**Xiaoling Zhang**

[](http://www.bumc.bu.edu/genetics/files/2016/02/Xiaoling-Zhang.jpg)

Assistant Professor of Medicine and Biostatistics  
Boston University School of Medicine, Section of Biomedical Genetics and  
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**Education**  
2010-2015 NIH Research Fellow, Cardiovascular Epidemiology and Human Genomics Branch  
the NHLBI Framingham Heart Study  
2009 PhD Boston University (Bioinformatics)  
2004 M.S. The State University of New York at Buffalo (Computer Science)

**Research Interests**

My primary research interests focus on: 1) develop methods for analyzing large-scale genetics and genomics data including microarray, genotype and next-generation sequencing data; 2) discover common, low-frequency and rare variants associated with complex diseases and their risk factors; 3) identify and prioritize putative causal genetic variants by integrative genomics study that involves combining biological information from a wide-range of data resources with eQTL analysis, pathway analysis and regulatory network analysis; and 4) translate the above computational findings to the molecular mechanisms underlying human genetic disorders by conducting experimental validation through collaborations.

**Selected Publications (\*Co-first authorship):**

1. M. Polfus, R.K. Khajuria, U.M. Schick, N. Pankratz, R. Pazoki, J.A. Brody, M.H. Chen, P.L. Auer, J.S. Floyd, J. Huang, L. Lange, F.J. van Rooij, R.A. Gibbs, G. Metcalf, D. Muzny, N. Veeraraghavan, K. Walter, L. Chen, L. Yanek, L.C. Becker, G.M. Peloso, A. Wakabayashi, M. Kals, A. Metspalu, T. Esko, K. Fox, R. Wallace, N. Franceshini, N. Matijevic, K.M. Rice, T.M. Bartz, L.P. Lyytikainen, M. Kahonen, T. Lehtimaki, O.T. Raitakari, R. Li-Gao, D.O. Mook-Kanamori, G. Lettre, C.M. van Duijn, O.H. Franco, S.S. Rich, F. Rivadeneira, A. Hofman, A.G. Uitterlinden, J.G. Wilson, B.M. Psaty, N. Soranzo, A. Dehghan, E. Boerwinkle, **X. Zhang**, A.D. Johnson, C.J. O’Donnell, J.M. Johnsen, A.P. Reiner, S.K. Ganesh, V.G. Sankaran.  *Whole-Exome Sequencing Identifies Loci Associated with Blood Cell Traits and Reveals a Role for Alternative GFI1B Splice Variants in Human Hematopoiesis*. *American Journal Human Genetics*. **2016** Aug 4;99(2):481-8. doi: 10.1016/j.ajhg.2016.06.016.  PMID: 27486782
2. Pankratz, U.M. Schick, Y. Zhou, W. Zhou, **X. Zhang**. CHARGE Consortium Hematology Working Group.  *Meta-analysis of rare and common exome chip variants identifies S1PR4 and other loci influencing blood cell traits. Nature Genetics*. **2016** Aug;48(8):867-76. doi: 10.1038/ng.3607. PMID: 27399967
3. R. Warren, J.F. O’Sullivan, M. Friesen1, C.E. Becker, **X. Zhang**, Y. Wakabayashi, J.E. Morningstar, X. Shi, J. Choi, F. Xia, D.T. Peters, M.H.C. Florido, J. Shay, K. Musunuru, S. Kathiresan, L. Daheron, J. Zhu, R.E. Gerszten, R. Deo, V. Ramachandran, C. J. O’Donnell, and C.A. Cowan.  *Induced Pluripotent Stem Cell Differentiation Enables Functional Validation of GWAS Variants in Metabolic Disease. Cell Stem Cell*. **2017** Apr 6;20(4):547-557. .e7. doi: 10.1016/j.stem.2017.01.010.
4. Joehanes, R\*., **Zhang, X\***., Huan, T., Yao, C., Ying, S., C.J. O’Donnell, P. J. Munson, D. Levy. *Integrated Genome-wide Analysis of Expression Quantitative Trait Loci Identifies Putative Disease-Related Genes and Pathways*. *Genome Biology* **2017** Jan 25;18(1):16. PMID: 28122634 (\* equal contribution)
5. Wu J. PD, Chung J., Lent S., Fisher V., Pitsillides A., Farrer L., **Zhang X\***. *An Efficient Analytic Approach in Genome-wide Identification of DNA Methylation Quantitative Trait Loci Response to Fenofibrate Treatment*. (\*Senior & Corresponding author, Accepted, in Press)
6. Chung J., **Zhang X.**, Allen M., Wang X., Ma Y., Beecham G., Montine TJ., Younkin SG., Dickson DW., Golde TE., Price ND., Ertekin-Taner N., Lunetta KL., Mez J., Alzheimer’s Disease Genetics Consortium, Mayeux R., Haines J., Pericak-Vance M., Schellenberg G., Jun G., Farrer LA. *Genome-wide Pleiotropy Analysis of Neuropathological Traits Related to Alzheimer Disease.* (*Alzheimer’s Research & Therapy*, in press)
7. Chung J., Wang X., Maruyama T., Ma Y., Kim M., **Zhang X**., Mez J., Sherva R.,  Takeyama H., The Alzheimer’s Disease Neuroimaging Initiative\*, Lunetta K.L., Farrer L.A., and Jun G.R. *Genome-Wide Association Study of Alzheimer Disease Endophenotypes at Preclinical and MCI Stages.* (Alzheimer’s & Dementia, in press)
8. Malhotra, A.C. Mauer, X. Guo, J. Yao, **X. Zhang**, F. Wunderer, A.V. Smith, Q. Wong, S. Pechlivanis, S. Hwang,  J. Wang, â€¦, D.B. Bloch#, W. S. Post#,, and C.J. O’Donnell#,, for the CHARGE Extracoronary Calcium Working Group. HDAC9 is Implicated in Atherosclerotic Aortic Calcification and Affects Vascular Smooth Muscle Cell Phenotype. (*Nature Genetics*, in revision)
9. “Genome-wide association study of carotid intima-media thickness and plaque identifies novel loci for atherosclerosis and cardiovascular outcomes, and downstream regulatory effects in vascular tissue” (*Nature Genetics*, under review)
10. C. Bis\*, X. Jian\*, B.W. Kunkle\*, Y. Chen\*…,**X. Zhang**, Alzheimer’s Disease Sequencing Project, C. Bellenguez, J. Lambert, M. Kurki, A. Palotie, M. Daly, E. Boerwinkle, K.L. Lunetta, J. Dupuis, E. R. Martin, G. D. Schellenberg, S. Seshadri, A.C. Naj\*\*, M. Fornage\*\*, L.A. Farrer\*\*. *Whole Exome Sequencing Study Identifies Novel Rare and Common Variant Loci Contributing to Risk of Alzheimer Disease*. (*Molecular Psychiatr*y, submitted)
11. **Zhang**, A. Frame, J. Williams, and R. Wainford. GNAI2 polymorphic variance associates with salt sensitivity of blood pressure in the Genetic Epidemiology Network of Salt Sensitivity study (*Physiology Genomics*, submitted)
12. Thiagalingam, A. Gower, C. Wong, J. Wu, **X. Zhang**, .. . Aberrant transcriptomes and DNA methylomes define pathways that drive pathogenesis and loss of brain laterality/asymmetry in schizophrenia (SCZ) and bipolar disorder (BD)” (American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, submitted)
13. Hwang,  O. Onuma, J.M. Massaro, **X. Zhang**, Y. Fu, U. Hoffmann, C. S. Fox, C. J. O’Donnell. Maintenance of Ideal Cardiovascular Health and Coronary Artery Calcification Progression in Low Risk Men and Women in the Framingham Heart Study. (*Circulation:Cardiovascular Imaging*, submitted)
14. **Zhang X.**, Ma Y., Lancour D.,  Farrell JJ., Chung J., Mayeux R., Haines J., Schellenberg G., Pericak-Vance M., Lunetta KL., Farrer L. Novel Genetic Variants Associated with Familial Late-Onset Alzheimer Disease in the Alzheimer’s Disease Sequencing Project. (in submission)