



Evaluation of Undiagnosed and Rare Conditions Course - June 12, 2025

Course Recording: [View Recording](#)

Overview

The goal of this training program was to teach methods of undiagnosed patient evaluation to students, clinicians and researchers so that they can apply them at their own institutions. After the course, they may refer particularly challenging cases to the Undiagnosed Diseases Network (UDN) but ideally will be able to diagnose and care for many patients with rare diseases locally.

The training was led by investigators from the UDN clinical sites and cores. Trainings focused on current methods, lessons learned, challenges faced and near-term advances that are on the horizon for patients with undiagnosed conditions.

The course was presented in a live online format with a combination of educational slides and other materials. The training focused on different aspects of the UDN methodology, including the clinical evaluation, challenges in genomics care, implicit bias considerations, the patient journey and foundation support, and sequencing analysis and functional study approaches.

Target Audience

- Primary Care Physicians
- Specialty Physicians

Learning Objectives

Upon completion of this activity, participants were able to:

- Determine appropriate methods for clinical evaluation and phenotyping of patients with undiagnosed conditions.
- Apply or refer patients to specialists for advanced analysis of sequencing data.
- Identify strategies for incorporating basic research into the diagnostic process or introducing appropriate research for patients.
- Recognize how patient engagement can be leveraged to improve rare disease diagnosis.

Course Contents

Welcome and Course Introduction	Emily Glanton, MS, CGC
Module 1: The Clinical Evaluation	Vandana Shashi, MD Camilo Toro, MD
Module 2: Advances and Challenges in Genomics Care	Rachel Mahoney, MS, CGC
Module 3: The Patient Journey and Foundation Support	Troy Evans Michele Herndon, DNP, RN
Module 4: Sequencing Analysis and Functional Study Approaches	Shilpa Kobren, PhD Michael Wangler, MD
Closing Remarks	

Faculty

Planners

[Emily Glanton, MS, CGC](#)

[Matthew Wheeler, MD, PhD](#)

[Jon Bernstein, MD, PhD](#)

Speakers

[Rachel Mahoney, MS, CGC](#)

[Shilpa Kobren, PhD](#)

[Michael Wangler, MD](#)

[Michele Herndon, DNP, RN](#)

[Troy Evans, patient and advocate](#)

[Camilo Toro, MD](#)

[Vandana Shashi, MD](#)