannot be — nor should it ever be — discounted, the potential future eradication of PIDDs and autoimmune diseases will likely be due to the combined effort and enlightened discoveries of medical research.

Groundbreaking scientific advances have taken place during the last 10 to 15 years that are leading to the enhanced identification and diagnoses of PIDDs and autoimmune diseases, as well as the development of new vaccines and new and improved therapies for the disorders and their symptoms. Yet, how many of the tens of millions of people worldwide with one of these diseases are aware of the research that is taking place? And, how many are actively participating, supporting and/or promoting the cause? The thousands, perhaps tens of thousands, of patients and families who are, are making a *big* difference; however, when considering the needless suffering, disability and loss of life that occurs every day due to these diseases, progress cannot come fast enough. It is one thing to hope, but quite another to continue that hope while getting involved. Only collectively can we bring change.

Advances in Research

One of the hottest topics globally in immunological research is the establishment of a connection between immune deficiencies and specific genes. In fact, the Centers for Disease Control and Prevention (CDC) reports that many PIDDs are due to a single gene defect. As a result of improved and expanded gene sequencing, “there are about 160 to 165 mutations in different genes that have been identified in PIDD,” explains Dr. Troy Torgerson of the University of Washington and Seattle Children’s Hospital, and co-director of the hospital’s Immunodeficiency Molecular Diagnostics Laboratory, who is participating in the clinical care of patients with immune deficiency and autoimmune disorders. “We’re learning why particular genetic defects make patients susceptible to certain infections or make patients develop autoimmune diseases. ... You can now sequence someone’s entire genome for a relatively reasonable cost, and it’s getting cheaper and cheaper.”

Dr. Torgerson is excited that, along with more specific diagnoses, comes the prospect of more effective treatments and potential cures for PIDDs and autoimmune diseases. In his estimation, one of the fruits of gene sequencing that is on the not-too-distant horizon involves stem cells that are taken from a patient’s own peripheral blood, bone marrow, preserved umbilical cord blood or other potential sites in their body. “With traditional gene repairs, in the

neighborhood of only one in a million cells is repaired and then put back," Dr. Torgerson explains. "But, it's not enough cells to completely fix the problem." The body's stem cells are unique because they are able to renew themselves by dividing and differentiating into specialized cell types, like the blood cells that are so important to our immune system. "We would like to take stem cells from a patient, select and repair them [with a normal copy of the defective gene], grow and expand them up in the lab and then put them back in the patient," he adds.

Whereas current therapies for PIDDs and autoimmune diseases focus on controlling symptoms, stem cell transplantation may offer a future cure for many of the conditions. The technique still faces several hurdles in the laboratory before it can become common practice in the clinic. However, society's best and brightest scientists are working hard to develop stem cell transplantation and other disease-specific therapies, and to both reduce the cost and increase the effectiveness of current treatments like immunoglobulin infusions. The greatest hurdle to the rate and height of their success may not be limitations on the human mind or the bounds of science but, instead, limitations on the pocketbook.

Funding Research

An overwhelming majority of funding for PIDD and autoimmune disease research (estimated by Dr. Torgerson to be at least 95 percent) comes from the federal government through grants approved by the National Institutes of Health (NIH). However, Dr. Hans Ochs, also of the University of Washington and Seattle Children's Hospital, who is widely considered a pioneer in the field of PIDD research and is a primary investigator for the NIH contract supporting the United States Immune Deficiency Network (USIDNet), elaborated that getting projects approved is "highly competitive" and "presently very difficult, with less than 10 percent being funded." During the Clinton years, the administration had a goal of doubling the NIH budget, with 20 percent to 25 percent of projects being funded. Increased funding continued into the first or second year of the Bush presidency, though budgets have experienced only modest, if any, increases since that time. "Consequently," declares Dr. Torgerson, "a lot more time is put into grant writing that could be used

otherwise. People are trying to find ways to scrape by and keep their labs open and keep going."

As disappointing as the current funding challenges may be, it's important to realize how far the funding of PIDD and autoimmune disease research has come. According to Dr. Ochs, "As PIDD became more prominent, the patient organizations started to make their voices heard, and the experts in PIDD banded together. There were very few grants from NIH supporting research directly related to PIDD until some 15 years ago when NIH started to support registries and began to notice that research fell behind in the U.S. compared with Europe." As a result, a task force recommended more funding and the USIDNet was charged with the tasks of providing small two- to three-year grants, establishing a new web-based PIDD registry, setting up a repository of DNA and cell lines and organizing meetings. Dr. Torgerson acknowledges one individual in particular, the late Dr. Josiah F. Wedgwood, MD, as a

powerful force behind specific funding opportunities for PIDD research. "Prior to his work with the NIH (as a program officer) in developing this specific area of interest, you would submit your grant to the general NIH and there wasn't much recognition," explains Dr. Torgerson.

Although most of the direct funding for PIDD and autoimmune disease studies comes from the government, several other organizations serve as significant sources of

monetary and non-monetary support for research. One group, the Clinical Immunology Society (CIS), just organized its first North American PIDD Conference in May 2010. The organization also is responsible for hosting an annual PIDD Summer School, which they began eight years ago, for future scientists who have a potential interest in clinical immunology. Companies that make immunoglobulin products also often give money to help fund research and to sponsor fellows in programs like the PIDD Summer School. And, other important organizations, including the Immune Deficiency Foundation (IDF), the Jeffrey Modell Foundation (JMF) and the American Autoimmune Related Diseases Association (AARDA), are nonprofit patient advocacy groups that provide invaluable opportunities for others to get involved in the promotion of PIDD and autoimmune disease research.

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Getting Involved

So, how can someone who doesn't put on a white lab coat to go to work each day make a difference in research? Fortunately, there are a number of opportunities to get involved, most of which don't include pulling out the checkbook.

One of the most important ways that anyone can advocate for research funding is to lobby their political representatives. "Patients can provide a tremendous role by getting to know their congress members and telling them their story, and letting them know what they struggle with and how medical research can really change their lives or has changed their lives," says Dr. Torgerson. "They [congressional representatives] have people from all sides telling them how to spend the money, and our voices need to be heard too. ... [They] need to hear about the importance of funding NIH and understand that for every dollar that they put in there, it generates more income in the long run because new discoveries generate new companies, new therapeutics and all kinds of things that generate jobs, etc."

The IDF and AARDA websites (www.primaryimmune.org and www.aarda.org, respectively) both have tools to help you contact your congressional representatives, as does the *IG Living* website (www.igliving.com). The IDF even has a "Grassroots Advocacy Toolkit" that teaches the basics of letter and email writing, telephoning legislators, writing letters to the editor and other skills. According to its website: "You don't need to be a professional lobbyist to influence how policy and legislation are created. ... All you need is personal experience, factual information to back up your personal experience, knowledge of who the key decision-makers [are] and what is most likely to influence them. IDF can provide the factual and political information necessary for a successful advocacy campaign."

Another way to support research is to participate directly by filling out questionnaires and surveys or taking part in clinical trials. The IDF, AARDA and JMF (www.info4pi.org or www.jmfworld.org) have online surveys, which can be completed anonymously, in support of gathering data that can be used for a number of purposes. The USIDNet (www.usidnet.org) also offers surveys and maintains a national patient registry for

the purposes of "providing a minimum estimate of the prevalence of each disorder in the United States, providing a comprehensive clinical picture of each disorder and providing a resource for clinical and laboratory research." To learn more about clinical trial opportunities in the U.S., visit the USIDNet or NIH-sponsored website at www.clinicaltrials.gov.

Finally, if individuals are able to donate money or participate in fundraising events, there are a number of options. For those interested in funding a specific area of research or a specific researcher, the best way to give may be directly to their institution. For instance, a gift fund can be given to a hospital and designated how it is to be spent. The JMF website has a map of the Jeffrey Modell Centers Network, a system of more than 50 diagnostic and research centers worldwide and physicians at 138 academic teaching hospitals and medical schools. At Children's Hospital in Seattle, the families and some of the older patients run a local immunodeficiency research guild.

They have several events each year to raise funds for research at the hospital. Activities in the past have included barbecues, Mariners baseball games and a day at the zoo. Individuals also can contribute to one of the aforementioned nonprofit organizations. Although there may not be a lot of input on how the money gets spent unless contributing a substantial amount, the foundations do lots of wonderful things with the donations.

"Just get involved," says Dr. Torgerson. "I know that it's difficult

and it's just one more thing to add to a busy schedule, but these things won't move forward unless people get involved. We are so incredibly grateful for the willingness of people to give of themselves whether it is research samples or money, or time to run a guild or participate in events, or write to their congress members."

There is power in numbers, and our voices need to be heard. ■

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