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Pupil size and pupillary light reflex in early infancy: heritability and link to neurodevelopmental and psychiatric genetic liability in the general population

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ABSTRACT:

The pupil regulates the amount of light reaching the retina and has basic visual sensory regulatory functions. Previous work links an atypical pupillary light reflex (pupil constriction to light) to neurodevelopmental and psychiatric conditions, which are highly heritable. However, there have been no twin studies of pupil responses in infancy or attempts to understand its genetic architecture. Here we estimated genetic and environmental influences on baseline pupil size and the pupillary light reflex in 510 infant twins assessed at 5 months (281 monozygotic and 229 dizygotic pairs), and the relationship with polygenic risk for neurodevelopmental and psychiatric disorders. Pupil size (h^2 = .64 CI: .54 to .72) and constriction response to light (h^2 = .62, CI: .51 to .70) were both highly heritable, and bivariate twin modelling indicated that the genetic factors showed modest overlap (rG = .38, CI: .23 to .52). When analyzing genome-wide polygenic scores (GPSs), a statistically significant association between pupil size and the GPS for schizophrenia (β = .15, p = .024, ΔR^2 = .016) was found but there were no significant associations with the GPS for autism (p > .25). Further, there were no associations between constriction response and GPSs. This study suggests that individual differences in pupil size and the pupil light reflex are heritable and provides initial evidence that pupil size may share some links with schizophrenia genetic liability. It underscores the possibilities of combining established behavioral genetic methods with experimental psychophysiological developmental assessments to further our understanding of human brain development.

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