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Research challenges in central nervous system manifestations of inborn errors of metabolism.

[Dickson PJ](#), [Pariser AR](#), [Groft SC](#), [Ishihara RW](#), [McNeil DE](#), [Tagle D](#), [Griebel DJ](#), [Kaler SG](#), [Mink JW](#), [Shapiro EG](#), [Bjoraker KJ](#), [Krivitzky L](#), [Provenzale JM](#), [Gropman A](#), [Orchard P](#), [Raymond G](#), [Cohen BH](#), [Steiner RD](#), [Goldkind SF](#), [Nelson RM](#), [Kakkis E](#), [Patterson MC](#).

Source

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Abstract

The Research Challenges in CNS Manifestations of Inborn Errors of Metabolism workshop was designed to address challenges in translating potential therapies for these rare disorders, and to highlight novel therapeutic strategies and innovative approaches to CNS delivery, assessment of effects and directions for the future in the treatment of these diseases. Therapies for the brain in inborn errors represent some of the greatest challenges to translational research due to the special properties of the brain, and of inborn errors themselves. This review covers the proceedings of this workshop as submitted by participants. Scientific, ethical and regulatory issues are discussed, along with ways to measure outcomes and the conduct of clinical trials. Participants included regulatory and funding agencies, clinicians, scientists, industry and advocacy groups.

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