

IDF ADVOCATE

THE NATIONAL NEWSLETTER OF THE IMMUNE DEFICIENCY FOUNDATION



Immune Deficiency Foundation Mission:

The national patient organization dedicated to improving the diagnosis, treatment and quality of life of persons with primary immunodeficiency diseases through advocacy, education and research.

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Newborn Screening for SCID *The Time is Now*

by J. Doug Gill

“This,” Carol Ann Demaret said to me during a recent phone conversation, “is a dream come true.”

The ‘this’ to which Carol Ann refers is the unanimous January 21, 2010 vote by the Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) to add screening for Severe Combined Immune Deficiency or SCID - commonly known as bubble boy disease – to the core panel for universal screening of all newborns in the United States.

For Carol Ann to have such a vested interest in the ACHDNC recommendation comes as no surprise: in 1971 she gave birth to David Vetter, the young man whose compelling “Boy in the Bubble” story catapulted SCID into the public consciousness.

Since his passing in 1984, no one could have predicted it would take more than a quarter century to go from David’s story to the recent Advisory Committee recommendation to screen newborns for this devastating – and often times deadly – disease.

With Severe Combined Immune Deficiency, affected infants lack T lymphocytes, the white blood cells that help resist infections due to a wide array of viruses, bacteria and fungi. Babies with SCID appear healthy at birth, but without early treatment, most often by a bone marrow transplant from a healthy donor, these infants cannot survive.

In January 2009, the Immune Deficiency Foundation (IDF) initiated a survey in conjunction with the SCID Forum and SCID Angels for Life, that reaffirmed the necessity for early diagnoses and treatments. Taking a national sample of 156 SCID families, IDF’s information gathering revealed that



James and Stacey Barrett with their son Liam

those patients who were diagnosed early and treated by 3.5 months had a 91% survival rate; those treated after 3.5 months had a 76% survival rate. Additionally, newborns that survived were found to have begun treatments at an average age of 29 weeks; where those newborns that perished began treatment at an average of 58.

Marcia Boyle, president and founder of IDF, presented her landmark survey data to a meeting of the ACHDNC in February 2009.

“This survey demonstrates that timely diagnosis and treatment makes the difference between life and death for these children,” Boyle testified. “If SCID is recognized only

(Continued on page 4)



Show your Stripes!
Request a free
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increase awareness of
primary immunodeficiency
diseases.
Call 800-296-4433
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A maximum of 5 buttons per household please.

IDF Advocate

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Primary Immunodeficiency Awareness Month April 2010

This April, the Immune Deficiency Foundation (IDF) is promoting Primary Immunodeficiency Awareness Month by telling the world to THINK ZEBRA!

In medical school, many doctors learn the saying, “when you hear hoof beats, think horses, not zebras.” Most physicians are taught to focus on the likeliest possibilities when making a diagnosis, not the unusual ones. However, sometimes physicians need to look for a zebra. Primary immunodeficiency patients are the zebras of the medical world. So IDF encourages all to THINK ZEBRA! this April!

In the United States, approximately 250,000 people are diagnosed with primary immunodeficiencies and thousands more go undetected. These diseases are chronic illnesses caused by hereditary or genetic defects in the immune system in which part of the body’s immune system is missing or does not function properly. These diseases are not contagious.

There are over 150 different primary immunodeficiency diseases and they affect people differently. For some, the body fails to produce any or enough antibodies to fight infection, while for others; the cellular defenses against infection fail to work properly. Throughout their lives, people with primary immunodeficiencies are more susceptible to infections, endure recurrent health problems and often develop serious and debilitating illnesses.

The Immune Deficiency Foundation has some great tools for you to help promote primary immune deficiency awareness in your community. Find our THINK ZEBRA! Press Kit, THINK ZEBRA! Fundraising Kit, advocacy tools and more at our Website www.primaryimmune.org. With your continued help and support, we can create awareness during the month of April and year round. We thank you for your efforts!



Lights, Camera, Action!

Look for the debut of the new *IDF Common Ground Talk Show* beginning in April 2010 on the teen and young adult IDF Common Ground site. A new featured video will be added every few weeks.

Talking about and dealing with certain issues that affect our young adult community can be difficult at times, so our teen hosts Isaac Antilla, Shane Oravetz, and Courtney Palmer, joined by a studio audience and licensed clinical social worker, Jodi Taub, have tackled a wide range of topics. The *IDF Common Ground Talk Show* discusses the facts, myths, hopes and fears that teens and young adults living

with primary immunodeficiency diseases face, while celebrating accomplishments, achievements and the ability to live life as a “normal” teenager. The episodes were created and produced with teens and young adults in mind and the segments will cover topics and issues that affect teens and young adults living with primary immunodeficiency diseases.

The videos will include the following topics:

- 0 Dealing with Transitions
- 0 Treatment Options, the Fear of Getting Sick and Missing School
- 0 Peer Pressure and Making Good Choices
- 0 Making Friends and When to Tell a Friend about your PIDD?
- 0 Dating, Privacy and Personal Boundaries

Make sure to visit IDF Common Ground (<http://www.idfcommonground.org>) in April to check out the first episode of the *IDF Common Ground Talk Show*!

IDF Common Ground is sponsored by an unrestricted educational grant from Baxter Healthcare.

THINK ZEBRA! and join us IDF Retreats

For Persons Living with Primary Immunodeficiency Disease & Their Families

**Doral Arrowwood
Conference Center**
Rye Brook, NY
June 25-27, 2010



Hotel Kabuki
San Francisco, CA
August 13-15, 2010

In medical school, many doctors learn the saying, "when you hear hoof beats, think horses, not zebras" and are taught to focus on the likeliest possibilities when making a diagnosis, not the unusual ones. However, sometimes physicians need to look for a zebra. Primary immunodeficiency patients are the zebras of the medical world. So IDF says **THINK ZEBRA!**

Join us for a retreat weekend of discovery, learning and fun!

IDF Retreats are for everyone in the primary immunodeficiency community – patients, parents, siblings, children and partners. IDF Retreats provide presentations by leading immunologists with the latest information about primary immunodeficiency diseases. Life management and everyday concerns will be featured in panel discussions led by experts in their fields. It is an atmosphere for people to build relationships with others who share common experiences, therapies and feelings. Come connect with the IDF community and have some fun while developing better approaches to live with primary immunodeficiency.

For Adults

- Learn more about primary immunodeficiencies, your immune system, and immunological testing
- Personalize your experience by choosing the Cellular, Combined & Phagocytic Defects or Antibody Production Defects options
- Gain knowledge about therapies – immune globulin replacement, antibiotic and antifungal therapies
- Find out how to manage living with a chronic illness, as well as managing chronic sinusitis and GI issues
- Get guidelines on working with your health insurance provider and share coping tips with your peers
- Take advantage of the *Ask the Doctor* session and get your questions answered

For Youth

- **Teen Escape** (ages 13 - 18 years) This activity-packed, fun-filled program is designed for teens to promote friendship, build leadership skills, develop coping skills and learn about primary immunodeficiency diseases from medical experts.
- **Kids Club** (ages 6 - 9 years) and **Tween Scene** (ages 10 - 12 years) These groups will enjoy crafts, games, and learn about their immune system and how to take care of themselves in between other fun activities.
- **Childcare** (6 months - 5 years) is available for the younger children.

And much more!

We hope you will treat yourself and family to one of these great weekend retreats!

Speakers*

Doral Arrowwood Conference Center

Shradha Agarwal, MD
Mount Sinai School of Medicine

Tony Bonilla, MD, PhD
Boston Children's Hospital

Rebecca Buckley, MD
Duke University Medical Center

David LaRosa, MD
University of PA, School of Medicine

Heather Lehman, MD
Women and Children's Hospital of Buffalo

Luigi Notarangelo, MD
Boston Children's Hospital

Hotel Kabuki

Laurence Cheng, MD
University of California, San Francisco

Mort Cowan, MD
University of California, San Francisco

Jennifer Puck, MD
University of California, San Francisco

E. Richard Stiehm, MD
UCLA David Geffen School of Medicine

Troy Torgerson, MD, PhD
Seattle Children's Hospital

Registration Information - New lower rates in 2010!

- **Individual Registration - \$100**
(1 person)
- **Family Registration - \$150**
(2-4 persons)
- **Family Registration - \$250**
(5-8 persons, includes additional hotel room)

Registration is all-inclusive and covers your hotel room for 2 nights, meals (Friday dessert reception, Saturday breakfast, lunch and dinner, Sunday breakfast) and educational and recreational programs.

For online registration, go to the IDF Web site, www.primaryimmune.org. For more information, please contact IDF at 800.296.4433 or retreats@primaryimmune.org.

Space is limited and will be assigned on a first-come, first-served basis so don't delay!

The IDF Retreats are generously sponsored by Baxter, CSL Behring, IgG America/ASD Healthcare, Octapharma, Talecris Biotherapeutics and Walgreens — IG Therapy Program.

Newborn Screening for SCID:

as a result of infection, it is often too late for effective treatment.”

“As the parent of a child who was diagnosed with SCID only after becoming critically ill, I am immensely pleased with the recommendation to make SCID screening a universal newborn test,” said Barbara Ballard, founder of the SCID Family Network and a member of IDF’s Board of Trustees.

Ballard’s son, Ray, was born with SCID, but the disease went undiagnosed for nearly 11 months. Now 16-years-old, her son is hearing impaired, has just 60-percent lung function and is fed via enteral and parenteral means.

“His long term, ongoing health issues are not a result of having SCID,” Ballard explained, “but because of the delay in diagnosis until he was critically ill with multiple infections.”

Ballard was also a driving force in the creation of IDF’s “SCID Initiative”, a program that supports education, awareness, diagnosis, screening and the search for a cure for this damaging disease.

“Unfortunately, I have to say there is still an incredible lack of awareness and knowledge when it comes to SCID,” says Heather Smith, co-founder of the non-profit foundation SCID Angels For Life, and member of the IDF SCID Initiative. Sixteen years ago, Heather lost her then seven-month-old son, Brandon, to SCID.

Smith’s second son, Taylor, was not only diagnosed with SCID in-utero, but also received bone marrow transplants during gestation. Today, Taylor is a thriving, healthy teenager.

“Unfortunately, a tragedy had to happen in our family, and in so many other families, in order for us to know about this

devastating disease,” Heather says, repeating the testimony she gave to the ACHDNC in 2008. “If Brandon could have been diagnosed right after birth - before the onset of a life threatening illness - his pain and suffering could have been stopped, his life could have been saved.”

In the years since David Vetter’s death, researchers, famed medical centers and world-renowned immunologists have worked diligently to both refine treatment options for children with SCID, and to develop a reliable, consistent testing process at birth.

Dr. Jennifer Puck, Professor of Pediatrics at the University of California, San Francisco and an expert in the field, nominated SCID for consideration by the Advisory Committee after pioneering development of a test that can detect SCID in the dried blood spot filter cards that are currently collected from all babies to screen for a variety of inborn disorders.

“Although this recommendation has been in development for two years,” said Dr. Amy Brower, researcher and former ACHDNC committee member, “and it may take several more years to implement screening in all 50 states and US territories, we must work to reduce the time from this recommendation to the widespread adoption of testing and treatment.”

Medical professionals cite numerous obstacles in this bid to bring testing to every state – among them a lack of funding and financial support for labs; each state’s willingness to adopt such procedures individually or an unwillingness to consolidate testing labs on a regional basis; a lack of awareness regarding the disease; and insurance company inclination to pay for it.

Still, celebrated immunologists such as Dr. Rebecca H. Buckley, Duke University Professor of Pediatrics and Immunology, emphatically contend that SCID-screening for newborns is more cost effective than the costs of testing, hospitalizations, and treatment before and after diagnosis.

“Such a blood test could pick up children with SCID that would not be apparent until the child developed an infection,” Dr. Buckley explained. “A simple blood test could allow us to treat, and most likely cure, SCID in an infant at a reasonable cost. If found later, less effective treatment can run into the millions.”

In January 2008, Wisconsin became the first state to screen all newborns for SCID, with Massachusetts following suit in February 2009. Hopes are now high that the ACHDNC action will encourage other states to pursue those models.

“SCID can now be cured with a relatively simple bone marrow transplant if diagnosed in the first weeks or months of life,” said Dr. John “Jack” Routes, medical director of Allergy and Clinical Immunology at Children’s Hospital of Wisconsin. “We believe routine screening of all newborns



Pictured are Taylor Dahley (son of Heather Smith) and David Singh. Both boys have SCID and lost an older brother to SCID. Because of that, they were diagnosed early, transplanted, and are both living healthy, productive lives.

The Time is Now

Continued from page 1

will find more SCID babies whose disease in the past may have been masqueraded as unexplained deaths in early infancy. If their problem had been identified correctly, early on, and treated properly through a screening program, those babies might be alive today.”

Adding to the urgency of nationwide adoption of SCID-screening is the growing risk posed by a vaccine that is routinely given to children beginning at 2-months-of-age: rotavirus.

The preventative rotavirus vaccine, used to treat the most common cause of gastroenteritis (diarrhea and stomach flu-like symptoms) in young children, has proven effective in reducing not only the number of infections in newborns, but has also been instrumental in reducing the number of hospitalizations and deaths associated with infection.

However, when the rotavirus vaccine is given to an infant who has an undiagnosed immune deficiency, like SCID—and therefore little or no immunity to any pathogen – there is an unforeseen risk for prolonged rotavirus infection, and even death, and a probability for other simultaneous and similar illnesses.

And while no one is suggesting the elimination of the rotavirus vaccine, advocates for screening say this aspect strongly reinforces the need to adopt widespread screening procedures for immune deficiencies before any vaccination series is begun.

Stacey and James Barrett, two young parents from Oregon, know first-hand the importance of a national-adoption of these procedures.

And if one doubts the power of advocacy, education and awareness, consider it was IDF’s “SCID Initiative” that brought the Barrett family from Oregon to testify before the ACHDNC.

The Barrett’s son, Liam, would have celebrated his first birthday on January 30 had he been born in Boston or Green Bay, but he entered the world in Oregon, a state that doesn’t test for SCID at birth.

“If our family were living in Wisconsin or Massachusetts at the time Liam was born,” Stacey Barrett testified before the ACHDNC panel in January, “Liam would have been diagnosed with an immune deficiency. If that were the case... statistics indicate our son would still be alive.”

Liam died on August 17, 2009, having endured multiple hospital stays, innumerable tests, four infections, a feeding tube, and ultimately a bone marrow transplant.

Describing her son as a “casualty in bureaucracy,” Stacey Barrett used her testimony to reiterate the desperate need to act – and to act quickly.

“Our family’s journey with SCID began when Liam was

admitted to the hospital for ‘failure to thrive,’” Barrett testified. “That was 8 months after this committee voted to delay acceptance of universal newborn screening for SCID, 10 years after the American Academy of Pediatrics (AAP) called for national newborn screening standards, 6 years after Dr. Buckley testified at the first meeting of this committee that SCID was a pediatric emergency and should be included in the uniform panel, 2 years after SCID was nominated and 18 months after Wisconsin began screening.”

And 26 years since the death of “the boy in the bubble.”

For now, SCID families and the tight-knit community of those touched by immune deficiency disease face yet another delay. The Advisory Committee’s policy recommendation will now be presented to Kathleen Sebelius, Secretary of Health and Human Services. Sebelius has 180 days to consider and respond to the committee’s proposal.

“Given this data,” Marcia Boyle said, citing the medical community’s advisement, the IDF survey and years of patient and family testimony, “It would be inconceivable to me that Secretary Sebelius would not agree with the committee.”

“The Advisory Committee has taken a significant step forward for the primary immunodeficiency community,” Boyle added, “and I want to thank our IDF SCID Initiative, the efforts of the Jeffrey Modell Foundation, Wisconsin and Massachusetts for pioneering the screening, the many immunologists who have advocated for this and most importantly, all the families who have lived with SCID and made the implementation of newborn screening their dream.”

For that dream to be realized, the momentum sparked by the committee’s action must be sustained in order to bring this screening program to all 50 states.

“People tend to call this disease rare,” Carol Ann Demaret said emphatically. “I say this disease is rarely diagnosed.”

But thanks to the ACHDNC’s recent recommendation, and a rising tide of support and advocacy, the ripple that began in David Vetter’s bubble is slowly showing signs of reaching tidal wave proportions.



Ray Ballard (Barb Ballard’s son)

Autoimmunity in Patients with

By R. Michael Blaese, MD

We are fast approaching the 60th anniversary of the report by Dr. Ogden Bruton of a patient with agammaglobulinemia, which is generally credited as the beginning of the modern era in the study of Primary Immunodeficiency Diseases (PID). Over the next couple of decades another 15 or so additional diseases were added to the list and physicians were beginning to learn some of the details of these rare disorders. It is common for the most severe forms of many diseases to be recognized first and this is certainly true of the primary immunodeficiency diseases. As greater experience was gained and more patients were studied, the range of defects in immunity and the breadth of symptoms that occurs in various disorders became more apparent.

The single feature that was found in all of these early patients was their increased susceptibility to infections.

However, it also became obvious that not all patients seemed to be susceptible to infections with the same kinds of microorganisms. In some, the predominant infections were in the respiratory tract from high-grade encapsulated pathogens like the pneumococcus and H. flu. Other patients were seen to have infections that often involved other areas of the body with organisms that did not commonly cause disease in the general population like certain types of fungi, viruses and even parasites. Just as the kinds of infections experienced by different groups of patients were unique, the medical community gradually began to recognize that these patients with immunodeficiency seemed to experience a number of disorders that were associated with autoimmune attacks. Here was a supreme paradox, individuals whose immune system dysfunction was severe enough that they were unable to defend themselves from the microorganisms with which we all live

- were themselves being attacked by their own immune systems gone awry.

Subsequently, as more and more distinctly different immunodeficiency disorders were discovered the realization that autoimmunity was common in some of these disorders while not being seen in others was also recognized. Recently a major review of immunodeficiency disorders by the International Union of Immunology Societies (IUIS) reported on a classification of immunodeficiency diseases that contained over 150 different disorders. Remarkably, over 35% of these conditions had autoimmunity as a part of their clinical disorder. Two of the eight classes of PID are not associated with an unusual frequency of autoimmune diseases, but up to 60% of the disorders in the other 6 classes of immunodeficiency are associated with symptoms of autoimmunity sometime during the course of the illness. Not every patient with most PIDs will experience autoimmunity,

Class of Immunodeficiency (Examples of types of disease in each class)	Number of disorders with autoimmunity		
	Total Number of Diseases in that Class of PID		
Combined T and B cell immunodeficiency syndromes RAG 1-2, Omenn syndrome, Hyper IgM	11 of 27	or	41%
Antibody deficiency syndromes CVID, Selective IgA deficiency	9 of 23	or	39%
Other well defined Immunodeficiencies Wiskott Aldrich, Mucocutaneous Candidiasis	4 of 16	or	25%
Diseases of Immune Dysregulation ALPS, Familial Hemophagocytic Lymphohistiocytosis	10 of 17	or	59%
Congenital defects in Phagocyte number or function CGD, congenital neutropenia, LAD	0 of 25		0%
Defects in Innate Immunity IRAK4, MyD88	0 of 9		0%
Autoinflammatory Disorders TRAPS, PAPA syndrome, CINCA syndrome	6 of 10	or	60%
Complement Deficiency C1-C8 deficiency	13 of 24	or	54%

Primary Immunodeficiency

but the high incidence seen across many different diagnoses certainly indicates that this problem is more than just coincidence and that it has major implications for the causes of autoimmune phenomena and impacts on the kind of treatments that can be used. The irony of the concurrence these two disorders is that the development of autoimmunity may require the use of immunosuppressive treatments in these patients who already have immune deficiency.

Autoimmune Conditions

The autoimmune conditions seen in patients with PIDD include examples of red blood cell destruction (autoimmune hemolytic anemia), platelet destruction (autoimmune thrombocytopenia) and even destruction of blood granulocytes (autoimmune neutropenia). Other examples include various forms of joint destruction (arthritis), inflammation of the eye (uveitis), inflammation of the blood vessels of the skin, heart or brain and other organs (vasculitis). Other autoimmune disorders that patients may experience include inflammatory bowel disease (Crohns Disease and ulcerative colitis), Sjogrens syndrome, lymphoid interstitial pneumonitis, thyroiditis, dermatomyositis - and the list goes on. Autoimmunity is found to occur in about 20% of patients with CVID, 50% of individuals with the Wiskott-Aldrich syndrome and essentially all patients with ALPS.

Immunodeficiencies and Autoimmune Diseases Scientific Colloquium

As a recognition of the growing importance of these autoimmune conditions to some patients with

primary immunodeficiency diseases, the Immune Deficiency Foundation (IDF) co-sponsored with the American Autoimmune Related Disorders Association (AARDA) a scientific colloquium held in Baltimore on November 13-15, 2009 on the topic of Immunodeficiencies and Autoimmune diseases

The Colloquium was a three-day meeting bringing together scientific investigators from diverse backgrounds and interest for discussion and conversation. Featuring a multidisciplinary approach, the meeting addressed the relationship between immunodeficiencies and autoimmune diseases through the following:

- Focus on the specific mechanisms by which immune dysregulation, both over and under, that might relate to the development of autoimmune disease and treatment failure.
- Identify new opportunities to study genetic mechanisms that help to explain the connection between immune deficiencies in autoimmune disease.
- Discuss research strategies to initiate collaborative research in the area of immunoregulation in autoimmune diseases and immunodeficiency.
- Adopt a multidisciplinary approach in meeting specific aims by bringing together specialists from many disciplines, including but not limited to immunologists, infectious disease specialists, and rheumatologists.
- Select critical research areas that represent unmet needs or are presently under-explored.

The Colloquium was attended by 88 scientists and physicians from 7 countries. Twenty eight speakers drawn from the United States, Canada, United Kingdom, Australia, France and Israel addressed a wide range of topics centered on the mechanisms of autoimmune disorders and how they might be influenced by immunodeficiency.

A common feature of the disorders characterized by increased autoimmunity is that they have defects in T and/or B lymphocyte function or activation that results in problems appropriately regulating their attempts to generate effective immune responses. This observation is supported by the finding that patients with defects that only involve the phagocytes, like chronic granulomatous disease (CGD), do not experience more autoimmunity than the general population. Studies of the processes of immune regulation have been greatly aided by the development of animal models in which the details of these immunoregulatory interactions can be carefully pieced together. Several of the presentations at the Colloquium addressed these immunoregulatory mechanisms and new insights are fostering rapid increases in understanding that should aid the development of effective new treatments for several of these autoimmune problems. The meeting should help to generate increased interactions between different groups of investigators and we hope to hold a follow-up conference in about 2 years to help maintain momentum in the development of treatments for autoimmune disorders in patients with PIDD.

Dr. Blaese is an active member of the IDF Medical Advisory Committee and the Consulting Medical Director for IDF.

Steps for Applying for

By William P. Leach

To paraphrase something that John Lennon once wrote, “Disability is what happens to you while you’re busy making other plans.” Most of us plan to work and live as others do, and then we are unexpectedly confronted with the possibility that chronic illness may lead us to choices we did not expect.

For anyone who is starting to consider the possibility that they may need to apply for disability benefits from the Social Security Administration, there are a number of things you may want to consider.

Considerations

The first thing to think about is your family’s financial situation. Both federal disability programs require that the person filing for benefits have no more than a very modest earned income (in 2010, less than \$1000 per month in gross earnings). Yet the process of being approved for benefits can be lengthy, stretching into two years or more if you have to appeal the denial of your claim.

The question becomes how do you survive financially while fighting for your benefits? The answers vary. Many people have disability insurance through their employer that can fill the gap until Social Security approves the claim. Others rely on their savings, their spouse’s income or support from other family members. Whatever your circumstances are, it is important to plan for the possibility that your disability claim may not be approved right away.

Another critical issue is maintaining health care coverage. Since many of us have our health insurance through our employer’s group health plan, once we stop working we need to look at how coverage will be



preserved. A federal law known as COBRA lets you keep your current coverage for a period of time after you stop working. However, the premiums can be quite expensive, particularly with significantly reduced family income. For this reason, you will want to explore all possible sources of health care coverage.

Talk With Your Physician

If you are considering applying for disability, it is a good idea to discuss this with your doctor. Often, he or she is in the best position to advise you as to whether your medical condition is likely to prevent you from working, and if so, for how long. It is also important that they understand that their medical records will carry great weight with Social Security in determining your disability claim. A letter from your doctor describing how your immune deficiency interferes with your ability to work can be very helpful as well.

You should be thorough in describing your limitations to your treating physicians and in the forms that you submit to the Social Security Administration. Activities that

you can no longer do, that now take longer to do or for which you now require help to do should be explained completely. Your claim is most likely to be approved when your medical care professionals document your restrictions in detail and support them with appropriate clinical data.

One of the important things to consider is the affect that treatment for your condition has on your ability to function. For example, if you are receiving intravenous immunoglobulin, you want to document how often you receive your infusions, how long it takes to administer, and any side effects and their duration after being infused. A related issue is the fatigue that many people experience in the week leading up to their next infusion. Being able to document these limitations can make it much easier for Social Security to adjudicate your claim favorably.

The other obvious issue is the frequency, severity and duration of infections that continue to occur despite compliance with prescribed therapy. One thing to think about is that most employers require regular attendance on a set schedule. To the extent that immune deficiency and its treatment would cause you to miss more than two or three days a month you are likely to be deemed disabled by Social Security. Statements from your treatment sources establishing problems with maintaining regular attendance can be very helpful.

Filing for Disability Benefits

Generally, you should file for disability benefits as soon as you feel you may no longer be able to work. Any delay in filing could result in receiving less than the full amount of benefits to which you might otherwise be entitled. To start an application for disability benefits from

Disability

Social Security Disability or Supplemental Security Income, you can:

- » Call the Social Security Administration toll-free at **(800) 772-1213**
- » Apply online at **www.ssa.gov**
- » Call or visit your local Social Security office.

There are a number of forms that Social Security will ask you to complete, but the date you first contact them will be treated as your protective filing date for purposes of determining when your benefits may begin.

If you do attempt to work while your disability application is pending, you will need to maintain careful records of your monthly gross earnings as well as where you work, when you started and stopped

each job, and your hours and duties. The rules governing work attempts while applying for disability can be complex, so having this documentation handy will make it easier to determine which rules apply to your situation.

It is not unusual to have your initial application for disability benefits denied. Many of these claims are later approved as part of Social Security's administrative appeal process. If you disagree with a disability determination, you should seriously consider appealing that decision. Our specialists at PSI-ACCESS are always happy to assist you with any questions you may have about federal disability programs. Our toll-free number is (888) 700-7010.

William Leach is a licensed Florida attorney from Tampa who has represented hundreds of people from all over the country on their claims for disability benefits before the Social Security Administration's Office of Disability Adjudication and Review. He is employed by PSI-ACCESS, a national non-profit organization that offers representation on disability claims to people with certain rare chronic conditions, including primary immune disorders. He has worked for ACCESS since 1995.

Patient Services Incorporated (PSI) is a premium and co-payment assistance foundation that has been helping people with certain chronic conditions for over 20 years. ACCESS became part of PSI in 2009. PSI-ACCESS services are provided at no cost to their clients.

Operation Outreach

Operation Outreach offers educational meetings in areas of the country where patients and families living with PIDD have not had the opportunity to experience an IDF patient meeting. For many attendees, these meetings offer the first opportunity to meet with other individuals affected by primary immunodeficiencies and learn about treatment options and life management skills. IDF Operation Outreach Meetings are sponsored by an unrestricted, educational grant from CSL Behring.



John Sleasman, MD, PhD and Elyse Murphy, RN take a break from presenting at the IDF Operation Outreach meeting in Gainesville, FL.



Carla Duff and son Steven greeted attendees to the Gainesville, FL IDF Operation Outreach meeting on February 13th. Carla is a member of the IDF Nurse Advisory Committee.

Faye Zwerling: *Defining Determination*

by J. Doug Gill

Challenges and struggles are part and parcel of life. The human spirit, for the most part, is strengthened not by simply meeting those challenges, but by the determination needed to overcome them.

“I’m not sure if ‘determination’ is the most accurate description,” Faye Zwerling tells me, “I’d go with ‘survival instinct.’ Without that, you’re not going to make it.”

Mrs. Zwerling, 78-years-young and a mother of three, doesn’t just talk of such resilience; she considers herself “living proof” that resolve and purpose are the secrets to longevity – even in the face of a primary immune deficiency disease.

Faye was born in 1931, years before the DPT (diphtheria, pertussis and tetanus) vaccine was given to newborns. As a result, she developed pertussis – or whooping cough – at a time when nearly 150,000 U.S. infants were diagnosed with the disease each year.

“I had pneumonia so many times that I stopped keeping track,” she confides, adding that she is also a cancer survivor and had part of her lung removed due to bronchiectasis, a condition often caused by recurrent inflammation or infection of the airways.

Still, Faye didn’t just cope with her health challenges; she mustered the tenacity needed to overcome them. After giving birth to three children, Mrs. Zwerling managed to return to college and earn a degree in social work, in spite of continued hospitalizations and what she called “being a guinea pig for all the latest and greatest antibiotics.”

But in 1961, Faye’s health picture became quite a bit clearer – from a diagnostic standpoint – when she was diagnosed with hypogammaglobulinemia, a primary immune deficiency of the main antibody defense against bacteria

“When I was diagnosed with PIDD,” Faye explains, “I was one of only two people at that hospital (National Jewish Hospital in Denver) that had been diagnosed with a primary immune deficiency. It was a disease hardly anyone knew anything about. I decided then that I would give back in any way possible to help people – especially children – who are trying to live their lives with chronic health problems.”

What followed were not only 45 years of monthly IMIg treatments (intramuscular immunoglobulin therapy), but also a renewed drive to live life to the fullest.



Faye Zwerling with her big brother, comedic actor, Harvey Korman

A few years ago Faye’s IMIg shots were replaced by IVIg (intravenous immunoglobulin) treatments, a move that to many patients proves far less painful.

“I had heard that before I started doing the IVIg,” she deadpans, “but I didn’t find the IMIg all that painful. You see, honey, I have been blessed with a big, round, firm, fully padded butt my whole life. Those ‘muscle shots’ never really were a problem.”

Today she gives back by volunteering her time reading to Las Vegas elementary schoolers, a task that – for a woman whose quality of life depends on an avoidance of

bacteria – borders on the precarious.

“Oh yeah, I think about what a breeding ground for germs that schools and schoolchildren can be,” she laughs. “So when the whole H1N1 scare arose I rushed to my doctor and got a flu shot – which, of course, made me sick.”

Spend a few minutes with Faye Zwerling and you’ll find yourself charmed by her sharp wit and sparkling sense of humor. And that should really come as no surprise: Faye is the younger sister of famed comedic actor Harvey Korman.

She and I spoke on what would have been the late comedian’s 83rd birthday.

“Harvey worked very hard to reach the level of success he enjoyed,” Faye says quietly, and when pushed about her desire to follow in her brother’s footsteps admits a certain fondness for stage and screen.

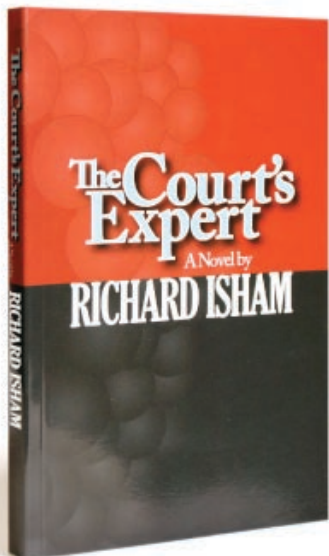
“I always wanted to be an actress,” she says confidently, “but I was too sick as a child. I was in and out of hospitals so much I barely made it through school, let alone any extracurricular activities.”

I bring up her ‘determination’ one more time, hoping for a concession on an apt word used to describe her life.

“Harvey used to tell me I had more determination than brains,” she chuckles, “but the way I see it is I’m just going to keep plugging away at everything and refuse to let anything deter me.”

Okay, Faye, we’ll go with firmness of purpose and unyielding resolve, but we won’t mention you just described the dictionary definition of ‘determination.’

A 'Novel' Approach to CVID



by J. Doug Gill

"I think each patient develops an individual philosophy," author/lawyer Richard Isham told me. "Once you're diagnosed, your disease is never off your mind."

Isham, after what he calls a "normal, healthy childhood," and after suffering from just "a couple of colds per year" well into adulthood, was diagnosed with common variable immune deficiency (CVID) 15 years ago.

"I was always considered a bit of an oddball anyway," he admits proudly, "My CVID just gave me another stripe."

The self-proclaimed "oddball" is celebrating his 44th year practicing law, and he recently returned to the Tulare County, California, deputy district attorney's office, where he currently serves as a consumer protection advocate.

Drawing from both his extensive legal knowledge and his understanding of CVID, Richard has just released his first novel. "The Court's Expert" not only includes intriguing characters and a gripping storyline, but also features a central character whose health condition – a primary immune deficiency disease – complicates the tale of a tangled legal system.

"I really just wanted to tell a story of 'junk science' testimony during the trial process," Isham explained, "but when one goes in for treatment every 28 days for 15 years, one tends to accumulate a lot of data."

That data not only led to the inclusion of a PIDD patient in Isham's book, but also to the efforts of the Immune Deficiency Foundation.

"I was impressed with IDF's work," Isham said. "When I first contacted them I was just a patient writing a book. But they were always encouraging," he laughed, "even after reading the rough draft."

That contact eventually led him to IDF president Marcia Boyle, who was so elated with his awareness-raising inclusion of a character with CVID that she penned a glowing paragraph for inclusion on the book jacket.

"I figured I had just enough juice left to make some sort of contribution," Isham added. "I hope this book will bring some much-needed attention to those affected by this disease."

You can order a copy of "The Court's Expert" by contacting Richard at rbisham@att.net

You can order a copy of "The Court's Expert" by contacting Richard at rbisham@att.net, or by visiting www.richardisham.com. To purchase multiple copies, please contact Richard.



Richard Isham

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