Transient Visual-Evoked Potentials as a Novel Biomarker for Autism and Phelan-McDermid Syndrome

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1 in 68 individuals is diagnosed with an Autism Spectrum Disorder. Currently, autism diagnosis is based solely on behavioral assessment; no objective, medically-based biomarkers have been identified. A biomarker is a biological characteristic that can be accurately measured and indicates a specific medical state. An autism biomarker could help diagnose children who are difficult to evaluate because of language or cognitive impairment, and could also assist in diagnosing children earlier in life, even during infancy. This is critical because early intervention is highly effective in helping children with autism gain valuable skills. This study used an EEG-based technique called short-duration transient visual-evoked potential (tVEP) to examine excitatory and inhibitory signaling in the brains of typically-developing children (TD), children with idiopathic autism (iASD) and children with Phelan-McDermid Syndrome (PMS), a subtype of autism caused by dysfunction of the SHANK-3 gene. Differences in signaling patterns could be the basis of a biomarker. As hypothesized, children with idiopathic autism showed smaller amplitudes in tVEP waveforms than typically developing children. Children with PMS showed even smaller amplitudes than children with iASD. Additionally, children with a SHANK-3 deletion showed smaller amplitudes than children with a SHANK-3 point mutation.

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