

UREA CYCLE DISORDERS ECHO *The Essentials*

Urea Cycle Disorders ECHO, The Essentials: Educating front-line care providers, expanding the expert network

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Background

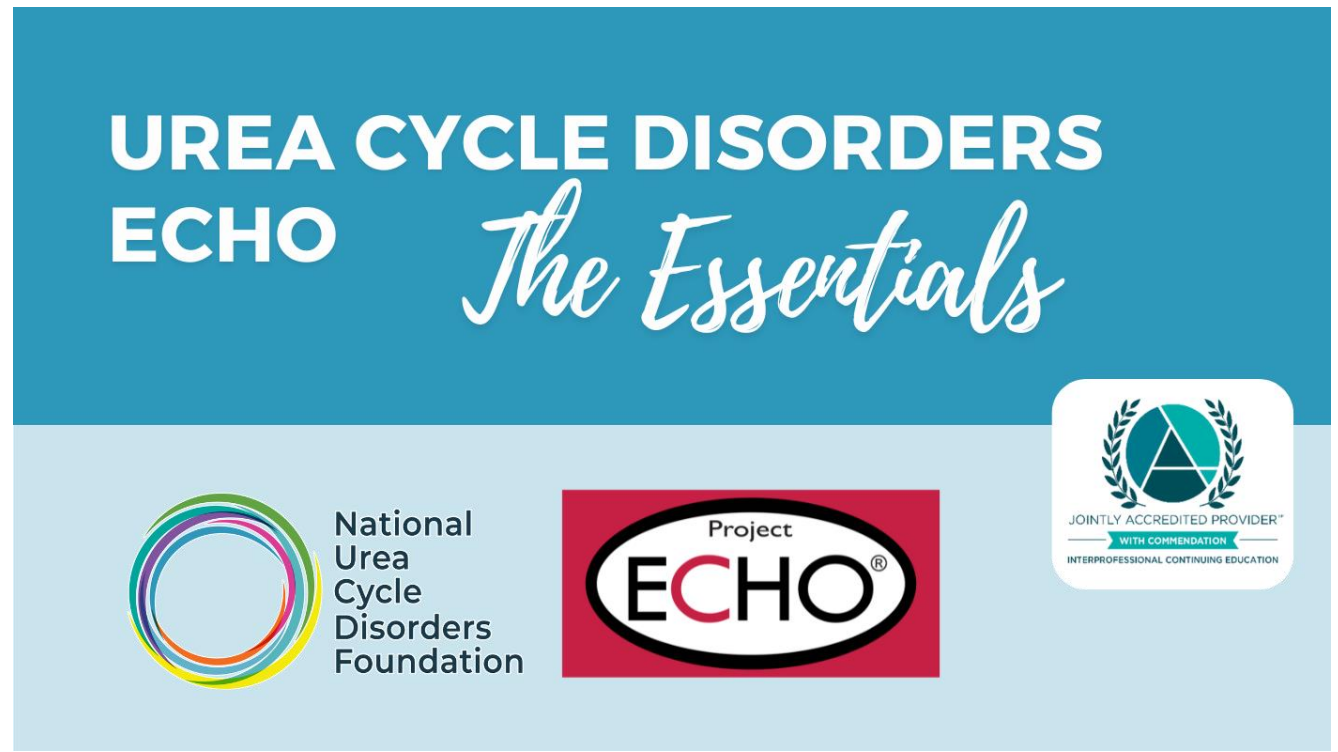
The National Urea Cycle Disorders Foundation (NUCDF) is a nonprofit patient group that advocates for people affected by urea cycle disorders (UCDs). Since 1988, NUCDF has been working to advance research, improve care, and raise awareness that saves lives.

Newly diagnosed patients frequently reach out to NUCDF for assistance in accessing care or in medical emergencies where local clinicians lack expert knowledge. Expert UCD care centers based in the U.S. are concentrated on the East and West coasts, leaving large geographic areas without access to needed expertise. Many children and adults with urea cycle disorders still go undiagnosed or misdiagnosed, with tragic consequences, including brain damage, coma, and death.

To address these gaps, NUCDF launched a virtual clinician education program in 2025 in partnership with Project ECHO® and the Urea Cycle Disorder Consortium (UCDC),

The UCDC, NUCDF's research partner, is a network of researchers and clinicians across 17 academic institutions in the U.S., Canada, and Europe. It was funded by the NIH's Rare Diseases Clinical Research Network from 2003-2025.

Project ECHO® (Extension for Community Healthcare Outcomes) is an innovative tele-mentoring program based at the University of New Mexico. The ECHO program creates virtual communities of learners by bringing together healthcare providers and subject matter experts using videoconference technology, brief lecture presentations, and case-based learning, fostering an "all learn, all teach" approach and democratization of knowledge.



Objectives

With this program, NUCDF aims to share essential information with frontline caregivers who may not encounter these disorders often, including emergency room physicians, nurse practitioners, pediatricians, and neurologists as well as medical students. It further aims create a community of practice, connecting UCD experts with frontline care providers and other experts in order to improve care for all patients.

To care for UCD patients, clinicians need to recognize the signs of high ammonia; know the diagnostic tests for UCDs; and learn the essentials in caring for UCD patients, including how to access patient care guidelines and ask advice.

Clinicians who do not routinely encounter urea cycle disorder patients need practice applying the principles of recognizing, diagnosing, and treating these rare disorders.

Following this series, front-line healthcare providers will be confident in applying the essentials of recognizing, diagnosing, and treating urea cycle disorders; understand when expert advice is needed; and be willing to share knowledge within their community.

Methods

To identify model programs for clinician education, NUCDF leadership tapped into its robust network of rare disease patient groups, which routinely share best practices. The Osteogenesis Imperfecta Foundation hosts two ECHO series for its rare disease community that became models for NUCDF's program. The Ehlers-Danlos Society similarly runs several robust series of ECHO programs and served as a second model for NUCDF during the early planning period.

Establishing the Program

NUCDF staff attended Project ECHO® Partner Launch Training in August 2024. This program is conducted by the ECHO Institute team and by global experts from the ECHO network. Through this training, NUCDF gained access to the iECHO platform, a web-based, proprietary teleECHO program management software and database developed and managed by the ECHO Institute. NUCDF was recognized as an ECHO Hub organization in late 2024.

NUCDF also partnered with the Project ECHO® accreditation office to offer continuing medical education (CME) credits for the series.

Program Planning

Three UCDC physician experts (Dr. Lindsay Burrage, Baylor; Dr. Sandesh Nagamani, Baylor; Dr. Andrea Gropman, St. Jude) volunteered to serve on NUCDF's ECHO planning committee. They established the following educational objectives for the series:

- Participants will understand the metabolic pathways of urea cycle disorders
- Participants will recognize the symptoms of high ammonia, understand the appropriate tests to diagnose urea cycle disorders, and be confident in interpreting results
- Participants will understand the basics of treating urea cycle disorders, including emergency and long-term management.

Additional UCDC experts were recruited as presenters. Four, 90-minute sessions were planned for the 2025 series (see schedule, below).

Recruitment

To recruit participants, NUCDF advertised the program across its communication channels (newsletter, blog site, social media channels); ran paid advertisements on LinkedIn and Facebook; engaged its patient community to invite their providers to attend; and forwarded recruitment materials to the UCDC expert network, who also assisted in recruiting participants.

Too often, patients die or suffer permanent injury because of a missed diagnosis or improper care. Our goal is to ensure that every front-line healthcare provider understands the essentials of recognizing, diagnosing, and treating urea cycle disorders.

*—Tresa Warner,
NUCDF Executive Director*



In support of improving patient care, this activity has been planned and implemented by the National Urea Cycle Disorders Foundation and Project ECHO. Project ECHO® is jointly accredited by the Accreditation Council for Continuing Medical Education (ACCME), the Accreditation Council for Pharmacy Education (ACPE), and the American Nurses Credentialing Center (ANCC), to provide continuing education for the healthcare team.

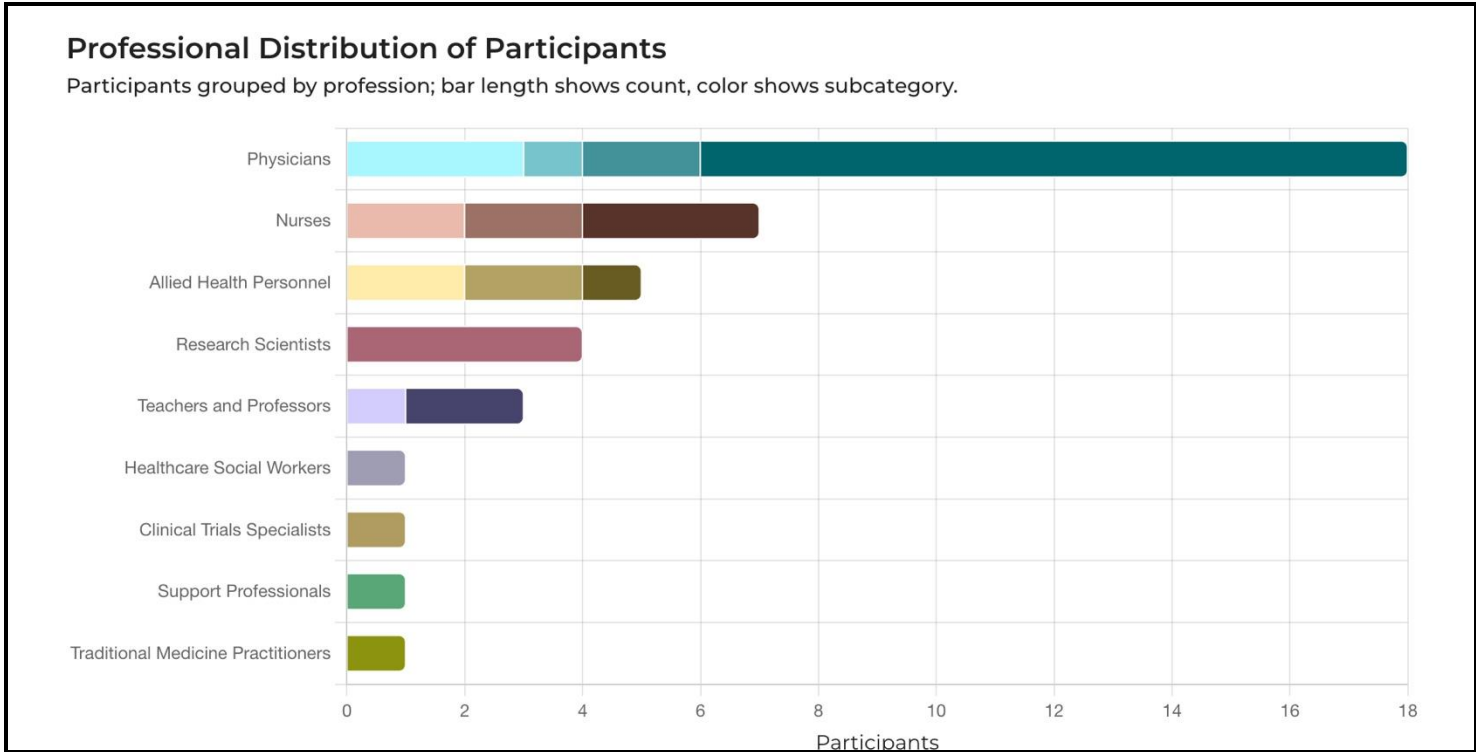
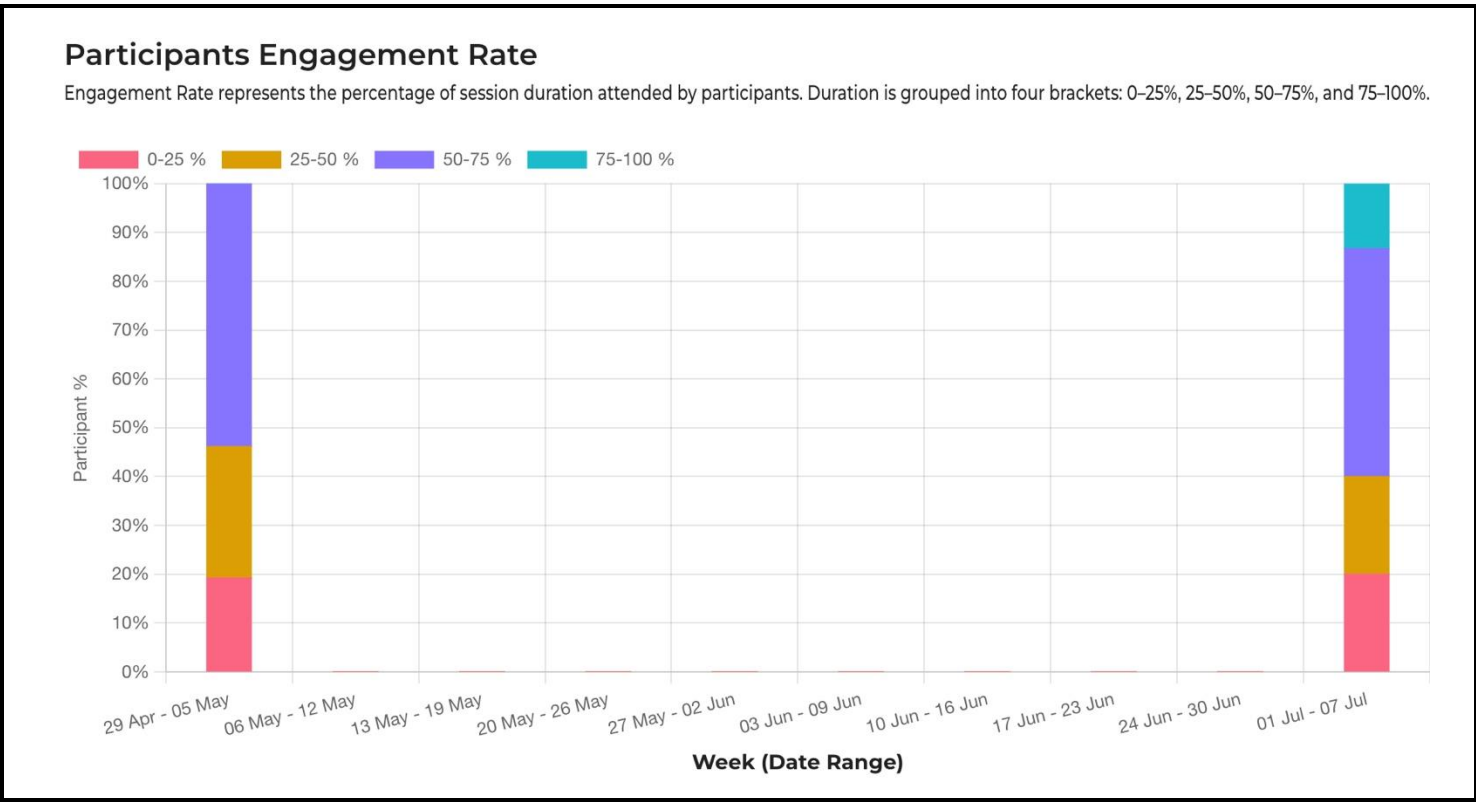
AMA Designation Statement
Project ECHO® designates this live activity for a maximum of 1.5 *AMA PRA Category 1 Credits™*. Physicians should claim only the credit commensurate with the extent of their participation in the activity.

Discussion

To date, NUCDF has hosted two ECHO sessions. A total of 96 participants representing 11 countries have registered for the series and 41 have attended. Recordings and slides from both sessions are now available at nucdf.org/echo.

The graphs below illustrate the engagement rate and professional distribution of participants. Post-session evaluations for both sessions showed an increase in attendee topic knowledge. On a scale of 1 (not knowledgeable at all) to 5 (extremely knowledgeable), mean responses for Session 1 showed a knowledge increase from 3.3 to 4; Session 2 responses showed an increase from 3.2 to 3.9. Almost all attendees said they plan to use what they learned from the sessions in their work.

Two more sessions are scheduled in 2025. Planning for 2026 sessions will begin later this fall.



Learn More and Register



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