

Endosomal Proteins in Neurodevelopmental Disorders

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Rett Syndrome is a neurodevelopmental disorder that primarily affects females and is detected at 6-18 months of age. Rett Syndrome results from a mutations in the methyl-CpG binding protein 2 gene (*MECP2*) which is found on the X chromosome. Its mutation results in impairment in cognitive, sensory, emotional, motor and autonomic functions. Schizophrenia is also a neurodevelopmental disorder with onset in adulthood, however there is no single genetic cause. Endosomal proteins have been implicated in both disorders through GWAS studies and animal models, suggesting a common molecular mechanism shared between these neurodevelopmental disorders. Previous research in our lab has demonstrated a disruption in endosomal trafficking in animal models of both disorders. This study explores the levels and localization of endosomal proteins in coronal brain sections for mice models for these neurodevelopmental disorders. Using immunohistochemistry, we will examine protein levels in the hippocampus and cortex for endosomal trafficking markers. We will also use whole-brain derived synaptosomes and western blots to examine the subcellular levels of these endosomal markers. Our data demonstrate a reduction of endosomal proteins in the hippocampus of mouse models of neurodevelopmental disorders. Future studies will include investigations into the affected cargo being mis-trafficked in these disorders.