Poster Presentation

Developing Zebrafish Models To Study the Link Between SoxC Transcription Factors and CHARGE Syndrome

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Presenter Information

Laura is a MD/PhD student at the University of Kentucky where she is currently conducting her graduate work. Her work focuses on the development of a zebrafish model to investigate the role that SoxC transcription factors play in CHARGE Syndrome and specifically the development of coloboma.

Presentation Abstract

The molecular mechanisms underlying the ocular birth defects observed in CHARGE patients are poorly understood. Our laboratory studies the development of the vertebrate visual system using zebrafish. Previous work from our lab has shown that knockdown of Sox11, a member of the SoxC family of transcription factors, in zebrafish results in microphthalmia, coloboma, brain, trunk, and heart defects, all phenotypes observed in CHARGE syndrome. Furthermore, a duplication of Sox11 has been identified in a patient clinically diagnosed with CHARGE syndrome, and CHD7 has been shown to directly interact with Sox11 and Sox4 in neural stem cells. Taken together, these data strongly suggest that loss of SoxC expression contributes to the ocular and other phenotypes observed in Chd7-associated CHARGE syndrome. In this study, we begin to further investigate the role that Sox11 plays in the phenotypes seen in CHARGE syndrome by generating Sox11-mutant zebrafish using the CRISPR-Cas system. The resulting Sox11 mutant lines will be characterized for phenotypes related to CHARGE and will be compared to an established CHD7 mutant line. These experiments will provide a better understanding of the potential role of Sox11 in the pathogenesis of CHARGE.