



Primary Immune Deficiency Treatment Consortium

NEWSLETTER

Winter/Spring 2022 | Issue 14



Greetings from Jennifer Puck, Chris Dvorak and Elie Haddad, Co-PIs

We hope everyone had a wonderful and safe holiday season.

The PIDTC begins the year with restarting the 6907 (SCID) protocol activation at all 47 PIDTC sties and with obtaining NIAID approval for our 6908 (CGD) and 6906 (PIRD) in the near future. We are very excited for these protocols to launch consortium wide and for everyone to see the efforts of our protocol teams come to fruition.

The PIDTC plans to publish more than 15 papers this year! This is thanks to the hard work of all our sites in helping with our legacy data cleaning. We are very excited to see what new insights these papers will deliver! We could not have done this without the help from our clinical research coordinators and investigators.

Thank you to everyone it the PIDTC for your dedication and hard work. We lookg forward to another great year together.

*With appreciation,
Jennifer, Chris and Elie*



Chris Dvorak, Jennifer Puck, and Elie Haddad enjoying a special night at the aquarium after the PIDTC Workshop.

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- Greetings from PIs
- PIDTC Education Day & Annual Workshop
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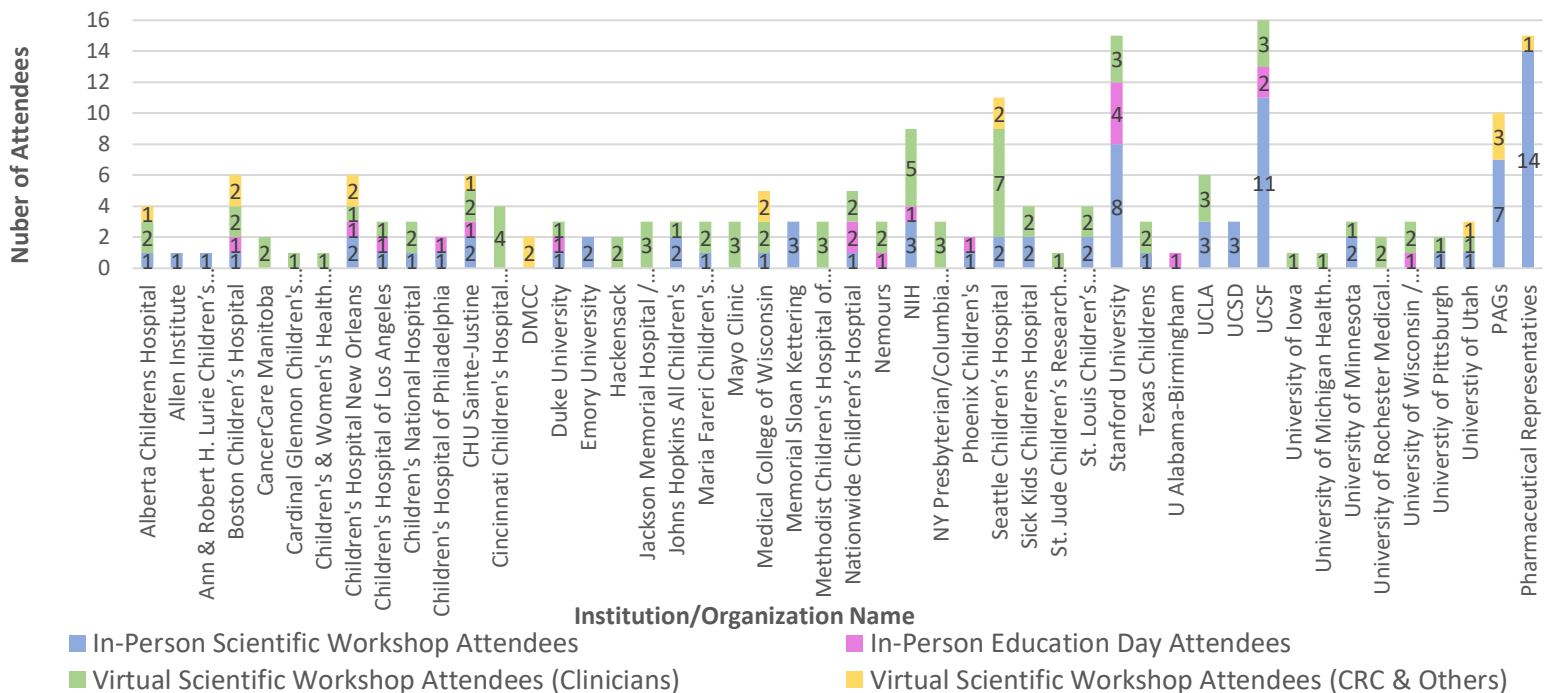
- Protocol Updates
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PIDTC Education Day & Annual Workshop (November 15-18, 2021)



This year’s PIDTC Education Day and Annual Workshop was hosted at Pacific Grove, California. This was the first year we offered both in person and virtual attendees to our annual meeting. We were thrilled to see 102 attendees in person as well as the 95 attendees virtually!

Number of In-Person and Virtual Attendees



The majority (95%) of the survey respondents (N=18) found the PIDTC Education Day to be very or extremely useful. The presentation led by Dr. Christopher Dvorak and Dr. Elie Haddad titled *Overview of Conditioning in HCT for PID* was found to be the most well-chosen, relevant, timely, and up to date presentation. All 3 clinical care topics were the most favorably received in terms of the descriptors above. Each Education Day attendee was provided a mentor to assist with their case report presentation. The survey responses indicated that the mentors were very helpful.

Ninety-four percent of the survey takers (N=126) stated the overall Scientific Workshop to be excellent. Many found the number of speakers (90%), scientific content overall (94%), representation of institutions (85%), representation of junior faculty (85%), and networking/consulting opportunities (68%) to have the perfect balance (neither too little nor too many). There were numerous attendees that have shown interest in wanting to be more involved in the PIDTC Protocols, including Severe Combined Immunodeficiency (SCID), Chronic Granulomatous Disease (CGD), Primary Immune Regulatory Disorders (PIRD), and Patient Advocacy Group (PAG) Related Research.

Thank you for attending our Ed Day and Workshop—we look forward to seeing everyone at our next one in April 2023!

We would also like to thank our wonderful keynote speakers for their great presentations.

Dr. Peter Hotez presented two talks: Science, Communication and Media and Vaccines; Immunity and the New World of Pandemics.



Dr. Lisa Westerberg presented: Approaches to restore WAS protein in platelets and immune cells of XLT and WAS patients.



Deciphering intracellular signalling by actin regulators and their role in immune cell homeostasis



Lisa Westerberg, Ph.D.
Karolinska Institutet
Dept of Microbiology Tumor and Cell Biology



A composite image featuring a small inset portrait of Dr. Lisa Westerberg in the top right corner. The main area contains three fluorescence microscopy images of cells. The left image shows cells with blue and red staining. The middle image shows a cell with bright blue staining. The right image shows a network of orange-stained filaments.

Here are some pictures from our event!



This is the first year the PIDTC had an option for virtual attendance—it was great to see our virtual attendees participating in real time.

Pictured: Dr. Luigi Notarangelo and Sung-Yun Pai (on screen, top row, left to right) with great questions and feedback to Dr. Don Kohn's (podium) presentation on Gene Therapy.



Dr. Parkman and Dr. Crooks enjoying the nice weather at Pacific Grove during a break.



Dr. Parkman and Robertson pictured with their mentees after the award ceremony.

(Bottom, Left to right) Drs. Robertson Parkman, Richard O'Reilly, and Neena Kapoor

(Top, Left to Right) Drs. Susan Prockop, Gay Crooks, Ami Shah, and Don Kohn



Asilomar had gorgeous views of the ocean! Many of our attendees were able to enjoy morning hikes and lunch at the beach.

Bonfire Night!

Our workshop attendees were able to relax, enjoy some smores and drinks at our Bonfire night! It was great seeing the results of everyone's skills at making s'mores.



Thank you to all our Education Day speakers and mentors for the creating an engaging learning environment. We look forward to the future of PID research.

A Night at the Monterey Bay Aquarium



Our guests were able to enjoy the many exhibits at the Monterey Aquarium during our dinner and reception.



What a fun night at the Monterey Aquarium! (Left to right) Drs. Farhan Murshed, Matthew Kan and Morna Dorsey in front of the Kelp Forest Exhibit.



Some of the amazing ladies in the PIDTC—they are Protocol PIs, site PI, sub-investigators, practicing doctors!



The PIDTC would like to thank Dr. Ami Shah, Beatrice Ochoa and the team at Stanford University as well as the UCSF program managers for a job well done!

Posters from the Workshop:

For an in-depth view of the posters, please click on the link below the poster:



Patient with Biallelic Variant in PI4KA with Multiple Intestinal Atresia and Combined Immunodeficiency

Sara W Van Meerbeke MD¹, Christin Deal MD¹

¹ Division of Allergy & Immunology, UPMC Children's Hospital of Pittsburgh, Pittsburgh PA, USA

Link: <https://ucsf.box.com/s/r9w423di7vujxcw6t9zn0zl58bvzzabm>



Spontaneous Resolution of Severe Idiopathic T Cell Lymphopenia

Saddiq Habiballah¹, Jennifer S. Whangbo^{2,3}, and Craig D. Platt¹

¹Division of Immunology, Boston Children's Hospital, Boston, MA. ²Division of Hematology-Oncology, Boston Children's Hospital, Boston, MA. ³Department of Pediatric Oncology, Dana-Farber Cancer Institute, Boston, MA.



Link: <https://ucsf.box.com/s/6ywdjumqiz6cvkd32nqug85a25qelab3>




Severe Combined Immunodeficiency (SCID) Screening in Arizona: Lessons Learned from the First 2 Years

Natalie A. Booth, DO^{1,2}, Catherine M. Freeman, MD^{3,4}, Benjamin L. Wright, MD^{3,4}, Christine Rukasin, MD^{2,3,4}, Priscila Badia, MD^{1, 5}, Cindy S. Bauer, MD^{2,3,4}, Holly Miller, DO^{1,2,5}

¹Center for Cancer and Blood Disorders, Phoenix Children's Hospital, Phoenix, AZ. ²Department of Child Health, University of Arizona College of Medicine – Phoenix. ³Division of Allergy, Asthma, and Clinical Immunology, Mayo Clinic, Scottsdale, AZ. ⁴Division of Allergy and Immunology, Phoenix Children's Hospital, Phoenix, AZ. ⁵Division of Hematology and Oncology, Mayo Clinic, Scottsdale, AZ.





Link: <https://ucsf.box.com/s/rjt0aznndvz0xgjsjxq2vs6149910z90>



Prevalence and Characteristics of Cytomegalovirus Ocular Disease in Children: A Multi-Center Study

Carmel L. Mercado MD^{1,2}, Colin P. Froines BA³, Eric D. Gaier MD PhD^{4,7}, Qinyun Wang MD⁸, Maanasa Indaram, MD⁸, Michael J. Wan, MD⁹, Ankoor S. Shah MD PhD^{4,5}, Euna B. Koo MD¹⁰

¹Department of Ophthalmology, University of Washington, ²Department of Ophthalmology, Seattle Children's Hospital, ³University of Washington, ⁴Boston Children's Hospital, ⁵Department of Ophthalmology, Massachusetts Eye and Ear Infirmary, ⁶Department of Ophthalmology, Harvard Medical School, ⁷Picower Institute for Learning and Memory, Department of Brain and Cognitive Sciences, Massachusetts Institute of Technology, ⁸Department of Ophthalmology, University of California San Francisco, ⁹Department of Ophthalmology and Vision Sciences, SickKids, ¹⁰Byers Eye Institute, Stanford Children's Hospital and Clinics



Link: <https://ucsf.box.com/s/j96nrxlz39k4c1i2nvrujwk6dz2svlb2>

XIAP Deficiency Successfully Managed with Monotherapy Tadekinig Alfa (IL-18BP)




Ashley V. Geerlinks¹, Michael B. Jordan², Andrea M. Dvorak³, Eduardo J. Schiffrin⁴, Edward M. Behrens⁵, Rebecca Marsh⁶

¹Children's Hospital London Health Sciences Centre, London, Ontario, Canada; ²Cincinnati Children's Hospital Medical Center, University of Cincinnati, Cincinnati, OH, USA; ³SUNY Upstate Medical Center, Syracuse, NY, USA; ⁴A2Bio Ltd, EPFL Innovation Park, Lausanne, Switzerland; ⁵Children's Hospital of Philadelphia, Philadelphia, PA, USA

Link: <https://ucsf.box.com/s/z6zsry77dax7t72i65y93yl4pxt5fsie>

Daratumumab for Management of Refractory Autoimmune Hemolytic Anemia in a Patient with Primary Immune Regulatory Disorder




Larisa Broglie, Rachel Phelan, Amy Moskop, Julie Talano, Sid Rao, Kristin Page, David Margolis, Jack Routes, James Verbsky

Link: <https://ucsf.box.com/s/vc3fs5hba8r53cjsrbh4rstgo9extrnv>

Posters from the Workshop:



FLIPPING THE COIN

AUTOLOGOUS VERSUS ALLOGENEIC TRANSPLANT FOR LYMPHOMA IN COMBINED IMMUNE DEFICIENCY

Andrea Sitek, M.D.; Avni Joshi, M.D., M.S.
Division of Allergy and Immunology, Mayo Clinic, Rochester, MN

Link: <https://ucsf.box.com/s/alzzvdp30sr9o2ouf0v86ccazrxre4ns>



Successful Transplantation Using TCR alpha beta Depleted Haploidentical Graft in a Female Patient with CGD and Skewed Lyonization of X Chromosome

¹Mary Ann Miranda MD, ²Jennifer W. Leiding MD, ³Deepak Chellapandian MD

1. Division of Allergy and Immunology, Department of Pediatrics, University of South Florida

2. Johns Hopkins University

3. Center for Cell and Gene Therapy for Non-Malignant Conditions, Johns Hopkins All Children's Hospital



Link: <https://ucsf.box.com/s/1p4i9jelciachjln8o6eedfxj38eprl2>

PEGylated Recombinant Adenosine Deaminase Maintains Detoxification and Lymphocyte Counts in Patients With ADA-SCID

Dorsey MJ¹, Rubinstein A², Lehman H³, Fausnight T⁴, Wiley J⁵, Haddad E⁶

¹University of California San Francisco Medical School, San Francisco, CA, USA; ²Montefiore Medical Center/Albert Einstein College of Medicine, Bronx, NY, USA; ³University of Buffalo Jacobs School of Medicine and Biomedical Sciences, Buffalo, NY, USA; ⁴Penn State Health Hershey Medical Center, Hershey, PA, USA; ⁵Leadiant Biosciences, Inc., Gaithersburg, MD, USA; ⁶University of Montreal, Montreal, QC, Canada

Link: <https://ucsf.box.com/s/d9gxzmswfoxy1vd5r1kudckdeyl0wdyn8>

Newborn Screening Enigmas

Talal I. Mousallem, MD and Rebecca H. Buckley, MD

Duke University School of Medicine, Durham, NC



Link: <https://ucsf.box.com/s/9es0ag3kt0v7e47ocai5v99enkmosdcu>



Persistent Mixed Chimerism post Bone Marrow Transplantation (BMT) in a Child with X-linked Chronic Granulomatous Disease (CGD)

Jeffrey R. Andolina MD, MS, Katherine L. Tuttle MD, and Geoffrey A. Weinberg MD

Department of Pediatrics, Divisions of Hematology/Oncology/Stem Cell Transplantation, Allergy/Immunology, and Infectious Diseases
Golisano Children's Hospital, University of Rochester, Rochester, NY



Link: <https://ucsf.box.com/s/p4vojgiky8ybxhbcttiaom4b7eyfpmjo>

CONGRATULATIONS!

The PIDTC presented our first Lifetime Achievement Awards at our Annual Scientific Workshop.



Dr. Rebecca Buckley and her family sporting Carolina Blue (Go Blue Devils!)

Dr. Rebecca Buckley

Dr. Rebecca Buckley is a pioneer in the field of immunology who trained many of today's immunologists and allergists. She developed SCID bone marrow transplantation using parental T-Cell depletion for unrelated donors, discovered numerous disease-causing genes, and championed Newborn Screening. Dr. Buckley is a huge advocate for research—her site, Duke University, has enrolled over 100 patients in our legacy protocols. Her devotion to her patients and clinical research is unparalleled.

Dr. Richard O'Reilly

Dr. Richard O'Reilly's contributions to PID are invaluable—he performed the first successful T-cell depleted HSCT and unrelated donor HSCT. For the transplant field in general, he is the founder of the ASBMT, one of the developers of FACT. He helped developed the PIDTC to address a need for this community. Today, Dr. O'Reilly sits as one of PIDTC's external advisors who is devoted to continuing the PIDTC's efforts beyond the current grant cycle. His guidance and expertise have been essential.



Drs. Parkman and O'Reilly were able to share this moment with their significant others.



(Left to Right) Drs. Robby Parkman and Richard O'Reilly posing with their awards!

Dr. Robertson Parkman

Dr. Robertson Parkman dedicated more than 50 years to clinical research in pediatric immunology, mentorship, and patient care. His contribution to immunology is instrumental for patient care. His lab first observed thymic dysfunction in the pathogenesis of chronic GvHD. Dr. Parkman sits at the PIDTC as one of our external advisors—his enthusiasm to continue the work PIDTC has done and will do is incomparable.

PAG UPDATES:

Immune Deficiency Foundation:



Funding Opportunity!

2022 IDF Research Grant—Deadline: February 28, 2022

The Immune Deficiency Foundation (IDF) Research Grants are funded in large part by proceeds from IDF's Walk for Primary Immunodeficiency.

The IDF Research Grant Program has been developed to encourage and support “Patient-Oriented Research on Primary Immunodeficiency Diseases”. The intent of the grant is to support well-defined research projects that have a specified benefit for improving the treatment, health, disease management or diagnosis of persons with primary immunodeficiency. Consideration will also be given to studies that contribute to the body of medical knowledge in primary immunodeficiency.

The program is open to applicants currently based in the U.S. and will consist of one-year grants.

Award Value: \$25,000 - \$50,000 (a somewhat higher level of support is available for a few exceptional proposals)

Link to apply: <https://primaryimmune.org/2022-research-grant-application>

Meet IDF's new President and CEO: Jorey Berry

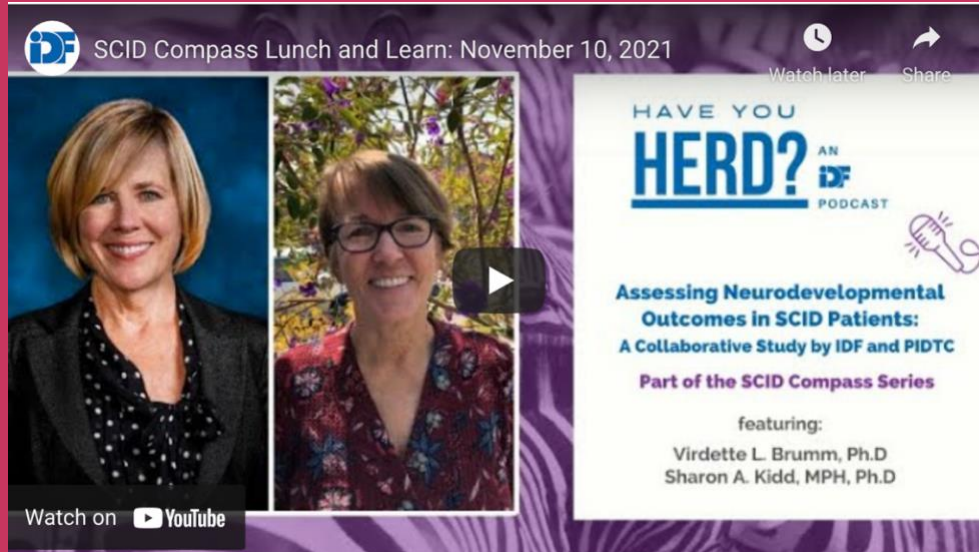


Jorey Berry is the new president and CEO of the Immune Deficiency Foundation. Her expertise with patient advocacy in her work at March of Dimes will be invaluable for our community. Also, she has some experience in this community in her help to push for Newborn Screening for SCID.

The PIDTC looks forward to working with her in 2022 and beyond.

Link: <https://primaryimmune.org/news/jorey-berry-announced-immune-deficiency-foundation-president-and-ceo>

SCID Compass Lunch and Learn: Assessing Neurodevelopmental Outcomes in SCID Patients: A Collaborative Study by IDF and PIDTC presented by Virdette L Brumm, Ph.D and Sharon A. Kidd, Ph.D



In this podcast, Dr. Brumm and Dr. Kidd share information about the goals of, and the need for this important PIDTC and IDF collaborative study. The PIDTC would like to thank them for spearheading the efforts in working tirelessly with our sites to activate this study as well as in managing this project.

Link: <https://youtu.be/g3oBK9OJB6g>

SCID Angels for Life:



Update!

- The SCID Angels for Life Foundation is proud to present the Alicia Chaudhury Scholarship to four brilliant awardees.
- The committee at SCID Angels are currently reviewing grant applications for their newly instated grant! Decisions will be out in January 14, 2022.

CGD Association of America:



The CGDAA developed a quick guide for female carriers of X-Linked CGD. This resource provides resources for CGD carriers to discuss options with their primary care providers.

Guide available for Download at:

<https://cgdaa.org/f/just-launched-cgdaas-guide-for-carriers-of-x-linked-cgd?blogcategory=CGD+carriers>

WAS Foundation & HyperIgM Foundation:



Open! WAS Carrier Survey

The WAS Foundation and Hyper IgM's X-Linked Carrier Survey is now open! Please help distribute this survey. For more information, please read the description below.

The Hyper IgM Foundation and the Wiskott-Aldrich Foundation's first-ever international X-Linked Carrier Surveys are now available! We are excited to launch this survey project in collaboration with the Primary Immune Deficiency Treatment Consortium (PIDTC) and the Immune Deficiency Foundation (IDF). We hope you will take a few moments to complete the survey at the following link:

Study Title: A study assessing the physical health and psychosocial well-being of female carriers of Wiskott Aldrich syndrome (WAS) and X Linked Hyper IgM Syndrome (X-HIGM)

<https://tinyurl.com/WAS-X-HIGM-CarrierSurvey>

Developed with immunologists who are experts in primary immunodeficiency diseases, including Wiskott-Aldrich syndrome (WAS) and X Linked Hyper IgM Syndrome (X-HIGM), this upcoming survey aims to learn more about the reported health outcomes of Carriers of WAS and X-HIGM and how this affects individuals' lives, including diagnosis, along with carries' overall well-being and quality of life.

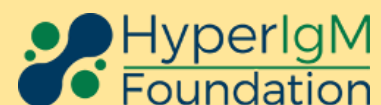
Carriers of X-HIGM and WAS are very rare and have never been part of a detailed international exploratory survey before. Therefore, participating is particularly important for all families affected by Hyper IgM and Wiskott-Aldrich syndromes as the survey results will help increase what is currently known about carriers of X-HIGM and WAS.

If you have any questions about this survey project, please email us at akiva@hyperigm.org or sumathi.iyengar@wiskott.org or cscalchunes@primaryimmune.org

Thank you for your help on this important project.



Thank you to all our PAGS!



Protocol Updates:

Severe Combined Immune Deficiency (SCID) - 6901/6902/6907

Updates:

Thank you to our PIs, **Drs. Chris Dvorak, Elie Haddad and Jen Heimall** for leading the SCID team finalizing the new 6907 protocol, and overseeing the numerous manuscripts that are in the works. We thank our outstanding statistics team, led by **Dr. Brent Logan**, for its efforts pulling together this data.

The modified consents are UCSF IRB-approved and we will begin onboarding all PIDTC sites in January.

Goals: Do not miss enrolling your 6901 Prospective SCID patients (while awaiting onboarding) during the DMCC transition period! Enter your data into the CRFs in the South Florida database and then email Elizabeth Dunn at Elizabeth.dunn@ucsf.edu, to finalize patient eligibility via email.

Chronic Granulomatous Disease (CGD)- 6903/6908

Updates: The CGD group is working on its first set of major papers on the overall dataset and survival, 2nd transplants, cord blood donors, and more. The data has been cleaned thoroughly and a final dataset is envisioned in the new year. The 6908 protocol, following the lead from the 6903 legacy protocol, will be enrolling patients with CGD, but this time focusing only on those patients going on to gene therapy or transplant. The protocol will likely be opened to the UCSF site (coordinating center) in January with all remaining 46 sites to follow soon after.

Enrollment: Do not miss enrolling your 6903 Prospective CGD patients. Enter in your Eligibility data into the South Florida database and then email Elizabeth Dunn at Elizabeth.dunn@ucsf.edu to finalize patient eligibility.

Wiskott-Aldrich Syndrome (WAS)-6904

Updates. The WAS team is now working on data clean-up, data analysis and manuscript writing for the second 6904 paper with a larger “N” of patients. We especially want to thank investigators **Drs. David Shyr, Blachy Davila, Jessie Barnum, and Ami Shah** and our talented statisticians **Dr. Ruta Brazauskas, and Joy Liu**. We would also like to thank **Dr. Sumathi Iyengar** for her advice and active participation in our protocol calls.

Primary Immune Dysregulation Disorder (PIRD)-6906

Updates: The PIRD group is planning on submitting its protocol to the central IRB in January for start-up at the UCSF site (coordinating center). This protocol will be enrolling all patients that meet its clinical and/or genetic criteria in both best available therapy and treatment arms. Unique to this protocol is a family member arm to focus on the same genetic defects in family members, with research sampling at baseline, and collecting annual reports from these individuals about their health. This is a new protocol without a legacy experience.

Industry Sponsors

Chiesa:

- **Abstract:**

- Pegylated Recombinant Adenosine Deaminase (Elapegedemase) Maintains Metabolic Detoxification in Patients With ADA-SCID
Dorsey MJ¹, Rubinstein A², Lehman H³, Fausnight T⁴, Wiley J⁵, Haddad E⁶

Link: <https://ucsf.box.com/s/xsk4a11s1cr02z78l9pnu9pbr5emrf53>

- Long-term immune reconstitution in ADA-deficient patients treated with elapegedemase: a real world experience

Luis Murguia-Favela¹, Sneha Suresh², Nicola Wright¹, Eyal Grunebaum³.

Link: <https://ucsf.box.com/s/acu45av2ptf53orh73rruug41nt7jv21>



Takeda:

- **Abstract:**

- T-Cell Receptor Diversity as a Biomarker for Immune Reconstitution after Bone Marrow Transplantation for Severe Combined Immunodeficiency

Matthew Kan, Jason Yu, Aidan Rossiter, Alison Yip, Wendy Chan, Chris Dvorak, Mort Cowan, Jennifer Puck

Link: <https://ucsf.box.com/s/bcf7dcio04g8fear7yixdzupqsuce38e>



X4:

- **Abstract:**

- Oral Administration of Mavoxifafor, a CXCR4 Antagonist, Increases Peripheral White Blood Cell Counts across Different Disease States

(<https://ash.confex.com/ash/2021/webprogram/Paper152990.html>)

- Mavoxifafor, an Oral CXCR4 Antagonist, for Treatment of Patients with WHIM Syndrome: Results from the Long-Term Extension of the Open-Label Phase 2 Study

(<https://ash.confex.com/ash/2021/webprogram/Paper145759.html>)



Path4ward Genetic Testing Program

X4 Pharmaceuticals, Inc. has partnered with Invitae to offer third-party sponsored genetic testing at no charge for individuals who may carry a genetic mutation known to be associated with congenital neutropenia. Primary immunodeficiencies are a group of inherited disorders of the immune system including congenital neutropenia and WHIM syndrome.

The PATH4WARD testing program helps eliminate barriers to genetic testing and increases the certainty in obtaining a correct diagnosis by enabling patients who may carry a genetic mutation known to be associated with congenital neutropenia, and their family members, to have access to genetic testing and counseling.

Website: <https://www.invitae.com/en/path4ward/>



Ongoing Clinical Studies

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@nih.gov); Los Angeles PI: **Donald Kohn, MD** (dkohn1@mednet.ucla.edu); Sponsor: **David A. Williams, MD** (david.williams2@childrens.harvard.edu).

LAD-I gene therapy trial

This Leukocyte Adhesion Deficiency Type I (LAD-I) gene therapy trial is currently enrolling patients at UCLA (US). Additional treatment centers will include UCL/GOSH (UK) and Hospital Infantil Universitario Niño de Jesús (Spain). The trial is sponsored by Rocket Pharmaceuticals, Inc., and funded by the California Institute of Regenerative Medicine (CIRM). For more information, please contact LADclinicaltrial@rocketpharma.com or visit <https://clinicaltrials.gov/ct2/show/NCT03812263?term=NCT03812263&rank=1> or <https://www.rocketpharma.com/lad-i-clinical-trial-for-health-care-providers/>

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 Artemis SCID Gene Therapy

In this trial, newly diagnosed or previously treated patients with insufficient immunity due to ART-SCID receive "lentiviral gene transfer," also called "gene therapy." A normal copy of the DCLRE1C gene is inserted into blood-forming stem cells that grow and develop into all blood lineages. The inserted gene provides correct instructions to the defective stem cells so that functioning T and B lymphocytes can develop. So far 10 patients have been treated.

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).

UPMC clinical trial: BOLT-BMT

The University of Pittsburgh, sponsored by NIAID, is conducting a study for patients with primary immunodeficiency (PID) and end-stage lung disease. In this study, patients receive a lung and bone marrow transplant (BMT) from the same donor. Lung transplant prior to BMT would allow for restoration of pulmonary function prior to BMT, allowing PID patients to proceed to BMT, which would be curative for the patient's underlying immunodeficiency. For more information, please contact Dr. Paul Szabolcs at paul.szabolcs@chp.edu or Shawna McIntyre at mcintyresm@upmc.edu or 412-692-5552.

Anti-c-KIT (JSP191) Transplant Protocol

This Phase I study is a single arm, open label, dose escalation trial being conducted at multiple PIDTC centers, including: UCSF Benioff Children's Hospital, Lucile Packard Children's Hospital at Stanford and Memorial Sloan Kettering Cancer Center in New York. The study objective is to evaluate the safety and tolerability of allogeneic CD34+ human stem cells (HSC) in patients with Severe Combined Immune Deficiencies (SCID) conditioned for transplantation with JSP191, a monoclonal antibody that targets human CD117. The trial is open for both patients in need of repeat HCT as well as newly-diagnosed patients undergoing first HCT.

For questions regarding the trial please contact Wendy Pang (wpang@Jaspertherapeutics.com).

Viral CTL Consortium (VIRCTL)

Principal Investigator Mitchell S. Cairo, MD and Study Chairs Julie Talano, MD and Nancy Bunin, MD, are studying (funding by the FDA) the safety, efficacy and biology of viral CTLs derived from related donors by the Cytokine Capture System using the Prodigy device in patients with immunodeficiencies either secondary to HSCT or primary immunodeficiencies with refractory CMV, ADV and/or EBV or intolerant to anti-viral therapy. If you and your institution are interested in participating in this clinical trial, please contact **Dr. Mitchell S. Cairo** (Mitchell_cairo@nymc.edu).

CSIDE

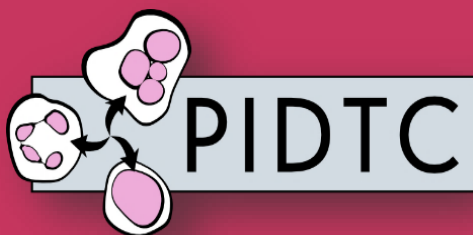
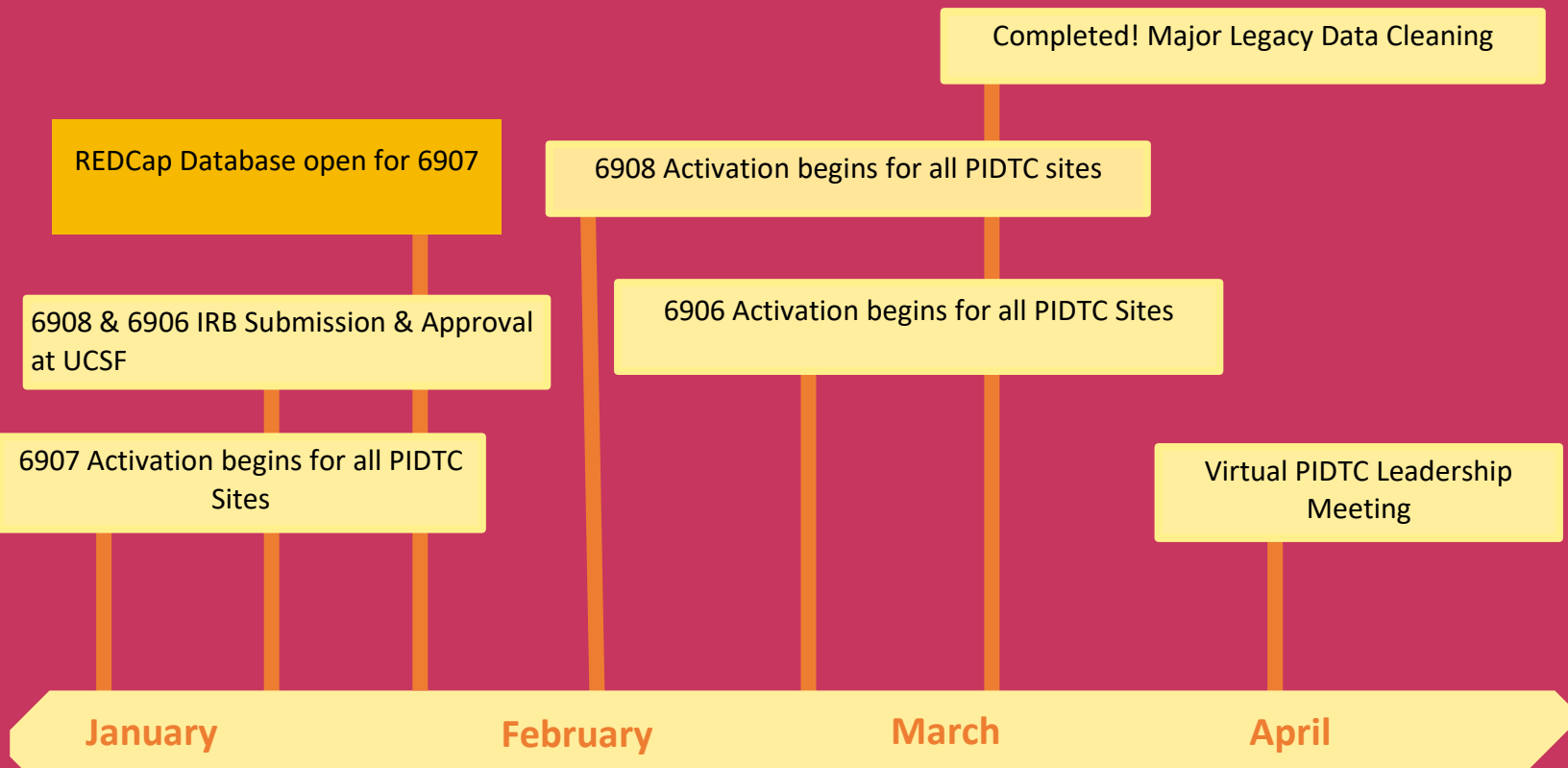
CSIDE is open to enrollment 34 sites and 13 patients have been enrolled to date. More centers are currently being activated! If you have any questions about getting your site on board, please email **Sung-Yun Pai, MD** (sung-yun.pai@nih.gov), **Mike Pulsipher** (mpulsipher@chla.usc.edu), and **Jenny Vogel** (jvogel@nmdp.org).

A Study of Mavoxifafor in Participants With Severe Congenital Neutropenia and Chronic Neutropenia Disorders

This Phase 1b study will determine the safety and tolerability of mavoxifafor in participants with severe chronic idiopathic neutropenia (CIN) and selected congenital neutropenia disorders. The anticipated enrollment is up to 25 participants.

For questions regarding the trial, please email clinicaltrialinfo@x4pharma.com or call 857-529-5779. Website: <https://www.x4pharma.com/patients/chronic-neutropenia/>

PIDTC: Winter/Spring 2022



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

Got announcements?
Email: alison.yip@ucsf.edu and kiana.soriano@ucsf.edu