ABSTRACT

GENETIC INFLUENCES ON SOCIAL COGNITION, EXECUTIVE FUNCTION, AND ASSOCIATED NEURAL NETWORKS

By

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Fundamental cognitive domains include executive function and social cognition. Both social cognition and executive functioning can be studied using neuroimaging techniques that allow direct observations to be made about brain structure and function. These techniques can also be applied to the study of brain development, revealing how circuits involved in executive function and social cognition change during important developmental periods such as infancy. Along with providing a window into brain maturation, neuroimaging can be used to study cases where cognitive domains are disrupted and make comparisons to learn about typical brain development and function. For my dissertation, I have explored these cognitive domains and associated neural circuits both in typically developing individuals and in individuals with Turner syndrome, a condition caused by the full or partial absence of the second sex chromosome. First, I used a classic twin design and demonstrated relatively low narrow-sense heritability estimates for neonatal resting-state functional connectivity phenotypes. I studied both between- and within- network connectivity in neonates and demonstrated that only 6 out of 36 phenotypes had heritability estimates greater than 0.10; no estimates were statistically significant. These within- and between-network phenotypes included networks heavily recruited for social cognition and executive functioning. I also showed statistically significant associations between neonate resting-state functional connectivity phenotypes and specific demographic and medical history variables. Second, I compared
structural and functional connectivity between typically developing male and female infants and infants with Turner syndrome. I saw no differences between the three groups in integrity of the superior longitudinal fasciculus or reduced connectivity between the right precentral gyrus and brain regions in the occipital and parietal regions involved with social cognition, visuospatial reasoning, and executive function. Fronto-parietal connectivity and integrity of the superior longitudinal fasciculus are disrupted in older individuals with Turner syndrome and these results suggested that these changes emerge after the first year of life. I conducted a further exploratory analysis of 54 fiber tracts and showed significant group differences that primarily reflected masculinization of white matter microstructure in TS. Other differences may have arisen due to hemizygosity of the pseudoautosomal region. Finally, I developed a browser-based online testing platform targeting domains such as executive functioning and social cognition, which are often disrupted in Turner syndrome. I then validated the battery via administration to neurotypical males and females and to adult women with Turner syndrome, who performed more poorly on tests of executive function and visuospatial reasoning. Taken together, the results presented in this dissertation contribute greatly to our understanding of the role of genetics in social cognition, executive function, and their related neural networks. These results can be further utilized in longitudinal studies of brain development and in future cognitive testing research.