Assessing Visual Processing with Gene Therapy in Cerebral Adrenoleukodystrophy

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Adrenoleukodystrophy is a genetic disorder and its most devastating form is cerebral adrenoleukodystrophy (cALD). This progressive disease manifests early in childhood and is potentially fatal. In the majority of these patients, the connections within the central visual system become compromised leading to a variety of visual perceptual deficits. These deficits are an important early marker for disease progression, but standard ophthalmic testing methods cannot detect and characterize these impairments effectively. Ex vivo lentiviral gene therapy has recently proven successful in restoring neuronal function, but intervention needs to be performed in the early stages of disease. The lack of appropriate functional visual assessments remains an obstacle in characterizing early disease and progression, as well as potentially saving critical neurological function and human lives. Given the importance and value of characterizing functional visual deficits in this disease, we have developed a highly innovative and ecologically valid virtual reality (VR) based testing platform that is well-suited for use with pediatric clinical populations. Combining this VR approach with enhanced structural and functional brain imaging techniques will allow for more comprehensive characterization of the neurophysiology associated with visual processing deficits, disease progression, as well as track the progress of therapy in patients with cALD. The results from this pilot study will serve as a basis to inform a large scale clinical trial assessing the therapeutic benefit of gene therapy in cALD.