

IG LIVING!

IMMUNE GLOBULIN COMMUNITY

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August-September 2007

**Genes, Heredity and CVID:
What Are the Odds?**

**CVID and Pregnancy:
Do We Have a Problem?**

**Bone Marrow Transplant:
A Chance for Health**

A woman with dark hair tied back, wearing a pink long-sleeved shirt, is shown in profile from the chest up, breastfeeding a baby. The baby is positioned in front of her, and she is holding it against her chest. The background is a soft, out-of-focus green. The overall mood is calm and nurturing.

**Breastfeeding
and PIDD**

Welcome Baby Softly

A community service from FFF Enterprises and NuFACTOR, its specialty pharmacy services division

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Learn About New IG Living Readers Groups — Page 3

Our mission is to support the IG community through education, communication and advocacy

A community service from FFF Enterprises and NuFACTOR Specialty Pharmacy

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Publisher **Patrick M. Schmidt**

Editor **Kit-Bacon Gressitt**

Assistant Editor **Cheryl Brooks**

Senior Copy Editor **Zachary Pugh**

Contributing Editor **Dayna Fladhammer**

Copy Editor and Proofreader **Jackie Logue**

Creative Director **Allan Bean**

Circulation Director **Carrie Craig**

Advertising Director **Barry Pawelek**

Graphic Artists **Allan Bean**

Caroline Carlson

Kim Kinman

Contributing Writers

Ever Fecske

Dayna Fladhammer

Lauren Gerstmann, MPH

Kit-Bacon Gressitt

Cheryl Haggard

Mark T. Haggard

Kris McFalls

Zachary Pugh

Jessica Schulman, PhD, MPH, RD

Melissa Schweitzer, MS, CGC

Jim Trageser

Michelle Vogel, MPH

Shirley German Vulpe, EdD

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Please direct editorial, advertising and marketing communications to
41093 County Center Drive
Temecula, CA 92591

Ph: 800-843-7477 Fax: 951-699-9655

Email: editor@igliving.com

www.igliving.com

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About IG Living

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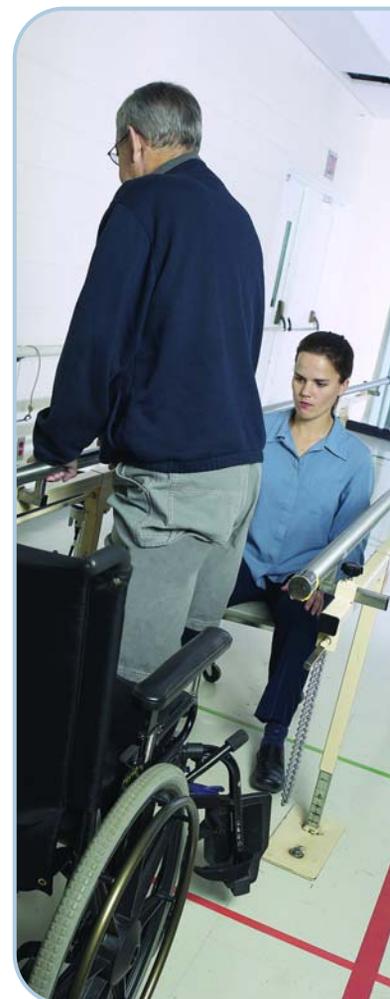
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Jeffrey Modell Cleveland Kids Day • Cleveland Metroparks Zoo
Saturday, September 1, 2007 • 10:00 am to closing
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Join an IGL Readers Group!

We hear from many, many patient and family member readers who would like to connect with others in their geographic areas—to share their experiences living with chronic diseases or maybe just to have a cup of coffee with folks who understand.

We can help you determine if there's a patient organization support group in your area or help you start an IG Living Readers Group.

To join a group or start one in your own area, visit www.igliving.com and click on IGL Readers Groups.

Ready for a Little Good News?



We have been reporting on the intravenous immune globulin (IVIG) access crisis since *IG Living's* inaugural issue in February 2006, and the news has not often been good. But in the last couple months, there have been two encouragingly bright lights.

First, the Department of Health and Human Services (HHS) released two studies on IVIG, one from the Office of the Inspector General (OIG) and the other from the Assistant Secretary for Planning and Evaluation (ASPE). The OIG and ASPE reports, along with some independent studies, confirmed what so many of our readers have been living: Since the January 1, 2005 implementation of the new IVIG reimbursement method in the Medicare Modernization Act, resulting in a reimbursement reduction that has since been emulated by private insurers, there has been a serious decline in quality of patient care.

The studies' findings include:

- Forty-two percent of Medicare patients receiving IVIG therapy in physicians' offices in 2004 had been shifted to other locations by the end of 2005.
- In 2006, only about half of hospitals and physicians could purchase IVIG below the Medicare reimbursement rate.
- An independent survey of hospital outpatient clinics and physicians found that 14.4 percent had already discontinued their IVIG infusion services and 45.4 percent planned to discontinue those services if reimbursement were not adequate.
- Another independent survey of hospital pharmacy directors showed that 32 percent of hospitals reported turning away patients for IVIG treatment at some point during 2006.
- Home infusion companies generally are not accepting new primary immune deficiency patients with only Medicare coverage, because healthcare providers are unable to acquire IVIG at prices at or below the reimbursement rate and they are not reimbursed for the infusion service.
- In 2006, *IG Living* was contacted by more than 150 readers (or their physicians) who reported reductions in frequency or volume of IVIG treatments, being at risk of losing access to IVIG therapy or actually being denied treatment.

With this new recognition that we do indeed have a national IVIG access crisis, we expect to see rapid movement

toward a solution, but IMIG community agreement on the solution is a prerequisite.

One patient organization did recently encourage a minority party member of Congress to sponsor a relevant bill, but it didn't garner much initial support—perhaps because it doesn't focus on restoring access to IVIG for all patients in all sites of care.

So, the second bright light is particularly hopeful: The new Alliance for Plasma Therapies announced its formation in May, with the mission to provide a unified, powerful voice of patient organizations, healthcare providers and industry leaders to advocate for fair access to plasma therapies for patients who benefit from their lifesaving and life-enhancing effects.

Two of the Alliance's initial objectives are especially encouraging: to ensure fair and adequate reimbursement for all brands of IVIG in all sites of care; and to advocate to Congress and HHS for fair access to IVIG.

The Alliance's founding board members include neurologist Jonathan Katz, MD; immunologist Roger Kobayashi, MD; Flemming Nielsen, general manager of Octapharma, a global manufacturer of products derived from human plasma; Patrick M. Schmidt, president and CEO of FFF Enterprises, a specialty distributor, and of NuFACTOR, a specialty pharmacy, and publisher of *IG Living*; and Chair Abbie Cornett, a common variable immune deficiency patient and Nebraska State Senator.

"Our vision is to make sure patients can access the treatment they need to stay healthy, to stay alive, to live the lives they deserve to live," Cornett said. "The Alliance looks forward to working with Congress, with the Department of Health and Human Services, with industry and, most important, with the patients and providers who are waiting for our collective leadership—to resolve our IVIG access crisis."

It's about time! ■

Kit-Bacon Gressitt, Editor

Please send your letters to the editor to editor@igliving.com.

Readers Write



Your feedback, opinions, suggestions and anything else you'd like to share are important! Email us at editor@igliving.com or visit www.igliving.com and go to the Contact Us page to send your comments.

I read your April-May 2007 article titled "Medicare Local Coverage Determinations Limit Access to IVIG" with interest, and I'd like to relay a recent experience I had with IVIG treatment for one of my Medicare patients with CVID. Because he has Medicare, I had to arrange for his IVIG to be given at his local hospital. The pharmacy charged about \$24,000 for one 40 gram dose of IVIG! Of course, Medicare did not cover this amount and reimbursed a more reasonable payment. Medicare has continued to be charged variable exorbitant amounts for his IVIG. Can you imagine what would happen if my patient signed an advance beneficiary notice?

—Peter Bressler, MD

I am a walking advertisement for successful IVIG treatment for myasthenia gravis (MG). Before I started treatment almost six years ago, I could not get up or walk across the room. With treatment, I graduated from that to a walker, then to a cane, and now I have only occasional need for a cane.

When given the opportunity to counsel MG patients, I tell them to never give up and to keep fighting to obtain treatment. This is their right. I will say that my hospital here in Texas is a valuable asset for us patients. I do not know of any patient who has been unable to receive treatment if referred by a doctor. However, IVIG is becoming hard to obtain, so I want to encourage people to donate plasma for our much needed treatment.

I am also very interested in fighting for IVIG access because my older daughter, a nurse, acquired my bad genes and has been diagnosed with an autoimmune neuromuscular disorder, multifocal motor neuropathy. She has to have a day of IVIG infusions every four weeks to be able to work in a profession she loves. So this cause is near and dear to my heart. Please know that it helps me to have someone listen to my fears, expectations and enthusiastic support for this magazine.

Thanks for all your efforts for IG patients.

—Peggy

I am not having reimbursement issues now. However, my real fear is that the hospitals here in Rhode Island are preparing to protect themselves for future IVIG payment cuts by non-Medicare insurance carriers.

Every three months I must have IgG trough levels tested at my hospital. This testing started in April 2006. I have private insurance and I'm 60 years old. If I refuse, the hospital will not treat me.

I asked my hematologist to do the IgG testing when I realized that the hospital's IgG levels were always higher than my MD's levels. However, testing of trough levels for me is not appropriate because I have an IgG3 deficiency. As my MD explained it to me, my trough levels were high when we started IVIG—except for IgG3, which is why I need IVIG. The hospital is doing a basic trough level test, not a specific IgG subclass test.

The problem with basic trough level testing is it makes some Medicare patients ineligible for IVIG. I see more of a problem here if other carriers start to follow Medicare guidelines.

—Betty

I am so pleased you put the article in your magazine about our disease [stiff-person syndrome (SPS), April-May 2007 issue]. I hope you will do a follow-up soon, describing more about the disease and how it affects each person differently. Each person I know of who has it takes a different medication to mask the problems it brings. Help us bring this to the attention of more of the medical field. It goes undiagnosed for so many and for a long time.

I believe if there was some way to take a count of the affected, we would find a larger count than what was quoted in your magazine. But it was great to have you give it space in your wonderful magazine.

Just another SPS person, living from day to day with the hopes of a cure,

—Myrna

Guillain-Barré Syndrome Treatable but Still Frightening

By Jim Trageser

It has to be one of the most terrifying moments—having your limbs go weak or numb, and then having that paralysis spread, perhaps to the point you can no longer breathe unassisted. You're at the emergency room, unsure of what's happening to you, being treated by doctors you've never met before.

And yet, what a relief the diagnosis of Guillain-Barré syndrome (GBS) must finally be: With a treatment regimen of intravenous immune globulin followed by physical therapy, the vast majority of patients will have a full or nearly-full recovery from GBS. Even those with lingering symptoms will have tremendous improvement from what they experience during the early stages of the disease.

Dr. Scott Carlson, a neurologist in practice near Spokane, Wash., who has treated GBS patients, says the knowledge that GBS is something every patient will eventually beat is an important tool in helping patients stay positive during the early, difficult days of diagnosis and treatment.

"The fact that everyone gets better and 85 percent have a full recovery is an optimistic point for patients," Dr. Carlson said.

It also helps that GBS is one of the easier of the rare diseases to diagnose,¹ so patients and their doctors know fairly quickly what is going on and how to treat it.

"The disease is at its worst within four weeks," Dr. Carlson said. "The rapidly progressive course usually allows a diagnosis at the first visit or within weeks of onset."

This was the experience of Mike Sutton, head basketball coach at Tennessee Tech University in Cookeville, Tenn. Sutton was stricken with GBS in April 2005. In an interview with *IG Living*, he described his experience with the onset of GBS:

"I was on a recruiting trip in Virginia, watching one of my players try out. I'd had a bad cough earlier in the week and experienced pain in my hands and feet. On the next Sunday, April 10, I fell down in the parking lot—I collapsed on my luggage.

"Dr. Richard Rosenblum, who is a friend, got me into the hospital. He diagnosed me with Guillain-Barré syndrome and started treating me. They put a trach in. I was on a ventilator and received IVIG treatment. Later on, I was transferred back to Vanderbilt, and they did plasmapheresis."

Early onset of GBS is typically marked by weakness or even paralysis of the arms or legs. Typically, but not always, the weakness and/or paralysis spreads, often to the chest and face muscles. Breathing assistance is often needed in the early stages of GBS. Yet the severity of the weakness or paralysis varies widely.



Still, Dr. Carlson said emergency room doctors—who he said see most GBS patients due to the severity of the early symptoms—are today very proficient at diagnosing GBS.

"Primary care practitioners are familiar with GBS, but rarely have to diagnose the problem, as many patients present to the emergency room," Dr. Carlson said. "Our ER physicians are quite good at diagnosing GBS, but almost always get a neurology consultant to confirm and treat."

Again, Sutton's experience was representative of what many GBS patients experience:

"I was not familiar with GBS," Sutton said. "The onset was very

¹ The GBS/CIDP Foundation International estimates that only one or two people in every 100,000 will contract GBS.

sudden. I did not know what was happening.

"I had no feeling. I don't remember a lot about the first week, because of the pain medication I was on. I just tried to figure out what was going on. For the longest time, it was very difficult. I couldn't do anything; I could only blink my eyes.

"I was hospitalized in Nashville after Vanderbilt; I was still on a ventilator until mid-July. Initially, I was completely paralyzed. When they took me off the ventilator, I went to rehab."

Dr. Carlson said that recovery from the symptoms of GBS begins fairly quickly. "Recovery starts within two weeks to two months, depending on the severity. Final recovery may take 12 to 18 months."

Intravenous immune globulin and plasmapheresis treatment are used during the first few weeks after the onset of symptoms. Physical therapy is then the primary tool in recovery, as patients regain their strength and use of their limbs.

As Sutton pointed out, though, contracting GBS is a "life-changing" experience.

"You go from being normal and healthy to being completely incapacitated," Sutton said. "I do outpatient therapy three days a week, strengthening muscles. I do have some limited movement in the hands and feet, but we're seeing improvement.

"Initially, there was a lot of uncertainty, because there's no concrete timetable for recovery or the extent of recovery. For my players and staff, that was a big issue for them."

And for Sutton, it was more than a year after the onset of GBS before he could really begin resuming his life as a collegiate basketball coach.

"As I got into the summer [of 2006], I could communicate more and talk, but I put a great deal of trust in

my coaches and staff. The athletics department has been very concerned and supportive. By the end of the season, I was still in a wheelchair, but I went to all the games. I depended on my associate head coach to do a lot of the coaching."

Guillain-Barré syndrome was first described by French doctor Jean Landry in 1859. (GBS is sometimes referred to as Landry's ascending paralysis.) In 1916, Georges Guillain, Jean Alexandre Barré and Andre Strohl—also physicians in France—discovered the underlying cause of Landry's ascending paralysis, and two of them ended up having the disease renamed after them. The disease is

*"For the longest time,
it was very difficult.
I couldn't do anything;
I could only blink
my eyes."*

—Coach Mike Sutton

also sometimes called Guillain-Barré-Strohl syndrome, but mostly Strohl gets left off—such is the unfair nature of recorded history.

GBS is an autoimmune disease in which the body's immune system attacks a part of the nervous system. It is triggered by the presence of a foreign antigen—infections or vaccines.

While there is no cure for GBS, it will eventually subside, and immune globulin therapy can accelerate that process.

Recovery for GBS patients can take from as little as a few weeks to a few years. About 15 percent of all GBS patients will still have some residual weakness, tingling or numbness after three years.

In addition to the immune globulin treatment, recovery from GBS also

involves physical therapy to regain full use of the limbs, as Sutton's example illustrates.

Doctors do not know why some people contract GBS and others don't, particularly in that it is triggered by fairly common antigens—widely used vaccines and common bacterial and viral infections.

Some doctors now believe that President Franklin D. Roosevelt had GBS rather than polio. Popular actor Andy Griffith contracted GBS in the early 1980s, and author Joseph Heller wrote about his own experiences with GBS in his novel, "No Laughing Matter." Even the popular TV show, "House," has featured diagnoses of GBS in several episodes, although each time the diagnosis proved wrong.

According to the National Institute of Neurological Disorders and Stroke at the National Institutes of Health, researchers are trying to isolate why some bacteria and viruses trigger GBS, to see if there is a consistent characteristic that could be targeted for future treatments or even a preventive approach.

For now, educating patients and beginning aggressive treatment remain the keys. While GBS is often fatal in undeveloped countries, due to the lack of ventilator equipment to assist breathing in those GBS patients who suffer chest paralysis, in the United States and other developed countries, GBS is serious but rarely fatal.

As Coach Sutton put it: "The recovery rate is very good. You have to maintain optimism about it. I've talked with a lot of people, and the odds are very good for recovery. ... Even though I have this condition, I'm still more fortunate than a lot of people. When you go through rehab, you see people every day who remind you of that." ■

One Step at a Time

By Cheryl Haggard



"Hey, Mom!" my 8-year-old PIDD kid bellowed. "This crazy dude in the newspaper wants to give you a second chance at Robie!"

The Race to Robie Creek is legendary in these parts of Idaho. Legendary, because it holds the dubious honor of being the toughest half-marathon in the Northwest. Dubious, because the first eight miles of the thing have runners ascending 2,500 feet into the Boise foothills, then descending to Robie Creek. Even the people who win have to walk in places. It's the only way one can survive Robie: one step at a time.

Knowing the sports editor of our local newspaper was giving away his well-fought-for race number should have deterred me, but Caleb's uttering "second chance" spurred me on to consider entering a 200-word essay about why I should be given the editor's ticket to run Robie. Our family understands second chances. IVIG affords us daily second chances at healthy lives. And the race offered an opportunity to put immune deficiencies, blood donations and hope on the radar screen.

So, recognizing that April was Primary Immune Deficiency Month, and wanting to join in the fun of promoting blood donations, I decided I would enter the contest to



win the sports editor's number to race Robie! Chances are, I thought, I wouldn't win and at least I could get our story out there—and maybe one person would be touched enough to give blood!



Two hundred words and an online poll later, I found myself in a puddle at my computer as the results began rolling in. Even the good folks at the Immune Deficiency Foundation got into voting, and my email account was burning with people all over the country wishing me well in a race that I had no intentions of running.

The chirping of the telephone interrupted my worrisome thoughts. "Is this Cheryl Haggard?" a stranger's voice questioned.

I contemplated having an identity crisis to avoid confirming who I knew was on the other end of the line. "Yes, this is Cheryl Haggard," I admitted with trepidation.

"This is Nick, the sports editor, and I have some very good news."

Having children with a chronic condition has set me up for this kind of a phone call. I am good at bracing myself when physicians call with lab results that bring emotional Goliaths to their knees. I love good news, but I suspected

Nick's definition of good news and mine were not jelling.

"You won the online poll! You get to run Robie!" he announced.

I dragged myself to the couch as my head slowly melted into my hands. The salty taste of sweat and tears burned my tongue, and I hadn't even started running yet.

"Are you OK, Mom?" Caleb's concern broke my pity party.

"Yeah, Babe. I'm OK. It looks like I'm going to run Robie for the immune deficiency community. They loved our story."

"Mom, that is so cool!" Caleb cheered. "And Mom," Caleb continued, "I qualified for the Miracle Mile at school, so if you're not feeling well, I'll run Robie for us."

This offer came from a child who was facing sinus and lung surgery within weeks of the toughest half-marathon in the Northwest.

Caleb's compassion gave me the confidence I needed to tackle the 13.1 mile mountain in front of me.

My kids and thousands of others like them run daily uphill medical marathons with their countless IVs, surgeries, illnesses and pain. If the chronically ill could face their mountains with smiles on their faces, I could too.

So, one crisp April afternoon in Boise, Idaho, I faced my mountain.

Because an unconventional disease deserved a daring diva, I had donned red from head to toe. I even dyed my beloved strawberry blond hair. I made a bold statement to the Robie Creek Community and our own patient community: Blood saves lives.

As I crossed the finish line, the Gatorade bath I received from my family felt so good. I got a taste of what it must feel like when my kids bravely face a needle stick and say, "Hey, that wasn't so bad after all!"

And perhaps the accolades I received from my family were because they know what it takes to be a champion: You've got to face that uphill battle—one step at a time. ■

Genes, Heredity and CVID: *What Are the Odds?*

By Melissa Schweitzer, MS, CGC

When I became pregnant with my first child four years ago, one of my biggest concerns was the chance that my child could also have common variable immune deficiency (CVID), as do I. As a genetic counselor, I know there is likely a genetic component to the development of CVID, but we don't know what it is, so it was difficult to determine how likely it would be that my child would develop CVID. I truly understood what my patients must have gone through when I would provide them genetic counseling on diseases that ran in their families but couldn't identify for them the genes associated with the diseases.

Unfortunately, this is still true today for the majority of patients with CVID considering parenthood. However, in the last five years, there has been some progress made in deciphering the genetic basis of CVID (the genes associated with CVID).

To understand how genetics plays a role in CVID, we will review some of the basic genetic concepts and the progress that has been made in genetic research of CVID, and we will discuss genetic testing and whether it may be an option for families.

Genetics 101: Basic Training

According to the Immune Deficiency Foundation, genes are the messages that determine the physical and chemical characteristics of an individual. They are the instructions that communicate to the body how to work and run efficiently. Typically, a human has two copies of every gene within each cell of his body. One copy of the gene is inherited from the mother and the other copy is inherited from the father. The immune system is made up of numerous genes to tell the body how to make the substances that will react to and fight infections. When a gene is

damaged or mutated, it doesn't work properly and this can ultimately affect the function of a whole system in the body, such as the immune system.

Chromosomes are long string-like structures that contain our genes. Humans have a total of 46 chromosomes, which come in 23 pairs. Half of these chromosomes come from the mother and half from the father. The first 22 pairs of chromosomes, called autosomes, are numbered. They are the same in both males and females. The 23rd pair is the sex chromosomes. Females have two X chromosomes and males have an X and a Y chromosome.

Inheritance Patterns

Genetic diseases follow different patterns of inheritance, how the disease is passed on in a family. Each pattern is determined by the gene defect that causes the disease and the chromosome on which the gene is located. The three most common patterns of inheritance for primary immune deficiency diseases are autosomal dominant, autosomal recessive and X-linked recessive. Autosomal dominant (AD) diseases can affect either gender because they are caused by a defect in a gene on one of the numbered chromosomes (those that males and females have in common). These diseases may appear to be passed on through a family from generation to generation. However, they may also occur for the first time in one individual as the result of a new genetic mutation that can happen by chance in anyone. A person affected by a known AD disease has a 50 percent chance of passing the gene mutation on to his or her children.

Autosomal recessive (AR) diseases can also affect either gender because they too are caused by gene defects on autosomes. However, these diseases occur only when both parents are carriers of the gene defect and both have passed

the defective gene on to their child. Typically, carriers of an AR gene defect do not show symptoms of disease. Couples who have a child with an AR disease have a 25 percent chance (1 in 4) of having another affected child. Siblings of an affected individual have a 25 percent chance of also being affected with the disease and a 50 percent chance of being a carrier, but not likely affected.

X-linked recessive (XLR) diseases typically affect males and are caused by gene defects on the X chromosome. Since males have only one copy of the X chromosome, they do not have another copy of the gene to balance out the damaged copy. Affected males have either inherited the gene defect from their mothers, who are carriers, or the gene defect occurred as a new mutation for the first time in the affected male. Regardless, the daughters of affected males will be carriers of the defective gene because they inherit their father's X chromosome. The sons of affected males inherit their father's Y chromosome, so they will likely be unaffected, depending on their mother's carrier status.

Female carriers of XLR diseases do not usually show symptoms of the disease. However, each of their sons has a 50 percent chance of being affected and each of their daughters has a 50 percent chance of being a carrier. For XLR diseases, it is important to determine whether the gene defect is inherited or is a new mutation, because this will affect the recurrence risk to other family members, including the couple's children, siblings and even more distant relatives. Families with inherited XLR defects typically show multiple generations of affected males, passing through unaffected female carriers. When the first affected person is a male with a new XLR mutation, family members may be negative.

Some primary immune deficiency diseases such as CVID and selective IgA deficiency may follow multifactorial inheritance, rather than a single gene inheritance pattern as those described above. Multifactorial inheritance is less defined and does not show the clear patterns when looking at the family history. In multifactorial diseases, it is thought that a person inherits a genetic predisposition to developing a disease that could be caused by one or more genetic components. However, the environment also plays a role in disease development and may act like a trigger. If a person has the genetic predisposition but is not in the right environment, he or she may never develop the disease. It has been difficult to determine the genetic components and

environmental factors for multifactorial diseases, and it is therefore difficult to determine actual recurrence risks for family members.

CVID Genetics Research

What does all of this jargon have to do with CVID? Current research shows that somewhere between 10 percent and 20 percent of individuals with CVID have an identified gene associated with their disease. These genes may follow autosomal recessive, autosomal dominant and even X-linked recessive inheritance, depending on the gene. Still, for the majority of patients with CVID, "there is not one single gene defect associated with the disease," says Hans Ochs, MD, professor of pediatrics and director of the Immunodeficiency Molecular Diagnostics Laboratory at the University of Washington in Seattle. Dr. Ochs explains that CVID is likely to be a group of disorders caused by multiple gene defects. Once certain genes are identified, this group of individuals will be pulled out of the CVID category and given a new diagnosis, even though they may still have the same clinical symptoms as others in the CVID category.

Recurrence risks for family members of CVID patients are most clearly defined for the following genes that have been identified with CVID:

- The ICOS gene is located on chromosome 2 and follows autosomal recessive inheritance. Therefore, carrier parents of a child with an ICOS gene defect have a 25 percent chance of having another affected child. Although this gene was an exciting discovery back in 2003, it has since been identified in only four families in the Black Forest region of Germany and Lienz, Austria. Moreover, the ICOS gene probably accounts for less than 1 percent of patients with CVID.¹
- The TAC1 gene is located on chromosome 17 and follows autosomal dominant inheritance. The original studies in 2005 suggested that defects in this gene may account for as many as 10 percent to 15 percent of patients with CVID in their study. However, additional studies have suggested that not all of the reported changes in the gene are associated with clinical symptoms of CVID and may be due to normal variation in the gene. Caution should be used when trying to predict risk in families with TAC1 gene alterations.² ➤

- The CD19 gene is located on chromosome 16 and follows autosomal recessive inheritance. Discovered by a group in the Netherlands, the CD19 gene is thought to account for less than 1 percent of patients with CVID.³
- The BAFF-R gene is located on chromosome 22 and follows autosomal recessive inheritance. It is thought to account for less than 1 percent of patients with CVID.⁴

Genetic Testing

Until the last couple of years, much of the genetic testing for primary immune deficiency diseases was done in the research laboratories that identified the genes. This is starting to change. Now, specialty commercial laboratories, along with university-based laboratories, are starting to offer clinical genetic testing. Many of these specialty labs also offer prenatal testing once a gene defect is identified in a family. However, health practitioners are not commonly familiar with such labs.

Two of the main reasons to consider genetic testing are to more clearly define a person's diagnosis and to help predict recurrence risks for other family members.

Dr. Ochs suggests that "many patients never get a genetic workup and may be misdiagnosed." For example, it is very

important that a male with CVID has a clinical laboratory workup or genetic testing to rule out X-linked and autosomal conditions that can be clinically similar to CVID, such as X-linked agammaglobulinemia, X-linked lymphoproliferative disease and hyper-IgM syndrome. Recurrence risks for someone with an X-linked primary immune deficiency will be much higher than for someone with standard CVID. Dr. Ochs believes that it is important for a patient to discuss his or her clinical and family history with an immunologist to determine if genetic testing is warranted. If so, the patient's immunologist or specialist can send a sample of the patient's blood to the appropriate genetic laboratory for analysis of the suspected genes.

It is worth noting that once a gene defect is identified in an individual, other at-risk family members can be offered this testing. In addition, prenatal testing may be an option through chorionic villus sampling (CVS) or amniocentesis, two procedures that can be performed on a fetus. However, there are risks of miscarriage associated with both procedures, and, if they are being considered, the risks should be addressed in genetic counseling. Genetic counseling is available in most hospitals and medical centers that offer high-risk obstetric services.

What Are the Odds?

Now, let's get back to the question: What is the chance my children might be affected?

The odds are all over the board. The bottom line is this: If a specific gene has been identified and is thought to be the cause of CVID in an individual, the odds will be based on the inheritance pattern of that gene. However, for the majority of us with CVID, we still don't have the definite answer to that question. And for those contemplating having children, it is a very personal choice that should be discussed with your spouse and your physicians.

Conclusion

If you have CVID and you are willing to take a chance that your child might also have it, keep in mind that any couple takes myriad chances when having a child. Despite the unknown, I go on about my life as I always have, thankful for a diagnosis and an effective treatment that allows me to enjoy every day with the wonderful children I am fortunate to have. ■



^{1,2,3,4} Gene Reviews: Common Variable Immune Deficiency Overview www.genetests.org/profiles/cvid.

CVID and Pregnancy: *Do We Have a Problem?*

By Zachary Pugh

*H*aving a child is one of the most important events in a person's life, if not the most. This said, we want to make certain that this replication of us, this life we have created and nurtured, is introduced to our world without flaw. More important, we want to be assured that we are informed and prepared for this new familial journey. For those with common variable immune deficiency (CVID), information is imperative and preparedness is paramount.

What We Know

CVID is a term that describes a heterogeneous cluster of disorders in people who have hypogammaglobulinemia, with variable degrees of T-cell dysfunction.¹ Among the information on CVID that has been accumulated within the medical community is the fact that it is not gender-specific. According to St. Jude Children's Research Hospital, diagnosis of CVID typically becomes prevalent in men and women in the third or fourth decade of life, but it is also seen in children. CVID is often suspected when a patient possesses "low levels of antibodies in the blood stream, a poor immune response to vaccines, and a history of recurrent infections."²

Although the medical community has reached a consensus that heredity does pose a role in the development of CVID, there seems to be no concrete agreement on what the genetic patterns are or are not. In its publication, "The Clinical Presentation of the Primary Immunodeficiency Diseases," the Immune Deficiency Foundation (IDF) clearly affirms "there is no recognizable pattern of inheritance, although CVID may cluster in some families." St. Jude Children's Research Hospital concurs that, although genetic factors do play a part in the development of CVID, "there is no single gene mutation (mistake) that can be identified."

As a board-certified genetic counselor, mother and CVID patient, Melissa Schweitzer is well-versed on pregnancy and CVID. "I tend to think of CVID as a disease that is inherited in a 'multifactorial' way," Schweitzer says,

"meaning that I think both genetic and environmental factors play a role in causing the majority of cases."

When discussing genetic factors, there is the obvious question of inheritance, which, as mentioned earlier, does play a role in the development of CVID. But how large of an influence do genes have? To date, there are no conclusive tests that have revealed a connection between one single gene and all CVID cases. However, Schweitzer does believe that single genes are responsible for subsets of the disease. Within these particular families, there are traditional patterns of inheritance such as autosomal recessive and autosomal dominant. While the autosomal recessive pattern involves the parents as unaffected carriers and a 25 percent chance of the child being affected, the autosomal dominant pattern involves affected parents and a 50 percent probability rate of inheritance.

"These subsets are caused by the single genes that have been identified...and probably other genes that have not yet been identified, but unfortunately still do not account for the majority of the cases of CVID," Schweitzer affirms.

Of these identified single genes, one is the TAC1 gene mutation.

In 2005, a group of researchers at the Children's Hospital Boston discovered a genetic mutation pattern in patients with CVID. Led by Raif Geha, MD, and Emanuela Castigli, PhD, this study concluded that certain mutations in the gene known as TAC1 contribute to the development ➤



¹ Immune Deficiency Foundation; Publications: The Clinical Presentation of the Primary Immunodeficiency Diseases www.primaryimmune.org/pubs/book_phys/phys_p06.htm.

² St. Jude Children's Research Hospital; Inherited Immunodeficiencies: Common Variable Immunodeficiency (CVID) www.stjude.org/disease_summaries/0,2557,449_2164_6532,00.html.

of CVID and other immune system conditions. In a press release on the findings, the National Institutes of Health (NIH) summarized that “[d]efects in TAC1 were found in four of 19 unrelated patients with CVID and in one of 16 unrelated patients with IgA deficiency.”

Upon further examination of the four individuals with TAC1 mutations, scientists discovered that all of them had relatives who possessed a similar mutation. “A test for TAC1 defects would enable the diagnosis of more children and their relatives with these immune deficiencies,” Dr. Geha said in a 2005 press release. Despite this discovery, there is no agreement in the healthcare community about the likely percentage of CVID patients with TAC1 mutations.

Genetic Counseling

During her 10-year career in genetic counseling, Schweitzer worked as the director of patient advocacy for the IDF. Although she mentions that she was not aware of any clear and concrete links between miscarriage and CVID, “infertility and pregnancy loss [was] a concern that many women with PIDD [primary immune deficiency disease] expressed when I talked with them at IDF.”

Howard M. Lederman, MD, PhD, and professor of Pediatrics and Medicine at Johns Hopkins University in Baltimore, echoes that there are no clear connections between CVID and infertility. “In general, I don’t expect CVID to cause problems with infertility.” He does add, however, that secondary effects of CVID, including chronic recurrent infections and autoimmune diseases such as various forms of colitis, could lead to pregnancy problems. The main thing to take into consideration is that antibody deficiency should not be an important issue.

Although Schweitzer recommends pre-pregnancy genetic counseling, if done during the pregnancy it still may be effective in determining whether or not the child will have a predisposition to CVID. “This can be done either by the patient’s immunologist or by a genetic counselor who would take a detailed family history in the form of a pedigree, which is like a family tree,” Schweitzer says. This method provides a way for the genetic counselor to pinpoint family inheritance patterns, even if the genetic basis for the person’s individual disease is not known.

Along with determining if the child would be predisposed to CVID, genetic counseling also provides women and couples with valuable advice on testing the fetus if there is a known familial genetic mutation. “Genetic counseling would also provide information on routine prenatal screening tests for other birth defects and diagnostic tests such as chorionic villus sampling (CVS) and amniocentesis,” Schweitzer suggests.

Pregnancy Management

As an expert in the field of genetics and an involved patient advocate, Schweitzer still needed guidance from her immunologist when it came to managing her own pregnancy. During her second trimester, Schweitzer’s immunologist began measuring her IgG levels every month. As a patient, she suggests that pregnancy management for those with CVID “should really be discussed by an immunologist who has managed several pregnant women with PIDD. However, the main concern is maintaining the IgG at a therapeutic level in both the woman and the fetus,” she explains.

In Schweitzer’s case, her immunologist began measuring her IgG trough levels (the levels just before the next infusion) once a month, beginning in her second trimester. “He had a baseline to start from and then when the level started going down... and my weight was increasing... he increased my dose of IVIG.” Toward the end of her term, her immunologist tried to give her an infusion as close to her delivery date as possible. “For my first pregnancy,” Schweitzer recalls, “I got lucky and had gotten my infusion just a couple of days before I ended up delivering my daughter by an emergency C-section. My second pregnancy was a planned C-section, so I was able to schedule my infusion right before the C-section.”

For women who plan to have a C-section, Schweitzer offers suggestions: “In order to minimize her risk of possible infection related to the surgery, she would want to have her infusion as close to her C-section as possible,” adding that women should always consult with their immunologist.

“[Women] need to be sure that the person that manages their gamma globulin knows they are pregnant,” Dr. Lederman says. There are “two things that will change the dose requirement for gamma globulin.” The first is that “pregnant women gain weight,” he explains, “and the dose of gamma globulin is based on weight. The other is that the placenta pumps gamma globulin from the mother into the baby, especially during the last trimester. You have to make sure that the dose of gamma globulin accounts for both of those things.” Dr. Lederman added that it’s a good idea to check IgG levels before each infusion, starting at about the third or fourth month of pregnancy so that the dose can be increased as necessary.

Schweitzer suggests that, if after your child is born and all of the tests reflect normal immunoglobulin levels, it is still important to watch for potential symptoms, because CVID and other immune deficiencies often manifest later in life. “I have always been told to keep a close eye on my children’s health, and, if they start experiencing more frequent infections or other symptoms of PIDD, then it’s time for more testing and evaluation.” ■

Breastfeeding and PIDD: *Welcome Baby Softly*

By Jessica Schulman PhD, MPH, RD



The importance of breastfeeding, especially in the first hour after birth, is highlighted in the theme for the 2007 World Breastfeeding Week, August 1 through 7: Breastfeeding, The 1st Hour—Welcome Baby Softly.

This theme celebrates the importance of early closeness between mother and baby in the first hour or two after birth, and calls on health professionals to establish a welcoming environment that greets babies softly.

Breastfeeding in the first hour or so after birth has been linked to positive health outcomes, including immediate protection from bacteria and viruses the baby is exposed to, less risk of jaundice in the baby, and improved milk production and less blood loss in the mother. A study published in *Pediatrics* in 2006 found that these health outcomes are so important that 41 percent of infants worldwide who die between two and 28 days of life could have been saved by the simple intervention of breastfeeding in the first hour.

According to Rebecca Mannel, president of the International Lactation Consultant Association (ILCA), "When mothers hold their babies skin-to-skin immediately after birth, their babies are kept warm, they regulate their heart, respiratory and oxygen saturation rates, and they do not feel pain as acutely. All of this means they are calmer, happier and cry less."

But what about mothers or newborns with immune system disorders?

Frequently Asked Questions

What is the consensus on breastfeeding and mothers with primary immune deficiency disease (PIDD)?

Mothers are encouraged to adhere to the American Academy of Pediatrics guidelines for breastfeeding. According to Dr. Jordan Orange of the Children's Hospital of Philadelphia, breastfeeding is encouraged unless the mother needs to be on medication that is transferred in breast milk and could be harmful to the baby or if she has certain chronic viral infections.

Is it safe for a mother on IVIG to nurse her baby?

According to Dr. Sean McGhee, a pediatric immunologist at UCLA's Mattel Children's Hospital, "It would be safe to breastfeed a baby whose mother receives IVIG." Dr. McGhee explains that human milk is rich in IgA but less concentrated in IgG, so most of the IVIG is likely to stay in the mother rather than in her breast milk. However, he points out, "it's a different answer if a woman is pregnant because IgG passes across the placenta to the baby." This is a normal and safe process,

unless the IVIG has caused an infection, but Dr. McGhee says, "infection from IVIG is extremely rare today."

What if the mother's antibody levels are low?

There is no guarantee that the breast milk of a mother with a PIDD will have the same protective antibodies as the breast milk of a mother without a PIDD. Regardless, there are numerous other benefits to human milk. It contains the right amount of fatty acids, lactose, water, amino acids, macrophages and beneficial bacteria for digestion and brain development.

Should a mother stop nursing when her baby is getting evaluated for a PIDD?

Breast milk has lymphocytes and anti-infective properties, but how much this impacts the infant's blood tests is not clear. Currently, there is no established protocol to stop breastfeeding solely for the purposes of testing the infant's immune system. Dr. Jennifer Puck, director of the new Jeffrey Modell Foundation Diagnostic Center for Primary Immunodeficiencies at University of California, San Francisco, explains: "Tests of the infants' immunity would, in general, not be rendered uninterpretable (or falsely appear normal), as far as I am aware." However, Dr. Puck recognizes that there is little research on this topic and more work needs to be done.

Are there concerns for infants with combined immune deficiencies?

For infants with severe combined immunodeficiency (SCID) awaiting treatment by bone marrow transplant, some centers discourage mothers from breastfeeding out of concern that maternal lymphocytes may be transferred to the baby and cause graft-versus-host disease. However, Dr. Puck says, "other centers let breastfeeding continue, believing that maternal lymphocytes in the baby are more likely to have been transferred at the time of delivery. SCID is so rare that this issue hasn't been amenable to study."

As always, *IG Living* recommends that you discuss your situation with your physician for individualized recommendations. ■

Editor's note: For additional information, visit www.babyfriendlyusa.org and www.ilca.org.



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Have a Very NICE Day

By Cheryl Haggard



It was a typical rainy spring Saturday in Seattle, but NICE Day In Seattle, an event sponsored by NuFACTOR and *IG Living*, brightened the lives of the caregivers and patients who came to learn about dealing with chronic immune and autoimmune disorders.

The atmosphere circulating the halls at the University of Washington, which hosted the event, encouraged a sense of camaraderie as the editor of *IG Living*, Kit-Bacon Gressitt, urged us to “pounce” on the various presenters, especially the resident experts, Dr. Troy Torgerson and Dr. Mark Hannibal, both immunologists at the University of Washington, and Michelle Vogel, a reimbursement expert.

Attendee Curtis Pease, 23, was diagnosed with X-linked agammaglobulinemia (XLA) at age 2. “I really liked the way Dr. Torgerson went in-depth in explaining our diseases, and after 20 years of having X-linked, I am glad to finally know what it is,” Curtis related.

Other attendees agreed with Curtis’ assessment of Dr. Torgerson’s NICE Day kick-off presentation. In it, the doctor made a hilarious comparison of the human immune system to a military operation, complete with phagocytes wearing combat helmets, B cells dropping smart bombs and T cells assassinating pathogens. Many in the audience had no clue that something as complicated as the immune response could be so entertaining.

Renee Burbank, whose 7-year-old son, Justin, is a patient of Dr. Torgerson, quipped, “He has a special way of explaining things [so] all can understand.”

In the second presentation, Dr. Mark Hannibal gave a fresh perspective on the history of immune globulin therapy, its production and the many advances that biopharmaceutical companies have made on behalf of patients.

Dr. Hannibal’s presentation encouraged Kathy Schultz to subsequently share with her physician her concerns about her IG treatment for chronic inflammatory

demyelinating polyneuropathy (CIDP). “I was able to chat with my doctor about what I had learned from Dr. Hannibal,” Kathy explained. “I feel a lot better about speaking with my doctor now because I truly understand what gamma globulin is all about.” That knowledge in turn has improved Kathy’s quality of life.

And this is the recurrent theme of the NICE Day events: to improve people’s quality of life while coping with chronic conditions.

Joe Schultz, Kathy’s husband and caregiver, especially enjoyed the time allowed for networking. “There are a lot of people to talk to, and that gives you a different perspective,” Joe said. “You realize that you are not floating around on your own.”

Kathy agrees: “I’ve been to other support group meetings like this and NICE is very different. The day was filled with very kind people who were trying to help or just give a hug.”

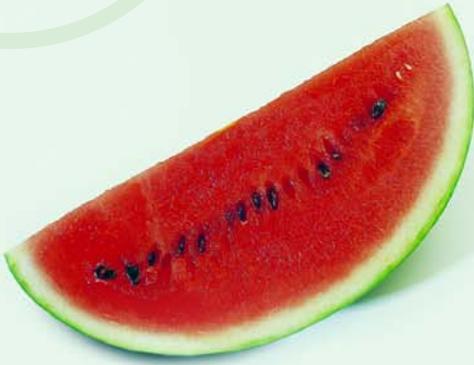
After a breakout session on sites of care, Desiree Cusicks, whose two sons, Cameron and Robert, have XLA, expressed enthusiasm for the event. “Each time I attend one of these meetings,” she said, “I learn more and more, and the information is sinking into my brain and my understanding has grown. It’s just wonderful to talk with someone who has been down the same road you are traveling.”

Michelle Vogel’s call to action on insurance reimbursement issues and the current Medicare crisis impressed on all of us the importance of getting involved. Michelle stressed that even though we might not have reimbursement issues now, we may someday. She emphasized that we are all in this together, and encouraged us to communicate with our elected officials. An air of urgency to advocate for fair reimbursement for rare diseases was felt by all.

“I’ve become a stronger advocate for my boys and a stronger mom because of the wonderful help that I have received,” Desiree said.

Renee may have captured the true essence of the NICE Day in her comment: “The best part for me was to... see that I am not alone in this everyday struggle.” ■





Understanding

By Jessica Schulman, PhD, MPH, RD

What is food to one man is bitter poison to others.

— Lucretius (99 BC – 55 BC)

It was a summery August night, and Emma was talking with friends and enjoying fresh-cut watermelon. The next morning, she woke up to get ready for work and could hardly open her eyes. Her eyelids were swollen and her neck was covered in hives. She knew that she was allergic to ragweed pollen, but she did not recall being allergic to watermelon. What do you think happened to Emma? Could it have been prevented? Was this a food allergy?

According to the Food Allergy and Anaphylaxis Network (FAAN), approximately 12 million Americans suffer from food allergies. As many as 30,000 individuals require emergency room treatment for severe food allergies or anaphylaxis each year. This impacts general health and emotional well-being.

Syed Arshad, MD, is director of the David Hide Asthma and Allergy Research Centre at St. Mary's Hospital, in Newport, Isle of Wight, U.K. He describes food allergies as a significant problem that is not going away. In particular, he says, "there has been an increase in peanut allergies... which tends to be the most dangerous." He suggests that food allergies are common, afflicting up to 8 percent of young children and 4 percent of older children and adults.

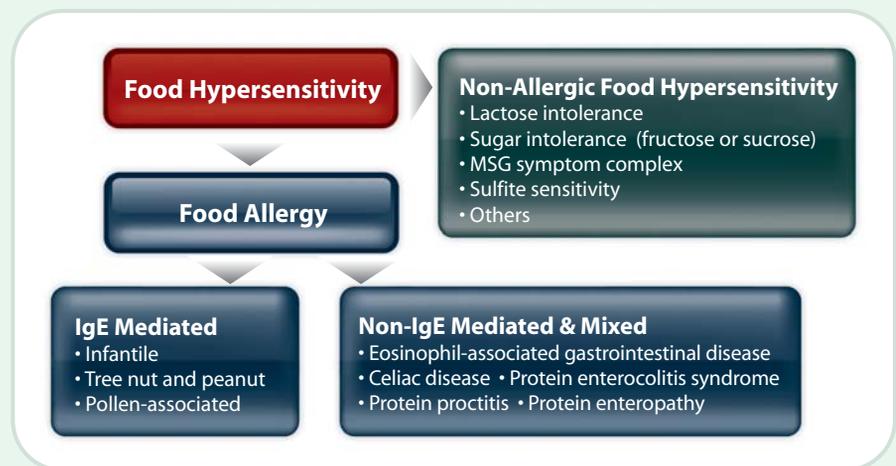
What Are Food Allergies?

Most of us have probably encountered someone with Emma's food allergy—the type that is provoked by exposure, and re-exposure, to a certain food ingredient or substance. The body mistakenly learns that a food component is harmful. Then, the immune system takes action by launching its weaponry. For example, the body may think that pollen is an infection that needs to be destroyed. What happens next? The body may respond with immunoglobulin E (IgE) antibodies and release compounds, such as histamine, that cause symptoms from mild itchiness to runny nose to atopic dermatitis to sudden death due to anaphylactic shock. Various

organs such as the skin, gastrointestinal tract and respiratory tract become irritated or injured by this process.

The European Academy of Allergy and Clinical Immunology Task Force suggests that adverse reactions to food should be called "food hypersensitivity" (see Figure 1). When the immune system is involved, the appropriate term is "food allergy." This is typically, but not always, driven by IgE antibodies. The task force suggests that other reactions, sometimes referred to as "food intolerance" (e.g., lactose intolerance), should be referred to as "non-allergic food hypersensitivity." Severe, generalized allergic reactions to food are classified as anaphylaxis.¹

Figure 1. Food Hypersensitivity and Food Allergy



¹ Adapted from: Nomenclature Revised by the European Academy of Allergology and Clinical Immunology, [Johansson et al. 2001] and the World Allergy Association [Motala, 2007].

Food Allergies



Ulrich Wahn, MD, director of the Department of Pediatric Pneumology and Immunology at the University of Berlin, suggests that, for the general population, eight foods account for 90 percent of all food-allergic reactions: milk, egg, peanut, tree nut (walnuts, almonds, cashews, pistachios, pecans, etc.), fish, shellfish, soy and wheat. Dr. Wahn provides a road map for understanding IgE-driven food allergies.

1. Infantile: Infantile food allergy begins early in life, around 6 to 12 months of age. It is generally transient, so it gets better over a period of months or a limited number of years. Most cases run their course by the baby's first birthday, but some cases last much longer. Symptoms may include vomiting, diarrhea, abdominal pain, hives, swelling, itching, eczema and difficulty breathing. The pediatrician will often recommend a special formula or diet. This involves removing one or more items from the "hit list" for the baby and the mother, if she is breastfeeding. Sometimes, specialized tests such as food challenge tests or IgE levels are ordered. The hit list includes hen eggs, cow's milk, soy and wheat. Most young children with atopic dermatitis from food allergies have reactions to milk, eggs or peanuts.

2. Tree nut and peanut: These nut allergies may be life-threatening, severe and persistent throughout life. Early signs include a runny nose, itchy skin, hives, or tingling in the mouth, tongue or lips. More severe signs include tightness in the throat, hoarse voice, wheezing, cough nausea, vomiting and anaphylaxis. Anaphylaxis may involve several symptoms at one time such as hives, blood pressure drop, narrowing of the breathing tubes and swelling of the tongue. The doctor may do specialized tests to confirm the allergy. The hit list includes cashews, almonds, pecans and walnuts among other tree nuts, and peanuts (really a legume). Although treatment for nut allergies is on the horizon, the doctor will probably recommend that the patient avoid the nut altogether.

3. Pollen-associated or pollen-food allergy syndrome: This syndrome can onset later in life. It may result from direct exposure to pollen on a fruit or vegetable or from a cross-reaction between a pollen allergy and a fruit or vegetable allergen. In a

cross-reaction, pollen-allergic individuals have IgE antibodies that also react to allergens coming from a fruit or vegetable (called cross-sensitization). For example, birch pollen allergies are associated with peach allergies (see Table 1). It has a wide range of severity, and symptoms are variable, including itching, urticaria or hives and welts, burning, swelling in the lips, tongue and palate, congestion, itchy watery eyes, wheezing, nausea, abdominal pain, diarrhea, chest or throat tightness, and difficulty swallowing or breathing. The syndrome can be persistent, and Dr. Wahn says, "The best cure is age, and you may have to wait for decades." The hit list includes fresh fruits and vegetables such as pears, apricots, melons, bananas, nuts such as hazelnuts or vegetables such as carrots. Raw and unprocessed foods tend to cause more severe reactions.

Which food allergy did Emma most likely experience? If you guessed pollen-associated, you're correct. She is allergic to ragweed pollen and developed a cross-reaction to watermelons. ➤

Table 1. Cross-reaction Allergies*

Pollen or Inhalant	Common Food Allergens
Birch pollen	Apple, raw potato, carrot, celery, hazelnut, pear, peach, plum, cherry
Mugwort pollen	Celery, apple, peanut, kiwi fruit, carrot, parsley, spices (fennel, coriander, aniseed, cumin)
Ragweed pollen	Melons (e.g., watermelon, cantaloupe, and honeydew), bananas
Latex (hevea brasiliensis tree, rubber tree)	Avocado, kiwi fruit, chestnut, papaya, banana

* Adapted from the World Allergy Organization

What Is Anaphylaxis?

Anaphylactic shock is the most severe type of food allergy: It causes death in 150 to 200 people annually.² If left untreated, it can lead to death in a matter of minutes.

On Halloween night in 2002, Gina's 2-year-old son, Conner, tasted his first chocolate-coated peanut candy. He did not like the texture and spit it



Conner and his mother, Gina

onto the floor. Within a few minutes, he was vomiting and developed fluid-filled hives that were growing and converging all over his body. By the time they reached the emergency room, the hives were around Conner's throat. Two epinephrine injections were administered, one into each of Conner's thighs. Gina says, "Within 10 minutes' time, Conner fell asleep on his daddy's shoulder, and the next morning the hives were gone." A pediatric allergist offered some unsettling guidance. "In all likelihood he will never be free of these life-threatening symptoms, so steer clear of nuts!"

Epinephrine (adrenaline) is used for controlling an anaphylactic reaction. It is available as a self-injectable device (EpiPen®, single dose, or Twinject®, two doses). In most cases, strict avoidance of the allergy-causing food is the only way to prevent a reaction. Early administration of the epinephrine is the key to treating anaphylaxis successfully, so, if the allergy is known, it is wise to carry epinephrine at all times.

Gina says, "Don't let the [epinephrine] be a source of worry. Instead, use it as a confidence builder, and help your children to live as normal a life as possible." However, cross-contamination is a problem so Gina does not take Conner to places such as ice-cream shops that may cross-contaminate with nuts. "You have to be vigilant. Sometimes people have a really difficult time grasping how a beautiful child can have a life-threatening food allergy," she says.



How Do You Confirm a Food Allergy?

A specialist may confirm an allergy with a food challenge in which a patient is given a small amount of the substance to which he or she might be allergic. Dr. Wahn explains that "allergy is the demonstration of a positive food challenge in an individual," but he warns that "it is unethical to challenge someone who has a clear-cut history of peanut allergy."

Today, numerous allergy tests are available, but the most commonly used are the skin prick test (a mosquito bitelike bump appears if the patient is allergic to the substance) and a blood test to check for elevated IgE to certain allergens ➤

² FAAN, 2002.

Can You Say Eosinophilic?

Allergic eosinophilic gastrointestinal disease is a mixed phenomenon involving IgE antibodies and other immune components, such as T cells. This is an emerging disorder that is linked to food allergies and an irregular immune system. Buddy's story, below, may serve as a model for understanding how other parts of the immune system interact to cause allergic disease. If there is suspicion of a problem, always work through the process of making the correct diagnosis with your doctor—and be persistent.

Buddy's Story

By 5 weeks of age, Buddy had projectile vomiting and diarrhea while being breastfed, a horrible skin rash on his bottom, and he was generally irritated. "He spent his first year strapped into his car seat next to my bed because we were afraid that if we laid him down he would throw up and suffocate on his own vomit," recalls Buddy's mother, Nancy. "Some days, he would cry and scream in pain for 14 hours straight."

When I asked Nancy what steps she took to get him correctly diagnosed and treated, she said, "For the next 14 months, we went to our pediatrician, ENT, pulmonologist, allergist, immunologist, surgeon and gastroenterologist." No one was able to see the whole picture, and the family continued to suffer. "I went through endless amounts of expensive medicines to no avail. I tried numerous formulas and restricted my own diet, since I was breastfeeding. Even simple things like going to the grocery store were impossible. He would scream in pain, and everyone would look at me as if I did something to hurt him."

Nancy was tireless and continued to advocate for her child. Eventually, Buddy was diagnosed with an immune deficiency and eosinophilic enteropathy.

When Buddy was 2 years old, the family traveled 860 miles from home to seek more specialized attention at Cincinnati Children's Hospital Medical Center. He is now properly treated with a very restrictive diet and special formula. "In the beginning, we went to no food for almost one year and even now, at four years later, we still have periods where he cannot consume anything but the formula," Nancy says.

If Buddy does not tolerate the introduction of a new food during a food trial, it is immediately removed from

his diet and they are back to the drawing board for a few weeks or months. Every few months, Nancy does food trials with Buddy with the hope that perhaps one day he will be able to eat a broader diet.

After Buddy was correctly diagnosed, he got almost immediate relief from his symptoms. Though he still depends on a special formula, he is a happy little boy and doesn't seem to mind his stringent diet. "The formula is bad tasting to me," Nancy said, "but he knows, and we know, that it is safe for his body, and that is what matters."

Buddy recently celebrated his fifth birthday. He attends preschool part time, plays tee ball and soccer, rides his bike without training wheels and loves to drive his big sister nuts. Nancy explains that, with immune problems and food allergies, the hardest part is "his problems are hidden inside of his body," adding that "if he eats the wrong food or tries to sneak something, his body attacks itself on the inside! At any moment we could be heading to the hospital."

What can other families learn from Buddy's experience? "I just want parents to know that they need to follow through with their gut feeling when something isn't right. There are many great doctors out there, but all doctors can't be specialized in everything. Rare issues make for rare proper diagnosis," Nancy said with conviction.

What Happened to Buddy?

An eosinophil is a type of white blood cell that occurs naturally in the human immune system—in very small numbers. Under certain conditions, such as when there is an infection, the body releases a lot more eosinophils and they accumulate in an organ such as the gut and release toxin. This is helpful, except when the cells incorrectly respond to a food ingredient as if it were a deadly parasite. When this happens, the toxin damages everything in its path, including healthy tissue. This is what happened to Buddy. Often, the allergic reaction is delayed, which makes it difficult to know what to eat or avoid. Symptoms vary by age. Babies will have problems eating, including vomiting and excruciating pain. Adolescents and adults may have trouble swallowing. Other reported symptoms include nausea, early satiety or fullness, diarrhea, slow emptying of the stomach, blood in the stool and anemia.

(e.g., radioallergosorbent tests or ImmunoCap®). Patch testing is a relatively new method that is used to determine if certain substances are associated with inflammation of the skin. Small amounts of food ingredients are placed directly on the skin, with an adhesive, for one to two days. Then, a specialist evaluates whether or not there has been a delayed reaction to the substance. Dr. Wahn considers patch testing to be useful, "but not to the extent that it is considered a gold standard for all food allergies," he adds.

An experienced allergist will also examine the context of a suspected food allergy, such as how the patient is feeling and any concurrent health conditions such as immune dysfunction. A negative IgE blood test does not always mean that you do not have a food allergy, as a positive test does not necessarily mean that you have a food allergy. "You have to correlate the positive test to the clinical exam or status of how the patient is doing in real life," Dr. Arshad emphasizes. A positive test might indicate a need for clinical challenges. Lack of a positive test might result in an allergist challenging the patient with the food that he or she seems to be allergic to, to reproduce an adverse reaction in a controlled setting. Then the allergist will watch closely to see if there is resolution of the symptoms by eliminating the offending ingredient from the diet. This is called an elimination challenge.

Food Allergy Testing

If individuals suspect that they have food allergies, they should contact a disease-specific organization and locate a physician that has

experience diagnosing and treating allergies (see Resources). Only a qualified specialist can attach the correct meaning to the test results. Self-diagnosing food allergies without proper guidance can lead to poor eating habits or mislead an individual to overlook a significant medical problem (see Can You Say Eosinophilic?).

Allergies and Pregnancy

At the American Academy of Allergy, Asthma and Immunology (AAAAI) 2007 Annual Meeting, Drs. Arshad and Wahn explained that they have not found clear evidence that allergen avoidance during pregnancy is useful. In fact, avoidance of certain foods during pregnancy has potential to compromise the mother's diet. This does not mean that avoiding allergic foods does not delay allergies in her baby. There is evidence showing that the immune system may be negatively affected by exposure to certain allergens before birth. Until more is known, however, these scientists agree that it may not be practical to restrict the diet of a mother during pregnancy.

Allergies and Breastfeeding

It is not realistic to say that breastfeeding is magic and prevents food allergies. "If there were a clear effect of breastfeeding on allergies, there would be no debate among scientists. Still, irrespective of food allergy, nursing is an outstanding source of nutrients," Dr. Arshad says. Although large-scale literature reviews and studies show a slight protective effect of breastfeeding for allergy prevention, Dr. Arshad calls attention to the fact that "not a single study on prevention of allergies and exclusive

breastfeeding has been for more than six months." The question of how long breastfeeding remains at all protective is unanswered.



Prevention

If the child is at high risk of developing an allergy (high risk meaning that at least one or more parents or siblings has atopic disease or a confirmed food allergy), then avoiding certain food ingredients can improve outcomes. Scientists report some benefits in preventing atopic dermatitis and food allergy by eliminating cow's milk, nuts and eggs, and sometimes wheat and tree nuts. The problem, however, is that restricting such a broad array of ingredients while keeping a balanced diet can be tricky. Dr. Wahn hesitates to give general recommendations. "In certain cases, if a mom observes a problem after nursing, then you can confirm cases where [food allergy] is plausible and make diet recommendations," but "a recommendation for a whole

Resources

Allergic Disorders:
Promoting Best Practices
[www.theallergyreport.com/
reportindex.html](http://www.theallergyreport.com/reportindex.html)

American Partnership for
Eosinophilic Disorders
www.apfed.org

National Institutes of Health, National
Institute of Allergy and Infectious
Diseases (2004). *Food Allergy: An
Overview* (NIH Publication No. 04-5518).
[www.niaid.nih.gov/
publications/pdf/
foodallergy.pdf](http://www.niaid.nih.gov/publications/pdf/foodallergy.pdf)

Sicherer, S.H. (2006). *The Complete
Peanut Allergy Handbook: Understanding
and Managing Your Child's Food Allergies*.
Johns Hopkins Press.

Food Allergy and Anaphylaxis Network
800-929-4040
www.foodallergy.org

World Allergy Organization
www.worldallergy.org

Find an Allergist

- American Academy of Allergy,
Asthma & Immunology
800-822-ASMA
www.aaaai.org/physref
- American Academy of Pediatrics
800-433-9016
www.aap.org/referral
- American College of Allergy,
Asthma & Immunology
800-842-7777
www.aaaai.org/LocateAllergist

group is not appropriate." The risks and benefits must be considered. "If there is a high risk and support from a registered dietitian, restricting the diet may be worthwhile," Dr. Arshad says.

If the mother's milk is insufficient through the sixth month, a child at risk for developing food allergies will benefit from a formula where the proteins have been broken down or hydrolyzed. In one study, babies given extensively hydrolyzed formula showed significant reductions in allergic manifestations compared to those who used regular cow's milk formula.³ In most cases, however, formula is not superior to breast milk.

The American College of Allergy, Asthma and Immunology (ACAAI) recommends that the optimal age for introducing new foods should be at least six months. For children at high allergy risk, foods should be introduced one at a time, in small amounts, and on an appropriate schedule (e.g., 12 months of age for dairy products, 24 months for hen's

eggs and at least 36 months for peanuts, tree nuts, fish and seafood). It is helpful to keep a food journal so that you can see if there is a pattern developing. If an allergy is suspected, consult with a qualified allergist and seek guidance from a registered dietitian. ■

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von Berg A., Koletzko S., Grubl A., Filipiak-Pittroff B., Wichmann HE., Bauer CP., Reinhardt D., Berdel D., German Infant Nutritional Intervention Study Group. The effect of hydrolyzed cow's milk formula for allergy prevention in the first year of life: The German Infant Nutritional Intervention Study, a randomized double-blind trial. *J Allergy Clin Immunol*, 2003;111;467-470.



³ The Journal of Allergy and Clinical Immunology, 2003.

IG Reimbursement



By Michelle Vogel, MPH, and Kit-Bacon Gressitt

What do you do when your doctor tells you she or he can no longer afford to treat you with your lifesaving IVIG therapy? What if your provider does not help you transition to a new site of care? Will you be able to continue your treatments? Where can you find help?

If you have been dumped and do not know where to turn, you are not alone. The continuing access-to-care problems associated with the 2005 IVIG Medicare reimbursement rate reduction appear to have resulted in similar situations with patients across the nation. While many physicians have continued to treat patients at a loss and many others have successfully transitioned patients to other sites of care, the extended IVIG access crisis is causing some doctors to essentially give up, leaving patients to fend for themselves in confusing and often unresponsive territory. *IG Living* receives an average of one report per week of such cases, and they have seemingly spiked in Arizona lately. In this article we review three Arizona cases and their potential solutions.

Medicare

Ellen arrived for her scheduled IVIG infusion, and her physician surprised her with the message that he would no longer be providing IVIG to patients covered by Medicare, United Health Care and Access, Arizona's Medicaid program. He explained he was losing too much money and could no longer afford to treat these patients. Ellen was shocked and demanded that she at least receive her scheduled infusion that day. Good for her! She did receive her treatment, but what next?

Ellen contacted *IG Living*, and we learned that she has a primary immune deficiency disease (PIDD) and is a Medicare patient. She is doing well on the IVIG product that she had been receiving in her physician's office and she wants to continue receiving it, but she doesn't know who will give it to her or where.

Since her last treatment at her physician's office, Ellen was notified she had been referred to a homecare company that would provide her IVIG infusions, supposedly because the lower reimbursement rates haven't affected homecare companies.

This is incorrect; just the opposite is true for Medicare beneficiaries with PIDD. Homecare companies receive reimbursement only for the IVIG product, but, unlike physician offices and hospitals, homecare companies are not reimbursed for the nursing services needed to administer the product to patients with PIDDs. Secondary—not supplemental—insurance is necessary to cover nursing services for PIDD patient homecare. (A secondary insurance plan is normally

provided by an employer. A supplemental or Medigap policy is purchased by the beneficiary as a supplement to Medicare. Availability of supplemental policies varies by state.)

Consequently, Ellen's best option was to find a hospital near her home that is still treating Medicare patients with IVIG. Luckily for Ellen, her other physician offered to help. His office contacted a hospital that is treating with IVIG on an outpatient basis. Unfortunately, the hospital contracts for only one brand of IVIG, which is not the brand that Ellen had been receiving with good outcomes. The doctor's office is working with the hospital's pharmacy to see if they can special order the brand that Ellen needs.

If your healthcare provider tells you he or she is no longer able to administer your IVIG, we recommend you do the following:

1. Ask the provider to keep you as a patient until you successfully transition to a new site of care. Be prepared to firmly demand this.
2. Ask your current provider to assist you in securing your preferred IVIG product at your new site of care. Many hospital clinics do not provide the full range of products, but your current provider can advocate for you.
3. If you have a PIDD, and your current provider is transferring you to a homecare company, remember your IVIG product is not covered under Medicare Part D, but, rather, under Part B, which reimburses at a rate lower than many homecare companies find inadequate. Other diagnoses are covered under Part D at a higher rate.
4. If you are transitioned to the homecare setting, be prepared for the homecare provider to discuss your switching to subcutaneous immune globulin therapy (SCIG), which is reimbursed more favorably under Medicare.
5. Again, if you are transitioned to homecare, Medicare does not cover homecare nursing services for PIDD patients' IVIG administration. Instead, you will need to pay for a secondary health insurance policy, not a supplemental or Medigap policy.

From Private Insurance to Medicare

Catherine has a PIDD and has been receiving her IVIG infusions in the homecare setting, reimbursed by her private insurance for about one year. She just won her disability case and will be transitioning to Medicare. She has been notified by her homecare company that they can continue to treat her under Medicare—if she switches to a subcutaneous immune globulin product (SCIG). However, Catherine does not want to self-infuse subcutaneously; she wants to stay on her current brand of IVIG with which she has been doing well. Catherine's health is further compromised by her having severe

asthma and chronic obstructive pulmonary disease (COPD), which limits her ability to leave her home. Catherine has one additional complication: Her immune deficiency diagnosis does not match the diagnostic code with which her physician has been billing her insurance company.

Catherine was diagnosed with hypogammaglobulinemia and common variable immune deficiency (CVID). Both are specifically FDA-approved uses for IVIG. However, her physician has been billing with a diagnostic code that reflects an "unspecified immune deficiency."

Medicare recognizes only five diagnostic codes for reimbursing for IVIG homecare for primary immune deficient patients, and the code Catherine's physician has been using is not one of the five. So, Catherine is not currently eligible for homecare under Medicare. What should Catherine do?

Catherine should contact her physician and discuss her diagnosis with him to determine why there is a discrepancy between her diagnosis and the diagnostic code used on her insurance forms. If the physician accurately specifies by diagnostic code Catherine's conditions, she will be eligible for homecare under Medicare. This would allow her to continue being treated in her home, which has been successful so far.

We still have the problem of coverage for Catherine's nursing services in the home. Because Catherine has been covered by private insurance, we discussed her keeping that insurance as a secondary plan. The secondary plan would normally cover her homecare nursing services.

However, we came across another obstacle: Catherine's secondary insurance was forcing her to switch to a supplemental plan when her Medicare takes effect. So, the final option that we can explore for Catherine to cover her homecare nursing services is to determine if she meets the criteria under Medicare to be declared homebound. Because Catherine has severe asthma, COPD and other healthcare problems, she must take a breathing machine with her when she leaves her home, which, consequently, she does not do often. She will need to discuss with her physician the possibility of her meeting the criteria for being considered a homebound patient. The homebound designation is difficult to obtain; a patient's health must be compromised by leaving the home. If Catherine does meet the criteria, then her nursing services would be covered under Medicare Part A.

Finally, Catherine is not alone in being asked to switch from an IVIG product to an SCIG product. The current IVIG access crisis is resulting in some patients having to switch products to continue their immune globulin therapy. However, *IG Living* maintains that no patient should be forced to switch product because reimbursement rates favor one product over another. Treatment decisions should be made by physicians and their patients, not dictated by reimbursement rates.

Private Insurance

Kristine has a PIDD, but it is a subclass deficiency. When she began her IVIG therapy in her physician's office in 2004, her private insurer initially denied coverage based on her diagnosis, but she successfully appealed the decision and received her prescribed treatment.

Recently, Kristine's physician informed her he will no longer treat her in his office, because her private insurer reduced the IVIG reimbursement rate below the physician's cost. Kristine's physician referred her to a homecare company, but the homecare company and the insurer notified Kristine that they will not cover her IVIG treatment because it is considered "experimental and medically unnecessary" for her condition.

In essence, the change in site of care rendered Kristine's original appeal moot. The effective period for a successful appeal of a coverage denial varies by insurance company. Some companies require patients to re-appeal annually; some accept a successful appeal for the entire time a patient is with the insurance company; and other companies require new appeals when patients change site of care.

So, Kristine had to submit a new appeal. She did this, but she faced a lack of treatment during the appeal process. Consequently, we recommended Kristine take the following actions:

1. Ask her physician to continue to provide her IVIG therapy until she wins her appeal.
2. If No.1 fails, provide the homecare company documentation of her initial successful appeal, and ask the company to provide her treatments until the new appeal is successful.

However, Kristine was highly motivated to assure her treatments continued, and she discovered a third option: She contacted her state insurance commissioner's office and requested an expedited appeal decision due to her need for ongoing, regular treatments. Her insurer immediately approved a single infusion.

Unfortunately, the insurance company subsequently determined that Kristine's IVIG treatment for her subclass deficiency was indeed experimental and, consequently, would not be covered. The denial letter erroneously indicated there is no clinical evidence to support IVIG treatment for a subclass deficiency.

We are now helping Kristine prepare another appeal with the necessary supporting medical literature. When the new appeal is filed, Kristine can again request an expedited decision from her state insurance commissioner's office, so she can continue treatment during the appeal process. ■

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You Are Not Alone

As long as the IVIG access crisis continues, *IG Living* will continue to advocate for individual patients experiencing challenges to receiving their IVIG therapy. We are here to help. Just email editor@igliving.com or call 800-843-7477 x 1143.

ASK KRIS



Kris has two adult sons with chronic diseases treated with IG. Formerly a physical therapist assistant, Kris is an avid patient advocate and now works with NuFACTOR, a sponsor of *IG Living*. Kris is eager to find answers to your questions. Email them to editor@igliving.com or go to www.igliving.com and click on Ask Kris to send your inquiries. Your confidential information will not be used for any purpose but communicating with you about your questions.

Michelle: I have an autoimmune disorder called polymyositis, and I was wondering what the success rate is for people with my condition who use IVIG?

Kris: I posed your question to Dr. Scott Carlson of the Rockwood Clinic in Spokane, Wash. He gave the following response.

Polymyositis is one of the inflammatory muscle diseases. There are good studies done in a placebo-controlled fashion that show IVIG effective for dermatomyositis, with a response often seen within 15 days of treatment. Polymyositis is a less common disease and there are fewer studies to review, but this inflammatory myopathy also will respond to IVIG. Polymyositis is sometimes overdiagnosed and if a patient does not respond to IVIG, one should reconsider the diagnosis. The other inflammatory myopathy is inclusion body myositis, and unfortunately this disease does not respond well to IVIG.

Betty: My physician has me on special testing of B subset. I do not understand what this is.

Kris: Dr. Richard Schiff, an immunologist, kindly answered your question regarding B subset testing. He said the following:

Standard lymphocyte testing includes the T cell markers, CD3, CD4 and CD8; a marker for NK cells, CD16/CD56; and usually one B cell marker, CD19 or CD20. Both B cell

markers usually show how many B cells are in the blood. However, they do not give a very good indication of whether the B cells are immature, transitional, mature or memory cells. If we are looking for a disease such as XLA, then CD19 or CD20 is adequate—either the B cells are present or they are not. However, in diseases such as CVID, B cells are present, but they may not work very well. Thus, it can be helpful to look at the different subsets of B cells. For example, B cells that express another marker, CD27, are memory B cells. They often are decreased in patients with CVID, even though the total number of B cells is normal. Small children and some adults with CVID have B cells that also express CD5; these tend to be more immature. Other subsets can be identified by looking to see if the B cells express the immunoglobulins on their surface, IgM, IgD, IgA and IgG, and activation/growth markers such as CD38.

Although CD27 and CD5 can be evaluated in most flow cytometry labs, more complex analysis must be done in research laboratories where they have the equipment to simultaneously look at five, six or even seven markers at a time. Although these detailed analyses are teaching us a lot about B cells, much of this is still research and subject to different interpretations.

Actually it isn't as mysterious as it all sounds! Now, for the translation from my friendly pharmacist:

There are two ways that a defect can be present. One, there is too little of the substance such as B cells or immunoglobulins, or two, there is enough, but they are not working properly. Measuring serum levels of a substance is easy; measuring whether or not they are working is more difficult. By looking at subsets of B cells (similar to looking at IgG1, IgG2, IgG3 and IgG4 for immunoglobulins), you can find out more specifically what is at fault. The subsets, in essence, tell you that B cells may be immature or not working properly, and, based on that, a more definitive diagnosis can be derived. Depending on the diagnosis, the therapy may be different.

In the case of immunoglobulin deficiencies, if the problem is simply too little immunoglobulins in the blood, replacement with IVIG is effective. But for some genetic disorders, even though IgG levels may be low, replacement IVIG therapy is ineffective because the genetic defect is causing all sorts of other problems unrelated to immunoglobulin levels. In such a case, the therapy required is often bone marrow transplant. More relevant to Betty's question, we had a patient once with normal IgG and subclass levels, but when he was challenged with a substance to trigger the immune system into action, it responded very slowly. He responded nicely to IVIG replacement therapy. Today, there are special markers, similar to what Dr. Schiff discusses, that can even pinpoint further where the problem area is in the chain from bone marrow to immunoglobulin creation.

I hope this helps, Betty. I would add that I have learned from personal experience these tests can be expensive and are not always covered by insurance. Therefore, I would add a caution that if you have concerns, to always discuss them with your doctor. Many times a preauthorization from your insurance company can be requested before testing. It helps eliminate the surprise of an unexpected bill.

Florence: My daughter, aged 9, has been suffering from myositis since she was 6, and her best friend is receiving IVIG treatment, thanks to which she always gets better. Can you tell me if there is any risk whatsoever to IVIG infusion, such as HIV or meningitis or anything else, and in what probabilities?

Kris: Dr. Richard Schiff responded to your question.

There has not been any transmission of any virus (HIV, hepatitis C, etc.) with any of the currently available IVIG or SCIG products. This was recently addressed on the FDA website. However, at the same time, all man-

ufacturers have to acknowledge that this is a blood product and thus the risk is never zero. The risk of viruses, and variant Creutzfeldt-Jakob disease, is extremely low due to testing and the viral inactivation steps taken by manufacturers, but it will never be zero. Her concern with meningitis may be due to hearing about aseptic meningitis. It is not an infection but might be a chemical irritation or some sort of allergic reaction.

We also recommend reading the package insert of the product you and your physician choose for your daughter, and visit the manufacturer's website regularly for any updated product safety information.

Cyrus: I have been told that I have to receive five infusions of IVIG every month for five months. I would prefer to take this treatment at home, especially since the infusions are over five hours, as long as they are safe.

Kris: Home IVIG therapy is considered safe for many patients, as long as you have no severe reactions to the drug. Most IVIG patients start with infusions in a clinical setting to ensure ready access to medical care, should it be necessary. Severe reactions are rare, but they can happen. Once the physician and patient are comfortable with the treatment, homecare with an infusion nurse (not the physician) can be set up, as long as your physician and your insurance company agree. It is important to communicate your interest in homecare with your doctor as soon as possible. You will also need to talk with a representative from your insurance company to find out what their reimbursement guidelines are.

To read further about sites of care you can go to <http://www.aaaai.org/members/resources/initiatives/ivig.stm> and click on "Guidelines for the site of care of the administration of IVIG therapy." Although this article is based on patients with a primary immune deficiency disease, the guidelines are generally followed by many physicians who treat patients with IVIG. ■

Have questions about living comfortably with your IG therapy?
Send them to editor@igliving.com or visit Ask Kris at www.igliving.com.

Snog Died

By Dayna Fladhammer

Snog died last night. We learned of his passing when Charlie came in and announced, “I have bad news for you guys: Snog’s dead.”

Snog is Charlie’s imaginary friend, or, should I say, was his imaginary friend. Charlie and Snog were superhero co-pilots in a race to save the universe from the evil ruler Cruton, who was intent on turning the world into cheese. Snog apparently died in a spacecraft crash while on a mission.

It’s not entirely surprising that Snog has passed on, especially when you consider what our family has encountered in the last few months.

First, two months ago our dog had to be euthanized when she became incontinent and unable to walk anymore from the pain of her arthritis. The following month, the longtime companion of one of the kids’ grandmothers died after a short illness. Just a few weeks later, a very close family friend, Jerry, was tragically killed in a helicopter crash.

Explaining death to the kids has been a trying process. Along the way, we’ve received support and advice; we’ve done well; and we’ve made missteps.

When the dog died, it was our children’s first real experience with death. They watched her leave the house and wanted to know when she would return. As we tried to figure out what to tell the kids, we could only come up with what we should not say. We knew not to tell them the dog went to sleep or was put to sleep or that she got an IV and then died or that the doctor gave her medicine to make her sleepy so she could die.

We figured all of these would traumatize them for years, because they go to their own doctors, get their own IVs and take a multitude of medicines, some of which make them tired or sleepy.

In the end, I sat on the floor of the bathroom, hiding out from little ears that seem to hear everything, except “clean your room,” and called our pediatrician. I knew she could help us navigate the scenario. I explained what was going on and asked her for a little advice. I felt a little silly calling the doctor on a Sunday morning to figure out what to say, but I was really glad I did.

A few scripted words helped us to explain to the kids that the dog had died, that she had died with the veterinarian, and that the veterinarian had used special tools to help her die. The pediatrician warned me not to use the words doctor or medicine because, at the kids’ ages, they would not be able to distinguish the difference between their doctor and medicine and the dog’s. Do not, she instructed, tell the kids the dog died from a shot.

Additionally, I was warned that because of his age and interest in all things medical, Charlie would likely want to know the exact mechanism of death, which, of course he did. We very simply explained that her heart stopped working and she died.

It was less than a month later that we received the call that my mother-in-law’s longtime companion, Herb, was in the hospital with low oxygen levels. It was not a surprise, as he had congestive heart failure. So, we explained to the kids that he was very sick and in the hospital. The next day he died, and we once again found ourselves having to explain death to a 6-year-old and a 4-year-old. Thankfully, 2-year-old Tommy simply said, “OK, Herb dead,” and walked away. However, Charlie and Kate had many more questions, each of which had its own pitfall we tried to sidestep.

“Did he die in the hospital?”

If we answer yes to this one, I wondered, will they be afraid to go to the hospital? Will they associate death with a hospital? Should we explain that sometimes hospitals are where people die, when in their little eyes hospitals are where you go to get better?

“Why didn’t the medicine make him better?”

Can we say sometimes medicine doesn’t work to a 6-year-old who relies on medicine to live a life outside his house? Do we say the doctors did everything they could, but they just couldn’t save him, to a child who relies on the knowledge of doctors to keep him well? Can we tell them sometimes, when people get older, they die and that everyone dies someday—without their becoming preoccupied with the impending death of every elderly person we know?

Grandma, when are you going to die?



In the end, we did explain that, yes, Herb died in the hospital and that, yes, sometimes medicine cannot make you better. We also spent days going over the idea that Herb had a different sickness than they have.

Oh, and in case you're wondering, yes, we used the "death happens when people get older" line and it did not work well for our kids. I'm slightly embarrassed to admit that each of my kids has since asked an elderly person when he or she expects to die. While this is slightly funny, it has caught my 90-year-old grandmother off guard more than once.

The death of our friend, Jerry, was tragic and horrible. He was the kind of guy who stepped up when you needed him. He had twice spent IVIG treatment day helping me with the kids when my husband was unavailable.

However, his death was much easier to explain to the kids. It didn't revolve around a hospital or an illness. It did not have the potential to bring about fears of how they would feel about their own bodies and their own treatments. Of course, as any death does, it brought questions and worries, but we were not forced to deal with the kids' concerns about their own health issues.

Explaining death to any child can be difficult. When you add in the child's own health concerns and issues, it can be a minefield. But I realized, as I watched Charlie play out the death of his imaginary friend, Snog, that we've helped him acquire the coping skills to work out his emotions. He demonstrated the same skills three years earlier, playing IVIG on my husband following his first infusion. ■



Duck, Duck, Mantis!

By Mark T. Haggard

in to your mechanic. I'd hate to have something leaking somewhere and—Boom!—Haggard Fricassee."

My father pulled my golf bag from the car and muttered, "I found your problem."

I've been told that the maternal instinct resides in all females, although some more than others. But I'm beginning to wonder if my daughter's pharmacist is slipping more than plasma into her IVIG: Mothering hormones ooze from my PIDD daughter. Putting Molly to bed is a 15-minute trial in patience, as she tucks in each of her babies and sings them a lullaby. There are stuffed animals all over the floor covered by towels and pillow cases. It's a big slumber party!

We were reminded of Molly's mothering instinct during a summer road trip. Despite our giving the SUV a good once over, it seemed that our mechanic missed something: An ever-so-slightly noxious smell began infesting the cab as we made our way through Nevada. An overnight in the desert baked the mysterious smell into the upholstery, permeating the passenger compartment. I searched for the source of the odor the next morning but came up empty, and I almost lost my continental breakfast.

"Is the engine running rich?" my wife asked, wrinkling her nose.

"That's not gasoline," I replied.

As I rolled the windows down, I watched my two boys giggling and pointing at each other in the rearview mirror. "Dad," Calvin teased, "Did you—?"

"No!"

The mysterious odor became quite entertaining during our labored drive to California. Twenty Questions became Match That Smell.

That afternoon we pulled into Nana and Papa's house, and we began the process of unloading the family vehicle.

"Can you smell that?" I asked my father, whose happy countenance turned sour as he approached our automobile.

"Mmm, that odor reminds me of the farm on Sundays. Like chicken gizzards on the barbecue."

I pulled the tailgate open. "I might need to get the car

I took the bag from him, put my nose inside and inhaled. The hard gag that followed let me know that I was alive. "Oh yeah," I gasped. "That's it!"

I dumped the bag's contents onto the front lawn: clubs, balls, tees, two empty soda cans, and the shell of a duck egg, the rotten contents of which had dripped onto everything.

My mind ventured back to the day in late spring when we noticed that all but one of Daisy Duck's eggs had hatched and she had led her new family to the canal behind our house.

The unhatched egg disappeared that evening. Apparently, a month of summer weather and the contents of the egg had become a pungent potion on my father's front lawn.

It was just like Molly, our mom in training, to try to rescue an unhatched duckling. Her compassion goes beyond the mothering instinct, though. She has seen plenty of waiting rooms, examination rooms, infusion clinics and once-every-four-weeks needle pokes. These things have caused her to have greater compassion than most little girls. She has a keen sense of when someone is hurting and takes action to alleviate that hurt, in all creatures, great and small, feathered or not.

Back in the chill of winter, Molly had come into the house with a green object wrapped in tissue paper. "What's that?" I'd asked.

"A bug. He's sleeping."

It was a praying mantis, frozen stiff by the first frost of the Idaho winter. "Molly, he's not, uh..." I decided to let it go.

"I think all he needs is a little love and he'll be OK," Molly gently replied as she tucked the tissue paper snugly around the bug's body.

As we prayed and sang to the newest of Molly's babies, she said, "He's gonna be all right. I know how he feels."

She surely does. ■

The Myositis Association Anatomy of a Donor

By Kit-Bacon Gressitt

About four years ago, Roger Stevens went to see his doctor for some problems he was having. The doctor didn't tell him much, just sent him for some tests.

Two weeks later Stevens' doctor diagnosed him with inclusion body myositis (IBM).

This could explain why Stevens has become a donor to The Myositis Association (TMA), but there's more to Stevens' story than his diagnosis. He literally worked hard for his money.

"I didn't inherit it," he said. "I had nothing, and I started a construction company when I was 27 years old. It grew to be a very successful construction company, and with that comes money. I have the ability to help. The Myositis Association needs help and the research community needs funding."

Understanding the need is a key incentive to making a donation, and having a particular disease or a loved one with it is a common motivation to give. But Stevens also understands that the ability to give is an equally important motivation—at any level of giving.

"In order to try to solve this problem you have to pitch in. We can't do the science but we can come up with the money. Short and sweet, I see a small organization with a lot of people needing help. ...If someone can give five dollars, they give five dollars. If I can give half a million, I give half a million."

And that is exactly what Stevens has done: He pledged half a million dollars to the association to help fund myositis research. According to Bob Goldberg, executive director of TMA, Stevens' donation is a matching challenge grant, intended to increase the level and volume of donations to TMA's research efforts, and Goldberg believes Stevens' gift will indeed encourage other donors.

"Roger is a very sincere, unpretentious guy who is able to help, and there are probably other donors who can also afford to give larger amounts," Goldberg said. He hopes other donors who can afford it will increase the level of their giving. "I think it's going to be a very good year for myositis research."

Goldberg has committed much time this year to meeting potential large donors around the United States, explaining Stevens' commitment and encouraging matching donations. Goldberg estimates the effort to match Stevens' donation is now about 25 percent complete.

Meanwhile, Stevens is continuing to live with IBM, run his company and nurture the heirloom vegetables in his garden—as he nurtures the motivation to give in others.

"I'm a very fortunate person—even with IBM I feel very fortunate," Stevens said. "And you can't take it with you. I want to motivate other people to help out." ■

The Myositis Association

The mission of The Myositis Association (TMA) is to find a cure for inflammatory and other related myopathies, while serving those affected by these diseases. For more information, visit www.myositis.org.

To make a contribution of any amount to TMA, visit www.myositis.org or contact TMA at 202-887-0082.



Bone Marrow Transplant: *A Chance for Health*

By Lauren Gerstmann, MPH

Step into Children's Hospital of Cincinnati, with its giant bright murals and its ever-available supply of sanitizing wipes, and it is as though you have entered a family-friendly universe. This is a good thing, because for several months the hospital will be the only world that Aidan, Ashley, Conner and their families will know. Aidan, Ashley and Conner are all getting ready to undergo bone marrow transplants (BMTs) to treat their primary immune deficiencies. With luck, they will go from a life of chronic illness to full health, but it is a long and risky journey.

These are not your "normal" transplant patients. A quick Internet search for immune deficiency and BMT reveals articles on transplant in infants with severe combined immune disease (SCID). For kids with SCID, transplant is the only hope of survival, and it is usually completed before 1 year of age. While Aidan, Ashley and Conner all have severe immune deficiencies, none has SCID, and they are older than the typical primary immune deficient BMT patient. Aidan is 5 and both Ashley and Conner are 12.

It seems that only disease would have brought these families together. Conner's family lives in Indiana near the

University of Indiana, Ashley's family lives in Indiana near the Ohio border, and Aidan's family lives in Illinois. But, for the past several years they have gotten to know and support one another through an informal email network and through the word-of-mouth that connects families living with immune deficiency. The three families share a strong faith in both their doctors and in a Higher Power, and they believe the transplants will save their children's lives.

Bone marrow transplant may be used to treat deficiencies in the blood such as those caused by leukemia or aplastic anemia, but it can also be used to replace a damaged or failing immune system. Testing prior to BMT can help determine how well the donor's marrow will genetically match the patient's, and it can give some insight into the potential success of the transplant. The most successful transplants are between nonidentical siblings who have an identical tissue type. Tissue type is determined through HLA typing, a blood test that characterizes how a person's antigens tell the difference between normal body tissue and foreign tissue.¹ Aidan, who will receive banked umbilical cord blood cells from his little brother, Liam, is receiving that "perfect" match.

¹ www.webmd.com/a-to-z-guides/Tissue-Type-Test#hw40264.

To understand the immune system, you need to know how the body makes blood. Blood cells originate in the marrow of the bone, and bone marrow cells have the capability to turn into red blood cells, white blood cells or platelets.² Lymphocytes are white blood cells that play a critical role in the immune system. Specific types of lymphocytes, called B cells, make antibodies that neutralize molecules that would set off an immune response. Other lymphocytes, called T cells, actually activate destructive cells in the body to kill any dangerous cells that are infected, mutated or cancerous. Some immune deficiencies affect B cells and T cells (as do Conner's, Ashley's and Aidan's), while other deficiencies affect only B cells. After they receive their transplants, all three children should have bone marrow that functions normally, creating the healthy B and T cells that will enable them to fight infections.

BMT Outcomes

According to data from the Center for International Blood and Marrow Transplant Research, an international organization that tracks transplant outcomes, 75 percent of SCID patients who received an identical sibling transplant and 59 percent of SCID patients who received an unrelated donor transplant between 1995 and 2005 survived at least three years.³

A European report, looking at non-SCID immune deficiency transplant success rates from 1968 through 1999, found a 78 percent survival rate three years or more post transplant for perfect sibling matches. The three-year or more survival rate with unrelated donors ranged from 42 percent to 59 percent, depending on other characteristics of the match.⁴

Patients who do not have a sibling donor can search for a matched, unrelated donor on the National Marrow Donor Program (NMDP) Registry of volunteer donors. Since 1987, the NMDP has facilitated more than 25,000 transplants, and roughly 260 patients receive matches through the registry each month.⁵ From 1999 through 2004, the NMDP facilitated 245 unrelated donor transplants for patients with SCID. Other immune deficient diseases have been treated with unrelated transplants, but such instances are rare.

Because the science of BMT continues to advance, it helps to talk with the doctors who are actively performing BMT on a regular basis. Some of the most experienced doctors are Dr. Alexandra (Lisa) Filipovich and Dr. Jack Bleesing at Cincinnati Children's Hospital Medical Center. Aidan, Ashley and Conner are all undergoing transplants with this team.

Dr. Bleesing firmly believes that the safety of BMT is continuing to evolve. At Cincinnati Children's Hospital, children are carefully evaluated and treated to make sure they are as healthy as possible when they undergo transplant. Drs. Bleesing and Filipovich believe that BMT should be considered before their immune deficiency has caused so many illnesses that their entire system is compromised. Healthier patients make for better outcomes. Also, doctors have improved the safety of donor-to-patient matches, and have gained insight into infection prevention during the recovery process. According to Dr. Bleesing, success rates for unrelated matches at Cincinnati Children's Hospital continue to improve and continue to approximate those for matched sibling transplants (keeping in mind that this estimate lumps together a variety of immune deficiency disorders that are fundamentally different in nature).

Bone marrow transplant is a risky procedure because, before you give these children a new immune system, you must first eliminate any immune function that they have. Only when there is no host immune system will the grafted immune system take over. Prior to the transplant, each child will undergo a preparatory treatment consisting of immunosuppressive and chemotherapeutic drugs to destroy his or her immune system. During recovery, the child will be vulnerable to new infections and to re-activation of infections that he or she may have had prior to the transplant.

Preparing for BMT

The preparatory regimen before BMT is called conditioning, and the type and degree of conditioning is specifically designed for each type of immune system disorder. Some research has shown that BMT is more effective if the patient is not weakened by intense ➤

² <http://children.webmd.com/Allogeneic-stem-cell-transplant>.

³ Based on raw data on characteristics and outcome for patients with severe combined immune deficiency registered with the Center for International Blood and Marrow Transplant Research (CIBMTR) from 1995 to 2005. The data presented here are preliminary and were obtained from the CIBMTR. The analysis has not been reviewed or approved by the Advisory or Scientific Committee of the CIBMTR. The data may not be published without the approval of the Advisory Committees.

⁴ Antoine, C. et al. Long-term survival and transplantation of haemopoietic stem cells for immunodeficiencies: report of the European experience 1968–99, *The Lancet* Volume 361, Issue 9357, 15 February 2003, Pages 553-560.

⁵ The National Marrow Donor Program www.marrow.org/ABOUT/History/index.html.

conditioning.⁶ Ashley is benefiting from this research and will undergo a reduced intensity conditioning regimen (RIC). It means she should feel better prior to the transplant and should bounce back sooner. But, it also means that some of her original immune system and other bone marrow-derived cells may remain after transplant. Aidan will also undergo RIC. In his case, the emphasis will be on immunosuppressive rather than on chemotherapeutic drugs, to ensure there is no possibility that his immune system will reject the new bone marrow cells. It will also reduce the chance that Aidan's original, defective immune system will overwhelm the healthy transplant.

Drs. Filipovich and Bleesing believe that RIC is not an option for some patients. For example, in Conner's case, it is likely that the medical team will use traditional conditioning, a method Dr. Bleesing believes is a better approach in some circumstances: "If we think that the immune system is more globally defective; if we think that we need to replace more than the B cell and T cell components early on after BMT...[or in] disorders where we are not sure how everything works (and has been affected by the genetic defect). ...It is important to remember that we have been doing these things for 30 years. We did not wait until we completely understood the disease to start transplanting because we learned that [intense conditioning

followed by BMT] could take care of our patients."

Because of the difficulty, risks and expense of BMT, it is never easy for a doctor to prescribe or for a family to choose. Everyone wants to look to logical science for a clear decision, but there is some art involved. An important part of the decision is choosing the most experienced medical team possible. Aidan, Ashley and Conner's families were referred to the team at Cincinnati Children's Hospital because of the doctors' skills and experience and their cutting-edge BMT research. Cincinnati Children's has one of the largest pediatric BMT programs in the nation and is recognized for expertise in treating unusual disorders. In the summer of 2005, the program performed its 1,000th BMT since the transplant program was established in 1981.⁷

In a perfect world, where the risks could be minimized, the benefits maximized and the costs contained, every person who needs curative therapy for an immune deficiency might receive a brand-new functional immune system through BMT. But with the current state of knowledge, doctors must determine whether BMT is feasible and whether the benefits outweigh the risks. In a simplified scheme, patients can be assigned to three categories: patients who should receive a BMT as soon as possible (for example, people with specific, known disorders such as SCID); patients for whom BMT would eliminate the need for lifelong symptomatic (non-curative) therapy and who continue to have significant health issues (for example, certain severe forms of common variable immune deficiency, CVID); and patients for whom BMT would not be feasible (for example, because their immune deficiency disorder is not posing a significant burden on their health or daily life issues). As BMT research evolves, the distinction between these categories may evolve as well.

Once a family has made the decision to forge ahead with BMT, the family members will need a lot of support. Aidan, Ashley and Conner's families have been in touch throughout the decision-making process, and will continue to support each other through the transplant process. Although the children will not be together during their initial months after transplant (BMT patients are kept in isolation), they will be able to spend virtual time together through email and web pages provided in the hospital, and their families will be able to meet face-to-face. Each

About the National Marrow Donor Program (NMDP)

The NMDP offers a wide range of services to help patients and their families every step of the way, from diagnosis through survivorship. The NMDP also recruits people to its donor registry. To become a volunteer marrow or blood cell donor, a person must be between the ages of 18 and 60, meet health guidelines and be committed to donating to any patient in need. It's an opportunity to save a life.

To learn more about patient services or becoming a donor, visit www.marrow.org or call 800-MARROW-2 (800-627-7692).

⁶ Rao, et al, Improved survival after unrelated donor bone marrow transplantation in children with primary immunodeficiency using a reduced-intensity conditioning regimen, *Blood*, 15 January 2005, Vol. 105, No. 2, pp. 879-885.

⁷ www.cincinnatichildrens.org.

family is also receiving tremendous support from their friends, families and religious communities who have been very helpful in raising funds for this expensive procedure.

Ultimately, the three families are able to remain optimistic in the face of great risk because successful transplants could give their children the chance to have normally

functioning immune systems.

Conner put it succinctly when he explained to his mother, "Even if I go to heaven, it is OK, because at least I have a chance."

BMT may just be the chance that results in a long and healthy life for all three children.

Conner

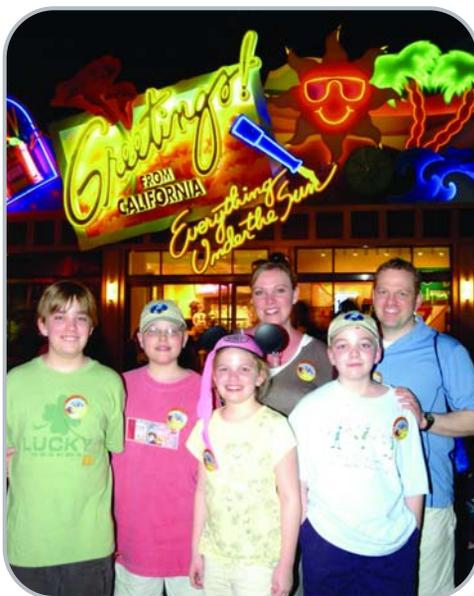
Conner is a triplet. He and his siblings were born almost two months early, and his repeated illnesses in infancy were attributed to his prematurity. He got sick a little more frequently than his brothers, and was always on and off antibiotics and breathing treatments, but it didn't seem unusual. When he was 9 years old, after having a cold, he suddenly became very sick and was hospitalized. He couldn't get enough oxygen, and the doctors never were able to discover why. Even after leaving the hospital, Conner was having significant trouble breathing. The hospital was a small community hospital in Lafayette, Ind., and the doctors were stumped.

A pulmonologist evaluated him, and immediately decided to measure his IgG levels, which were significantly low. A few months prior to his hospitalization, Conner had developed seizures and had gone on anti-seizure medication. When the family consulted a hematologist-oncologist to follow up on the low IgGs, he suggested that this was a transitory immune deficiency caused by the new medication. Conner went into semi-isolation to try to reduce his exposure to infection and build his immune system back up. Three months after he stopped the medication, Conner's IgG levels had still not risen. The family consulted with an immunologist, who advised that Conner's life would be in danger until he began IVIG therapy. CVID was a possible diagnosis.

The family then saw Dr. Melvin Berger, an immunologist at Cleveland Children's Hospital, who concurred with the CVID diagnosis. Dr. Berger mapped out a course of

treatment, including subcutaneous immune globulin therapy, but the family continued to search for more answers. When Conner was 10, the family decided to take him off treatment to see if there had been any improvement with his immune function. Within a few weeks, Conner developed bronchitis and then pancreatitis. His IgG levels had bottomed out again. Realizing that this was no transitory deficiency, they decided to consult Dr. Bleesing at Cincinnati's Children's Hospital. He found a defect in one pathway of Conner's complement system (a system that causes inflammatory response, eliminates pathogens and enhances the immune response⁸), and he also demonstrated that his natural killer cells and B cells weren't functioning normally.

While the family was trying to understand Conner's immune deficiency, his younger sister, Kelsey, became very ill with an unusual pneumonia. At that point, concerned about a genetic relationship, the family took all the kids to Cincinnati. When Dr. Bleesing tested the whole family, he discovered only slight depressions in IgG levels and questionable antibody responses, but all the kids had the defect in their complement system. So does Chris, Conner's dad. It is unclear if and how this deficiency affects people, or whether it is actually consistent with a clinical immunodeficiency disorder. Kelsey also has non-functional natural killer cells. Since she is doing well clinically, she has not yet started IVIG. The natural killer cell malfunction put Dr. Bleesing on the trail of some more specific immune deficiencies. Eventually, he found ➤



Conner (front row, right) with his family

⁸ www.emedicine.com/med/topic419.htm.

that Conner has a specific deficiency known as NEMO, a rare immunodeficiency that affects only boys. (No one knows why Kelsey is also having some symptoms.) This was a shock to the family since most other children with NEMO are much sicker than Conner. NEMO has been discovered only in the past decade, and it is believed to be fatal without treatment with bone marrow transplant. Even though Conner appears to have a less severe presentation, the doctors believed Conner might not be able to lead a full and normal life without a transplant. NEMO patients have been found to be vulnerable to damaging infections like tuberculosis. Development of a serious infection would greatly complicate BMT, so doctors at NIH urged the family to go through transplant while Conner is still healthy.

Conner has been looking for a cure since he first got sick and has been asking for BMT since he learned that he had an immune deficiency. He is willing to risk his life for a chance to have a normal life. He was thrilled with the news from NIH, and was really excited to talk with Dr. Filipovich. His family warned him about the chemotherapy conditioning and the risks in graphic detail, but Conner has never wavered, actively participating in his treatment decisions. He is already planning his DVD selection for his time of isolation in the BMT unit. His biggest concern is missing a year at school and his friends. Cincinnati is three hours from his hometown, but whether and when he will be able to have visitors depends on how he is doing.

It is helpful for Conner to go through this with other school-age kids so they can cheer each other along. Ashley is the same age as Conner. They met at the immunology department at Cincinnati Children's Hospital and have gotten together a few times since then. Conner is very open about transplant, but it is harder for Ashley to talk about it. Sharing it with him has made it easier for her to voice her fears. They plan to email and keep in touch by phone.

Ashley

Ashley was born full-term and healthy. But as an infant, she had chronic ear infections and had tubes placed when she was 6 months old. Her condition was then stable until she was a year old and she developed a cellulitis infection around her eye, landing her in the hospital. Right before she turned 2, she had a streptococcal and two blood infections. Her white blood cell and platelet counts were low, so the family went to Children's Hospital of Cincinnati, where they saw Dr. Karen Kalinyak, a hematologist. She asked for an immunology consult, and when Ashley was 3, she met Dr. Filipovich. Dr. Filipovich confirmed that

Ashley's immune system was compromised. Ashley's respiratory symptoms continued but responded well to antibiotics. When Ashley was 6, Dr. Filipovich told her family that she had CVID. Along with CVID, Ashley has severe lymphopenia—low levels of lymphocytes (white blood cells), which are important in regulating the immune system—caused by the T cells' inability to mature properly. Since she had already been hospitalized a few times with pneumonia, Ashley started IVIG.

As Ashley has grown older, she has done well, but she has had many side effects from the IVIG. When Ashley was 9, she developed debilitating headaches. At 10, she developed aseptic meningitis after an infusion. She missed five weeks of her treatments, and then switched to subcutaneous therapy, which helped alleviate the side effects. When frequent home infusions proved to be too long and painful, Ashley asked to switch back to office-based infusions. She immediately developed aseptic meningitis again. Now back on subcutaneous therapy, Ashley is doing really well, but her headaches have returned. The headaches, probably an inflammatory response to the infusions, are eased by regular steroid use. However, the amount of steroids Ashley needs to control her symptoms is not sustainable long-term.

Although Ashley is stable now, without a transplant soon, she will start having more health problems. Discussion about transplant started four years ago, when Dr. Filipovich first identified Ashley's immune deficiency, and she accepts the idea. A while ago, when Ashley asked if she would need infusions forever, her parents let her know that the infusions would continue unless Ashley received a new immune system via transplant. A few weeks after hearing it was time for the transplant, she was



Ashley with her parents and brothers

very concerned. But her strong spirit and spiritual faith are carrying her through. She does not let her disorder define her; it is only a small part of who she is.

Dr. Filipovich is optimistic that there will be a great outcome for Ashley. Shawna, Ashley's mother, says the doctors' biggest concerns are Ashley's social challenges, and the challenges that Ashley will face in adapting to the restrictive post-transplant environment. But Ashley has a matched donor from the national donor registry, so everyone is very positive about her long-term medical prognosis.

In the meantime, the family has developed rituals that help distract Ashley from her medical regimen. Every Friday night several friends come over to watch movies, eat takeout and keep Ashley company while she has her infusions. It takes a lot of the pressure off Ashley's parents, as she has so many wonderful cheerleaders.

Aidan

Aidan was a healthy infant until he developed a rare form of autoimmune anemia at 10 months. Although the family hoped this was an isolated diagnosis, Aidan soon lost platelet function and was diagnosed with Evans' syndrome (an autoimmune disease where the immune system attacks both red blood cells and platelets). When Aidan was a toddler, he developed Guillain-Barré syndrome, an acute neurological disorder that paralyzed him from the neck down. He immediately started treatment with IVIG.

In addition to the surprising number of autoimmune diseases that Aidan developed before he was 3, he also suffered an unusual number of infections. He saw an immunologist who diagnosed CVID and treated him with IVIG, even though his blood work was not typical of CVID. About a year ago, just before Aidan turned 5, the family consulted Dr. Gulbu Uzel, an immunologist at the National Institutes of Health, to learn if there was something amiss other than CVID. As soon as the family returned home from the appointment, Dr. Uzel called them and said, "I think he needs a bone marrow transplant." In reviewing Aidan's genetic tests, the doctor found that Aidan had a recombination activating gene (RAG) deficiency; she referred the family to Cincinnati Children's Hospital.

At Cincinnati Children's, Aidan is in good hands with Drs. Filipovich and Blessing. They are familiar with Aidan's autoimmune symptoms and his immune deficiency. Noting the RAG mutations, both doctors concur that Aidan needs a transplant. He has low levels of T cells, and his thymus does not output T cells as it should. Up until this point, Aidan has been very lucky that he has not suffered any of



Aidan (center) surrounded by his family

the more severe infections associated with a T cell defect. But, he has been developing new autoimmune conditions annually, and his T cell function continues to decline, leaving him increasingly vulnerable. So, the doctors want to transplant before his health spirals downward.

When Aidan's baby brother, Liam, was born, his bone marrow turned out to be a perfect genetic match, yet fortunately he did not have the same genetic defect. Liam's saved umbilical cord blood will provide the necessary cells for Aidan's transplant.

Aidan's mother, Amy, says family and friends have been a huge support. Aidan just wants to be a normal kid. He goes to school, plays with his brothers and cousins, and even participates in karate. He gets upset when kids tease him about some of his more obvious symptoms, like his vitiligo (a sun-sensitive skin autoimmune condition). To help, a teacher at his school presented a talk about respecting kids who are different and kids who have medical problems. Now Aidan is not teased so much, but he is scared. He knows he is going to feel "crummy" for a long time before he feels better. He understands he will be receiving blood from his baby brother to make him healthy, but he doesn't know the risks involved. His parents don't feel he is old enough at 6 to deal with that. But clearly, he is trying to comprehend it all. Aidan doesn't ask questions during his doctor appointments. He quietly listens and then fires questions at his parents on the ride home. His parents try to answer him as honestly as possible without distressing him even more.

Amy has high hopes the BMT will eliminate Aidan's autoimmune issues and that his B and T cells will return to normal. She hopes he'll end up with the normal immune system of a healthy child. ■

Yes, We Can!

By Ever Fecske

I never cease to be amazed by the many adventures on which our so-called condition takes us. Let me tell you about my latest travels in Common Variable Immune Deficiency Land...

Did you know the government has all sorts of benefits for people like us? They just keep it a secret or at least restrict it to a need-to-know basis. Well, I needed to know and you may need to know, too. I decided to go back to school, and it turns out the government has a budget just for furthering the education of people with disabilities.

Now, there are times when I have a hard time accepting this designation. I mean, are we really disabled? I am pretty self-sufficient. I guess I have had the wrong vision of "disabled" in my mind. Apparently, the definition of disabled isn't restricted to having a handicap placard in your car window. Well, in order to move forward, I needed to have a clear idea of what disabled means.

According to Webster it means incapacitated by injury or illness. That seems accurate: Sometimes, because of my illness, I am incapacitated. It's almost impossible to keep a regular job because of the way I feel on a daily basis and because of all of my weekly doctor appointments. But I still needed to see how the government defines disabled. So I made an appointment with the vocational rehabilitation office.

Vocational rehabilitation services programs (VRSPs) assist people with disabilities in many ways, it turns out, starting with helping a disabled person obtain and retain employment and maximize their ability to live independently. And the agency provides funding for training and education.

The first thing that happened when I went to my appointment was that I was assigned a counselor, Jackie. She is a 40-something straight shooter who finds joy in helping others succeed. She started our meeting by asking me to fill out an application.

Then she asked me about my career goals. This was going to be harder than I thought. Did she want me to decide right then and there what I want to do for the

rest of my life? The answer is yes. You see, when you use vocational rehab to get a college education, you have to indicate a college major on your application.

I told Jackie that I would like to be a writer. I got a sideways glance. Turns out, there was a catch. Being a freelance writer, like I am, is a respectable career, but there is no definite income or employer. There has to be a demand and a market for any career that you choose when it comes to qualifying for VRSP assistance. Nevertheless, where there is a will there is a way. I learned another secret: Even though you may choose to go to school to become a teacher or a nurse, because those fields are high in demand, that doesn't mean you have to actually become a teacher or a nurse.

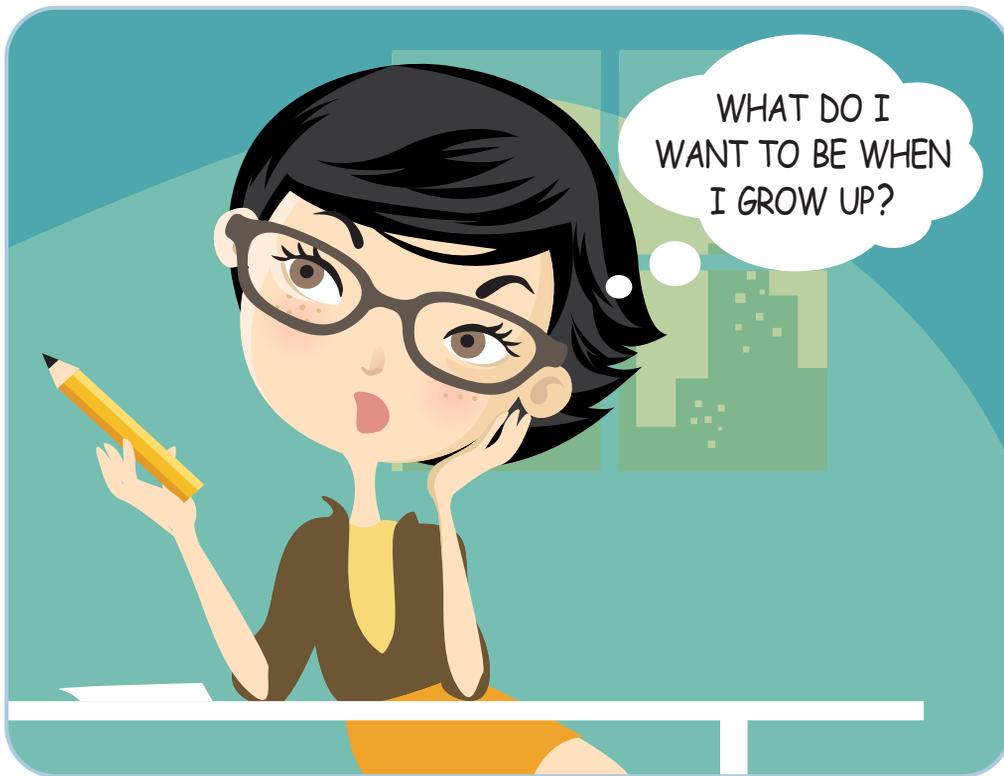
I wasn't able to come up with a career goal in the hour I spent with Jackie, so she scheduled an appointment with a social worker for me. And the adventure continued. They have social workers who give you tests to determine, based on your interests, your best possible choice for a major.

I took the tests during an hour-and-a-half meeting with my social worker, Richard. He is a very nice and patient man in his 60s. During our meeting, I found out he is a jazz pianist and he lived in Switzerland for eight years. He said the point of our meeting was for him to get to know me. He wanted to know my favorite author and books, my favorite movie, what kind of music I like. He wanted to know about my hobbies and what my ideal job would be if there were no restrictions.

So I told him that if there were no restrictions, I would want to be on Broadway!

I actually had fun in this meeting. He was supportive and encouraging. I will be the first person to admit that sometimes I need that little extra push and boost of confidence. That is something the VRSP offers, the confidence that I can get it done!

But the meeting didn't just end there; it got harder. I had to take a series of tests to determine my grade levels in math, reading, spelling and problem solving. I struggled



programs do make taking these important steps a lot less scary than I thought they'd be. They really are there to make sure we succeed. I believe I could succeed on my own, but when you throw all my health problems into the mix, it becomes so much more challenging and stressful. So why not get some wheels under our loads as we travel the same path as those with perfect health?

I was assigned a disability counselor at my school, Diane, and she helped me determine a tentative school schedule for all the required classes I need in order to transfer to a four-year

with some subjects, but I surprised myself: My results weren't that bad at all.

So, I went back to Jackie, who told me she needed a note from my doctor stating my diagnosis, and she asked me to briefly explain why I needed their help.

I told Jackie that I haven't been working because it's hard to keep a consistent schedule between how I feel on most days and my doctors' appointments, along with the many surprises of the illness I am fighting right now. I also told her that I was concerned about keeping a school schedule and that I might start with a light course load.

That's when she suggested online classes and setting up a meeting with a disability counselor at my school.

Listen to this: Let's say I have to miss an exam because I have my infusion on the same day. My disability counselor will contact my professor and ask him or her to schedule another time for me to take the exam. Pretty cool, don't you think?

Going through this adventure has made me realize how much there really is out there in the world to help us. We just have to be willing to seek it out.

By the way, Jackie and the other people who run these

university. Oh, and did I mention that vocational rehab will also pay for any state school I choose to go to? Really awesome! Diane and I discussed my school and career goals. She let me know that in most cases disabled students get priority when it comes to class schedule. That means I will never have to stress out about getting the classes I need. But I do have to inform my instructors of my condition. I don't really feel that will be necessary in every situation, but if it will help me get the best accommodations possible, then I guess I will have to suck it up!

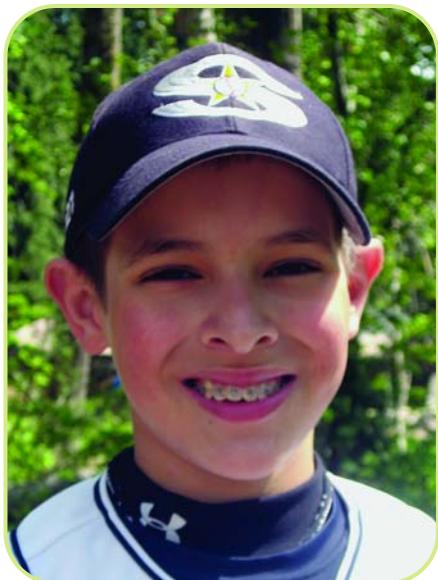
It has been two years now that I have been out of work and school and just dealing with doctors. I am ready to start again, and having supportive people at my side will only make it better.

If you are thinking about going back to school or if you need help finding a career, I strongly recommend that you apply to establish your own team of supporters. Contact the vocational rehabilitation services and disability counseling department at your school. It is so refreshing to hear someone say, "Yes, you can do this!"

Yes, we can! 📌

Let's Talk!

By Shirley German Vulpe, EdD



In this column, I interviewed 13-year-old Joshua and his mother, Ede. Their family goal is to celebrate normalcy!

Shirley: Thank you both very much for agreeing to this interview. Ede, can you tell me a little bit about Joshua's illnesses?

Ede: Sure, he was diagnosed three-and-a-half years ago with common variable immune deficiency (CVID). Thirteen months later a lung biopsy diagnosed a lung disease, often called lymphocytic interstitial pneumonitis.

Shirley: Was he sick a lot as a child?

Ede: Actually, not too much. Six months before his diagnosis, he had many ear and sinus infections. He took multiple antibiotics, and no sooner would one infection clear than another would start.

Shirley: That was a very quick diagnosis.

Ede: The diagnosis happened so quickly because his sister had hives. The medical investigation of the hives led to a diagnosis of IgA deficiency.

If your life depends on immune globulin, this column is for you! It is an opportunity to network and share our experiences, because it behooves us to learn as much as possible about all of the ramifications of our illnesses. This column allows us to learn from one another. If you have a story you'd like to share, please email us at editor@igliving.com.

Her main symptoms are multiple allergies and gastrointestinal. We have another child who has Down syndrome. The allergist decided to test the IgG levels of the whole family because of the genetic features of many allergic illnesses. Thus, the discovery that Joshua had CVID.

Shirley: What was the treatment?

Ede: He started IMIG therapy three months after diagnosis. He switched to subcutaneous [SCIG or subQ] two years later. His lung disease is treated with IV prednisone. The dose is slowly being decreased.

Shirley: Joshua, how do you like the SCIG?

Joshua: I prefer it to the IV. I feel more normal—not the ups and downs I had when I was treated with IVs. I can do treatment when I want. It lets me live a more normal life.

Shirley: Normal life?

Joshua: Yes, like other 13-year-old boys. You know, school and sports. I'm in grade seven. My favorite subjects are math and reading, but I like to play baseball more than anything.

Shirley: What position do you play?

Joshua: Shortstop and pitcher.

Shirley: What a coincidence. I played those same positions in grade school. What team do you play on?

Joshua: Cool! I play for the Seattle Stars, a select travel team.

Shirley: Wow! Sounds like fun!

Joshua: Yes. Everyone in our family tries to live a really normal life, even though we realize that our normal is different than most families. I try not to let my illness run my life or define who I am!

Shirley: Do you do your SCIG for yourself?

Joshua: I order supplies, prepare supplies and start the pump. My mom usually puts the needles in. I prefer to avoid that, but I do it if I need to when mom's not available.

Shirley: Has taking the medication helped?

Joshua: Very much. I get a cold occasionally, but it only lasts a short time. Just like the other kids. The subQ administration lets me plan my IgG treatments around my baseball.

Shirley: Wow, Joshua, I have certainly been impressed by your maturity. You set a great example for me! Any final words about what having a chronic illness means to you?

Joshua: It's taught me responsibility, to take care of myself and to appreciate life more! An illness does not take you over. It is part of you, and you do what you have to do. Getting shots is no big deal. They are just a part of my normal life. ■

Resource Directory

Guillain-Barré Syndrome (GBS)

Websites and Chat Rooms

- The GBS/CIDP Foundation International, www.gbsfi.com, has 23,000 members in 160 chapters on five continents. 610-667-0131
- The GBS Foundation Discussion Forums provide the opportunity to talk to other GBS patients and learn more about ways to manage the illness: www.guillain-barre.com/forums.
- Miller Fisher syndrome, considered to be a variant of GBS, is explained on the National Institute of Neurological Disorders and Stroke, NINDS Miller Fisher Syndrome Information Page: www.ninds.nih.gov/disorders/miller_fisher/miller_fisher.htm.

Online Pamphlets

- The National Institute of Neurological Disorders and Stroke has a GBS Fact Sheet at www.ninds.nih.gov/disorders/gbs/detail_gbs.htm.
- The Mayo Clinic has an overview of Guillain-Barré syndrome at www.mayoclinic.com/health/guillain-barre-syndrome/DS00413.
- The National Institute of Neurological Disorders and Stroke has an information page about CIDP: www.ninds.nih.gov/disorders/cidp/cidp.htm.

Online Peer Support

- GBS & CIDP Discussion Forum – UK Bulletin Board – For Ireland and England www.gbs.org.uk/cgi-bin/ikonboard3/ikonboard.cgi
- GBS Support group – UK • Chat room – requires registration <http://www.jsmarcussen.com/gbs/uk/chat.htm>
- GBS Foundation Discussion Forums: www.guillain-barre.com/forums
- Yahoo Support Group Discussion Board http://health.groups.yahoo.com/group/GBS_CIDP

Books and Articles

- "A Handbook for Care Givers," by Helen Rummelsburg, gives an account of caring for her husband and offers helpful hints for patients and caregivers. Available at the GBS website bookstore at www.gbsfi.com.
- "Bed Number Ten," by Sue Baier, provides a view of long-term care through the eyes of a patient totally paralyzed with GBS.
- "Caring for a Child With GBS," by Patricia Schardt, is a short guide written by a mother of a child with CIDP. Available at the GBS website bookstore at www.gbsfi.com.
- "No Laughing Matter," by Joseph Heller (the best-selling author of Catch-22), who teamed up with Speed Vogel, his best friend, to describe Heller's battle with and triumph over GBS.

ITP (Idiopathic Thrombocytopenic Purpura)

Websites

- ITP Support Association, UK: www.itpsupport.org.uk
- Platelet Disorder Support Association: www.ITPpeople.com 87-PLATELET (877-528-3538) or 301-770-6636

Online References

- Idiopathic thrombocytopenic purpura http://en.wikipedia.org/wiki/Idiopathic_thrombocytopenic_purpura
- Idiopathic Thrombocytopenic Purpura: Michael A. Silverman, MD www.emedicine.com/EMERG/topic282.htm

- Immune Thrombocytopenia: Current Understanding and Future Research, Robert S. Mocharnuk, MD www.medscape.com/viewarticle/459249
- Infusion Network Systems article: The Expanding Use of IVIG provided by ZLB Bioplasma, Inc. www.infusionsystems.net/article-ExpandingUseofIVIG.html
- ITP: Idiopathic Thrombocytopenic Purpura: <http://familydoctor.org/113.xml>

Kawasaki Disease

Websites

- Kawasaki Disease Foundation: www.kdfoundation.org PO Box 45 • Boxford, MA 01921 Tel: 978-356-2070 • Fax: 978-356-2079 • Email: info@kdfoundation.org
- Kids Health discusses Kawasaki disease at www.kidshealth.org/parent/medical/heart/kawasaki.html.
- Overview from the American Heart Association focuses on how the disease affects the heart. www.americanheart.org/presenter.jhtml?identifier=4634

Mitochondrial Disease

Websites

- United Mitochondrial Disease Foundation promotes research and education for the diagnosis, treatment and cure of mitochondrial disorders and provides support to affected individuals and families. www.umdf.org
- The Cleveland Clinic website provides many articles when searched by the topic, "mitochondrial disease." www.clevelandclinic.org/health
- The National Institute of Neurological Disorders and Stroke provides an informative Mitochondrial Myopathies Information Page. www.ninds.nih.gov/disorders/mitochondrial_myopathy/mitochondrial_myopathy.htm

Multiple Sclerosis (MS)

Websites and Chat Rooms

- The mission of the National Multiple Sclerosis Society is to end the devastating effects of MS. www.nationalmssociety.org/
- All About Multiple Sclerosis provides accurate and comprehensive medical information about MS written in plain English by people living with the disease and its symptoms. www.mult-sclerosis.org/index.html
- Multiple Sclerosis Foundation works for a brighter tomorrow for those affected by MS. www.msfacts.org
- Multiple Sclerosis Association of America seeks to enrich the quality of life for individuals with multiple sclerosis. www.msaa.com
- MSWorld's Chat and Message Board features patients helping patients. www.msworld.org

Online Peer Support

- Friends with MS website: <http://friendswithms.com> forum: <http://health.groups.yahoo.com/group/FriendsWithMS>
- My MSViews website: www.mysviews.org forum: http://health.groups.yahoo.com/group/MSViews_Multiple_Sclerosis
- MS Support Group: <http://health.groups.yahoo.com/group/mscured>

- The MS Carousel—A Place to Meet With People Who Understand MS!
<http://health.groups.yahoo.com/group/themscarousel>

Myasthenia Gravis (MG)

Websites and Chat Rooms

- The Myasthenia Gravis Foundation of America (MGFA) is the only national volunteer health agency dedicated solely to the fight against (MG). www.myasthenia.org
- Myasthenia Gravis Fact Sheet prepared by National Institute of Neurological Disorders and Strokes.
www.ninds.nih.gov/disorders/myasthenia_gravis/myasthenia_gravis.htm
- Mayo Clinic's overview of myasthenia gravis:
www.mayoclinic.com/health/myasthenia-gravis/DS00375

Online Peer Support

- MGFA's Forum: <http://health.groups.yahoo.com/group/MGnet>
- Bette's Myasthenia Gravis Support:
<http://health.groups.yahoo.com/group/bettesmyastheniagravisupport>
- Maddy's MG Support: <http://health.groups.yahoo.com/group/maddysmgsupport>
- Autoimmune Information Network Inc.: www.aininc.org
PO Box 4121 • Brick, NJ 08723 • 877-246-4900
Email: autoimmunehelp@aol.com

Myositis

Websites

 <p>THE MYOSITIS ASSOCIATION</p>	<p>The mission of The Myositis Association, www.myositis.org, is to find a cure for inflammatory and other related myopathies, while serving those affected by these diseases. 202-887-0088</p>
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- International Myositis Assessment and Clinical Studies Group is a coalition of healthcare providers and researchers with global approaches to improved treatments and understanding of myositis:
<https://dir-apps.niehs.nih.gov/imacs/index.cfm?action=home.main>.
- The Cure JM Foundation was created specifically to find a cure for Juvenile Myositis (JM), while also providing support and information for families affected by JM. <http://curejm.com>
- Johns Hopkins Myositis Center is a new patient treatment center that brings the expertise of rheumatologists and neurologists into a single clinic for patients with inflammatory (autoimmune) and toxic (drug induced) muscle conditions.
www.hopkinsmedicine.org/rheumatology/clinics/myositis_center.html

Online Peer Support

- Juvenile Myositis Family Support Network:
www.curejm.com/family_support/index.htm
- Myositis Association Community Forum: www.myositis.org
- Myositis Support Group: www.myositisupportgroup.org
- Myositis Support Group UK: www.myositis.org.uk
- Yahoo Myositis Support Group Discussion Board:
<http://health.groups.yahoo.com/group/OurMyositis>
- The California Myositis Symposium held in 2005 was captured on DVD. It contains information about polymyositis, dermatomyositis and inclusion body

myositis, including doctors' discussions and detailed slides and explanations of muscle biopsies, skin rash, and tools used to diagnose these diseases. Other presentations offer valuable lessons in maintaining a positive attitude, exercises for physical therapy and innovative tools to aid in everyday activities. The DVD is available at no charge by sending an email to Richard Gay at rgay@socal.rr.com.

Books and Articles

- "Coping With a Myositis Disease," by James R. Kilpatrick, is written by myositis patients telling their personal stories.
- "Inclusion-Body Myositis and Myopathies," by Valerie Askanas (Editor), Georges Serratrice (Editor) and W. King Engel (Editor), is devoted to discussing the two forms of inclusion-body myositis.
- "Living With Myositis," edited by Jenny Fenton, is an accessible, realistic and sympathetic guide to facts, feelings and future hopes.
- "Myositis—A Medical Dictionary, Bibliography, and Annotated Research Guide to Internet References," by ICON Health Publications, is a three-in-one reference book: a complete dictionary of terms relating to myositis, a list of bibliographic citations about the disorder and a guide to Internet resources.
- "Myositis and You: A Guide to Juvenile Dermatomyositis for Patients, Families, and Healthcare Providers" is an indispensable resource for families of children with myositis and their healthcare teams is available at www.myositis.org.
- "The Official Patient's Sourcebook on Inclusion Body Myositis," by James N. Parker (Editor) and Philip M. Parker (Editor), is a reference manual for self-directed patient research.

Peripheral Neuropathy (PN)

Websites

 <p>The Neuropathy Association</p>	<p>The Neuropathy Association, www.neuropathy.org, is devoted exclusively to all types of neuropathy, which affects upwards of 20 million Americans. The Association's mission is to increase public awareness of the nature and extent of PN, facilitate information exchanges about the disease, advocate the need for early intervention and support research into the causes and treatment of neuropathies. 212-692-0662</p>
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- To learn about PN, how it is classified, the symptoms, causes and treatments, see the Peripheral Neuropathy Fact Sheet available at www.ninds.nih.gov/disorders/peripheralneuropathy/peripheralneuropathy.htm.
- The Neuropathy Action Foundation, at www.neuropathyaction.org, educates, empowers and informs patients and physicians about neuropathy.

Support Groups

- Click on the Member Services tab of the website, www.neuropathy.org, for listings of support groups across the nation.
- The Neuropathy Action Foundation, at www.neuropathyactionfoundation.org, educates, empowers and informs patients and physicians about neuropathy.

Online Peer Support

- Calgary Neuropathy Support Group: www.calgarypnrs.org/index.htm
- MSN Support Group Discussion Board: <http://groups.msn.com/PNPARTNERS>
- The Neuropathy Association Bulletin Board: www.neuropathy.org
- Yahoo Neuropathy Support Group Discussion Board:
<http://health.groups.yahoo.com/group/neuropathy>
- Yahoo Support Group – Australia Discussion Board:
<http://au.groups.yahoo.com/group/LifeWithPN>

Books and Articles

- "If You're Having a Crummy Day, Brush Off the Crumbs!," by Mims Cushing, is a how-to book that offers more than 75 ways to help people get through the days when neuropathy (or other ailments) is particularly difficult.
- "Medifocus Guide to Peripheral Neuropathy," is a guide to current and relevant PN research, organized into categories for easy reading.
- "Numb Toes and Aching Soles," by John Senneff, discusses the symptoms, causes, tests, treatments and coping strategies for peripheral neuropathy.
- "Numb Toes and Other Woes," by John Senneff, is the second in a series of three books. It focuses on clinical findings and treatment strategies for PN.
- "Nutrients for Neuropathy," by John Senneff, the third in the Numb Toes series, is focused exclusively on nutrient supplementation as a means for managing PN.

Primary Immune Deficiency Disease (PID)

Websites and Chat Rooms



The Immune Deficiency Foundation (IDF), www.primaryimmune.org, is dedicated to improving the diagnosis and treatment of PID through research and education. 800-296-4433



The Jeffrey Modell Foundation, www.info4pi.org, is dedicated to early and precise diagnosis, meaningful treatments and, ultimately, cures for primary immunodeficiency. 212-819-0200

- The National Institute of Child Health and Human Development (NICHD), www.nichd.nih.gov, is part of the National Institutes of Health. Go to the "Health Information and Media" tab on the website and do a search under "primary immunodeficiency."
- The American Academy of Allergy, Asthma & Immunology, www.aaaai.org, has a helpful Q&A section on its website, with resources and tips for those with various immune deficiencies.
- The Michigan Immunodeficiency Foundation, www.midf.org, seeks to improve the quality of life for Michigan residents affected by PID.
- The International Patient Organization for Primary Immunodeficiencies (IPOPI), www.ipopi.org, promotes the worldwide improvement in the care and treatment of PID patients.
- To connect to a PID message board, go to www.info4pi.org.
- To chat with peers on IDF's Forum, go to www.primaryimmune.org/forums/forum_intro.htm.
- Chat with parents of children affected by primary immune deficiency at <http://health.groups.yahoo.com/group/PedPID>.
- Chat with peers with PID at <http://health.groups.yahoo.com/group/PIDsupport>.
- A group of family and friends of patients with primary immune deficiencies maintains a nonprofit network in the New England area: www.nepin.org
- Baxter's website, www.immunedisease.com, offers in-depth information on immunology, PID and treatment with intravenous immune globulin. Click on "European" to see SCIG information.

Online Pamphlets and Education

- Go to the National Institute of Allergy and Infectious Diseases site at www.niaid.nih.gov and search for "primary immune deficiency."
- "Understanding the Immune System: How It Works," by the U.S.

Department of Health and Human Services, is found at www.niaid.nih.gov/publications/immune/the_immune_system.pdf

- "NIAID Initiative Addresses Primary Immune Deficiency Diseases by National Institute of Allergy and Infectious Diseases" is located at <http://www3.niaid.nih.gov/news/newsreleases/2003/pirc.htm>
- The "Immunodeficiency in Pediatrics" program (PREP®) Audio series is a new pediatrician education program that can be obtained by contacting the American Academy of Pediatrics at 866-843-2271 or visiting www.prepaudio.org.

Online Peer Support

- Chat with parents of children affected by PID <http://health.groups.yahoo.com/group/PedPID/>
- Chat with peers with PID: <http://health.groups.yahoo.com/group/PIDsupport/>
- Immune Deficiency Foundation Forum www.primaryimmune.org/forums/forum_intro.htm
- Jeffrey Modell Foundation Message Board: www.info4pi.org

Books and Articles

- "21st Century Complete Medical Guide to Primary Immune Deficiency, Severe Combined Immunodeficiency (SCID), Chronic Granulomatous Disease (CGD), for Patients and Physicians," by PM Medical Health News, contains federal government clinical data and practical information for patients and physicians.

Stiff-Person Syndrome (SPS)

Websites

- American Autoimmune Related Diseases Association Inc., www.aarda.org, is the only national organization dedicated to addressing the problem of autoimmunity. 800-598-4668 aarda@aarda.org
- Autoimmune Information Network Inc., www.aininc.org, helps patients and family cope with the disabling effects of autoimmune diseases. 732-262-0450 autoimmunekhelp@aol.com
- National Association for Rare Disorders (NORD), www.rarediseases.org, promote awareness of rare diseases and the need for research. 800-999-6673 orphan@rarediseases.org
- National Institute of Neurological Disorders and Stroke (NINDS), www.ninds.nih.gov, offers treatment, diagnosis and research information for rare diseases. 800-352-9424 braininfo@ninds.nih.gov
- Diagnosed with SPS in 1994, Debra Kemery recounts her experience and offers practical information about coping with the disease at www.stiffman.org.

General Resources

Product Information

- Influenza and the influenza vaccine www.cdc.gov/flu or call 800-CDC-INFO (800-232-4636)
- IVIG Carimune NF www.carimune.com
- IVIG Flebogamma www.grifolsusa.com/flebogamma.htm
- IVIG Gammagard Liquid www.gammagardliquid.com
- IVIG Gammagard S/D www.immunedisease.com/US/therapies/gammagard/index.html
- IVIG Gamunex www.gamunex.com
- IVIG Octagam www.octapharma.com/corporate/03_products_and_therapeutic_areas/01_immunoglobulin_product_line/03_octagam.php
- SCIG (subcutaneous immune globulin) Vivaglobin www.vivaglobin.com

Other Organizations

- For suggestions on how to deal with the medical and emotional impact of caring for an ill child, go to www.kidshealth.org/parent/system/ill/seriously_ill.html.
- The National Committee for Quality Assurance provides free access to detailed report cards on health plans, clinical performance, member satisfaction, access to care and overall quality on its Health Plan Report Cards Online at www.ncqa.org.
- The nonprofit Patient Advocate Foundation, www.patientadvocate.org, seeks to assure patient access to care, maintenance of employment and financial stability. 800-532-5274



The nonprofit Patient Services Incorporated, www.uneedpsi.org, specializes in health insurance premium, pharmacy co-payment and co-payment waiver assistance for people with chronic illnesses. 800-366-7741

- WebMD, www.webmd.com, is a handy medical reference that helps consumers take an active role in managing their health by providing objective healthcare and lifestyle information.
- For a pediatrician's guide to your child's health and safety, visit www.keepkidshealthy.com.
- The National Organization for Rare Diseases, at www.rarediseases.org, provides links to numerous other organizations that have disease-specific support groups and virtual communities for patients and caregivers.
- American Autoimmune Related Diseases Association (AARDA) www.aarda.org brings national focus to autoimmunity through research, education and patient services. 800-598-4668
- Band-Aides and Blackboards, www.lehman.cuny.edu/faculty/jfleitas/bandaides, started by a nurse working on her dissertation, focuses on children with chronic illness.

Education and Disability Resources

- Continuation of Health Coverage—Consolidated Omnibus Budget Reconciliation Act (COBRA): www.dol.gov/dol/topic/health-plans/cobra.htm
- Social Security: www.ssa.gov/disability
- California State Disability Insurance (SDI): www.edd.ca.gov (Please note that each state has a different disability program.)
- IDEA 2004 Resources: www.ed.gov/policy/speced/guid/idea/idea2004.html
News and information on the Individuals with Disabilities Education Improvement Act of 2004 (IDEA), the nation's law that works to improve results for infants, toddlers, children and youth with disabilities.
- The National Disabilities Rights Network: www.ndrn.org. This website offers a search tool to find resources in your state to assist with school rights and advocacy.
- U.S. Department of Education Website: www.ed.gov. This website, a U.S. federal government website, offers a parents section that has a subsection titled "My Child's Special Needs" that can be most helpful.
- U.S. Department of Health and Human Services, Office of Civil Rights, www.hhs.gov/ocr/504.html, spells out your rights under Section 504 of the Rehabilitation Act.
- Wrightslaw: www.wrightslaw.com. Parents, educators, advocates, and attorneys come to Wrightslaw for accurate, reliable information about special education law and advocacy for children with disabilities.
- The Americans with Disabilities Act of 1990
Provides protection for people with disabilities from certain types of discrimination and requires employers to provide some accommodations of the disability. For more information, visit www.usdoj.gov/crt/ada/adahom1.htm.

Additional Reading

- "Anatomy of an Illness," by Norman Cousins, is a best-seller about overcoming illness and the triumph of the human spirit. The premise is that the human mind is capable of promoting the body's capacity for combating illness and healing itself even when faced with a seemingly hopeless medical predicament.
- "A Parent's Guide to Special Education: Insider Advice on How to Navigate the System and Help Your Child Succeed," by Linda Wilmschurst and Alan W. Brue, is available on Amazon.com.
- "The Confused Consumer's Guide to Choosing a Health Care Plan: Everything You Need to Know," by Martin Gottlieb, helps consumers through the confusing maze of choosing a healthcare plan.
- "The Everyday Guide to Special Education Law," by Randy Chapman, Esq., makes the law accessible to parents so they can be more effective advocates for their children. Available at www.thelegalcenter.org/thelegalcenter-cgi-bin/shop?item=15.
- "Living Creatively With Chronic Illness: Developing Skills for Transcending the Loss, Pain and Frustration," by Eugenia G. Wheeler, is a self-help book specifically designed to help the chronically ill, their families, friends, counselors, medical personnel and the clergy.
- "Managing Pain Before It Manages You," by Dr. Margaret A. Caudill, is a wellspring of wisdom and practical approaches that can help transform your life and your pain.
- "Not Dead Yet: A Long Strange Trip From Doctor to Patient and Back Again," by Dr. Robert Buckman, an oncologist and comic writer, is a witty account of his life as a doctor and autoimmune disease survivor.
- "Pride and the Daily Marathon," by Jonathan Cole, describes how Ian Waterman was suddenly struck down at work by a rare neurological illness that deprived him of all sensation below the neck, and how he reclaimed a life of full mobility.
- "Proneia Is the Antidote for Paranoia," by Rob Breznsny, explores the best way to attract the blessings that the world is conspiring to give us.
- "When You're Ill or Incapacitated" comprises one-half the booklet it shares with "When You're the Caregiver," both written by James E. Miller, suggesting 12 things to remember or do in each role.
- "YOU the Smart Patient: An Insider's Handbook for Getting the Best Treatment," by Michael F. Roizen, MD, and Mehmet C. Oz, MD, with the Joint Commission on Accreditation of Healthcare Organizations, shows you how to tackle such healthcare decisions as picking the best doctors and hospitals for you, knowing when to get a second opinion, and more.

IG Manufacturer Websites

- Baxter: www.baxter.com
- CSL Behring: www.cslbehring.com
- Grifols: www.grifolsusa.com
- Octapharma: www.octapharma.com
- Talecris: www.talecris.com

Pump and Infusion Sets Websites

- EMED Corporation: www.safetymedicalproducts.com
- Graseby Marcal Medical: www.marcalmedical.com
- Intra Pump Infusion Systems: www.intrapump.com
- Repro Med Systems, Inc: www.repro-med.com
- Norfolk Medical: www.norfolkmedical.com

Medical Research Studies

- Check out the official website for the National Institutes of Health patient recruitment program. This site provides summaries and criteria for studies as well as the ability to search for studies being conducted for a specific disease or disorder. <http://clinicalstudies.info.nih.gov>

- This website provides a wealth of information about clinical trials and volunteer participation. It gives you the ability to specify the disorder you are interested in, the location of the study, and the medication names or research protocols. www.centerwatch.com
- This site has a registration form to request that you be notified about recruitment for future studies. www.clinicaltrials.com
- WebMD has a service that matches volunteers with trials. There is an online questionnaire to complete and you will be notified via email of upcoming studies that match the criteria of your questionnaire. You can also search for specific studies. www.webmd.com

Food Allergies

- Allergic Disorders: Promoting Best Practice www.theallergyreport.com/reportindex.html
- American Partnership for Eosinophilic Disorders: www.apfed.org
- National Institutes of Health, National Institute of Allergy and Infectious Diseases (2004). Food Allergy: An Overview (NIH Publication No. 04-5518). www.niaid.nih.gov/publications/pdf/foodallergy.pdf
- Sicherer, S.H. (2006). "The Complete Peanut Allergy Handbook: Understanding and Managing Your Child's Food Allergies," Johns Hopkins Press.
- Food Allergy and Anaphylaxis Network: 800-929-4040 www.foodallergy.org
- World Allergy Organization: www.worldallergy.org

Nutrition and Food Safety

- American Board of Physician Nutrition Specialists: www.ipnec.org
- American Dietetic Association: www.eatright.org
- American Gastroenterological Association: www.gastro.org
- North American Society for Pediatric Gastroenterology Hepatology and Nutrition: www.naspgn.org/
- Bovine spongiform encephalopathy (BSE) www.aphis.usda.gov/newsroom/hot_issues/bse.shtml
- Childhood choking prevention: www.ific.org/publications/brochures/index.cfm
- American Academy of Allergy, Asthma & Immunology (AAAAI): www.aaaai.org Patient Information and Physician Referral Line: 800-822-2762
- The Food Allergy & Anaphylaxis Network: www.foodallergy.org 800-929-4040
- U. S. Food and Drug Administration, Center for Food Safety and Applied Nutrition www.cfsan.fda.gov Food Information Line (24 hours): 888-SAFEFOOD
- USDA Meat and Poultry Hotline: www.IsItDoneYet.gov 888-MPHotline 888-674-6854 · TTY: 800-256-7072
- Dietary Guidelines for Americans, 2005 Edition: www.health.gov/dietaryguidelines
- Drug Nutrient Interactions (MedlinePlus & the Physician Desk Reference): www.nlm.nih.gov/medlineplus/ or <http://www.pdrhealth.com>
- Electronic menu planner: <http://hp2010.nhlbi.nih.net/menuplanner/menu.cgi>
- Modified Food Pyramid for Older Adults (Tufts): <http://nutrition.tufts.edu/consumer/pyramid.html>
- The American Dietetic Association or to find a registered dietitian in your area: www.eatright.org or 800-877-1600
- USDA: MyPyramid personal nutrition tracker: www.mypyramid.gov

Nutrition Assistance Programs for the Elderly

- Community-based services can be located through the Eldercare Locator: www.eldercare.gov 800-677-1116.
- Nutrition, Aging, and Assistance: <http://nutritionandaging.fiu.edu>
- Meals on Wheels Association of America (MOWAA) provides home-delivered meals services to older adults, homebound, and at-risk individuals. For help, or to give a gift, go to: www.mowaa.org 703-548-5558

Resources Just for Kids

- "Germs Make Me Sick," by Melvin Berger, explains with colorful illustrations how your body fights germs.
- "Little Tree: A Story for Children With Serious Medical Illness," by Joyce C. Mills, is a comforting fable for young children facing serious life challenges.
- "My IVIG Book," written from a 3-year-old's perspective about his infusions, comes with a kit for other children to create their own personalized book. Free from Baxter at www.immunedisease.com/US.
- "Our Immune System," enables children who are immune deficient and their families to explore together the immune system. Available from the Immune Deficiency Foundation at www.primaryimmune.org.

Working Caregivers

- Caregivers USA <http://caregivers-usa.org/db/index.html> Provides an index of local and state caregiver support services.
- Home Based Careers <http://edirectoryofhomebasedcareers.com/dir2.htm> Download an eDirectory of home based careers.
- Families USA www.familiesusa.org 202-628-3030 A nonprofit organization dedicated to the achievement of high-quality, affordable health and long-term care for all Americans. At www.familiesusa.org/resources/program-locator, they offer a program locator that will direct you to a local program to answer questions and assist you in obtaining health insurance. Programs may also be able to refer you to low-cost or free healthcare, including prescription drug assistance.
- Patient Advocate Foundation www.patientadvocate.org 800-532-5274 A national nonprofit organization that serves as an active liaison between the patient and his insurer, employer, and/or creditors to resolve insurance, job discrimination and/or debt crisis matters relative to his diagnosis through case managers, doctors and attorneys. They offer a pharmaceutical co-pay assistance program to patients who qualify medically and financially.
- Work Options www.workoptions.com Templates and tips for writing a proposal to telecommute, work part time, job share, and/or switch to a compressed workweek schedule.

Have something to add to these pages? Please send your suggestions for additions to the IG Living Resource Directory to editor@igliving.com. In this case, more is indeed better!

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