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Key words

Strabismus, esotropia, association

Application lay summary

1a: Strabismus is the pathological misalignment of the eyes. Childhood-onset strabismus affects up to 4% of the population. Congenital strabismus may be differentiated into comitant (CCS) and incomitant forms (ICS). CCS accounts for approximately 98% of all childhood-onset strabismus. Population, family-based, and twin studies support a genetic contribution to CCS. CCS is thought to be inherited as complex trait. To identify common variants providing susceptibility to CCS we have conducted a Genome Wide Association Study (GWAS) for congenital esotropia. We have replicated our results in a Caucasian cohort and we are now trying to identify another cohort for second replication.

1b: Our research aims to identify the genetic susceptibility underlying a common childhood-onset disease (congenital concomitant strabismus). Improved understanding of the genetic contributions to this disease will likely provide insight into its neuro-developmental basis and could have a profoundly positive impact on the health and well-being of the population.

1c: We will obtain genotypes of individuals from the UK Biobank who are affected with strabismus, and compare their genotypes to ethnically matched controls to identify if significant differences exist between cases and matched controls.

1d: A subset of the cohort will be included. We are interested in participants who reported a diagnosis of strabismus (including but not limited to s/p strabismus surgery,

wearing glasses for strabismus (data field 6205), etc,). We believe the cohort could be 4000-5000 participants.

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