Collaborative Studies with the Clinic for Special Children

Heather Madara
Regulatory and Outreach Manager
Roy S. Small, MD
Medical Director of Clinical Research
Penn Medicine Lancaster General Health Research Institute


Small


Madara

Editor's note: This is the 13th in a series of articles from the Penn Medicine Lancaster General Health Research Institute that describes ongoing research studies. Other active studies have been described in previous issues of this journal.

Located in the heart of Lancaster County, the Clinic for Special Children (CSC) is a nonprofit medical clinic for children and adults with genetic disorders and other complex medical needs, serving primarily the Amish and Mennonite (Plain) communities. CSC dedicates its clinical research efforts to the genetic diseases that impact these communities; often it is the case that these disorders occur at a much higher rate among the Plain than they do in the general population due to isolation and shared genetic heritage.

While many in these communities are less likely to seek medical treatment from outside the community, the relationship CSC has built with them over the years has allowed the clinic to make a tangible impact in the health of patients and their families.

CSC started in 1989 in a single building built by the Plain communities it still serves today. Since then, an expansion and updates have allowed for the growth of the team and for medical advancements. The building includes a CLIA-certified clinical laboratory, patient exam rooms, staff offices, and community spaces.

These resources enable the CSC's four physicians and one nurse practitioner to provide services that community members would otherwise need to travel much greater distances and incur much higher costs to receive. As a result of their proximity to the patients and their indefatigable efforts, CSC has been able to "pioneer innovative treatments and gain insights that are broadly applicable to genomic medical practice as a whole."

Kevin Strauss, MD, performs the roles of both medical director for CSC and practicing pediatric physician. Under his direction, the clinic has developed many collaborations across Lancaster County and beyond, Lancaster General Hospital among them. His research accomplishments include coauthoring more than 80 peer-reviewed journal articles and serving as the principal investigator for more than 15 studies.

LGH partnered with CSC in the past for studies investigating community health and understanding the specific conditions affecting the communities it serves. These studies have examined familial hypercholesterolemia due to Apolipoprotein B-100 mutations in the Amish, novel gene-modifying therapies for spinal muscular atrophy in Mennonites, and community attitudes toward certain medical procedures such as medical photography.

Dr. Strauss notes that, "given the fruitful nature of this important collaboration between two community-centered medical facilities in Lancaster County, we anticipate and hope for future innovative research opportunities that will improve the lives of the patients we both serve."

The LG Health Research Institute is excited to announce the approval of two new studies through collaborative efforts by LGH, the University of Pennsylvania, and CSC. Anyone interested in these studies (outlined below) or the crucial work being done by CSC can reach out to the Research Institute at 717-544-1777 for more information.

Safety and Efficacy of HMI-103, a Gene Editing Development Candidate in Adults with Classical PKU Due to PAH Deficiency

Sponsor: Homology Medicines, Inc.
Principal Investigator: Kevin Strauss, MD

This Phase 1 trial is evaluating the safety and efficacy of a single intravenous administration of HMI-103, a gene editing development candidate, in participants aged 18 to 55 years with classical Phenylketonuria (PKU) due to Phenylalanine hydroxylase (PAH) deficiency. These patients must have been following a low phenylalanine diet and yet still exhibit uncontrolled disease.

The study will implement sequential ascending dose-escalation, investigating up to three dose levels of HMI-103 in the cohort. A long-term extension study lasting 13 years is planned as well.

The three dosing cohorts could include up to three participants each. LGH and CSC plan to enroll up to three participants in total.

A Randomized, Sham-controlled, Double-blind Study to Evaluate the Efficacy and Safety of Intrathecal OAV101 in Type 2 Spinal Muscular Atrophy (SMA) Patients Who Are ≥2 to <18 Years of Age, Treatment Naive, Sitting, and Never Ambulatory

Sponsor: Novartis
Principal Investigator: Kevin Strauss, MD

This Phase 3 trial will enroll treatment naive Type 2 SMA participants ages 2 through 17. The study team will screen potential participants during a rigorous screening period (lasting 45-60 days) that involves data collection and clinical assessments.

If patients meet the eligibility criteria, they enter Treatment Period 1. This period involves randomization to either study drug (intrathecal OAV101) or sham (placebo) treatment during inpatient hospitalization and monitoring for up to three days. The next phase, Follow-Up Period 1, involves a 52-week outpatient follow-up schedule with regular safety and efficacy assessments.

A final period, Treatment Period 2, will be offered to eligible participants after completing the follow-up period in which they cross over the treatment randomization from Treatment Period 1. (Those who receive OAV101 will receive placebo, and those who received placebo will receive OAV101.) They will then enter the long-term extension study, which is being planned now.

The sponsor plans to enroll 125 participants. LGH plans to enroll up to three participants.

A complete list of active clinical studies at Lancaster General Health is available online. To access the most current list, scan the QR code at left, or find the link on the JLGH.org Resources/Links page. To make a referral to any study on the list, call the LG Health Research Institute at 717-544-1777.

Heather Madara
Penn Medicine LG Health Research Institute
131 E. Frederick St.
Lancaster, PA 17602
717-544-1777
Heather.Madara@pennmedicine.upenn.edu

Roy S. Small, MD
The Heart Group of Lancaster General Health
217 Harrisburg Ave.
Lancaster, PA 17603
717-544-8300
Roy.Small@pennmedicine.upenn.edu

Update from the Lancaster Medical Heritage Museum

Meagan Schulman, a recent Millersville University graduate and winner of the 2022 Penn Medicine Lancaster General Health Summer Internship, spent her summer researching the Lancaster County Vaccine Farm, also known as the Marietta Vaccine Farm. She discussed the project during the Lancaster Medical Heritage Museum's September "Lunch and Learn." (Scan the QR code below to view Schulman's full lecture, which will be published as a paper in the future.)

To briefly overview: In 1882 Dr. H.M. Alexander founded the Lancaster County Vaccine Farm, where he produced and supplied the entire country with smallpox vaccine. The Vaccine Farm drastically shaped not only Pennsylvania's ability to eradicate smallpox, but also that of the nation. It was beyond its years in biological products regulation and vaccine standards. What started out as one man and a calf had lasting effects on millions of people across the globe.

Smallpox was considered a "dread" disease: even if a person was spared from death, they could be scarred for the rest of their lives. Until the rise of inoculation and eventually vaccination, there was no means possible for the prevention of such a fate. Even after Dr. Edward Jenner's vaccine discoveries, it took doctors nearly another century to discover the most safe and effective way to mass produce a regulated, sanitary vaccine.

Dr. Alexander and the Lancaster County Vaccine Farm may have been lost to history, but society is still discovering the strides made in Marietta which have become a cornerstone in learning about mass vaccine production and regulation.