Primary Immune Deficiency Treatment Consortium

NEWSLETTER

Winter 2025 | Issue 22

The St. Petersburg Pier Building At Night (https://floridatraveler.org/2020/07/18/ready-or-not-saint-petersburgs-new-downtown-pier-is-open/)

Greetings from Drs. Rebecca Marsh, Chris Dvorak, Elie Haddad, and Troy Torgerson, Multi-PIs

Hello PIDTC Members,

We are looking forward to seeing everyone next month in St. Petersburg, Florida for the upcoming 14th Annual PIDTC Education Day and Scientific Workshop!

We have a number of exciting initiatives under the R24 grant. We are working on making current data more publicly accessible on the CHIRON platform. Additionally, a new PIDTC website is underway, as well as implementing new 'tumor board' style webinars. We will continue to update you on these developments.

Congratulations to all of our PIDTC sites for getting the 6906, 6907, and 6908 protocols open. Thank you to your teams and our program managers for making this huge feat possible. Please continue to enroll in the 3 active protocols.

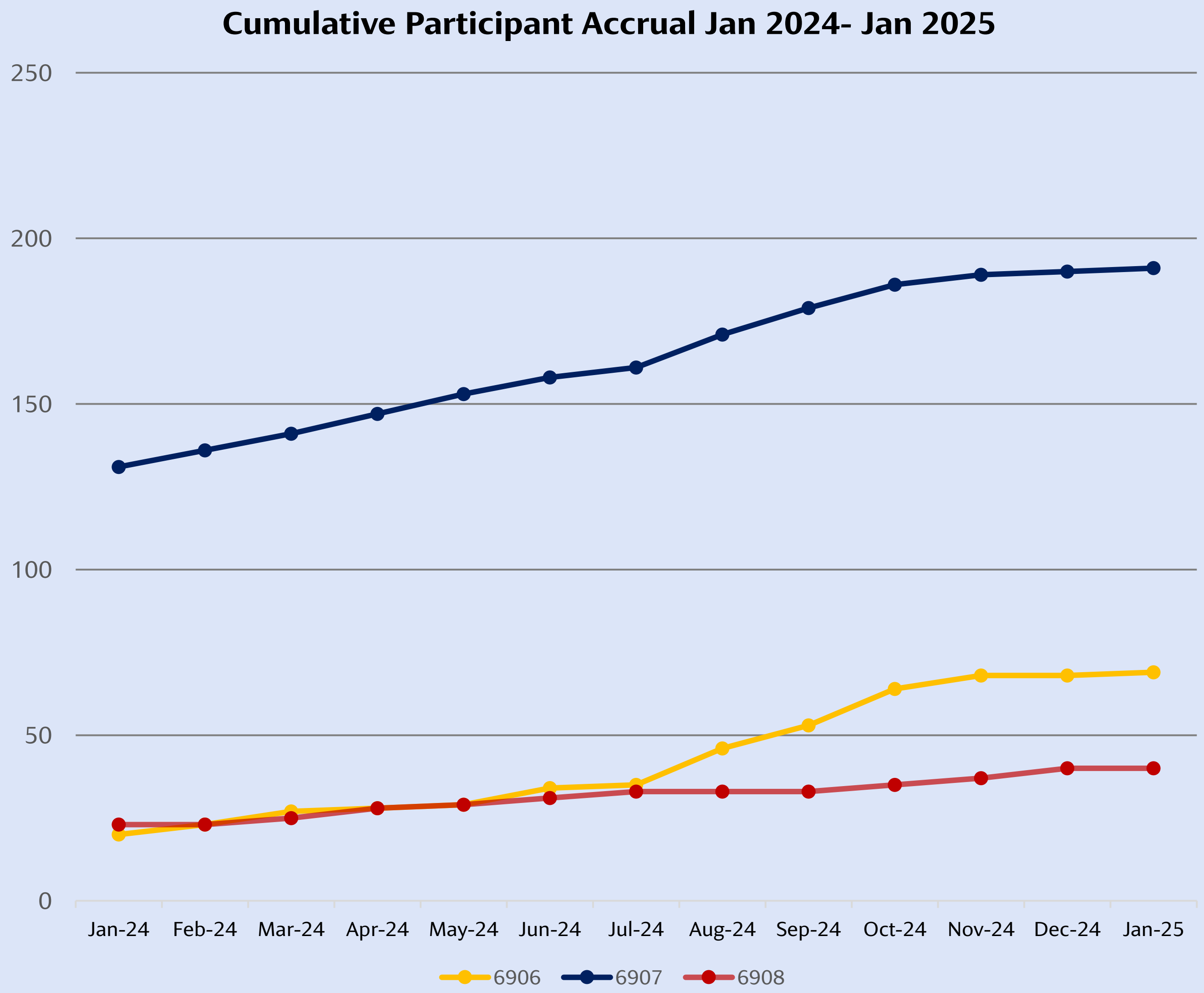
Cheers!

Rebecca, Chris, Elie, and Troy

IN THIS ISSUE

- **Greetings from the PIs**
- **Enrollment Update**
- **2025 Annual Workshop**
- **PAG Updates**
- **Ongoing Clinical Studies**

Enrollment Update



All three current PIDTC protocols are activated at most of our participating sites. The 6906 PIRD Protocol has enrolled 49 new participants in the last year with a total of 69 enrolled. The 6907 SCID protocol has enrolled 60 participants including 18 new SCID patients. The 6908 CGD Protocol enrolled 17 participants over the past year including 8 new CGD patients.

Let’s continue to keep up the enrollment!

PIDTC Education Day & 14th Annual Scientific Workshop in St. Petersburg, Florida

The PIDTC host our **Education Day** on March 9th-10th, where we will once again explore unique aspects of diagnosis and management of primary immunodeficiencies with leaders in the field, followed by our **Scientific Workshop** from March 10th-13th. The workshop program includes invited lectures from leading experts in the IEI field, updates regarding PIDTC working group efforts, and new and exciting developments for the PIDTC, all with the chance to enjoy the Florida sun with friends and colleagues.

We look forward to discussing the progress we have made as researchers and healthcare providers with the diseases we investigate, the challenges and opportunities ahead of us, and our achievements as a consortium!

Thank you to Jennifer Leiding and Johns Hopkins All Children's Hospital for hosting us!

PAG Updates

Jeffrey Modell Foundation

jmTranslational Research

The JMF is thrilled to have awarded two grants of $200,000 each in the twelfth cycle of its Translational Research Program! Cycle 13 is now underway. Interested applicants should submit a Letter of Intent through ProposalCentral by March 12, 2025. Learn more here: https://info4pi.org/medical/research-grants/

jmGENIE

The Global Education and Information Exchange (G.E.N.I.E.) grant funds programs with a focus on PI to encourage global collaboration, meaningful dialogue, and ongoing education to improve the quality of life for patients worldwide. Funds may be used to support continuing education programs, grand rounds, scientific publishing, the creation of local meetings and seminars, and other physician education events. Awards are granted on a rolling basis throughout the year, following review and approval by the Advisory Board. Applicants must be enrolled in a fellowship or above. Learn more and apply here: https://info4pi.org/medical/genie/.

For more information about JMF click here: https://www.info4pi.org/

Wiskott-Aldrich Foundation

Global Hematologic Rare Disease Alliance

The Wiskott-Aldrich Foundation was recently invited to speak at the Global Hematological Rare Disease Alliance (GARDEN). “GARDEN (Global Hematological Rare Diseases Alliance) is a global alliance dedicated to transforming the lives of individuals affected by rare blood diseases. We unite healthcare professionals, researchers, policymakers, and patient advocates in a collaborative effort to address the challenges posed by hematological rare diseases to drive innovation, advance research, and advocate for equitable policies that ensure access to care.”

Management Consensus

The Wiskott-Aldrich Foundation will move forward with a management consensus statement on WAS using the Delphi method. Stay tuned for details!

For more information about the WA Foundation click here: https://www.wiskott.org/

Immune Deficiency Foundation

IDF Research Grant Program

IDF is now accepting research grant applications until June 30, 2025! IDF's Research Grant Program supports patient-centered research focused on improving the treatment, health, disease management, or diagnosis of people with PI, as well as research that contributes to the general body of PI knowledge. Applications are due June 30, 2025.

- Awards range from $25,000-50,000. Exceptional proposals are considered for higher levels of support.
- The *Michael Blaese Research Grant Award* is given to the highest-scored application each cycle.
- Currently unavailable for individuals employed by federal government agencies or departments.

Compromised: Life Without Immunity

Join the Immune Deficiency Foundation (IDF) this spring during one of our community online streaming events to view our documentary Compromised: Life without Immunity https://go.primaryimmune.org/2025-online-compromised-screenings

For more information about the IDF: https://primaryimmune.org/

CGD Association of America

CGDAA Upcoming Events

Coffee and Clinicians: CGDAA is hosting a series of Zoom talks on a variety of topics. The latest one featured Dr. Donald Kohn of UCLA, sponsored by PRIME medicine. Please email Felicia Morton at felicia@cgdaa.org if you are interested!

The CGDAA is currently developing a workshop and education day for X-Linked SCID and a Lunch and Learn with Dr. Christin Deal in collaboration with the Immune Deficiency Foundation. Sign up for their newsletter to stay updated with details about these events and the CGDAA: https://cgdaa.org/blog-%26-e-newsletter

For more information about the CGDAA: https://cgdaa.org/

SCID Foundation

VA 59th House District Delegate "Buddy" Fowler and Barb Ballard

The **SCID Foundation**, a patient advocacy organization for Severe Combined Immune Deficiency (SCID), began operating at the beginning of 2025. Barb Ballard is the President and Founder of this newly created non-profit. Barb's personal journey with SCID began in 1994 when her son, Ray, was diagnosed with X-linked SCID.

In 1996, Barb began searching for other families affected by SCID. The following year she established an email listserv to connect with the increasing number of families she had encountered. The listserv eventually grew to include several hundred families.

In 2001, Barb joined the Immune Deficiency Foundation's Board of Trustees, where she served for 18 years. She also served on The Board of the SCID Angels for Life Foundation from 2008 to 2021. She was named Director of SCID Angels in 2021 after losing Ray in 2019. In each of these roles, she strongly advocated for improved clinical outcomes including a 10-year effort to make universal SCID Newborn Screening a reality.

At the end of 2024, Barb founded the SCID Foundation to honor Ray's legacy and continue the advocacy, awareness, and support for SCID families that she has been providing for 30 years.

On January 30th, Barb joined IDF staff and advocacy volunteers for Virginia State Advocacy Day. The Virginia House was considering HB1782, a bill to establish a process for evaluating and implementing new newborn screening tests added to the Recommended Uniform Screening Panel (RUSP), and to set timelines for implementing tests deemed suitable by the Virginia Department of Health. On January 31st, the bill passed the Virginia House and will proceed to the state Senate. Upon passage, this legislation will set limits on the time it takes for new newborn screening tests to be implemented.

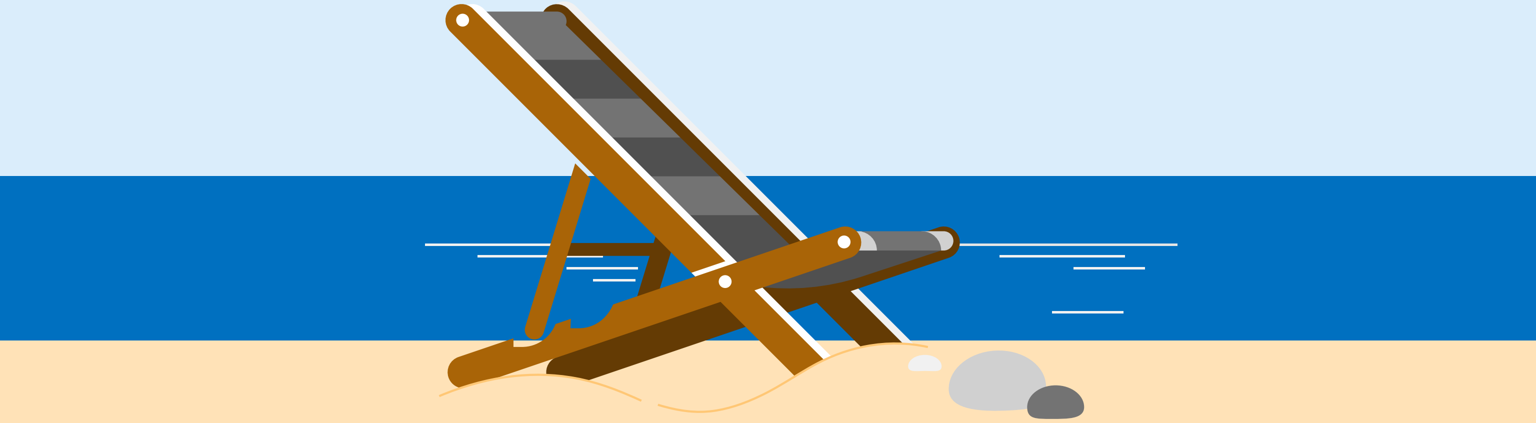
The SCID Foundation continues to support SCID families on their journey through the [Facebook Group](#) and associated [Facebook Page](#). When directing families to the SCID Foundation, be sure to emphasize that they should search for the "SCID Foundation". When searching for the term "SCID", families may come across other groups which do not have the safety protocols SCID Foundation uses to protect the group's privacy. More information on the SCID Foundation can be found at https://SCIDFoundation.org.

Ongoing Clinical Studies

C-SIDE (aka PIDTC 6905)

The purpose of this trial is to test the efficacy of regimens containing busulfan targeted to 30 mg*h/L vs 60 mg*h/L in patients with X-linked SCID, JAK3 SCID, RAG1/RAG2 SCID. To date 13 IL2RG/JAK3 and 10 RAG1/RAG2 patients have been enrolled. The vast majority of patients have done very well with reconstitution of T cells and varying degrees of humoral immune reconstitution. We encourage all sites to be sure to offer enrollment on CSIDE to every eligible patient.

If you have any questions, please email **Sung-Yun Pai, MD** (sung-yun.pai@nih.gov), **Mike Pulsipher** (mpulsipher@chla.usc.edu), and **Janelle Olson** (jolson@nmdp.org).



UCSF Artemis SCID Gene Therapy

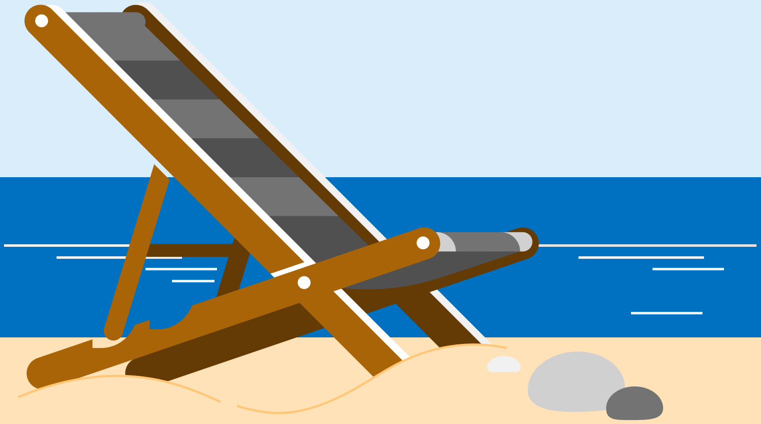
In this trial, newly diagnosed or previously treated patients with insufficient immunity due to ART-SCID receive “lentiviral gene transfer,” also called “gene therapy.” A normal copy of the DCLRE1C gene is inserted into blood-forming stem cells that grow and develop into all blood lineages. The inserted gene provides correct instructions to the defective stem cells so that functioning T and B lymphocytes can develop. So far 17 infants 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).

Ongoing Clinical Studies

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 visit contact **Donald Kohn** (DKohn1@mednet.ucla.edu) or visit https://clinicaltrials.gov/study/NCT03812263



Clinical Trial at Mayo for LADII Deficiency

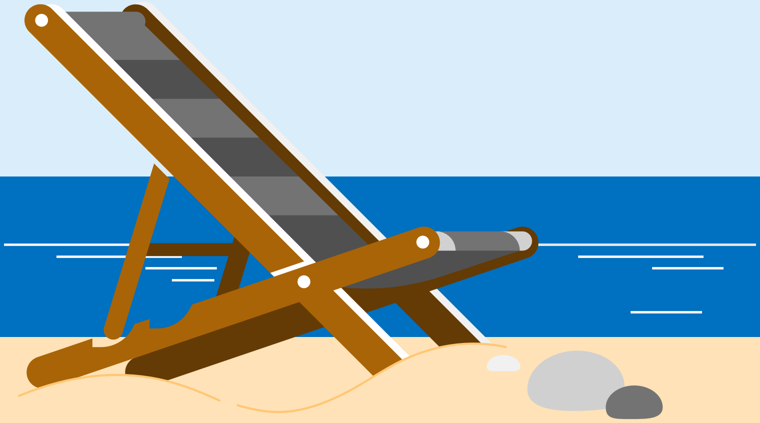
A Phase 3 Randomized, Double-blind Crossover Study to Assess the Efficacy and Safety of AVTX-803 in Subjects with Leukocyte Adhesion Deficiency Type II (LAD II; also called SLC35C1-CDG) enrolled the first patient at Clinical Genomics, Mayo Clinic Rochester.

The study enrolls LAD II patients older than 6 months receiving dietary supplements containing L-Fucose and randomizes patients into a two-period study with a withdrawal phase (placebo) and treatment phase (L-Fucose) with crossover. Patients with abnormal sialyl-Lewis antigen and a history of recurrent infections are eligible for the study. Please see the link for more information: https://clinicaltrials.gov/study/NCT05462587

Ongoing Clinical Studies

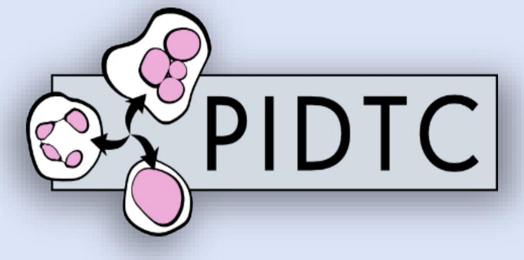
A Study of Mavorixafor in Participants With Severe Congenital Neutropenia and Chronic Neutropenia Disorders

This Phase 1b study will determine the safety and tolerability of mavorixafor in participants with severe chronic idiopathic neutropenia (CIN) and selected congenital neutropenia disorders. For more information, please visit https://www.x4pharma.com/patients/chronic-neutropenia/



Viral CTL Consortium (VIRCTLC)

Principal Investigator **Mitchell S. Cairo, MD** and Study Chair **Julie Talano, 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. For more information, please visit https://viraltcelltherapy.org/



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

Got announcements?
Email: rafael.ricon@ucsf.edu