Disorders of chromatin regulation: bridging genes, brains and behaviour

Chromatinopathies are neurodevelopmental disorders arising from rare, pathogenic variants in regulators of DNA structure, impacting on gene transcription. Collectively, chromatinopathies are a relatively common cause of intellectual disabilities, autism spectrum disorders and other associated cognitive and behavioural characteristics. In our previous functional networks study of autism spectrum characteristics, we found that individuals with chromatinopathies had elevated symptoms of behavioural inflexibility. We have now repeated this analysis in a larger independent cohort, and found again that the chromatinopathy group have a distinctive dimensional profile involving social relating / communication and sensory / motor flexibility. This project will build on these findings by investigating the cognitive and neural systems characteristics of chromatinopathies. Methods may include cognitive testing (using our recently published remote, dynamic cognitive testing app) and neuroimaging (using MRI and MEG). We will apply advanced analysis methods such as generative modelling and HMM to investigate dynamic aspects of brain structure and function.

References and URL(s)

URL
https://doi.org/10.1080/09297049.2022.2054968

URL
https://doi.org/10.1093/cercor/bhx027