

Medical Journal Publishes HemoShear’s Pilot Study of Neuropsychological Assessments for MMA and PA

Molecular Genetics and Metabolism Reports has published a study sponsored by HemoShear evaluating how different assessments can be used to measure verbal communication, processing speed, life skills, quality of life and sleep in people with MMA and PA. The study, titled [“Neuropsychological Endpoints for Clinical Trials in Methylmalonic Acidemia and Propionic Acidemia: A Pilot Study”](#) was conducted in collaboration with Children’s National Rare Disease Institute.

The study enrolled 21 patients with MMA and PA who were remotely administered several tests measuring relatively discrete neuropsychological domains to inform the selection of clinical outcome assessments for clinical trials. Tests and questionnaires were selected for their possible relevance to MMA and PA and potential sensitivity to modest changes in functioning and behavior.

“Many children and adults with MMA and PA have difficulties learning, communicating and participating in activities of daily living,” says Pat Horn, MD, PhD, chief medical officer at HemoShear. “As potential new treatments like HST5040 are evaluated in clinical studies, we need effective ways to measure changes in these areas. This study provides insights into the types of assessments that have the sensitivity and range to detect impact, potentially within the short-term timelines of clinical trials.”

“Language and motor deficits in some participants impacted scoring, especially in the areas of verbal performance and processing speed. Overall, test and questionnaire results differed between participants with MMA and PA, suggesting that neuropsychological assessments should be studied independently in these diseases,” said Susan Waisbren, PhD, professor of pediatrics at Harvard Medical School and world-wide expert in neuropsychological evaluations in children with metabolic disorders. “In addition, patients and caregivers gave high scores on the metabolic quality of life test, indicating that families had adapted to their challenges.”



Author Susan Waisbren, PhD,
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Dr. Waisbren continued, “This research demonstrates the value of conducting a pilot observational study to select appropriate neuropsychological instruments before initiating an interventional clinical trial. It also provides a rationale for focusing on discrete neuropsychological domains rather than global measures of functioning, which tend to be undifferentiated in their results and may fail to uncover important changes in one domain while other functions remain unchanged. Finally, this pilot study reinforces the need to carefully define the sample cohort to ensure that differences in diagnoses and baseline functioning are considered.”

The study was conducted by Dr. Waisbren and Kimberly Chapman, MD, PhD, medical geneticist at Children's National Rare Disease Institute.

The HemoShear clinical team will apply the insights generated from this study to the design of future clinical trials of HST5040 for MMA and PA.



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