

TITLE:

Using dual Adeno associated virus (AAV) vectors to treat inherited retinal disease

FACULTY MENTOR NAME, EMAIL PHONE NUMBER

Shannon E. Boye, PhD
shaire@ufl.edu; 352 273-9342

FACULTY MENTOR DEPARTMENT

Dept. of Ophthalmology

RESEARCH PROJECT DESCRIPTION

Many inherited retinal diseases (IRD) are associated with mutations in genes that are too large to fit within a standard Adeno associated virus (AAV) vector (carrying capacity ~5kb). My lab investigates dual AAV platforms for their ability to deliver genes >5kb to animal models of inherited retinal disease. I am currently applying dual vector technology to a mouse model of *MYO7A* Usher syndrome 1B (the most common and severe form of this deaf and blinding disorder) and *CEP290*-LCA (the most common form of a severe pediatric retinal dystrophy, Leber Congenital Amaurosis). We have already shown that two AAV vectors (the first containing a promoter and the N' terminal portion of *MYO7A*, the second containing the C' terminal portion of *MYO7A*) are capable of recombining within the target cell leading to the expression of full length *MYO7A* protein. Importantly, sequence fidelity of the reconstituted gene is 100% accurate. Ongoing research aims to characterize therapy in a new mouse model of *USH1B* and, ultimately, to develop a paradigm shift in AAV-mediated gene therapy by opening the door to treat retinal (and potentially systemic) diseases associated with mutations in large genes.