Gaze Aversion Detection Software in Fragile X Syndrome

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Introduction: Fragile X Syndrome (FXS) is caused by a CGG trinucleotide repeat within the FMR1 gene leading to methylation of the gene and loss of synthesis of FMRP. Gaze aversion is a well-known hallmark of FXS and recent social eye tracking findings have shown reduced eye gaze and increased pupillary response when viewing emotional faces. However, there remains ambiguity as to the exact mechanisms underlying social gaze abnormalities with inattention, social disinterest, and social anxiety as potential explanations.

Hypothesis: Gaze aversion detection software developed utilizing eye tracking gaze data and OpenFace facial recognition will validly and specifically detect gaze aversions comparable to evaluation of gaze aversions by manual assessment of subject video and gaze recordings.

Methods: 36 individuals with FXS completed an emotional faces eye tracking paradigm and a battery of neuropsychological testing and caregiver-report measures. Gaze and OpenFace data were processed in SPSS and Matlab with a variety of criteria for gaze aversion detection.

Results:

1. The gaze aversion detection software implemented in this study was able to detect gaze aversions with a sensitivity of 83.9%, specificity of 94.6%, positive predictive value of 70.27%, and negative predictive value of 97.5%.
2. Gaze aversions were found to correlate with clinical measures of irritability, hyperactivity, and obsessive-compulsive behavior.

Conclusions: Gaze aversions were successfully detected using the gaze aversion detection software developed in this study. Contradictory to our hypothesis, gaze aversive behavior was not found to correlate with social anxiety but rather irritability, hyperactivity, and compulsivity. Few if any studies have directly correlated social eye tracking and clinical measures in FXS so it is possible gaze aversive behavior in FXS is less related specifically to social anxiety and more to neural hyperexcitability and overall FXS-related impairment.

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