

Human Clinical Phenotyping Core

The Human Clinical Phenotyping (HCP) Core provides investigators at Albert Einstein College of Medicine and Montefiore with comprehensive recruitment, clinical phenotyping, and participant management services to support research in intellectual and developmental disabilities (IDD). As a Scientific Core of the NIH-funded Rose F. Kennedy Intellectual and Developmental Disabilities Research Center (IDDRC), the HCP helps investigators efficiently recruit, characterize, and retain research participants while ensuring high-quality, standardized phenotyping across studies. The Core serves investigators studying individuals with neurodevelopmental disorders including autism and related disorders and rare genetic neurodevelopmental disorders across the lifespan.

Core Services

Participant Recruitment

The HCP provides comprehensive recruitment support, including:

- Access to a well-curated participant registry of more than 2,000 individuals and families
- Recruitment through community outreach and engagement activities
- Recruitment through Einstein and Montefiore clinical programs, including the Children's Evaluation and Rehabilitation Center (CERC) and Children's Hospital at Montefiore (CHAM)
- Study advertising and participant communications
- Participant scheduling and retention support

Clinical Phenotyping

The Core provides standardized clinical and cognitive assessments for children and adults across a broad range of developmental and cognitive abilities, including individuals who are minimally verbal or have significant intellectual disabilities. Services include:

- Diagnostic evaluations
- Cognitive assessments
- Adaptive behavior assessment
- Behavioral and developmental characterization
- Research-specific clinical phenotyping
- Consultation on selection of clinical outcome measures

Participant Registry

The HCP maintains a secure longitudinal research database containing demographic, clinical, cognitive, and research participation information, including available neuroimaging, electrophysiological, and genetic data when applicable. This resource enables investigators to:

- Identify eligible participants
- Reduce redundant testing
- Facilitate longitudinal follow-up
- Support cross-project collaboration
- Improve study efficiency while minimizing participant burden

De-identified participant information is available to approved investigators to facilitate study planning and recruitment.

Community Engagement

The HCP maintains strong partnerships with local families, clinicians, advocacy organizations, and community groups. Through educational events, outreach activities, and ongoing

communication, the Core promotes participation in research while helping ensure that research remains responsive to the needs of individuals with IDD and their families.

Who We Serve

The HCP supports investigators conducting research on:

- Autism spectrum disorder
- Genetic and rare neurodevelopmental disorders
- Intellectual disability
- Language and communication disorders
- Neurodevelopment across the lifespan

The Core welcomes collaborations with investigators across Einstein/Montefiore and assists both established investigators and new users in incorporating rigorous human phenotyping into their research programs.

Leadership

The Human Clinical Phenotyping Core is led by Director **Dr. Sophie Molholm** with Associate Directors **Dr. Melissa Wasserstein** and **Dr. Erica Weiss**. The multidisciplinary Core team includes experts in clinical genetics, neuropsychology, developmental assessment, behavior analysis, participant recruitment, community engagement, and research coordination. Together, the team provides investigators with the expertise and infrastructure needed to conduct rigorous, participant-centered clinical research.