



Mary Anna Carbone



### ***Drosophila* as a model system for the study of disease**

#### **Research emphasis:**

The focus of Dr. Carbone's research is to develop the fruit-fly, *Drosophila*, as a model system for the study of disease. In humans, mutations in the gene myocilin (MYOC) have been associated with cases of juvenile-onset and primary open-angle glaucoma. We have developed a transgenic fly model that over-expresses human MYOC and 4 variants previously associated with glaucoma. More recently, the *Drosophila* Genetic Reference Panel has been utilized to uncover candidate genes contributing to age-related visual senescence. Currently, we are using *Drosophila* as a model-system to study glaucoma in dogs and hypertrophic cardiomyopathy in cats.

#### **Application:**

- Transgenic flies
- Genome-wide Association Studies
- Transcription Profiling
- Crispr/Cas technology in flies

#### **Collaboration potential:**

- *Drosophila* as a disease model
- Validation of candidate genes and polymorphisms associated with human and animal disease.

#### **Selected publications:**

Carbone MA, Ayroles JF, Yamamoto A, Morozova TV, West SA, Magwire MM, Mackay TF, Anholt RR. Overexpression of myocilin in the *Drosophila* eye activates the unfolded protein response: implications for glaucoma. PLoS One. 2009;4(1):e4216. doi: 10.1371/journal.pone.0004216. Epub 2009 Jan 16.

Carbone MA, Chen Y, Hughes GA, Weinreb RN, Zabriskie NA, Zhang K, Anholt RR. Genes of the unfolded protein response pathway harbor risk alleles for primary open angle glaucoma. PLoS One. 2011;6(5):e20649. doi: 10.1371/journal.pone.0020649. Epub 2011 May 31.

Anholt RR and Carbone MA. A molecular mechanism for glaucoma: endoplasmic reticulum stress and the unfolded protein response. Trends Mol Med. 2013 Oct;19(10):586-93. doi: 10.1016/j.molmed.2013.06.005. Epub 2013 Jul 19. Review.

Huang W, Carbone MA, Magwire MM, Peiffer JA, Lyman RF, Stone EA, Anholt RR, Mackay TF. Genetic basis of transcriptome diversity in *Drosophila melanogaster*. Proc Natl Acad Sci U S A. 2015 Nov 3;112(44):E6010-9. doi: 10.1073/pnas.1519159112. Epub 2015 Oct 19.

Research Assistant Professor

#### **Education:**

B.Sc. University of Toronto, Canada

M.Sc. University of Toronto, Canada

Ph.D. University of Toronto and the Hospital for Sick Children, Canada

#### **Address:**

Department of  
Biological Sciences  
North Carolina State University  
Program in Genetics  
Room 3531 Thomas Hall  
110 Derieux Place  
Raleigh, NC 27607

**Phone:** 919-515-5789

**Email:** macarbon@ncsu.edu