

Primary Immune Deficiency Treatment Consortium

# NEWSLETTER

JUN 2019 | Issue 09



*Introducing Stanford University's SCID patient- Lucas, 2.5 yrs. old*

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### GREETINGS FROM DR. JENNIFER PUCK, PIDTC PI

I want to thank everyone who joined our 9<sup>th</sup> Annual Workshop this past May and contributed to our lively and thoughtful discussions on primary immunodeficiencies.

The past few months, our consortium has been busy completing outstanding case report forms across all four protocols. We appreciate every site's hard work, dedication, and teamwork. Our protocol teams look forward to analyzing and sharing this data with the entire PIDTC community and putting out several important peer-reviewed papers in the coming year. Have a wonderful summer!

Thank you all,

*Jennifer Puck, MD*

*Left: Heather and John Smith of SCID Angeles, alongside Dr. Jennifer Puck, enjoy a beautiful New York City sunset while cruising the Hudson River.*



### >>patient spotlight: Emmett





# Patient Advocacy Groups (PAGs)

## IDF 2019 Conference and SCID Angels

The Immune Deficiency Foundation 2019 National Conference took place in Maryland on June 20-22. It is the world's largest gathering of families affected by Primary Immunodeficiencies. **SCID Angels** proudly showed support.



**Top Left:** Jake and Tyler, brothers with XSCID from Duke, smile with TZ, the IDF Zebra mascot.

**Top Right:** Several PID families gather to talk and unwind following a full day of learning and sessions.

**Bottom:** We are thrilled that so many PID families were able to attend this conference!

**Felicia Morton**, PIDTC PAG liaison, just launched **The CGD Association of America**, a nonprofit organization committed to advocating on behalf of patients, carriers, and families by providing clear, accurate, and independent news and information about CGD and advancing CGD research. There are many ways to get involved and support this important mission. For more information, please visit: <https://cgdaa.org> or send an email to [felicia@cgdaa.org](mailto:felicia@cgdaa.org).

## The CGD Association of America

## Patient Advocacy Groups (continued)

### Jeffrey Modell Foundation's "Do Something: The Jeffrey Modell Story"

After a rare disease took the life of their son, Vicki & Fred Modell's enduring love for each other gave them the strength to fulfill their promise to him and 'do something' by looking for better diagnosis and treatment for children with primary immunodeficiency.

#### **Do Something** **THE JEFFREY MODELL STORY** A Film by Lisa Binns

From loss comes love  
From love comes purpose  
From purpose comes inspiration

Available NOW on all Digital & VOD platforms

SEAL FILMS PRESENTS "DO SOMETHING" BY JASON SCHMIDT WITH SVEN FAULCONER AND JONATHAN BARENBOIM  
JOAN LYNN MELISSA PALZER MATTHEW WATERS SHANTON STANLEY JAY TIPTON  
BY LISA BINNS ROBBY FOOTLOCK BY LISA BINNS

jm www.dosomethingdoc.com @dosomethingdoc

We encourage everyone to watch!

Available on DVD, Digital, and

Video on Demand: [dosomethingdoc.com](http://dosomethingdoc.com)

(Now!) iTunes, Google Play, Amazon, Vudu, Vimeo, YouTube,

(July 2nd!) Vubiquity, DirecTV, InDemand



Jeffrey never knew how he would change his parent's lives forever, and the lives of so many he would never meet. But in the spirit of his optimism and courage, Vicki & Fred Modell created the Jeffrey Modell Foundation... not in memory of his death, but in celebration of his life.





# PIDTC 9<sup>TH</sup> Annual Workshop & Education Day

Thank you to everyone who joined us in New York City for our largest PIDTC Workshop and Education Day to date. We extend a huge thanks to our excellent hosts, **Drs. Susan Prockop** and **Richard O'Reilly** from Memorial Sloan Kettering Cancer Center! The workshop had over 120 attendees, 30 Education Day case presentations, and 50 fabulous talks by faculty. It was a full four days of learning and collaboration among leaders in the field of primary immune deficiencies (PID). As one Education Day mentee aptly said, "My brain is so saturated—it is overflowing with new knowledge about PID!"

Highlights included: **Dr. Schattman's** Prepubertal Fertility Preservation talk, several informative presentations on Gene Therapy, and presentations from our best abstract winners **Ami Shah, Elizabeth Kang, Dani Arnold, and Katie Harris**. We even capped off the workshop with a cruise of the Hudson Bay! Additionally, thanks to the efforts of **Elizabeth Dunn**, this was the first year we offered CME Credit!



**Top:** During the workshop, we honored **Dr. Richard O'Reilly** for his outstanding contributions to the field of PID.

**Bottom Left:** During a break, attendees enjoy refreshments and discuss the day's presentations.

**Bottom Right:** Everyone listens in as **Dr. Mort Cowan** gives a presentation on Artemis SCID Gene Therapy.



# PIDTC 9<sup>TH</sup> Annual Workshop & Education Day (cont.)



Left: Elie Haddad and Linda Griffith enjoying the New York skyline.

Right: Lauri Burroughs and Sumathi Iyengar of the WAS Foundation smile under the Brooklyn Bridge.

## 2020 PIDTC Workshop

Save the Date!

**Where:** Asilomar Conference Grounds in Pacific Grove, CA

**When:** April 28-30, 2020 (Ed Day April 27-28)

**Hosted by:** Dr. Ami Shah from Stanford University





# Protocol Updates-6901, 6902, 6903, 6904

## Severe Combined Immunodeficiency (SCID) - 6901/6902

**Updates:** PIs **Chris Dvorak** and **Elie Haddad**, and the SCID team are working on cleaning up the 6901 and 6902 datasets. Be on the lookout for queries from **Tara Bani**! Numerous manuscripts are in the works and investigator help is always needed. Also, now that case reports form completion for 6901 is more up to date, we are shifting our focus to completing 6902 outstanding case report forms. Be on the lookout for emails from **Katie Chang** regarding what is still outstanding at your site. Lastly, we thank our wonderful statistician, **Brent Logan**, for his efforts pulling together this data!

**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** with any questions.

## Chronic Granulomatous Disease (CGD)-6903

**Updates:** Congratulations to **Rebeca Marsh**! The Protocol team's Inflammatory Bowel Disease manuscript was recently accepted for publication. **Jen Leiding**, **Suhag Parikh**, and **Dani Arnold** have been busy cleaning the data for the 6903 overall paper and thank every site for their efforts to complete outstanding CRFs. Also, we thank our PIs **Elizabeth Kang** and **Harry Malech**, and statisticians **Kadam Patel** and **Rachel Wu**!

**Deadlines & Goals:** Stay on top of completing prospective 6903 CRFs, and continue to resolve 6903 queries for the larger CGD overall paper. Continue to enroll your prospective and cross sectional patients.

## Wiskott-Aldrich Syndrome (WAS)-6904

**Updates:** The 6904 WAS team has been busily writing the WAS manuscript on the 129 patients transplanted 2005-2015. We especially want to thank **David Shyr**, **Blachy Davila**, **Jessie Barnum**, and **Ami Shah** for starting to clean the rest of the retrospective cohort. We thank our talented statisticians **Ruta Brazauskas**, and **Joy Liu**, and our wonderful PI, **Lauri Burroughs**!

**Deadlines:** We recently closed enrollment in the 6904 cross sectional study and already closed enrollment in the 6904 prospective and retrospective studies in 2018.

**Goals:** Please complete outstanding Prospective CRFs as time point windows approach. We need at least one year of follow up data on every prospective patient.

# Clinical Studies

## CSIDE

CSIDE is open to enrollment at Vanderbilt, CHLA, Boston, Methodist, Minnesota, Hackensack, and 3 patients have been enrolled to date. More centers are currently being activated! If you have any questions about getting your site on board, please email **Sung-Yun Pai, MD** ([sung-yun.pai@childrens.harvard.edu](mailto:sung-yun.pai@childrens.harvard.edu)), **Mike Pulsipher** ([mpulsipher@chla.usc.edu](mailto:mpulsipher@chla.usc.edu)), and **Jenny Vogel** ([jvogel@nmdp.org](mailto:jvogel@nmdp.org))

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

## 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](mailto:Mort.Cowan@ucsf.edu)) or **Jennifer Puck, MD** ([Jennifer.Puck@ucsf.edu](mailto:Jennifer.Puck@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](mailto:sung-yun.pai@childrens.harvard.edu)); Los Angeles PI: **Donald Kohn, MD** ([dkohn1@mednet.ucla.edu](mailto:dkohn1@mednet.ucla.edu)); Sponsor: **David A. Williams, MD** ([david.williams2@childrens.harvard.edu](mailto:david.williams2@childrens.harvard.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](mailto:jshizuru@stanford.edu)), or **Christopher Dvorak** ([Christopher.dvorak@ucsf.edu](mailto:Christopher.dvorak@ucsf.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](http://stjude.org/LVXSCID-ND), or contact **Ewelina Mamcarz, MD** ([ewelina.mamcarz@stjude.org](mailto:ewelina.mamcarz@stjude.org)), **Aleksandra Petrovic, MD** ([Aleksandra.Petrovic@seattlechildrens.org](mailto:Aleksandra.Petrovic@seattlechildrens.org)), or **Mort Cowan, MD** ([Mort.Cowan@ucsf.edu](mailto:Mort.Cowan@ucsf.edu)).



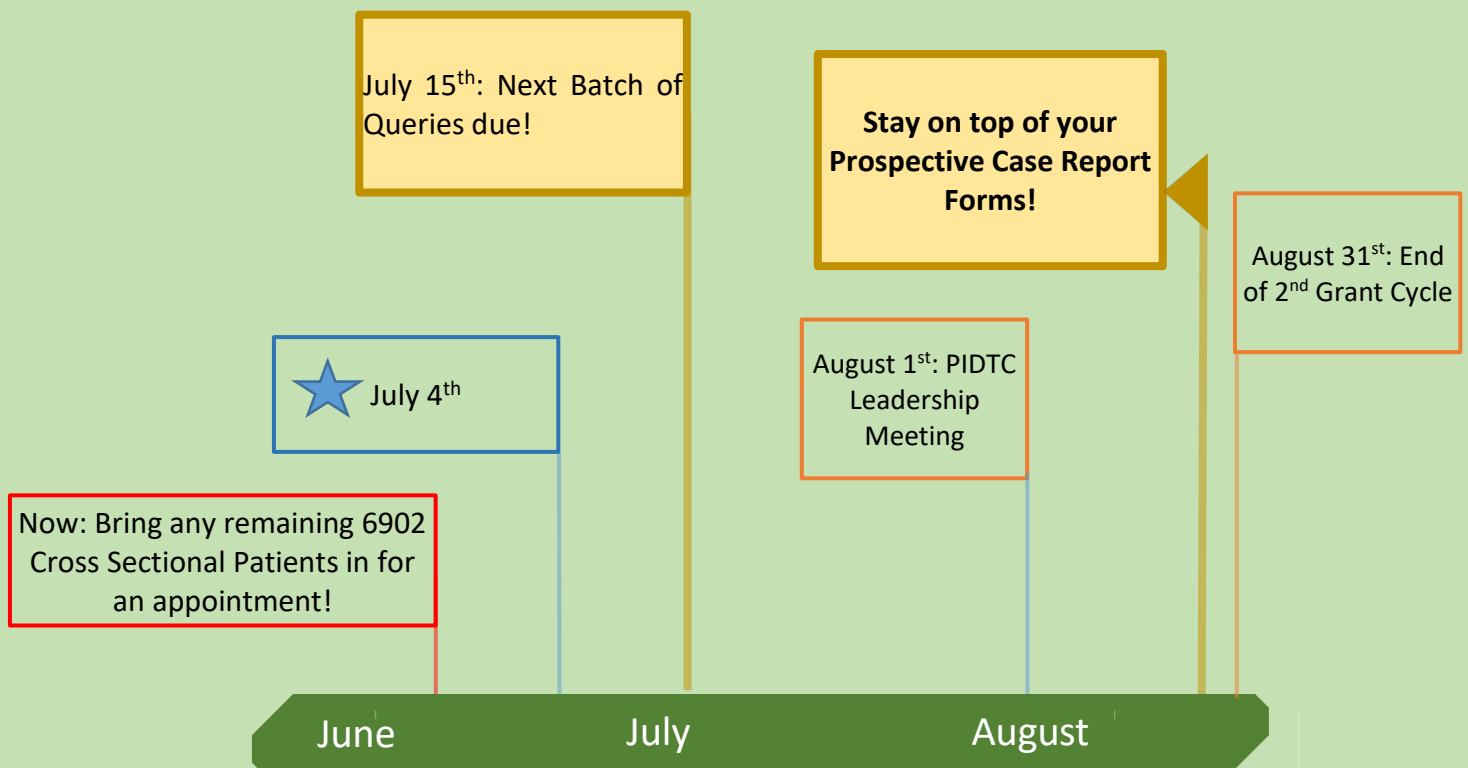


## RDCRN/DMCC Update

We bid farewell to **Amoy Fraser**, who served as PIDTC's DMCC Project Manager since April 2017. During her time with PIDTC, Amoy helped coordinate the RDCRN training for numerous new coordinators and investigators. We are happy for her as she moves on to new career adventures.

This month, we welcomed **Shannon Dotzel (left)**. Shannon earned her Bachelor of Science degree in Biology from Ursinus College in 2015. She participated in research involving gene mapping during this time. Shannon has 4 years of clinical trial experience, including phase 2 and 3 studies and has worked for both a pharmaceutical company and a contract research organization. Shannon has been with the RDCRN since February of 2018 and is the project manager for two additional RDCRN consortia. Be sure to contact her with any DMCC questions at [Shannon.Dotzel@epi.usf.edu](mailto:Shannon.Dotzel@epi.usf.edu).

## PIDTC: Summer Timeline 2019



# Attention Families!

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

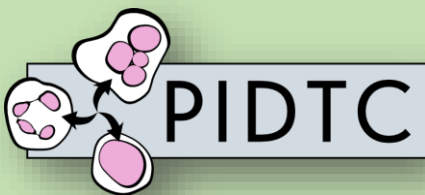
## Join the RDCRN PIDTC Contact Registry!

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>



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



***Left to Right: Katie Chang, Tara Bani, and Elizabeth Dunn***

Got announcements or photos you'd like to share in the next newsletter? Email: [catherine.chang@ucsf.edu](mailto:catherine.chang@ucsf.edu)