



Poster Presentation

Conserved Roles for CHD7 in Transcriptional Elongation of Genes Involved in Neural, Neural Crest, and Inner Ear Development

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Presenter Information

Emily Mathis is a doctoral student in School Psychology at Mississippi State University and a Board Certified Behavior Analyst (BCBA). Mathis holds a Master's degree in Educational Psychology from Mississippi State University and a Bachelor of Science degree in Psychology from Mississippi College. Mathis has been a member of the Bulldog CHARGE Syndrome Research Lab at Mississippi State University for three years. Mathis has worked on various CHARGE related research projects including sexuality education and the quality of behavioral services. In addition to her research in CHARGE, Mathis is a graduate assistant and clinical coordinator for the Autism and Developmental Disabilities Clinic at Mississippi State University where she provides behavioral services rooted in applied behavior analysis to individuals with disabilities.

Presentation Abstract

Central and peripheral nervous system impairment are common in CHARGE, yet the underlying mechanisms of CHD7 action in early neural and neural crest development are not well understood. *Chd7Gt/+* mice exhibit dysplastic semicircular canals and mild mixed conductive-sensorineural hearing loss and are an excellent model for CHARGE syndrome. Reduced CHD7 function in the ear disrupts expression of genes involved in developmental patterning and neurogenesis. We explored potential roles for CHD7 in global regulation of gene transcription in the nervous system, including neural crest cells and inner ear tissues. Transcriptome analysis via RNA-Seq on E10.5 otocysts from *Chd7+/+* and *Chd7Gt/+* mouse embryos showed a length-dependent effect of CHD7 on gene transcription, with preferential effects on longer (>100kb) genes. Notably, a similar but milder length-dependent effect was observed in heterozygous CHD7 mutation-positive human induced pluripotent stem cells (iPSCs) differentiated into neural crest cells (NCCs) when compared to patient derived iPSCs in which the variant was corrected via CRISPR/Cas9 gene editing. Collectively, our findings support roles for CHD7 in chromatin events that regulate transcriptional elongation.