People are talking about the Immune Deficiency Foundation’s new electronic personal health record. Just introduced at the IDF 2011 National Conference in June, and many are already energized about how it will help them manage their health.

Terry Halper, a patient and IDF Board of Trustees Member, has been a volunteer with IDF for the last 20 years and has been helpful in the development of the IDF eHealthRecord from the very beginning. His experiences along with other patients’ have helped customize the IDF eHealthRecord specifically for the primary immunodeficiency community.

Terry knows that if you or a loved one lives with a primary immunodeficiency disease, one of the biggest challenges is dealing with the mounds of paperwork involved in keeping track of providers, insurance information, diagnoses, symptoms, treatments, and appointments. Organizing all of your health information can feel insurmountable, but this program allows you to do that and more in one centralized location.

A setup wizard guides you through the process of entering old and current information step-by-step. A calendar feature allows you to schedule and review important events, as well as track doctor visits and prescription refills, record symptoms and monitor your progress. Other features help you organize all of those pieces of paper you have in files and boxes.

However, Terry is especially excited about the Health and Life Log. This feature encourages users to evaluate their general health and well-being by recording symptoms, infections, emotions and lifestyle behaviors. There is even a Journal Notes section to type in personal comments or observations. Users can print these pages or export them to physicians prior to an upcoming visit.

“This feature is even better than I could have imagined,” he said with great enthusiasm. “I love the way it walks me through step by step—it makes it so easy! I like all the prompts within the Log because it helps me remember how I was feeling on a particular day, and that is so important. Even better is the flexibility of the program and I can skip through parts and complete them at my own convenience,” Terry expounded.

Sue Manley, an IDF volunteer and patient from Illinois, already liked to keep her information updated on a computer. “However until the IDF eHealthRecord was developed, I always ended up with the most current version of my medication list on the “wrong” computer. Now my list is accessible from any computer, and I can keep all of my medical information up to date without worrying about which computer I am using.”

Sue also likes that she can scan reports from labs and providers so all of her medical information will be in one place. She believes that using the eHealthRecord will help her to get better medical care.

continued on page 3
IDF’s website, www.primaryimmune.org, is one of the most frequently visited websites on the internet for people seeking information on primary immunodeficiency diseases. For that reason, IDF recently launched a new, revised version of the site that improves usability and navigation, making the information you are looking for, much easier to find. Additionally, the new site features more dynamic content, images, and videos, that will help promote awareness and provide a valuable resource of information to patients and families.

“With all the new programs and initiatives we are managing, the increasing amount of disease information we are capturing, and our growing use of web 2.0 technology, it was time for us to restructure the website,” said IDF President and Founder Marcia Boyle. “The new www.primaryimmune.org is much easier to use, and provides increased flexibility for future growth. We are very proud of it.”

In addition to increased navigation, better usability, and updated information on primary immunodeficiency diseases; there is also new information on IDF programs and events, new banner links that link to IDF programs and initiatives and, a new IDF TV guide with all IDF videos housed in one place. There’s even a new shopping cart feature which makes ordering publications much easier.

Be sure to visit www.primaryimmune.org and see for yourself all that the new website has to offer!

The revised www.primaryimmune.org, and ongoing sponsorship for the site was made possible by Grifols.

IDF Online Continuing Education Course for Nurses
Primary Immunodeficiency Diseases and Immunoglobulin Therapy

IDF is proud to offer this Online Continuing Education Course, developed as an initiative of the IDF Nurse Advisory Committee. This free, accredited course enhances the knowledge of the nurse clinician by providing an update on primary immunodeficiency diseases, immunoglobulin therapies by defining the differences between intravenous immunoglobulin therapy (IVIG) and subcutaneous immunoglobulin therapy (SCIG) and the nurse’s role with these therapies.

IDF would like to thank the presenters featured in this course Mark Ballow, MD; Kristin Epland, MSN, FNP-C; Jordan Orange, MD, PhD; and M. Elizabeth Younger, CRNP, PhD for their commitment to advancing education for better patient care.

Any nurse who is involved with administration and management of immunoglobulin therapy or the disease states where immunoglobulin is used would find this program very informative and applicable to practice.

For more information or to take the course, visit www.primaryimmune.org/healthcare-professionals/continuing-education-course-for-nurses

The IDF Online Continuing Education Course for Nurses was made possible by an unrestricted educational grant from CSL Behring.
continued from page 1

Sue explained, “Having a printout of recent infections or a log of my daily temperatures from my eHealthRecord to hand to my physicians will allow them to see patterns that they might otherwise miss by looking at their notes.”

It is not just patients who will benefit from the IDF eHealthRecord; it will help organize the health profiles of entire families. Melody Medellin, of San Antonio, Texas, is a mom with two children. Her oldest, Avery, was diagnosed with CVID seven years ago, and was hospitalized 33 times in just his first year of life. His medical information has continued to pile up to the point that Melody has a separate room where she keeps all of his paperwork.

Melody is excited about the recent release of the IDF eHealthRecord. “With two children, one of whom has a primary immunodeficiency, it is hard to stay on top of all of the medical information for my entire family,” she said. “I love the way I can create new health records within the same account and manage the information of both of my children. Now I can access all of my family’s health information in one place and it doesn’t get easier than that.”

On the rare occasion when she is away from Avery, Melody prepares a file of emergency information, including documents, physicians, medications and all of the information that an emergency room physician might need. According to Melody, “The ICE (In Case of Emergency) Report is awesome and will save so many steps for so many busy people.”

For many people, the fact that there is no cost to use the IDF eHealthRecord and that it can be personalized to fit your individual needs make it extremely appealing. Whatever your reason is for using the IDF eHealthRecord, we hope you will agree with Sue Manley about why the IDF eHealthRecord is the right choice for her.

“|I already have a trust relationship with IDF, and they understand the needs of patients with primary immunodeficiencies. Who else would I trust to help me manage my health records?”|

To explore the IDF eHealthRecord and set up your account, go to https://idfehealthrecord.org The IDF eHealthRecord is made possible by a generous, unrestricted educational grant from CSL Behring.
Over a thousand people from 10 countries, 40 states and Washington DC gathered in Phoenix to participate in the IDF 2011 National Conference. They were there to learn more about primary immunodeficiency diseases, meet others who live with these diseases and this year, celebrate the 30th Anniversary of IDF.

As the first IDF National Conference held in the West, attendees were welcomed at the Opening Session by the Lone Ranger, one of the world’s best-known cowboys, who recognized the real heroes – the people with primary immunodeficiencies.

"Your fighting spirit and heroic efforts have led to tremendous progress in improving the lives of people with this disease," the Lone Ranger praised those in the room. "Before IDF stepped forward with a small, committed cadre of parents of patients and doctors and formed IDF, the face of primary immunodeficiency was hidden behind a mask like the one I’m wearing. Few if any Americans knew about this disease, but fortunately, that is no longer the case."

The conference was just getting started! Over the three days of the conference, there were over 59 educational sessions, presented by 70 healthcare and life management professionals. The 13 disease specific sessions were led by world-renowned clinical immunologists and there was time for questions and sharing experiences. The two breakfast symposia featuring expert panels, one focused on the future of primary immunodeficiencies, while the other tackled some of the current policy issues our community faces. And specifically for physicians, there was even a Continuing Medical Education program that was presented by notable immunologists.

And that was just for adults! There were youth programs for ages six months to 17 years, divided into four tracks of age appropriate activities. Teens had their own gathering, while the Tweens, (ages 10-12) and the Kids Club, (ages 6-9) were led through both educational and fun sessions. All three of these groups visited the Arizona Science Center on Friday and had a wonderful time!
When not in sessions, there was plenty for attendees to do. Most attended the lively exhibit hall where they had the opportunity to talk to representatives of industries that produce products or offer services that may be helpful in the management of their primary immunodeficiency. Many registered with the United States Immunodeficiency Network (USIDNet) national patient registries for primary immunodeficiencies. These patients are allowing their medical information to be combined with that of other patients to help establish a valuable comprehensive database for researchers. Others visited the “Reel Stories Studio” and taped their account of living with a primary immunodeficiency disease for the IDF website Reel Story channel, a collection of patient and family generated videos about their experiences of living with these diseases.

However, the fun really started when the sun went down! Thursday evening’s “Welcome to the Wild, Wild West!” sponsored by CSL Behring, was a great time to renew old friendships, meet new acquaintances and help “kick-off” the conference with western music and great grub!

Friday night was the opportunity to celebrate with “Happy 30th Anniversary IDF!” sponsored by Grifols. Along with wonderful food and music, the crowd enjoyed a sing-along that many turned into a dance-along! The THINK ZEBRA! Silent Auction was in full force and many enjoyed the thrill of bidding on over 151 items. Awards for sponsors and special volunteers were given out and IDF was toasted in honor of its 30th year.

There was a Southwestern Family Festival sponsored by Baxter Healthcare on Saturday night. All ages enjoyed the dinner, games and entertainment that highlighted the exotic blend of Native American, Mexican and the colorful Western cultures.

The 2011 IDF National Conference was an extraordinary event thanks to all the amazing people who helped in so many ways. IDF’s 30th Anniversary was celebrated as it was intended—with the remarkable members of the primary immunodeficiency community.

Now to the future. It is “Back to Baltimore!” with plans for the 2013 IDF National Conference June 27 - 29, 2013 in the beautiful Inner Harbor area of Baltimore, MD.
Sixty-seven patients and family members from 24 different states made the trip to Washington, DC on May 26, 2011 to make the rounds with their Congressional offices. Their mission – to bring awareness to primary immunodeficiency diseases and ask their Members of Congress to solve the problems of access to IVIG for Medicare beneficiaries by supporting HR 1845/S 960, the Medicare IVIG Access Act.

As a result of current Medicare law, patients with primary immunodeficiency diseases are denied IVIG in the home setting, forcing them to seek care in hospitals or infusion centers despite concerns about increased exposure to germs. HR 1845/ S 960 was introduced by Rep. Kevin Brady (R-TX) and Rep. Doris Matsui (D-CA) in the House and Sen. John Kerry (D-MA) and Sen. Lamar Alexandar (R-TN) in the Senate. The bill authorizes a three year demonstration project allowing for the payment of home infusion services for patients with PIDD. The bill also authorizes government studies of other obstacles to access IVIG care. Instead of costing Medicare, the bill will save Medicare over $210 million over the next 10 years because it includes a budget offset or “pay-for” provision.

This year, IDF included a training program specifically for teens and young adults called Raise Your Voice! Fourteen young people from 11 states participated in the training and accompanied their parents on their Congressional meetings. These teens and young adults were just some of the many fresh faces that participated in Advocacy Day this year. With about 30 individuals participating in Advocacy Day for the first time, many Members of Congress and their staff heard about primary immunodeficiency diseases for the first time! IDF works to identify volunteers living in key districts for Advocacy Day meetings, targeting Members who are on the committees of jurisdiction for health legislation so that their support will be more impactful to progress the bill through the legislative process.

The IDF 2011 Advocacy Day participants met with 92 Congressional offices! Already these meetings have translated directly into 17 additional cosponsors of the Medicare IVIG legislation (bringing the current total to 39 co-sponsors for the House, and 8 for the Senate).

This is a fabulous start to this year’s Medicare IVIG Access campaign, but we need more support from the PIDD community! Access to IVIG treatment is a serious problem affecting more and more patients every day. Please visit the IDF Action Alert http://primaryimmune.org/idf-advocacy-center/action-alerts to tell your legislators how important IVIG is to you and your loved ones and ask your Members of Congress to cosponsor and support HR 1845/ S 960. Members of Congress need to hear from their constituents to learn that something is an issue. Your voice does matter and makes a difference!

IDF 2011 Advocacy Day was made possible by an unrestricted educational grant from Baxter Healthcare, CSL Behring and Grifols.

For more information about the bill please visit the IDF website at http://primaryimmune.org/idf-advocacy-center/ivig-reimbursement
IDF is founded. Marcia Boyle is first president and chair of the Board of Trustees.

First local IDF groups are established in Ohio and California. Today, IDF has a strong volunteer network comprised of several hundred volunteers, nationwide.

IDF sponsors its first research symposium. Since that time, IDF has sponsored countless medical symposia for healthcare professionals.

IDF National Newsletter Issue #1 is printed — It later became the IDF ADVOCATE and 67 issues have been published and distributed to hundreds of thousands of readers.

First annual Fellowship competition takes place. Twenty fellowships have been awarded to date.

The IDF Scholarship for post-secondary education of patients with primary immunodeficiency diseases is initiated. In 2007, it was named the Eric Marder Scholarship Program.

488 scholarships totaling $600,125 have been awarded to date.

IDF receives a donation of its first computer.

IDF Medical Advisory Committee provides consensus statement that IVIG is the treatment of choice for most patients with primary immunodeficiency diseases requiring immunoglobulin supplementation.

IDF exhibits for the first time at the American Academy of Allergy, Asthma, and Immunology (AAAAI) annual meeting.
1992
Dr. Robert Good receives IDF’s first annual IDF Boyle Scientific Achievement Award.
IDF publishes *Primary Immune Deficiency Diseases, A Guide for Nurses*. IDF publishes *Clinical Presentation of the Primary Immunodeficiency Diseases: A Primer for Physicians.*
The second, expanded edition, *IDF Guide for Nurses on Immune Globulin Therapy for Primary Immunodeficiency Diseases* was printed in 2007.

1993
Dr. Max Cooper, Howard Hughes Medical Institute, Birmingham, AL and Dr. Fred Rosen, The Center for Blood Research, Boston, MA, receive the IDF Boyle Scientific Achievement Award.
IDF conducts the first national patient survey.
Since then, more than 25 surveys of patients and physicians have been completed.

1994
IDF initiates its first edition of *Clinical Updates*, a professional monograph which later becomes *IDF Clinical Focus*. Six more have been published.
NIAID awards IDF a contract to establish a national registry of patients with Chronic Granulomatous Disease.
Dr. Rebecca H. Buckley, Duke University, Durham, NC, receives the IDF Boyle Scientific Achievement Award.

1995
1996
Dr. Richard Hong, Vermont Regional Cancer Center, Burlington, VT and Dr. E. Richard Stiehm, UCLA School of Medicine, Los Angeles, CA, receive the IDF Boyle Scientific Achievement Award.

1997
IDF embarks on its first public policy program.
IDF plays a significant role with the Centers for Disease Control and Prevention (CDC) in changing polio vaccine protocol to benefit patients with primary immunodeficiency diseases.
IDF launches the *LeBien Visiting Professor Program*. 125 Visiting Professorships have reached over 7,000 physicians to date.
IDF initiates annual funding for a Molecular/Genetic Diagnostic Laboratory.
IDF spearheads a consumer coalition of plasma users that results in the establishment of a Patient Notification System.
NIAID awards IDF a contract to conduct eight patient registries.
John Boyle appointed to serve on the FDA Blood Products Advisory Committee.

1998
National IVIG Shortage: IDF creates an internal advisory committee on blood safety and availability, provides Congressional and FDA testimony, and conducts surveys that quantify the shortage.
The IDF Consulting Immunologist Program is created.
Dr. Hans D. Ochs, University of Washington School of Medicine, Seattle, WA receives the IDF Boyle Scientific Achievement Award.

1999
The IDF Nurse Advisory Committee is established to improve the quality of healthcare and education provided by nurses for patients.
IDF launches Safety Net, a program to distribute emergency supply of IVIG to patients.
IDF develops Operation Outreach, patient education meetings designed to strengthen underserved areas.
FDA Panel supports IDF recommendation to revise and expedite new IVIG products.
The first IDF website, www.primaryimmune.org is constructed.

First IDF Family Retreat is held. Conducted every two years, there have been sixteen retreats to date.
Dr. R. Michael Blaege receives the IDF Boyle Scientific Achievement Award.
IDF provides testimony to Congress, the FDA, and HHS on the impact of Medicare reimbursement on patient access to IVIG and health consequences.

IDF Diagnostic & Clinical Care Guidelines for Primary Immunodeficiency Diseases is developed by expert immunologists. The second edition was published in 2009.

IDF Plasma Center Program is launched.

IDF Center of Excellence at Duke University Medical Center is established.

IDF conducts landmark surveys to quantify the effect of changes in Medicare reimbursement on patient access to IVIG.

The first online IDF Discussion Forum is launched. Today there are over 2,000 active participants.

The inaugural “Capitol Hill Advocacy Day” brings IDF volunteers to the nation’s Capitol to meet with their Members of Congress. Five more have successfully taken place in 2006, 2008, 2009, 2010 and 2011.

IDF holds its second National Conference in Baltimore, Maryland.

IDF School Guide, Information about Students with Primary Immunodeficiency Diseases is developed and published, with an updated edition following in 2007.

IDF Action Alert System, an online advocacy tool, is launched as a means for the community to instantly reach out to their Members of Congress.

The United States Immunodeficiency Network (USIDNET) is started through a contract with IDF and NIAID.

IDF Blue Jeans for Healthy Genes, a fundraising and awareness program, is kicked off during Primary Immunodeficiency Awareness Week.

The first IDF School Guide, Information about Students with Primary Immunodeficiency Diseases is developed and published.

Dr. Alain Fischer, Hopital Necker-Enfants Malades, Paris, France, receives the IDF Boyle Scientific Achievement Award.

Dr. Jerry Winkelstein, Johns Hopkins University School of Medicine, Baltimore, MD, receives the IDF Boyle Scientific Achievement Award.

Jerry Winkelstein, MD, is appointed to serve on the HHS Advisory Committee on Blood Safety and Availability.

IDF, helps sponsor clinical trials to bring new IVIG brands to the U.S. market.

Rebecca Buckley, MD is chosen as the second chair of the IDF Medical Advisory Committee.

2001
IDF convenes its first National Conference in Baltimore, Maryland.

IDF launches two pilot comprehensive care centers.

IDF begins Compassionate Care Program to help patients experiencing difficulties finding product.

IDF protests the Touchstone/Disney movie “Bubble Boy,” a gross-out comedy mocking individuals born without a functioning immune system.

Dr. Mary Ellen Conley, St. Jude Children’s Research Hospital, Memphis, TN, receives the IDF Boyle Scientific Achievement Award.

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Dr. Jerry Winkelstein, Johns Hopkins University School of Medicine, Baltimore, MD, receives the IDF Boyle Scientific Achievement Award.
The Immune Deficiency Foundation (IDF) is a national patient organization dedicated to persons living with primary immunodeficiency diseases. The IDF advocates for the introduction of policies that will improve treatment options and provide patients with the care they need. The IDF is dedicated to spreading awareness and making health care providers better understand the complexities of primary immunodeficiency diseases. The IDF is the first national patient organization dedicated to primary immunodeficiency diseases and is the leader in research and patient advocacy. As the national patient organization dedicated to persons living with primary immunodeficiency diseases, IDF says THINK ZEBRA! The stigma of primary immunodeficiency diseases is that it is just an infected animal. However, it is critical to get an early diagnosis and treatment to avoid the development of serious and debilitating diseases. Anyone, regardless of age or sex, can be affected by primary immunodeficiency diseases. If any of these words describe your infection, you should be suspicious: Common, Persistent, Recurrent, Severe, Infection, Antibiotics, Pain, Temperature, Fatigue. People with primary immunodeficiency diseases are the zebras of the medical world. As the national patient organization dedicated to persons living with primary immunodeficiency diseases, IDF says THINK ZEBRA!

IDF is awarded a five-year US Immunodeficiency Network grant from the NIAID. The IDF Nurse Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children recommends that SCID be added to newborn screening panels. The IDF’s SCID Newborn Screening Campaign is launched.

IDF introduces three blogs: Policy Matters, Community in Action and IDF SCID Newborn Screening.

IDF develops its own Twitter and Facebook account.

IDF convenes its sixth National Conference in Phoenix, Arizona and celebrates its 30th Anniversary!

IDF launches the eHealthRecord, an electronic personal health record for the primary immunodeficiency community.

IDF Nurse Advisory Committee develops the IDF Online Continuing Education Course for Nurses: Primary Immunodeficiency Diseases and Immunoglobulin Therapy.

IDF’s website is updated to better address the needs of the community and the increased use of Web 2.0 capabilities.

IDF spearheads legislation for new Medicare Demonstration Project for home infusion.

Steven Holland, MD, National Institutes of Health, Bethesda, MD, receives the IDF Boyle Scientific Achievement Award.
Primary Immune Deficiency Treatment Consortium (PIDTC)

The Primary Immune Deficiency Treatment Consortium (PIDTC) brings together scientists, transplant physicians and immunologists with a strong commitment and expertise in the diagnosis and management of primary immunodeficiency diseases (PIDD) to consider different treatment methods and provide improved results to future patients.

The objectives of the research will include characterizing the long term outcomes and late effects in children with Severe Combined Immunodeficiency (SCID), Wiskott-Aldrich Syndrome (WAS) and Chronic Granulomatous Disease (CGD) who undergo Hematopoietic cell transplant transplantation (HCT). Additionally, the project will incorporate the design and implementation of prospective clinical trials that improve care for children with PIDD and allow for training to be provided to physician scientists in the understanding and treatment of PIDD. The research will determine critical questions concerning HCT for these disorders and aid in the development of prospective future clinical trials.

The PIDTC consists of 13 major centers in North America:
- Cardinal Glennon Hospital St Louis, Missouri
- Cincinnati Children's Hospital, Ohio
- Children's Hospital Boston, Massachusetts
- Children's Hospital Los Angeles, California
- Children's Memorial Hospital Chicago, Illinois
- Children's Hospital Philadelphia, Pennsylvania
- Children's Hospital Seattle, Washington
- Duke Medical Center Durham, North Carolina
- Memorial Sloan Kettering Cancer Center NYC, New York
- NIH-NIAID Bethesda, Maryland
- Texas Children's Hospital Houston, Texas
- UCSF Benioff Children's Hospital San Francisco, California

An additional 19 centers within the Pediatric Blood and Marrow Transplant Consortium (PBMT) also are part of the PIDTC: British Columbia Children's Hospital, Cancer Care Manitoba, Children's Healthcare of Atlanta/Emory University School of Medicine, Children's Hospital Denver, Children's National Medical Center, Children’s Hospital/LSUHSC, New Orleans, CHU St. Justine, Hackensack University Medical Center, Medical College of Wisconsin, Methodist Children's Hospital of South Texas/Texas Transplant Institute, Oregon Health and Science University, Primary Children's Medical Center/University of Utah, Stanford University, The Steven and Alexandra Cohen Children's Hospital of New York (previously Schneider Children's Hospital), University of California, Los Angeles, University of Alabama at Birmingham, University of Michigan Health System, University of Texas Southwestern Medical Center/Children's of Dallas.

There are now two active PIDTC research protocols:
- **PIDTC 6901**: A Prospective Natural History Study of Diagnosis, Treatment and Outcomes of Children with SCID Disorders. The purpose of this study is to evaluate all children newly diagnosed with SCID who are undergoing hematopoietic cell transplantation, enzyme replacement (for ADA SCID), or gene therapy. Patients are entered at the time of diagnosis before the initiation of definitive therapy and followed for up to 4 years post transplant. In addition to the more standard evaluations that every child with SCID would have, additional studies are being done to identify the genetic defects as well as evaluate more sophisticated measures of correction of the immune defects.
- **PIDTC 6902**: A Retrospective and Cross-Sectional Analysis of Patients Treated for SCID (1968-2010): This study will enroll all children with SCID who were treated with hematopoietic cell transplantation, PEG-ADA, or gene therapy between 1968 and 2010. Patients are being actively enrolled in order to review charts and collect data regarding types of donors, treatment regimens, immune correction, complications and outcomes. In addition, surviving patients will be asked to participate in the cross-sectional evaluation in which they will be seen in their clinic for a history, physical, blood tests and evaluations of developmental status and quality of life. In those patients for whom no genetic diagnosis has been made and who consent, special efforts will be made to identify the specific cause of SCID. Finally, there will be four research studies done in which selected patients and donors will be asked to participate by providing an extra sample of blood. These studies will look at the degree of bone marrow stem cell engraftment, B cell recovery post transplant, the role of Natural Killer Lymphocytes post transplant, and how the patient's body accepts the donor cells post transplant.

All patients with SCID will be eligible for either protocol 6901 or 6902 provided they received or will receive their therapy at a PIDTC center. If a patient is being followed at a non-PIDTC center it is possible that they can participate in one of the studies IF that center joins the PIDTC. Unfortunately, the funding for the PIDTC limits the total number of centers that can participate.

The PIDTC is actively developing two more protocols, one for Chronic Granulomatous Disease (CGD) and the other for Wiskott Aldrich Syndrome (WAS). It is anticipated that these protocols will be activated within the next 4-6 months. While only patients who have undergone or plan to have a hematopoietic cell transplant for WAS will be enrolled, for the CGD study there will be a group of patients enrolled who have not and do not plan to undergo a transplant.

For questions regarding the PIDTC please contact:
- Elizabeth Dunn, PIDTC Clinical Coordinator, UCSF (dunne@peds.ucsf.edu)
- Morton J Cowan, M.D. PI, PIDTC (mcowan@peds.ucsf.edu)
- Luigi Notarangelo, M.D. co-PI, PIDTC (luigi.notarangelo@childrens.harvard.edu)

Or go to the PIDTC website: [http://rarediseasenetwork.epi.usf.edu/PIDTC/index.htm](http://rarediseasenetwork.epi.usf.edu/PIDTC/index.htm)
Andy & Sofia:
A Lesson in Health, Life and Hope

— by J. Doug Gill

It took all of 48 hours for young Andy Treviño to develop his first life-threatening infection. Andy, the then-newborn son of Andrés and Pauline Treviño, would – between the Mexico City hospital in which he was born and the Boston Hospital where doctors provided his cure – eventually log nearly 1,000 days of hospitalization.

“There were many times that Andy was so sick,” Andrés tells me, “that I was pretty sure this story was going to have a different ending.”

Andy was diagnosed with NEMO (nuclear factor kappa B essential modulator), a rare form of immunodeficiency caused by a mutation of the gene on an X-chromosome. NEMO causes sufferers to develop bacterial infections early in life and leads them to increased susceptibility to mycobacterial infections later in what is normally a very short life.

“Andy had infections and viruses all over his body,” Andrés explains, “and his body had no way to fight them. He had infections in his central nervous system, stomach, blood – even in a bone on one of his fingers.”

While the doctors in Mexico City could offer no answers, a friend of the family suggested the possibility of a primary immunodeficiency disease, sending Andrés on a path of painstaking research that eventually led him to Boston’s Children’s Hospital and immunologist, Dr. Jordan Orange.

“The first time I spoke to Children’s Hospital they suggested – given Andy’s vulnerability to disease – that we not fly to Boston. I told them I’d be there the next day.”

The survival rate for children born with NEMO is approximately 50-percent – and that’s with a successful bone marrow transplant – a complex, potentially life-saving procedure made even more difficult by the lack of candidates to provide a perfect match.

According to the National Marrow Donor Program as many as 6,000 people (worldwide) search for a bone marrow match every day, and less than 30-percent find one. Those who don’t find a compatible donor either die waiting for one, or find an alternative approach.

In 2002 the Treviños chose the latter, and by doing so not only saved their son’s life, but also created a controversy that still rages today.

“We searched public registries worldwide,” Treviño tells me, describing a futile process that would play out for two-and-a-half years.

“We not only consulted with doctors,” Treviño tells me. “We talked to friends, family and priests who spoke to other priests and all of them had a positive response. Our decision was based on finding a solution that would save my son.”

Yet, Andrés and Paulina didn’t just wade into the contentious waters of stem cells.

“We knew we had to deal with the moral and ethical aspect of our decision,” Andrés confides, “but I also knew what my family was facing so we had to make a life-changing choice.”

“Instead of seeking a match, the Treviños could make one of their own.”

“We didn’t really know what in-vitro fertilization (IVF) was,” Andrés adds, “but Dr. Orange explained the process – including the fact that only 1 in 4 siblings would be a perfect match. But having new bone marrow seemed the only way Andy was ever going to get a chance at life.”

In addition to learning about IVF, the Treviños also became familiar with pre-implementation genetic diagnosis (PGD), a procedure that would assure Andrés and Paulina that another baby would be free of the inherited disease that was killing Andy.

“PGD allowed us to create a baby with the right genetic profile – a baby whose umbilical cord blood was free of NEMO,” Andrés says, “a baby that would give us the medical miracle with which we could save our son.”

“We knew we had to deal with the moral and ethical aspect of our decision,” Andrés confides, “but I also knew what my family was facing so we had to make a life-changing choice.”

Once in motion, it took five IVF cycles before the Treviños were blessed with a healthy daughter they named Sofia.
"It was absolutely the best day of my life," Treviño says joyously, "when I first heard Sofia crying."

Even with Sofia's birth it would still take months before her healthy cells would be able to help heal her brother.

"It was a couple weeks after Sofia's birth when we found out the number of cells obtained from the procedure would not be enough for Andy's transplant," Andrés says, "so we waited six more months until Sofia could donate an additional amount from her bone marrow."

Today, the Treviños have a third child, two-year-old Tania, and Andy and Sofia are not only thriving, but are the stars of a book – "Andy & Sofia" – that was written by Andrés and Kate Kruschwitz of the Children's Hospital Foundation.

"Andy doesn't like the spotlight," Andrés explains, "but Sofia loves it. She wishes she were a superhero."

In some ways the now seven-year-old is a superhero – especially to her parents and brother.

Superhero status may even be reached by Andrés, as the father-turned-author is donating part of the proceeds from "Andy & Sofia" to the Immune Deficiency Foundation's NEMO Initiative in hopes that Andy's story (and the Treviño's financial contribution) may help other patients.

"It was Dr. Orange giving us a copy of the 'Patient & Family Handbook','" Treviño admits, crediting that IDF publication with his first exposure to the Foundation. "I was so impressed by the work IDF was doing that I couldn't wait to attend the 2007 National Conference in St. Louis."

Andy, meanwhile, is just like any other rambunctious 11-year-old. He plays soccer, loves to dance to Michael Jackson music and is working his way through scouting ranks having just reached the Weblos level.

"Andy's treatment was about disease disability and potential death," Andrés concludes, "and nothing else. If medical science offers an opportunity to heal the sick and relieve suffering I find no morality in blocking the path. Hope is a huge part of the human condition, and I'm all for restoring health, life and hope to all who need it."

A portion of the proceeds from the sale of the book "Andy & Sofia" will be donated to the IDF NEMO Initiative. To order your copy from Amazon, go here for the paperback version: http://amzn.to/idfnemoinitiative, and here for the Kindle version: http://amzn.to/idfnemokindle.

For Teens Only!

In Tune with Your Immune System, Battle of the Bands!

Have you ever had trouble explaining to your friends how your immune system works? Then, you should check out "In Tune with Your Immune System, Battle of the Bands." This presentation compares the immune system to a rock band and how all the parts need to function to make great music.

"In Tune with Your Immune System” sets the stage with “The Immunos,” who represent the immune system, (the good guys) and “The Invaders,” who represent bacteria and fungus, (the bad guys). Once all of the band members have been introduced, and their roles to the band are described, a “Battle of the Bands” takes place. To see how “The Immunos” compete with the shredding power of “The Invaders,” stop by IDF Common Ground www.idfcommonground.org. Sorry parents, but this battle is for teens only. But maybe your teen will give you a backstage pass to see the show!

The “In Tune with Your Immune System” presentation was made possible by an unrestricted educational grant from Baxter Healthcare.
New Model Provides Insights into Intravenous and Subcutaneous Immunoglobulin Pharmacokinetics in Patients with Primary Immunodeficiencies

CSL Behring announced the development of an innovative pharmacokinetic (PK) model that allows the absorption, distribution, metabolism, and elimination of subcutaneous (SC) immunoglobulin G (IgG) following administration to be simulated with a high degree of accuracy and precision. The new PK model provides a novel means of simulating the mechanism by which SC IgG is transported after it is injected into the subcutaneous tissue. The current understanding of the clinical implications of SC versus intravenous (IV) dosing of IgG in primary immunodeficiency (PI) patients is limited. In addition, little is known about where SC IgG travels within the body after it is administered and how long it remains there. This information defines IgG’s pharmacokinetic (PK) profile and could affect the volume and frequency of IgG dosing for PI patients.

Excerpted from CSL Behring news release March 19, 2011

Study demonstrates relationship between subcutaneous IgG (SC IgG) dosage and clinical outcomes with Hizentra® treatment in patients with primary immunodeficiencies

Data presented by CSL Behring suggest that treatment with higher dose Hizentra® (IgPro 20) correlates with reduced risk of infection and missed school or work among patients with primary immunodeficiencies (PI). These data, presented at the 2011 American Academy of Allergy, Asthma and Immunology annual meeting, derived from two recent trials of Hizentra, one performed in the United States and one in the European Union, and aimed to show the relationship between subcutaneous IgG (SC IgG) dosage and clinical outcomes. Earlier studies have shown that higher immunoglobulin G (IgG) doses by intravenous (IV)g infusion result in higher serum IgG and therefore fewer infections. Hizentra is indicated for the treatment of patients with PI. Although both studies demonstrated clear evidence of effectiveness with zero acute serious bacterial infections (aSBIs), patients on the higher of the two doses of SC IgG experienced a lower rate of infection (2.76 vs. 5.18 infections/patient/year) and fewer days missed from school/work (2.06 vs. 8.0 days/patient/year).

Excerpted from CSL Behring news release March 21, 2011

Early Diagnosis and Treatment the Focus of International Dialogue on Primary Immunodeficiency Disease

Thought leaders from the medical, scientific and patient advocacy communities gathered in New York and London for CSL Behring’s Key Issues Dialogue—Immunoglobulin to examine challenges facing patients with primary immunodeficiencies (PIDD). They found common ground between the US and Europe on access-to-care issues such as early diagnosis and treatment of PIDD and explored possible ways of improving patient care.

The participants examined barriers to accessing immunoglobulin (Ig) therapy, which is available through subcutaneous and intravenous dosing, including challenges by some insurance plans that impede patient care. The problem has been further compounded by cuts to healthcare benefits. Another challenge involves rigid guidelines for managing patients, particularly with respect to dosage. Areas of common ground included a universal emphasis on sharing information to ensure that patients receive optimal care. Marcia Boyle said an IDF Internet survey found that patients who are connected to a patient organization receive better care than those that are not.

Excerpted from CSL Behring news release April 20, 2011

Baxter and Halozyme Announce Top-Line Results of Phase III Study of HyQ in Patients with Primary Immunodeficiency

Baxter International Inc. and Halozyme Therapeutics, Inc. announced top-line results of a phase III study of HyQ, an investigational facilitated subcutaneous immune globulin (Ig) product for use in patients with primary immunodeficiency (PI). The subcutaneous administration is facilitated by recombinant human hyaluronidase, an enzyme that increases dispersion and absorption of the Ig. The data confirm the interim results presented late in 2010 and support the recent submission of a biologics license application to the United States Food and Drug Administration.

The phase III prospective, evaluated the effectiveness of HyQ in the prevention of infections and measured other secondary endpoints including tolerability. The objective of the study was to infuse a 3 week or 4 week dose of 10% HyQ in a single infusion site. In the study, the acute serious bacterial infection rate was 0.025 per patient per year, which is below the required efficacy threshold of 1.0. In the tolerability assessment of HyQ, the most frequently reported adverse reactions were infusion site reactions (20% of infusions), headache (3% of infusions), fatigue (1% of infusions) and pyrexia (fever) (1% of infusions).

Excerpted from Baxter news release July 8, 2011

Baxter Announces FDA Approval of Subcutaneous Route of Administration for GAMMAGARD LIQUID for Patients with Primary Immunodeficiency

Baxter International Inc. announced that the U.S. Food and Drug Administration (FDA) has approved the subcutaneous administration of GAMMAGARD LIQUID 10% (Immune Globulin Infusion (Human)) for patients with primary immunodeficiency (PI). The approval of this new route of administration will allow physicians and PI patients to work together to determine which route of administration of GAMMAGARD LIQUID is most appropriate. Subcutaneous use of GAMMAGARD LIQUID allows patients to self-administer their therapy at home on a weekly basis.

Excerpted from Baxter Healthcare news release July 25, 2011
Special Recognitions at the IDF 2011 National Conference

BOYLE SCIENTIFIC ACHIEVEMENT AWARD
Steve Holland

OUTSTANDING ACHIEVEMENT AWARD
Heather Smith

OUTSTANDING ACHIEVEMENT AWARD
Barbara Ballard

VOLUNTEER PEER SUPPORT AWARD
Holli Jo Bess

IPOPI’s 2010 LUCIANO VASSALLI AWARD
Isaac Antilla

WITH GRATITUDE
Honorary and Memorial Gifts – 2/15/11 to 7/20/11

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Richard DuFour
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Bryan Skill
Scott Solberg
Dylan and Nicholas Sutton
Kim, Jade, Savannah, and
Serenity Vickers
Casey and Andrew Voss

These donations help IDF to improve the diagnosis and treatment of patients with primary immunodeficiency diseases through advocacy, education and research. If you would like to make a donation, please go on our Web site, www.primaryimmune.org and click the “please donate today” picture in the top right corner. You can also contact us in any of the following ways:

Phone: 800.296.4433 or 410.321.6647
E-mail: idf@primaryimmune.org
Mail: IDF, 40 W. Chesapeake Ave., Suite 308, Towson, MD 21204

Pregnancy and PIDD

IDF has received an unrestricted educational grant to perform a groundbreaking survey.

If you are the mother of a child with PIDD, or a woman, of any age, with a PIDD, IDF needs your help!

IDF will be conducting this survey in the Fall of 2011 and we need to make sure we can get in touch with you in the most effective manner. If you are not sure that you have provided IDF with your current e-mail address, please go to the “Get Connected” page on www.primaryimmune.org and fill out this contact form.

Contribute to this important research and help IDF make a difference!

If you have any questions regarding this upcoming study, please contact IDF’s Director of Survey Research at: 800.296.4433
Visit primaryimmune.org to download or order copies of the new IDF Brochure, designed to increase awareness of primary immunodeficiency diseases and services provided by IDF.

For an updated IDF Calendar of events, visit www.primaryimmune.org/event-calendar.

Leading the Way

Visit primaryimmune.org to download or order copies of the new IDF Brochure, designed to increase awareness of primary immunodeficiency diseases and services provided by IDF.

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IDF Advocate
Immune Deficiency Foundation
40 West Chesapeake Avenue
Suite 308
Towson, Maryland 21204

Toll-Free 800.296.4433
Direct 410.321.6647
Fax 410.321.9165
E-mail idf@primaryimmune.org
Web Site www.primaryimmune.org

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