Genetic associates of a visual endophenotype of autism and schizophrenia

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ABSTRACT

Evidence of abnormal contrast sensitivity in autism[1–3] and schizophrenia[3–4].
Past genetic work has used cognitive and neurophysiological endophenotypes[9–13].
Basic visual functions are ideal endophenotypes as mechanisms are well characterised.

1) BACKGROUND

- Sensitivity defined as inverse of 82% 2-AFC detection threshold contrast.
- Basic visual functions are ideal endophenotypes as mechanisms are well characterised.

2) BEHAVIOURAL METHODS AND RESULTS

- Sensitivity defined as inverse of 82% 2-AFC detection threshold contrast.

3) GENETIC METHODS

- Sensitivity analysed for association with genotype at 642 758 single-nucleotide polymorphism (SNP) markers distributed across the genome.

4) GENETIC RESULTS

- Significant association with SNP marker rs1797052 ($P = 7.9 \times 10^{-9}$).
- Permutations to account for multiple testing show significance at $\alpha < .005$.

5) DISCUSSION

- Each copy of minor allele associated with > 0.5 SD sensitivity increase.
- SNP situated in the 5’ untranslated region of the gene PDZK1.

REFERENCES & ACKNOWLEDGEMENTS