

## Michael L. Stitzel, Ph.D.

The Jackson Laboratory for Genomic Medicine  
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### POSITIONS HELD:

2013-present **Assistant Professor**, The Jackson Laboratory for Genomic Medicine, Farmington, CT

### EDUCATION & TRAINING:

2007-2013 **Postdoctoral Fellowship**, National Human Genome Research Institute  
Postdoctoral Advisor: Francis S. Collins, M.D., Ph.D.  
2001-2007 **Ph.D., Human Genetics & Molecular Biology**, The Johns Hopkins University  
Graduate Advisor: Geraldine Seydoux, Ph.D.  
2000-2001 **Fulbright Fellowship**, Zentrum für Molekulare Biologie Heidelberg (ZMBH), Germany  
Research Advisor: Renato Paro, Ph.D.  
1996-2000 **B.S., Biochemistry & Molecular Biology**, The Pennsylvania State University  
Honors Thesis Advisor: Joseph C. Reese, Ph.D.

### FUNDING:

2014-2015 **NIH/NIAID HIPC Pilot Grant (U19AI089987; co-Investigator, 10% effort; PI: Banichereau)**,  
"Epigenetics of Human Blood Dendritic Cell Subsets", \$275k.  
2011-2016 **NIH/NIDDK K99/R00 Pathway to Independence Award (K99DK092251; Principal Investigator)**, "Investigation of noncoding variation in human pancreatic islets and their developmental precursors", \$750k total over three years of the R00 phase.  
2014-2016 **TJL Director's Innovation Fund (co-Investigator; co-PIs Howell and Carter)**, "Maximizing human and mouse resources to identify novel variants for Alzheimer's Disease"  
2014-2016 **TJL Director's Innovation Fund (co-Investigator; Carter, PI)**, "Genetics of Molecular Epigenetics"

### COMPLETED FUNDING:

2012-2013 **NIH/NHGRI/NISC Flagship Project Sequencing Award**, "Determining effects of diabetes variants on the transcriptome of human pancreatic islets", \$175k.

### AWARDS AND HONORS:

2014 American Diabetes Association Young Investigator Travel Grant Award  
2013 Genome Technology Young Investigators of the Year Award  
2012 Endocrine Society Early Investigator's Workshop for Trainees

2011	NIH Fellows Award for Research Excellence
2005	Best Graduate Presentation, Johns Hopkins McKusick-Nathans Institute of Genetic Medicine Retreat
2002, 2003	Honorable Mention, NSF Graduate Research Fellowship
2000	William J. Fulbright Scholarship
2000	National Institutes of Health Summer Research Fellowship
1998-2000	Schreyer Scholar, Schreyer Honors College, Penn State University
1999	Pennsylvania State University Life Sciences Consortium Summer Research Fellowship
1998	National Institutes of Health Summer Research Fellowship
1998	Penn State Undergraduate Faculty Senate Scholarship for Academic Excellence
1997	National Institutes of Health Summer Research Fellowship
1996-2000	Dean's List, Pennsylvania State University

#### **INVITED TALKS:**

2014	Epigenomics, Sequencing and SNiPs-2014, Cambridge, MA
2014	Innovative Approaches to Diabetes Research and Therapies, New Haven, CT
2014	Endocrine Grand Rounds, UMass Med Diabetes Center of Excellence, Worcester, MA
2013	Vanderbilt University, Nashville, TN
2013	The Jackson Laboratory, Bar Harbor, ME
2013	The Jackson Laboratory for Genomic Medicine, Farmington, CT
2012	National Institute of Environmental Health Sciences, NIH, Raleigh-Durham, NC
2012	Program in Personalized and Genomic Medicine, University of Maryland School of Medicine, Baltimore, MD
2012	Biology Department, Dickinson College, Carlisle, PA

#### **ABSTRACTS SELECTED FOR TALKS:**

1. **Stitzel ML**, Huyghe JR, Morken MA, Parker SCJ, Fuchsberger C, Welch R, Jackson AU, Erdos MR, Kuusisto J, Laakso M, Boehnke M, Collins FS. Fine-mapping and functional genomic analysis link an intergenic islet stretch enhancer in the *C2CD4A/B* locus to Islet Dysfunction. American Diabetes Association 74<sup>th</sup> Scientific Sessions. San Francisco, CA
2. **Stitzel ML**, Parker SCJ, and Collins FS. 2014. Stretch enhancers, cell identity, and GWAS. 2014 Keystone Symposium: Transcriptional Regulation. Santa Fe, NM
3. **Stitzel ML**, Morken M, Chines PS, Erdos MR, Narisu N, Sethupathy P, and Collins FS. 2012. Cis-regulatory variation in islet dysfunction and diabetes. Keystone Symposium: Advances in Islet Biology. Monterey, CA
4. **Stitzel ML**, Pearson DS, Chines PS, Sethupathy P, Song L, Erdos MR, Crawford GE, and Collins FS. 2009. Global analysis of chromatin marks in human pancreatic islets provides insights to type 2 diabetes susceptibility loci. NHGRI Annual Scientific Retreat. Gettysburg, PA
5. **Stitzel ML** and G. Seydoux. 2007. The meiotic cell cycle regulates the EGG-3/MBK-2 cortical complex essential for the oocyte-to-zygote transition. 16th International *C. elegans* Conference. Los Angeles, CA
6. **Stitzel ML**, Pellettieri J, and Seydoux G. 2005. A clean start: coordinate degradation of maternal proteins during the oocyte-to-embryo transition. 2005 Gordon Research Conference on Fertilization and the Activation of Development. Holderness, NH
6. **Stitzel ML**, Pellettieri J, and Seydoux G. 2005. MBK-2 and the coordinate degradation of maternal proteins during the oocyte-to-embryo transition. 15th International *C. elegans* Conference. Los

Angeles, CA

7. **Stitzel ML**, Pellettieri J, and Seydoux G. 2005. A clean start: coordinate degradation of maternal proteins during the oocyte-to-embryo transition. Johns Hopkins McKusick-Nathans Institute of Genetic Medicine 2nd Annual Scientific Retreat. St Michaels, MD

### **TEACHING AND MENTORING EXPERIENCE:**

- 2007-2013      **Preceptor**, NIH Post-Baccalaureate Intramural Research Training Award Program
- Jose Orozco Segrera (currently enrolled in Harvard's MD/PhD program)
    - Awarded Outstanding Poster award at 2013 NIH Postbac Poster Day
  - Damien Abreu (currently enrolled in Washington University's MD/PhD program)
    - Awarded Outstanding Poster award at 2012 NIH Postbac Poster Day
  - Daniel S. Pearson (currently enrolled in Harvard's MD/PhD program)
  - Parimal Deodhar (currently Pediatric Resident, Yale University)
- 2010            **Course Lecturer**, "Pancreatic Stem Cells", FAES NIH Graduate School
- 2006            **Visiting Undergraduate Student Mentor**, Seydoux Lab
- Romain Levayer (currently PhD student at IBDML, UMR, Marseille, France)
- 2003-2006     **Rotation Student Project Mentor**, Seydoux Lab
- Rachel Webster
  - Kayam Chak
  - Kristi Hohenstein
  - Laura Koontz
  - Chih-Chien "Ken" Cheng
- 2005            **Intern**, Maryland Science Center
- 2004            **Teaching Assistant**, Fundamentals of Genetics, Johns Hopkins Graduate Students
- 2003            **Teaching Assistant**, Advanced Topics in Human Genetics, Johns Hopkins Graduate Students
- 1999-2000     **Tutor**, Department of Chemistry, Penn State University

### **SCIENTIFIC SERVICE:**

- 2013            Presenter, "Genome Geeks", Smithsonian's National Museum of Natural History
- 2013            *Ad hoc* reviewer for Juvenile Diabetes Research Foundation (JDRF)
- 2012            Trainee Panel Member, NHGRI Blue Ribbon 10-year Review
- 2011-            Manuscript reviews for *PLoS One*, *Genome Research*, *PNAS*, *Cell*, *EJHG*
- 2010-            Member, NHGRI Genome Trainee Advisory Committee
- 2010-2011     NIH Fellows Editorial Board
- 2008-2009     DNA Day Ambassador, NHGRI
- 2004-2006     Advisory Committee, Johns Hopkins Student Assistance Program
- 2004            Johns Hopkins School of Medicine (JHSOM) Minority Student Science Day
- 2003-2006     JHSOM Community Science Day
- 2002-2006     JHSOM Graduate Student Association Representative

### **PROFESSIONAL MEMBERSHIPS:**

- 2012-            The Endocrine Society
- 2009-            American Diabetes Association
- 2007-            American Society of Human Genetics

**PEER-REVIEWED PUBLICATIONS:**

1. Kulzer, JR, **Stitzel ML**, Morken MA, Huyghe JR, Fuchsberger C, Kuusisto J, Laakso M, Boehnke M, Collins, FS, Mohike KL. 2014. A Common Functional Regulatory Variant at a Type 2 Diabetes Locus Upregulates ARAP1 Expression in the Pancreatic Beta Cell. *Am J Hum Genet.* Feb 6; 94(2):186-97. PMID: 24439111.
2. Wang Y, Wang, JT, Rasoloson D, **Stitzel ML**, O'Connell KF, Smith HE, Seydoux G. Identification of Suppressors of mbk-2/DYRK by Whole-Genome Sequencing. 2014. *G3 (Bethesda).* 19;4(2):231-41. PMID: 24347622.
3. Parker SCJ\*, **Stitzel ML\***, Taylor DL, Orozco J, Akiyama JA, Chines PS, Narisu N, Erdos MR, Pennacchio LA, and Collins FS. Chromatin stretch enhancer states drive cell-specific gene regulation and harbor human disease risk variants. 2013. *Proc Natl Acad Sci U S A.* 110(44):1792106 PMID: 24127591 \*Equal contribution.
4. Bonnycastle LL, Chines PS, Huyghe JR, Swift AJ, Heikinheimo P, Mahadevan J, Peltonen S, Huopio H, Nuutila P, Narisu N, Goldfeder RL, **Stitzel ML**, Lu S, Boehnke M, Urano, Collins FS, Laakso M. 2013. Autosomal Dominant Diabetes Arising from A Wolfram Syndrome 1 Mutation. *Diabetes.* 62(11):3943-50. PMID: 23903355.
5. Gartner JJ, Parker SC, Prickett TD, Dutton-Regester K, **Stitzel ML**, Lin JC, Davis S, Simhadri VL, Jha S, Katagiri N, Gotea V, Teer JK, Wei X, Morken MA, Bhanot UK; NISC Comparative Sequencing Program, Chen G, Elnitski LL, Davies MA, Gershenwald JE, Carter H, Karchin R, Robinson W, Robinson S, Rosenberg SA, Collins FS, Parmigiani G, Komar AA, Kimchi-Sarfaty C, Hayward NK, Margulies EH, Samuels Y. 2013. Whole-genome sequencing identifies a recurrent functional synonymous mutation in melanoma. *Proc Natl Acad Sci U S A.* 62(11):3943-50 PMID:23901115
6. **Stitzel ML\***, Sethupathy P\*, Pearson DS, Chines PS, Song L, Erdos MR, Welch R, Parker SCJ, Boyle AP, Scott LJ, NISC Comparative Sequencing Program, Margulies EH, Boehnke M, Furey TS, Crawford GE, and Collins FS. 2010. Global epigenomic analysis of primary human pancreatic islets provides insights into type 2 diabetes susceptibility loci. *Cell Metabolism* 12(5): 443-455. \*Equal contribution. PMID: 21035756.
7. **Stitzel ML\***, Cheng KCC\*, and Seydoux G. 2007. Regulation of MBK-2/Dyrk kinase by dynamic cortical anchoring during the oocyte-to-zygote transition. *Current Biology.* 17(18): 1545-1554. \*Equal contribution PMID: 17869113
8. **Stitzel ML** and Seydoux G. 2007. Regulation of the oocyte-to-zygote transition. *Science* 316(5823): 407-408. PMID: 17446393
9. **Stitzel ML**, Pellettieri J, and Seydoux G. 2006. The C. elegans DYRK kinase MBK-2 marks oocyte proteins for degradation in response to meiotic maturation. *Current Biology* 16(1): 56-62. PMID: 16338136.
10. Swanson DA, Liu ML, Baker PJ, Garrett L, **Stitzel M**, Wu J, Harriss M, Banerjee R, Shane B, and Brody LC. 2001. Targeted disruption of the methionine synthase gene in mice. *Molecular and Cellular Biology* 21(4): 1058-1065. PMID: 11158293.
11. **Stitzel ML**, Durso R, and Reese JC. 2001. The proteasome regulates the UV-induced activation of the AP-1-like transcription factor Gcn4. *Genes & Development* 15(2): 128-133. PMID: 11157770.

**References**

**Francis S. Collins, M.D., Ph.D.**

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