Vision and Hearing Loss
Resulting from a Mutation in Ush2a gene

Ryan Crane

Abstract
Usher syndrome (USH) is the leading cause of combined deafness and blindness worldwide, with mutations in USH2A being the most causative. Studying the mechanism of this debilitating sensory loss remains elusive due to the inability to recapitulate patients’ phenotype in animal models. To overcome this, a USH2A knock-in mouse expressing a common human disease-mutation in Usherin was generated and evaluated. This model exhibited a significant reduction in visual function consistent with late-onset retinitis pigmentosa phenotype seen in USH2A patients. Further, evaluation of the model showed reduced auditory function beginning at early age and persisting throughout the life of the animal, again, consistent with the phenotype regularly seen in patients.

Biosketch
Ryan Crane is a Ph.D. candidate in biomedical engineering at the University of Houston. He received his B.Sc. in Chemistry and in Cellular, Molecular and Physiological Biology from Christopher Newport University in 2016. His current research focuses on characterizing the Usherin mutant mouse models to better understand the mechanism behind the Usher syndrome. His research focuses on the effect of the mutation on both the retina and the cochlea, with primary focus in the cochlea. His ultimate goal is to develop a non-viral gene therapy for both organ systems. His research interests include non-viral gene therapy and improved delivery methods.