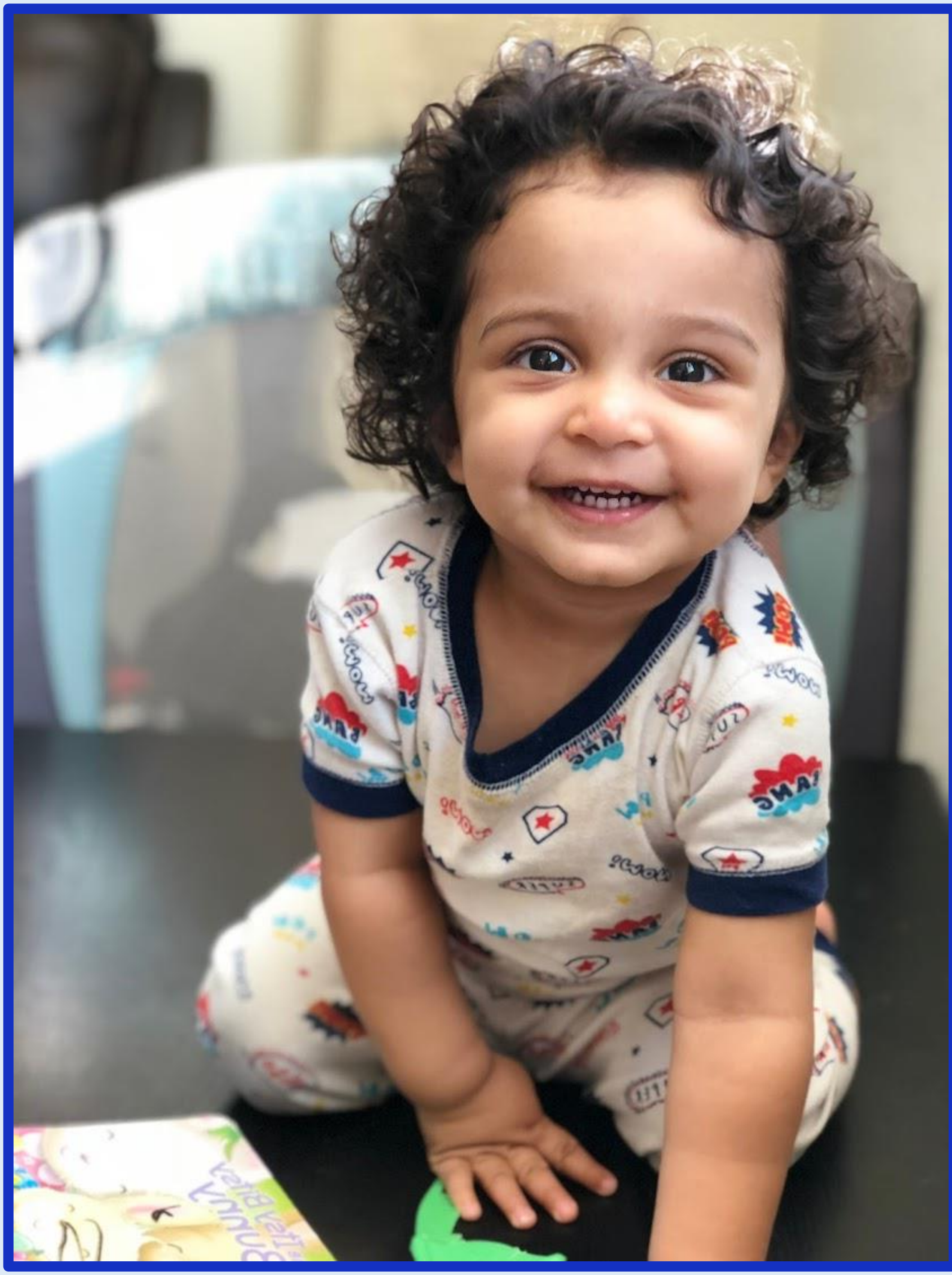
Primary Immune Deficiency Treatment Consortium

NEWSLETTER

JAN 2019 | Issue 08



Meet UCSF's Ronav (Page 2)

IN THIS ISSUE

GREETINGS FROM DR. JENNIFER PUCK, PIDTC PI

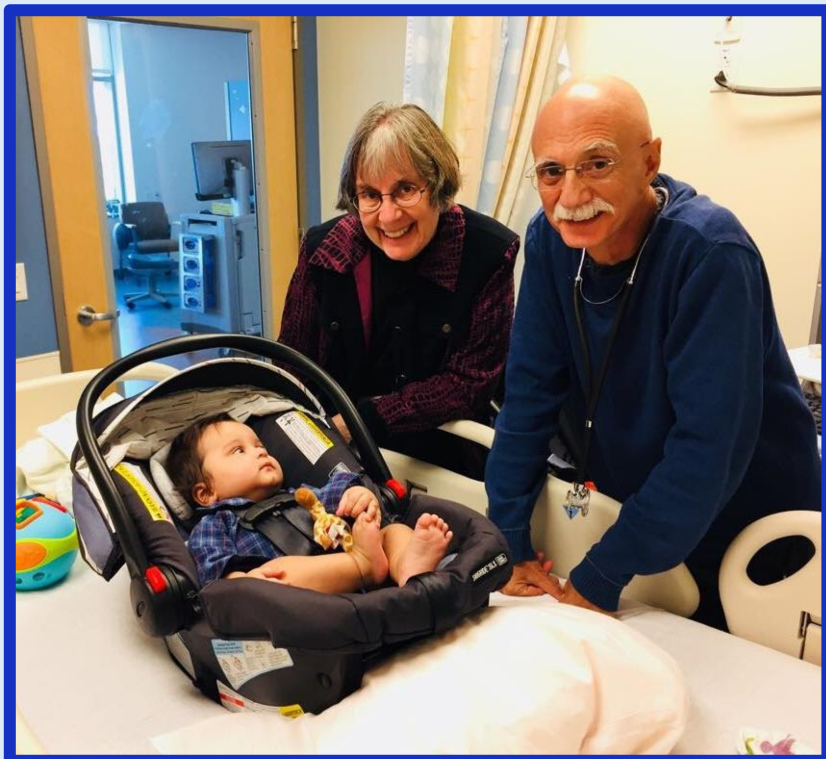
I hope everyone had a wonderful holiday season and was able to recharge for all the work that our 44 centers will accomplish together in 2019!

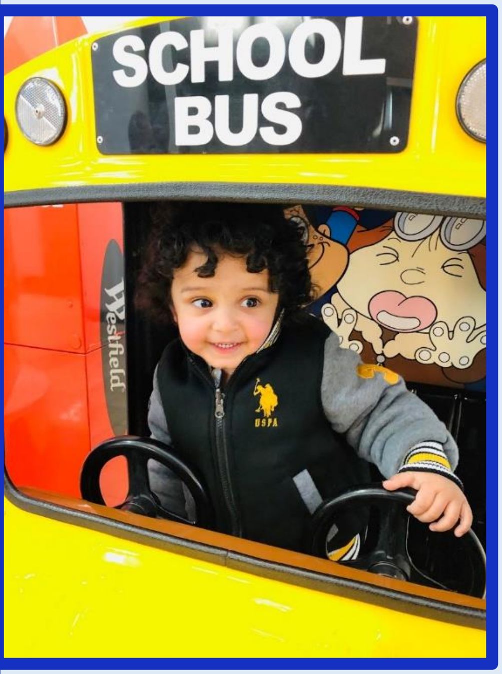
PIDTC recently welcomed the addition of two new Steering Committee Members, **Troy Torgerson, MD, PhD** from Seattle Children's Hospital (top right) and **Mike Pulsipher, MD** from Children's Hospital of Los Angeles (middle right). We appreciate their experience, enthusiasm and sage advice.

I look forward to seeing everyone at our 9th Annual Workshop and Education Day in New York City in May!

Thank you all,

Jennifer Puck

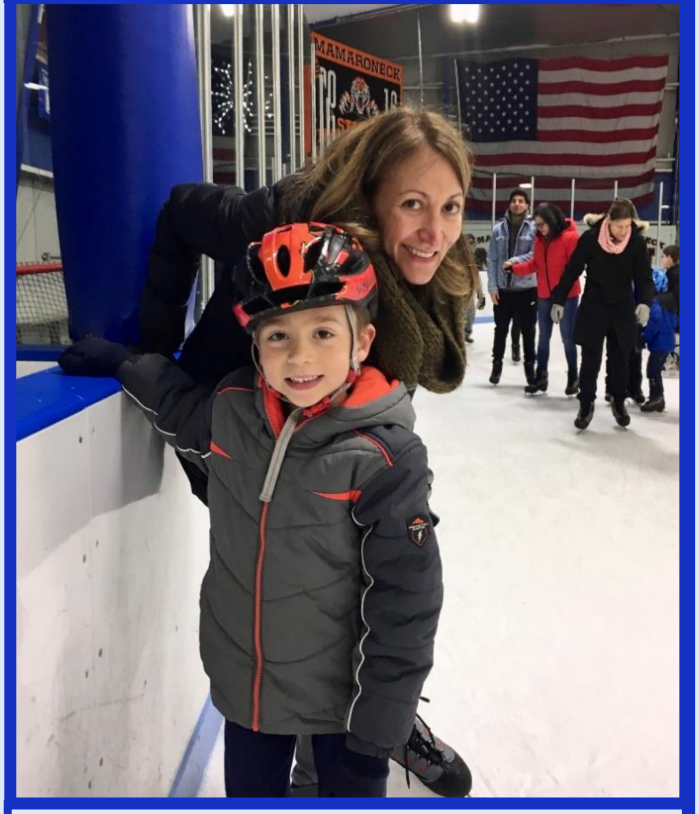




Dr. Jennifer Puck and Dr. Morton Cowan with their patient, Ronav, who received gene therapy at UCSF for XSCID. Today, Ronav is almost two years old and loves to play!

Introducing our new CGD Patient Advocates!

Felicia Morton



Felicia Morton with her son, Sebastian, age 6, at an ice skating rink in Larchmont, New York.

PIDTC welcomes **Felicia Morton** as a Patient Advocate for CGD! As the mother of Sebastian, a former patient with CGD, Felicia offers different perspectives on the disease, from managing it with daily prophylaxis to researching and undergoing a curative option. As a public relations consultant she currently works with the Immune Deficiency Foundation (IDF) to develop content for "Living With CGD." The webpage launched in 2015, and it has since become a top global resource for news and information about CDG. Articles can be found at: https://protect2.fireeye.com/url?k=b8c54006-e4852d50-b8c5671b-0cc47adb57f0-58d0df1f8f27db65&u=https://www.livingwithcgd.org/blog/

"I am committed to working with PIDTC to increase our understanding of the disease and the best treatments. I am fortunate because my son had a successful cord blood transplant at Duke University Hospital in 2016. Today he is a happy and healthy first-grader who enjoys going to school, engaging in as many playdates as possible, playing soccer, ice skating, and designing intricate railroad systems with his Thomas the Train sets. He hopes to be an engineer when he grows up, because he loves fixing and building things."

Keith Russell

PIDTC extends a warm welcome to **Keith Russell**, a new CGD Patient Advocate. He became involved after speaking personally with Dr. Harry Malech about this consortium. He has encouraged Keith to become an advocate for this group. He has been treated for various maladies at the National Institutes of Health for over 25 years and has been receiving care from Dr. Harry Malech ever since. He is currently working as a Pediatric Hematology/Oncology, Bone Marrow Transplant Nurse at Yale New Haven Hospital. *"I am motivated by the progress of the gene therapy and transplant program at the National Institute of Allergy and Infectious Disease. I hope that someday there will be a safe cure for CGD and look forward to collaborating with PIDTC in the future."*

Robert Karp

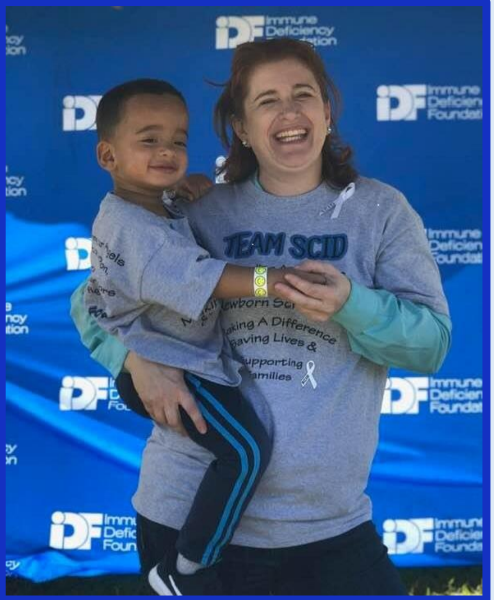
PIDTC is excited to welcome **Robert Karp** as a CGD Patient Advocate. He became involved after connecting with Dr. Harry Malech at the NIH.

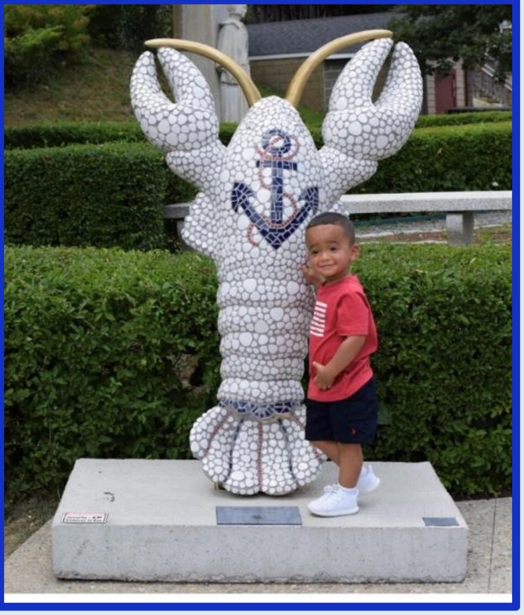
"As a CGD patient, I think I can provide valuable input on the medical issues of importance to me and our patient community and I will be glad to work with the other PAG representatives."

Patient Advocacy Groups (PAGs)

SCID Angels show their support at the IDF Walk in 2018

SCID Angels for Life showed their support of **IDF** and the PID community by organizing a team in most of the cities that had “IDF Walk for PI” events in 2018. Here are just a couple of the photos from Team SCID during their walk in Tampa, FL, including an adorable picture of **Dr. Jolan Walter** with **Kai**, one of her SCID patients!





*Above: **Team SCID** at the IDF Walk in 2018.*
*Top Right: **Dr. Jolan Walter**, of University of South Florida, with her with patient **Kai**.*
*Lower Right: **Kai** exploring New England after his one year post-gene therapy appointment in July 2018.*

PIDTC wants to hear about your patients! If you would like one of your patients featured in next quarter’s issue, please send a photo and a brief blurb to Katie at: Catherine.Chang@ucsf.edu

Patient Advocacy Groups (continued)

Genetic Sequencing Program, Jeffrey Modell Foundation

The **Jeffrey Modell Foundation** is conducting a new and exciting Foundation-sponsored pilot program. In anticipation of a broader global outreach, JMF is offering diagnostic genetic sequencing to patients with a suspected Primary Immunodeficiency (PI) at no cost to physicians or patients. Access to these types of tests can be a barrier to diagnosis for PI patients. Through this program, JMF aims to improve diagnosis, disease management, and ultimately quality of life, all while justifying greater access to Next Generation Sequencing in the right context. We thank Vicki and Fred Modell, Co-Founders of the Jeffrey Modell Foundation, for this exciting update!

New at IDF, The Immune Deficiency Foundation

In July, **IDF** was awarded a two-year grant from the U.S. Health Resources and Services Administration (HRSA) to design and implement an advanced screening and education program for people with Severe Combined Immunodeficiency (SCID) in rural areas or underserved communities. Since August, IDF staff along with its partners at the Association of Public Health Laboratories (APHL) and RTI International have begun the systematic approach to evaluate current conditions and needs—and to build upon those family and clinician needs—to develop communication strategies and create new materials related to SCID. IDF has also been hard at work developing programming for the upcoming IDF 2019 National Conference, including diagnosis-specific tracks in SCID, CGD, and WAS. For more information on the conference, visit: www.idfnationalconference.org

PIDTC THANKS OUR PATIENT ADVOCACY GROUPS FOR THEIR CONTINUED SUPPORT AND COLLABORATION

PIDTC 9TH Annual Workshop & Education Day

RSVP TODAY!! Attention site PIs and Patient Advocacy Groups: RSVP and book your hotel

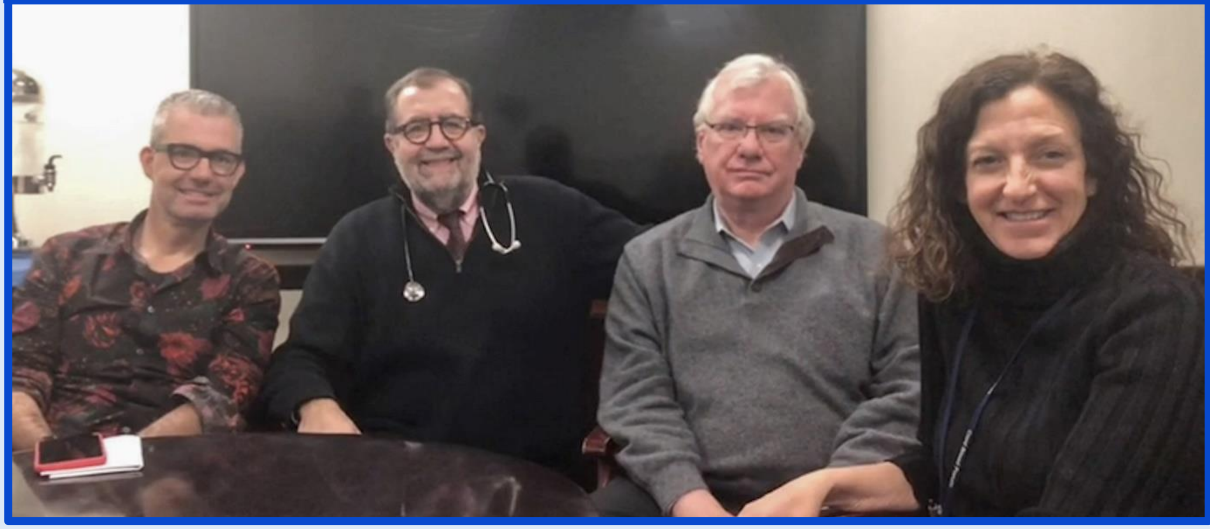
Quick Facts:

- **Workshop Dates**: May 16-18, 2019; **Education Day** (by application only—see below): May 15-16, 2019
- **Hosted by:** Memorial Sloan Kettering Cancer Center; **Location**: Warwick Hotel, New York City, NY, USA

Details:

- Rooms blocked for Tuesday, May 14th - Saturday, May 18th at the Warwick New York Hotel
- In order to facilitate active interaction among attendees, PIDTC's Annual Workshop is by **invitation only.**
- **Funding**: We are in the process of obtaining funding for the meeting costs. Leftover funding will be prioritized to support travel expenses for trainees and speakers at the PIDTC workshop, followed by junior faculty. We cannot guarantee that we will be able to cover the costs and all others should plan on covering their own travel to the meeting!
- For the first time, PIDTC is working on granting **CME accreditation** for this upcoming meeting. If you are interested in CME credits, please contact **Tara Bani** (Tara.Bani@ucsf.edu) so she can send you information on registering once accredited. Registration fee will be $45 per registrant.

Questions? Contact: **Dr. Susan Prockop**, (prockops@mskcc.org 212-639-6715) or **Rachel Corke** (corker@mskcc.org; 212-639-2285)



Meet MSKCC's Clinical Team- thank you for hosting!

L-R: Jaap-Jan Boelens, MD, PhD; Farid Boulad, MD; Richard J. O'Reilly, MD; Susan E. Prockop, MD

*Attention Junior Faculty and Fellows!

PIDTC invites you to apply to Education Day taking place on May 15-16, 2019, prior to the start of the Annual Workshop. The purpose of the PIDTC Education Day is to present and discuss unique aspects of diagnosis, management and definitive treatment for severe primary immune deficiencies aimed at fellows in training and junior faculty with either immunology or hematology/oncology backgrounds. The format will be a combination of didactic lectures and case report discussions on the diagnosis and management of SCID, WAS and CGD, focusing on treatment with hematopoietic stem cell transplantation (HSCT) or gene therapy (GT)

APPLY NOW: Email Tara.Bani@ucsf.edu no later than February 28, 2019 with your: (1) completed application form and (2) single-page Case Report on typical SCID, leaky SCID, WAS or CGD raising provocative HSCT/GT issues.

Protocol Updates-6901, 6902, 6903, 6904

Severe Combined Immunodeficiency (SCID) - 6901/6902

Updates: Our PI's **Chris Dvorak**, **Elie Haddad**, and the SCID team are working on cleaning up the 6901 dataset so that it can be merged with and analyzed alongside the 6902 dataset. The team is also focusing on completing 6902 cross sectional recruitment. We also extend a big thank you to centers that contributed patients to the T-Cell O'Reilly study; we believe we have recruited all of our consortia's eligible participants at this time. Also, thank you to wonderful statistician, **Brent Logan**!

Goals: Please continue to enroll patients in 6902 cross sectional (must be at least two years post-transplant). As a reminder, these cross sectional visits can be done over the phone--please email Tara.Bani@ucsf.edu for any questions. Also, Katie Chang has been sending 6901 queries to centers, so please be on the lookout for center-specific emails on what needs to be resolved.

Chronic Granulomatous Disease (CGD)-6903

Updates: The 6903 CGD team has been hard at work completing their Inflammatory Bowel Disease paper—we thank **Rebecca Marsh** for all her efforts! Additionally, **Liana Falcone's** presentation on the microbiome at ESID this past fall received favorable reception. She plans to sequence more samples this spring and submit an abstract to the workshop. Also, we thank **Jen Leiding, Dani Arnold, and Suhag Parikh**, who have been leading the protocol working team call discussion! Thank you to our PIs **Elizabeth Kang** and **Harry Malech!**

Deadlines: Retro enrollment deadline has already passed. Please contact Katie ASAP if you have an additional patient to enroll. Retro CRFs must be completed no later than due date of **February 28th**.

Goals: Please continue to enroll patients in the cross sectional arm!

Wisckott-Aldrich Syndrome (WAS)-6904

Updates. The 6904 WAS team has done a fantastic job cleaning up the retrospective data for patients and thanks all sites that have responded to the autoimmune queries sent out in December. The manuscript will feature 129 patients transplanted 2005-2015. The WAS team plans to analyze the rest of the retrospective cohort and thanks **Blachy Davila, David Shyr**, and **Jessie Barnum** for joining the WAS Sub-Committee team! Special thanks to statisticians **Ruta Brazauskas**, and **Joy Liu**, and our wonderful PI, **Lauri Burroughs!**

Deadlines: We will close enrollment to the 6904 cross sectional study in the next few months. We already closed enrollment for prospective and retrospective studies on September 1st, 2018.

Goals: Please continue to register patients for 6904 Cross Sectional and complete outstanding Cross Sectional and Prospective CRFs.

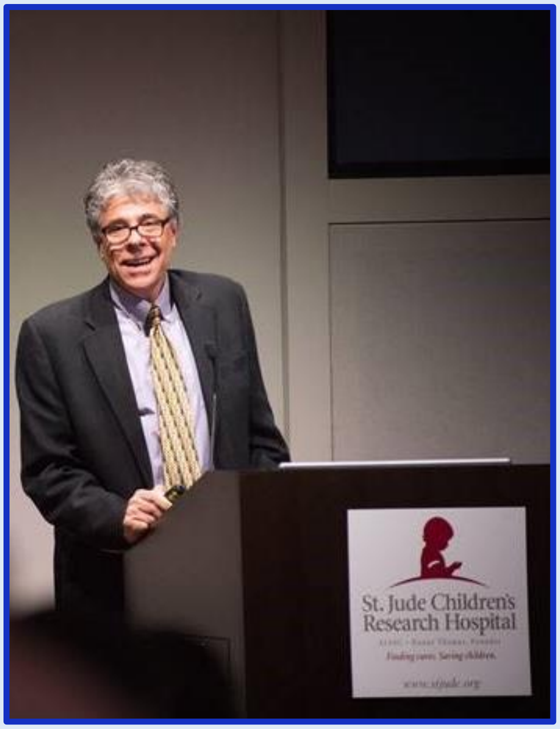
*Attention CRCs

Please focus on 1) Completing ALL outstanding CRFs and 2) Bringing all eligible new patients in for 6902, 6903, and 6904 cross sectional visits

Remembering

Dr. Shearer and Dr. Sorrentino

Dr. William T. Shearer, MD, PhD was a pillar of the PIDTC since its beginning. He served as the Principal Investigator for the PIDTC at Baylor/Texas Children's and led Texas Children's Allergy and Immunology Service from 1978-2012. He was best known for his compassionate and innovative care of David, "the boy in the bubble," which led to new immunologic discoveries. Professor Shearer was dedicated to clinical research in primary and secondary immunodeficiency. As a preeminent clinician, mentor, and educator, he published over 400 journal articles, 86 book chapters, and was Chief Editor for Clinical Immunology: Principles and Practice since 1996, and Co-Editor of the Journal of Allergy and Clinical Immunology. During his distinguished career, Dr. Shearer received numerous awards and accolades and held leadership roles in immunology and pediatric HIV professional organizations, including the American Board of Allergy and Immunology (ABAI) and the American Academy of Allergy, Asthma and Immunology (AAAAI). We greatly miss our dear Dr. Shearer but are confident that his legacy will live on in the lives of all who had the privilege of knowing and working with him.



Dr. Brian Sorrentino, MD was an exceptional colleague and physician and served as a Co-Investigator for PIDTC trials at St. Jude's Hospital. After spending five years at the National Institutes of Health, he arrived at St. Jude's in 1993 and went on to become the Director of Experimental Hematology. In 2001, he discovered the "universal "stem cell marker, ABCG2/Bcrp1. He worked for nearly a decade aiming at curing infants with X-linked SCID and created a multicenter study, *LVXSCID: gene therapy trial utilizing lentiviral vector and low dose of Busulfan for treatment of X-SCID.* He served on the Advisory Council for the American Society of Cell and Gene Therapy. He was elected to the American Society of Clinical Investigation and received the Till & McCulloch Award from the International Society of Experimental Hematology. His contributions to the field are many, and it is with great sadness that we announce his passing last fall. He will be dearly missed.

Clinical Studies

Got news to share? Email:
Catherine.chang@ucsf.edu

Lentiviral gene transfer for SCID-X1 with low dose targeted Busulfan conditioning

This trial is open and enrolling at Boston Children's Hospital and Mattel Children's Hospital UCLA, as well as at Great Ormond Street Hospital in London. For eligibility or more information about the study, please contact: Overall PI: **Sung-Yun Pai, MD** (sung-yun.pai@childrens.harvard.edu); Los Angeles PI: **Donald Kohn, MD** (dkohn1@mednet.ucla.edu); Sponsor: **David A. Williams, MD** (david.williams2@childrens.harvard.edu)

Gene Therapy Trial to Treat X-linked Severe Combined Immunodeficiency

This trial is currently enrolling at St. Jude’s, Seattle, and UCSF Benioff Children’s Hospital. In this research study, boys with SCID-X1 will receive a treatment called “lentiviral gene transfer,” also called “gene therapy.” This method inserts a normal copy of the SCID-X1 gene into blood-forming cells or “stem cells” from bone marrow that grow and develop into all blood cell types. The inserted gene will provide correct instructions to the defective stem cells in SCID-X1 so that functioning lymphocytes can develop.

For eligibility or more information about the study, please visit: stjude.org/LVXSCID-ND, or contact **Ewelina Mamcarz, MD** (ewelina.mamcarz@stjude.org), **Aleksandra Petrovic, MD** (Aleksandra.Petrovic@seattlechildrens.org), or **Mort Cowan, MD (**Mort.Cowan@ucsf.edu)

UCSF / Stanford Transplant Anti-c-KIT Transplant Protocol

This Phase I study is a single arm, open label, dose escalation trial being conducted at 2 centers: UCSF Benioff Children’s Hospital and Lucile Packard Children’s Hospital at Stanford. The study objective is to evaluate the safety and tolerability of tandemly purified allogeneic CD34+CD90+ human stem cells (HSC) in patients with Severe Combined Immune Deficiencies (SCID) conditioned for transplantation with AMG 191, a monoclonal antibody that targets human CD117.

For questions regarding the trial please contact **Julie Shizuru, MD** (jshizuru@stanford.edu), or **Christopher Dvorak (**Christopher.dvorak@ucsf.edu)

UCSF Artemis SCID Gene Therapy

In this research study, children with ART-SCID receive a treatment called “lentiviral gene transfer,” also called “gene therapy.” This method inserts a normal copy of the DCLRE1C gene into blood-forming cells or “stem cells” from bone marrow that grow and develop into all blood cell types. The inserted gene will provide correct instructions to the defective stem cells in ART-SCID so that functioning T and B lymphocytes can develop.

For eligibility or more information about the study, please contact: **Mort Cowan, MD** (Mort.Cowan@ucsf.edu) or **Jennifer Puck, MD** (Jennifer.Puck@ucsf.edu)

CSIDE

CSIDE is open to enrollment at Vanderbilt University and CHLA, and 2 patients have been enrolled to date. Boston Children's Hospital is expected to activate shortly. If you have any questions about getting your site on board, please email **Sung-Yun Pai, MD** (sung-yun.pai@childrens.harvard.edu), **Mike Pulsipher** (mpulsipher@chla.usc.edu), and **Jenny Vogel** (jvogel@nmdp.org)

**The PIDTC does not endorse these studies, but provides this information to our readers as a courtesy.

PIDTC on the Horizon: Timeline 2019

Feb 28: Education Day application due

Feb 28: 6903 Retrospective CRFs must be completed.

March 31: Education Day case report slides due

April 15, 2019: Workshop Abstracts due. All centers must submit an abstract!

April: 6904 Cross Sectional Enrollment is closing soon—bring in your patients!

February

March

April

May

Attention Families!

If you are a PID patient and would like to participate in a PIDTC study…

Join the RDCRN PIDTC Contact Registry!

The Contact Registry is a way for patients with primary immune deficiency and their family members to learn about PIDTC research studies and find out if they may be eligible to participate in one of our studies. Registration is voluntary and you may choose to withdraw at any time. There is no cost to join the Contact Registry. Visit the link to join today: *https://www.rarediseasesnetwork.org/cms/pidtc/Get-Involved/ContactRegistry*

Join the RDCRN Contact Registry

PIDTC

Newsletter brought to you by Katie Chang and the PIDTC Management Team. Thank you to our partners at the RDCRN/DMCC!