THE VOCKLEY LAB AT UPMC CHILDREN'S

An Update on Your Philanthropy

Spring 2026

With your support, Dr. Vockley strives to improve outcomes for children living with rare metabolic disorders, like Maddie.

UPMC CHILDREN'S HOSPITAL FOUNDATION

THIS MOMENT
PUT A CHILD'S FUTURE FIRST

THE VOCKLEY LAB AT UPMC CHILDREN'S

A Visionary Leader

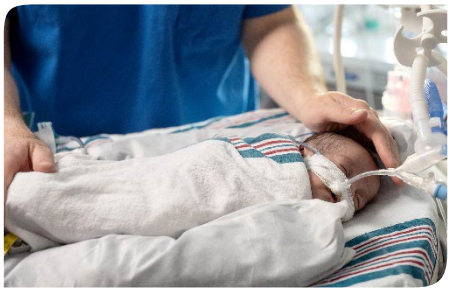
With your philanthropic support, Jerry Vockley, MD, PhD, is advancing care, education, and discovery for children and families affected by rare diseases. A global leader in inborn errors of metabolism, Dr. Vockley has devoted his career to improving diagnosis and treatment for rare genetic conditions, including fatty acid oxidation disorders (FAODs). **His work includes the discovery of previously unrecognized disorders. He has published more than 400 scholarly articles and is the founding director of the Center for Rare Disease Therapy at UPMC Children's Hospital of Pittsburgh.** As the Cleveland Family Endowed Chair, he co-founded and co-chairs INFORM, an international network shaping care standards for FAODs. He additionally serves on several high-profile advisory boards.

Earlier this year, Dr. Vockley was elected to the Association of American Physicians (AAP). Election to the AAP is an honor reserved for esteemed physician-scientists whose careers reflect sustained, nationally recognized contributions to medical research and the advancement of patient care. This is a tremendous honor, and we celebrate Dr. Vockley's achievement and the many donors who support his significant work!

Your generosity advances Dr. Vockley's leadership as a compassionate physician, a dedicated educator, and a visionary researcher.

Advancing Rare Disease Care

Fast PACE Dx: Improving NICU Diagnoses



As previously reported, in 2025, Dr. Vockley partnered with colleagues across UPMC Children's to implement a new precision medicine initiative for infants in the Neonatal Intensive Care Unit (NICU) and Cardiac Intensive Care Unit (CICU). More than 30% of unexplained illnesses in NICU patients are linked to underlying genetic disorders. Research led by Dr. Vockley and others has demonstrated that

One Children's Hospital Drive | 4401 Penn Avenue | Pittsburgh, PA 15224

1-877-247-4483 | gifts@chp.edu | givetochildrens.org

broad-based genetic testing can significantly improve care for these vulnerable children. Earlier diagnosis enables more targeted treatment, reduces unnecessary testing and procedures, and results in substantial health care savings. Importantly, timely genetic results provide clarity and direction for families facing uncertainty.

For the past year, the Fast Precision Acute Care Expert Diagnosis (Fast PACE Dx) Program has offered whole genome sequencing to every NICU and CICU baby admitted to UPMC Children's without a definitive diagnosis. This advanced testing can analyze a newborn's DNA and return results in several days, accelerating diagnosis and guiding critical early-care decisions.

While the Fast PACE Dx Program has only been operational for about a year, the team has already uncovered exceptional results. In an article published in *Genetics in Medicine* in March, Dr. Vockley and his collaborators describe early outcomes. Between April and October 2025, the Fast PACE Dx team evaluated the impact of early whole genome sequencing in the NICU and CICU at UPMC Children's. **During this six-month period, 61 critically ill infants with unexplained medical conditions were evaluated through the program, and 45 babies ultimately received rapid genome sequencing.** Genetics teams were consulted early, typically within the first two days of admission, allowing testing to begin while babies were still receiving intensive care.

The results underscore the power of early genetic testing. **More than half of the babies tested had genetic findings that helped explain their illness, and for nearly 40% of patients, the results directly informed or changed medical care.** In many cases, sequencing led to faster diagnoses, more targeted treatment plans, and the avoidance of unnecessary tests or procedures. Even when test results did not arrive before the end of a child's life, families received valuable information that could guide future care decisions and genetic testing for relatives.

One Children's Hospital Drive | 4401 Penn Avenue | Pittsburgh, PA 15224

1-877-247-4483 | gifts@chp.edu | givetochildrens.org

These promising early findings demonstrate that the Fast PACE Dx Program can improve clinical decision-making and reduce overall hospital costs for some of the most medically complex newborns. Dr. Vockley and his colleagues are now looking to expand Fast PACE Dx across additional UPMC neonatal and cardiac intensive care units, with the long-term goal of establishing early genetic diagnosis as standard of care for critically ill infants.

Reducing Uncertainty in Newborn Screening Results

A related project that has proven highly successful is Dr. Vockley's work developing a clinical test to clarify genetic findings for babies identified through newborn screening (NBS) with possible very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency. Beginning in 2022, Dr. Vockley partnered with Meena Sethuraman, MD, then a medical student at the University of Pittsburgh, to address one of the most persistent challenges in newborn screening: how to interpret genetic changes known as variants of uncertain significance (VUS), which can delay diagnosis and treatment for infants and leave families without clear answers.

VLCAD deficiency is a rare metabolic condition that prevents the body from properly converting certain fats into energy, particularly during fasting or illness. **Although newborn screening can identify babies at risk, follow-up DNA sequencing often reveals genetic variants with unclear clinical impact.** Until recently, confirming whether a VUS was disease-causing required invasive skin biopsies and lengthy laboratory testing that could take weeks to complete, and were not always definitive.

With philanthropic funding, Dr. Vockley and Dr. Sethuraman developed a faster, less invasive laboratory assay that allows clinicians and researchers to test whether specific genetic variants disrupt enzyme function. Using engineered cell models rather than patient biopsies, this approach can distinguish harmful variants from benign ones more quickly and reliably. What began as a medical student research project has resulted in a usable clinical assay that will help care teams reach confident diagnoses sooner and tailor treatment plans for infants identified through NBS.

Since completing this work, Dr. Sethuraman has continued her medical training and is now an ObGYN resident at UPMC Magee-Womens Hospital.

One Children's Hospital Drive | 4401 Penn Avenue | Pittsburgh, PA 15224

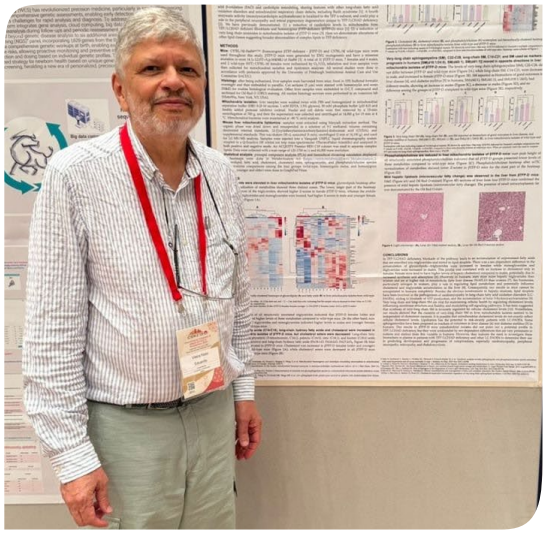
1-877-247-4483 | gifts@chp.edu | givetochildrens.org

Researcher Spotlight: Dr. Eduardo Vieira Neto

Eduardo Vieira Neto, MD, PhD, is a physician-scientist in the Vockley Lab whose work focuses on improving diagnosis and treatment for children and adults living with rare metabolic and mitochondrial disorders. **With more than two decades of experience spanning clinical genetics, newborn screening, laboratory science, and health policy, Dr. Vieira Neto brings a uniquely comprehensive perspective to research that bridges patient care and therapeutic development.** Thanks to philanthropic funding, his work examines how rare diseases affect metabolism at a cellular level. He is particularly interested in translating these insights into meaningful advances for patients and families.

Dr. Vieira Neto's research journey began in Brazil, where he trained in medical genetics and clinical pathology. He completed graduate studies focused on phenylketonuria (PKU), a rare inherited metabolic condition. His early career included leadership roles in newborn screening laboratories and genetic clinics, where he helped expand screening programs and improve early identification of inherited disorders. **Over time, his work increasingly centered on translational research, combining clinical data, molecular genetics, and laboratory models to better understand disease mechanisms and treatment response.**

In 2020, Dr. Vieira Neto joined the Vockley Lab, where he developed a research program examining the bioenergetic and lipid abnormalities underlying mitochondrial trifunctional protein and long-chain 3-hydroxyacyl-CoA dehydrogenase deficiencies.



Appointed research assistant professor in Pediatrics in 2025, he now leads studies aimed at identifying disease mechanisms and testing new therapeutic strategies, including approaches to improve mitochondrial function. **This important work would not be possible without the tremendous philanthropic support of generous patient families, whose commitment accelerates discoveries that advance care for individuals affected by rare metabolic diseases.**

We remain deeply grateful to our many donors for supporting Dr. Vockley and his remarkably talented team!

One Children's Hospital Drive | 4401 Penn Avenue | Pittsburgh, PA 15224

1-877-247-4483 | gifts@chp.edu | givetochildrens.org

Maddie's Spark

This is Maddie's story, as told by her mom, Ashley. The family started Maddie's Spark Foundation to raise awareness and funds for VLCAD and related long chain fatty acid oxidation disorders.

When our daughter Maddie was born, we stepped into parenthood with the joy and excitement every first-time parent dreams of, but within just one day, that joy turned to fear when she suffered her first metabolic crisis.

Through the newborn screening program, we learned she had VLCAD deficiency, a rare genetic metabolic condition that affects how her body processes certain fats for energy.

The past four years have been filled with uncertainty, countless questions, and fear as Maddie endured 17 hospitalizations, including spending her 4th birthday weekend in the hospital, most often due to painful rhabdomyolysis.

By her 14th hospitalization in November 2024, we knew we needed more specialized care, which led us to Dr. Jerry Vockley and his incredible team at UPMC Children's Hospital of Pittsburgh.

For the first time, we felt a sense of hope and clarity about Maddie's condition, and in the spring of 2025, we made the life-changing decision to move to Pittsburgh so she could be under Dr. Vockley's full-time care.

His team's knowledge, compassion, and experience have brought us renewed strength and peace, knowing we are not walking this journey alone.

Since coming to Pittsburgh, the support we've received from Dr. Vockley and his extraordinary team has exceeded every expectation. Their deep understanding of Maddie's more severe form of VLCAD deficiency has not only guided us in managing her condition but has also given us confidence and peace in the face of uncertainty. They've shown us compassion at every step, and we finally feel like we are not alone in this journey.

With their care, Maddie is getting the best chance at a brighter, fuller life, and as her parents, that is the greatest gift we could ever ask for.

One Children's Hospital Drive | 4401 Penn Avenue | Pittsburgh, PA 15224

1-877-247-4483 | gifts@chp.edu | givetochildrens.org

Our Gratitude

Thank you for your continued support of Dr. Vockley's work here at UPMC Children's. Your generosity strengthens both patient care and scientific discovery that improve outcomes for children like Maddie and many others. We are grateful for your commitment to the Vockley Lab and for the tremendous impact of your partnership.

Dr. Vockley received the prestigious NORD Rare Impact Medical & Scientific Trailblazer Award in 2025.

UPMC CHILDREN'S HOSPITAL FOUNDATION | THIS MOMENT PUT A CHILD'S FUTURE FIRST

One Children's Hospital Drive | 4401 Penn Avenue | Pittsburgh, PA 15224

1-877-247-4483 | gifts@chp.edu | givetochildrens.org