

## Michael L. Stitzel, Ph.D.

The Jackson Laboratory for Genomic Medicine  
10 Discovery Drive  
Farmington, CT 06032  
Email: michael.stitzel@jax.org

### ACADEMIC APPOINTMENTS

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2019-present **Associate Professor**, The Jackson Laboratory for Genomic Medicine, Farmington, CT  
2013-2019 **Assistant Professor**, The Jackson Laboratory for Genomic Medicine, Farmington, CT  
2013-present **Affiliated Faculty**, Department of Genetics and Genome Sciences, UConn Health

### EDUCATION & TRAINING

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2007-2013 **Postdoctoral Fellowship**, National Human Genome Research Institute, NIH  
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.

### ACTIVE FUNDING

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*Funding for 2020-2024*

**1 R01 DK118011-01A1, NIH/NIDDK (Stitzel, PI)** 12/1/2020 - 11/30/2024  
20% Effort Annual Direct: \$521,964 Total Costs: \$4,654,015  
*“Genetic programming of human islet metabolic and endoplasmic reticulum (ER) stress responses in diabetes”*

Our overall objective in this proposal is to elucidate the genetic regulation of islet stress responses and to determine how genetic variants, including SNPs associated with T2D and other metabolic traits (T2D SNPs), modulate these responses to contribute to islet dysfunction and T2D pathogenesis.

**PR202461, DoD/PRMRP (Stitzel, PI)** 5/1/2021 – 4/30/2022  
Percent Effort: 10% Annual Direct: \$199,845 Total Costs: \$371,044  
*“Proximity proteomic identification of trafficking and secretory markers of islet (dys)function”*

The goal of this project is to determine protein trafficking/processing defects in stressed islets and to identify secreted biomarkers of islet (dys)function and failure in diabetes.

**R01DK117137-01A1, NIH/NIDDK R01 (Stitzel, PI)** 01/01/2019-12/31/2023  
Percent Effort: 25% Annual Direct: \$250,000 Total Costs: \$2,362,500  
*“Regulation and function of the type 2 diabetes-associated C2CD4A/B locus”*  
Percent Effort: 25%  
Direct costs per year: \$250,000  
Total costs for project period: \$2,362,500

**W81XWH-18-0401, DoD IIRA (Stitzel, Partnering PI)** 09/01/2018-08/31/2021  
Stitzel (Partnering PI)

*“Dissecting the Heterogeneity of Islet Stress Responses in Type 2 Diabetes (T2D)”*

Percent Effort: 25%  
 Direct costs per year: \$500,000  
 Total costs for project period: \$2,835,000

**1-18-ACE-15, American Diabetes Association Pathway to Stop Diabetes Accelerator Award** 1/01/2018-12/31/2022

Stitzel (PI)

*“Deciphering Longitudinal Cell Type-Specific Defects in Diabetes Pathogenesis”*

Percent Effort: 25%  
 Direct costs per year: \$250,000  
 Total costs for project period: \$1,625,000

**PENDING FUNDING**

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1 R01 DK128514-01 (Stitzel, Ucar, Tewhey) 4/1/2021 - 3/31/2026 1.80 Calendar  
 NIH/NIDDK Annual Direct: \$499,810 Total Costs: \$4,710,165

*Dissecting the functional effects of cis-regulatory type 2 diabetes variants on islet and (pre)adipocyte cell identity and function*

The goal of this proposal is to decode the functions of non-coding, cis-regulatory T2D SNPs and the target/effector genes they impact in adipose and islet cells, which contribute respectively to the insulin resistance and insulin secretion defects contributing to diabetes risk and progression.

Role: Principal Investigator

Status: Priority Score 34, 26<sup>th</sup> percentile; planned 7/5/2021 A1 submission

R01 (Herold) 7/1/2021 - 6/30/2026 0.6 Calendar  
 NIH/NIDDK Annual Direct: \$87,503 Total Costs: \$859,378

*Adaptive epigenetic mechanisms of beta cells to immune responses*

The goal of this proposal is to delineate the molecular mechanisms by which Tet2 contributes to pathophysiologic beta cell responses and immune cell-mediated beta cell destruction.

Role: Consortium PI

Status: Pending Scientific Review, March 2021

R01 (Stitzel, Soleimanpour PIs) 12/1/2021-11/30/2025 2.4 Calendar  
 NIH/NIDDK Annual Direct: \$499, 987 Total Costs: \$4,342,230

*Genetic regulation of islet mitochondrial health and dysfunction in type 2 diabetes*

The goal of this project is to test the hypothesis that T2D-associated SNPs and their effector genes in 17 loci alter mitochondrial contribute to islet failure by modulating mitochondrial health or function.

Role: Co-PI

Status: Submitting 6/5/2021

**COMPLETED FUNDING**

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*Funding for 2013-2018*

**W81XWH-16-1-0130, DoD Discovery Award.** 01/01/2016-11/30/2018 (NCE)

Stitzel (PI)

*“Single-cell Dissection of Human Pancreatic Islet Dysfunction in Diabetes”*

Percent Effort: 5%  
 Direct costs per year: \$150,000  
 Total costs for project period: \$350,000

**R00DK092251, NIH/NIDDK R00 Pathway to Independence Award** 08/01/2014-07/30/2017

Stitzel (PI)

*“Investigation of noncoding variation in human pancreatic islets and their developmental precursors”*

Direct costs per year: \$142,857

Total costs for project period: \$750,000

**R00DK092251 03S1, NIH/NIDDK R00 Supplement**

01/01/2015-12/31/2015

Stitzel (PI)

*“Investigation of noncoding variation in human pancreatic islets and their developmental precursors”*

Direct costs per year: \$26,857

Total costs for project period: \$47,000

**U19AI089987, NIH/NIAID HIPC Pilot Grant**

07/01/2015-09/30/2016

Stitzel (Co-Investigator), Banchereau (PI)

*“Epigenetics of Human Blood Dendritic Cell Subsets”*

*Funding prior to 2013*

**K99DK092251, NIH/NIDDK K99 Pathway to Independence Award**

10/01/2011-09/30/2013

Stitzel (PI)

*“Investigation of noncoding variation in human pancreatic islets and their developmental precursors”*

**NIH/NHGRI/NISC Flagship Project Sequencing Award**

04/01/2012-09/30/2013

Stitzel (PI)

*“Determining effects of diabetes variants on the transcriptome of human pancreatic islets”*

## **AWARDS AND HONORS**

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### ***National / International:***

2018 Investigator, American Diabetes Association Pathway to Stop Diabetes Program  
 2017 Invited to Faculty of 1000 Prime, Genomics  
 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 Trainee  
 2002, 2003 Honorable Mention, NSF Graduate Research Fellowship  
 2000 William J. Fulbright Scholarship  
 1997, 2000 National Institutes of Health Summer Research Fellowship

### ***Institutional:***

2011 NIH Fellows Award for Research Excellence  
 2005 Best Graduate Presentation, Johns Hopkins McKusick-Nathans Institute of Genetic Medicine Retreat  
 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  
 1996-2000 Dean’s List, Pennsylvania State University

## **INVITED TALKS**

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### ***National / International:***

2021 81<sup>st</sup> Scientific Sessions of the American Diabetes Association (Virtual)  
 2020 NIEHS Division of Intramural Research Seminar Series, Research Triangle Park, NC  
 2019 Department of Molecular and Cell Biology, University of Connecticut, Storrs, CT

2019 Diabetes Obesity Metabolism Institute, Mount Sinai School of Medicine, New York, NY  
2019 79<sup>th</sup> Scientific Sessions of the American Diabetes Association, San Francisco, CA  
2019 20<sup>th</sup> Anniversary Servier-IGIS Symposium, St. Jean Cap Ferrat, France  
2018 Genome Sciences Seminar Series, University of Virginia, Charlottesville, VA  
2018 78<sup>th</sup> Scientific Sessions of the American Diabetes Association, Orlando, FL  
2016 76<sup>th</sup> Scientific Sessions of the American Diabetes Association, New Orleans, LA  
2015 Keystone Symposium, Mechanisms of Pro-Inflammatory Diseases, Olympic Valley, CA  
2015 Navigating the Sea of Genomic Data, American Dental Association, Chicago, IL  
2014 Center of New Technologies, University of Warsaw, Warsaw, Poland  
2014 Medical University of Bialystok, Bialystok, Poland  
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

**Regional:**

2018 26<sup>th</sup> Annual Boston Ithaca Islet Club Meeting, Worcester, MA  
2014 Illumina Sequencing Users' Meeting, New Haven, CT  
2014 Endocrine Grand Rounds, UMass Med Diabetes Center of Excellence, Worcester, MA  
2014 Epigenomics, Sequencing and SNIps-2014, Cambridge, MA  
2014 Innovative Approaches to Diabetes Research and Therapies, New Haven, CT

**Local:**

2017 Connecticut Children's Medical Center Endocrinology Department, Farmington, CT  
2015 University of Connecticut Health Center (UHC) Grand Rounds, Farmington, CT  
2014 Wesleyan University Seminar Series, Middletown, CT  
2014 Institute for Systems Genomics Workshop, Farmington, CT

**ABSTRACTS SELECTED FOR TALKS:**

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1. Khetan S, Kursawe R, Youn A, Lawlor N, Jillette A, Marquez-Campos E, Ucar D, and **Stitzel ML**. 2018. Chromatin accessibility profiling uncovers genetic and T2D disease state-associated changes in *cis*-regulatory element use in human islets. Boston-Ithaca Islet Club. Worcester, MA
2. Kycia I, Huyghe JR, Wolford B, Piecuch E, Vadlamudi S, Kursawe R, Kuusisto J, Collins FS, Boehnke M, Mohlke KL, Ruan Y, Parker SCJ, and **Stitzel ML**. 2015. Fine-mapping and functional genomic analyses link an evolutionarily conserved *C2CD4A/B* locus islet stretch enhancer to islet dysfunction and type 2 diabetes. Gordon Research Conference on Human Genetics and Genomics. Newport, RI
3. **Stitzel ML**, Huyghe JR, Morken MA, Parker SCJ, Fuchsberger C, Welch R, Jackson AU, Erdos MR, Kuusisto J, Laakso M, Boehnke M, Collins FS. 2014. 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
4. **Stitzel ML**, Parker SCJ, and Collins FS. 2014. Stretch enhancers, cell identity, and GWAS. 2014 Keystone Symposium: Transcriptional Regulation. Santa Fe, NM

5. **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
6. **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
7. **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
8. **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
9. **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
10. **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:**

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- 2016-present **Thesis advisor**, University of Connecticut Health Center Department of Genetics and Genome Sciences
- 2016-2020: Shubham Khetan, PhD candidate
    - Current position: Postdoctoral fellow, Bulyk lab, Harvard Medical School
  - 2019-present: Redwan Bhuiyan, MD/PhD candidate
- 2014-2016 **Postdoctoral advisor**, Ina Kycia
- Ina's abstract was selected for a talk by the organizing committee for the 2015 Gordon Research Conference on Human Genetics and Genomics
  - Current position: Research Laboratory Supervisor, Boston Children's Hospital
- 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, PhD (currently completing clerkships in Harvard's MD/PhD program)
  - Parimal Deodhar (currently Pediatrician at Seattle Children's Hospital)
- 2010 **Course Lecturer**, "Pancreatic Stem Cells", FAES NIH Graduate School
- 2006 **Visiting Undergraduate Student Mentor**, Seydoux Lab
- Romain Levayer (currently Group Leader, Department of Developmental and Stem Cell Biology at Institut Pasteur)
- 2003-2006 **Rotation Student Project Mentor**, Seydoux Lab

- Kayam Chak (currently Scientific and Medical Writer, DAVA Oncology)
- Kristi Hohenstein Elliott (currently Director, Human Therapeutics Division at Intrexon Corporation)
- Laura Koontz (currently member of Personalized Medicine Staff at the FDA)
- Chih-Chien “Ken” Cheng (currently Staff Scientist at NCATS)
- Rachel Webster

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

## PROFESSIONAL SERVICE

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### ***National/International Peer Review Groups/Grant Study Sections:***

2020,2021      NIH (NIDDK) Diabetes, Endocrinology, and Metabolic Diseases Catalyst Award, Stage 1 Reviewer

2020      INSERM ATIP-Avenir 2020

2019-2022      Standing member, Research Grants Review Committee, American Diabetes Association

2019,2020      University of Michigan Diabetes Research Center

2019      NIH (NIDDK) Special Emphasis Panel, ZDK1 GRB-J (M2), Mechanisms Underlying the Contribution of Type 1 Diabetes Disease-associated Variants

2019      Department of Defense (DOD) Peer-Reviewed Medical Research Project (PRMRP) Pre-application Diabetes 1 Peer Review Panel

2019,2020      University of Washington Diabetes Center

2018      The Wellcome Trust Investigator Award in Science Program

2018      Programme “Actions de recherche concertée” 2018-2023 ARC Consolidator Projects, Université Libre Bruxelles (ULB)

2018      Juvenile Diabetes Research Foundation (JDRF), *Biomarker Analysis Centers for Mass Cytometry and Transcriptome Analysis of TrialNet Samples*

2018      Department of Defense (DOD) Peer-Reviewed Medical Research Project (PRMRP) Pre-application Diabetes 1 Peer Review Panel

2017      DOD PRMRP Discovery Award Diabetes Peer Review Panel

2017      Great Ormond Street Hospital Children’s Charity (largest charitable funder of research dedicated to pediatric research in the United Kingdom)

2017      Israel Science Foundation

2017      NIH ZDK1 GRB-N (M2) Special Emphasis Panel Inflammatory Bowel Disease Genetics Consortium

2016      Department of Defense (DOD) Peer-Reviewed Medical Research Project (PRMRP) Pre-application Diabetes 1 Peer Review Panel

2013      Juvenile Diabetes Research Foundation (JDRF), *Biomarkers of Beta Cell Stress*

### ***Regional Peer Review Groups:***

2017      Boston Area Diabetes Endocrinology Research Center, Peer Reviewer

### ***National/International Meeting Planning and Abstract Review:***

2018, 2019      American Diabetes Association (ADA) 78<sup>th</sup>, 79<sup>th</sup> Scientific Sessions, Abstract Reviewer

2017      Islet Biology/Insulin Secretion subcommittee for 78<sup>th</sup> ADA Scientific Sessions, Member

- 2016 American Diabetes Association (ADA) 76<sup>th</sup> Scientific Sessions, Abstract Reviewer  
2015 Genetics and Gene Regulation subcommittee for ADA 76<sup>th</sup> Scientific Sessions, Member

***Institutional Service:***

- 2014- UCONN Health thesis committee member  
-PhD candidates (Dinesh Uthaya Kumar, Zukai Liu, Rachel Gilmore, Ardian Ferraj)  
-MD/PhD (Grace Kwon, Alexandra Goetjen)  
2015- Single Cell Genomics, The Jackson Laboratory, Faculty Partner  
2016-2018 UCONN MEDS 5369 Advanced Genetics Course, Guest Instructor  
2016 UCONN/JAX Postdoctoral Fellow Forum, “Pathways to your own lab”, Panel Member  
2015 The Jackson Laboratory Scientific Advisory Council (elected by faculty peers), Member  
2012 NHGRI Blue Ribbon 10-year Review, Trainee Panel Member  
2010-2013 NHGRI Genome Trainee Advisory Committee Member  
2010-2011 NIH Fellows Editorial Board Member  
2004-2006 Johns Hopkins Student Assistance Program Advisory Committee Member  
2002-2006 JHSOM Graduate Student Association Representative

***Scientific Community Service/Outreach:***

- 2019 Focus on Fellows, “Careers in Research” session, American Diabetes Association  
2018 JAXtosition TED-style talk, “Targeting Type 2 Diabetes: Precision Approaches to a Global Disease”, The Jackson Laboratory for Genomic Medicine  
2016 Community Health Discussion Series, “Diabetes and Genetics: A Researcher’s Quest”, Connecticut Children’s Museum and The Jackson Laboratory  
2014-2016 Forum for Discovery, The Jackson Laboratory  
2013 Presenter, “Genome Geeks”, Smithsonian’s National Museum of Natural History  
2008-2009 DNA Day Ambassador, NHGRI  
2004 Johns Hopkins School of Medicine (JHSOM) Minority Student Science Day  
2003-2006 JHSOM Community Science Day

***Journal Reviews:***

- 2011- Manuscript reviews for *Cell*, *Cell Metabolism*, *Nature Genetics*, *Nature Reviews Endocrinology*, *eLife*, *Genome Research*, *Nature Communications*, *Nature Metabolism*, *PNAS*, *AJHG*, *EJHG*, *PLoS Genetics* (also guest Editor), *Cell Reports*, *Diabetes*, *Diabetologia*, *Development*, *Genome Medicine*, *Scientific Reports*, *PLoS One*, *JoVE*

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**PROFESSIONAL MEMBERSHIPS**

- 2009- American Diabetes Association  
2007- American Society of Human Genetics

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**MANUSCRIPTS IN PREPARATION**

Khetan S, Bhuiyan R, Kursawe R, Jillette A, and **Stitzel ML**. Genome-wide identification of genes controlling beta cell viability and insulin content. *In preparation for submission to Nature Metabolism* (12/2020)

Eroglu A, Lawlor N, Kursawe R, Ucar D, and **Stitzel ML**. Single nucleus ATAC-seq defines precise *cis*-regulatory circuits of each islet cell type and identifies type 2 diabetes variants altering their use. *In preparation for submission to Nature Genetics*.

Thibodeau A, Eroglu A, Nehar-Belaid D, Kursawe R, Marches R, Banchereau J, **Stitzel ML**, and Ucar D. A read count-based method to detect multiplets and their cellular origins from snATAC-seq data. *Under review, Bioinformatics*.

## MANUSCRIPTS UNDER REVIEW / IN PRESS

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Khetan S, Kales S, Kursawe R, Jillette A, Reilly SK, Ucar D, Tewhey R\*, **Stitzel ML\***. Functional characterization of thousands of type 2 diabetes-associated and chromatin-modulating variants under steady state and endoplasmic reticulum stress. *Second revision, Nature Communications*. bioRxiv deposit (2020 Feb 12): doi:10.1101/2020.02.12.939348 (\*co-senior authors)

Rui J, Deng S, Ponath G, Kursawe R, Lawlor N, Sumida T, Levine-Ritterman M, Perdigoto AL, **Stitzel ML**, Pitt D, Lu J, Herold KC. Tet2 Controls  $\beta$  cells Responses to Inflammation in Type I Diabetes. *First revision, Nature Communications*. bioRxiv deposit (2020 Sept 2): doi.org/10.1101/2020.09.01.278028.

## PEER-REVIEWED PUBLICATIONS

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### Original Research:

Period 2013-2020

1. Varshney A, Kyono Y, Elangovan VR, Wang C, Erdos MR, Narisu N, Albanus RD, Orchard P, **Stitzel ML**, Collins FS, Kitzman JO, Parcker SCJ. A transcription start site map in human pancreatic islets reveals functional regulatory signatures. 2021. *Diabetes*. Apr13;db201087. PMID:33849996.
2. Lawlor N, Nehar-Belaid D, Grassmann JDS, Stoeckius M, Smibert P, **Stitzel ML**, Pascual V, Banchereau J, Williams A, Ucar D. Single cell analysis of blood mononuclear cells stimulated through either LPS or Anti-CD3 and Anti-CD28. 2021. *Front Immunol*. Mar 17;12:636720. PMID:33815388.
3. Viñuela A, Varshney A, van de Bunt M, Prasad R, Asplund O, Bennett A, Boehnke M, Brown A, Erdos MR, Fadista J, Hansson O, Hatem G, Howald C, Iyengar AK, Johson P, Krus U, MacDonald PE, Mahajan A, Manning Fox JE, Narisu N, Nylander V, Orchard P, Oskolkov N, Panousis NI, Payne A, **Stitzel ML**, Vadlamudi S, Welch R, Collins FS, Mohlke KL, Gloyn AL, Scott LJ, Dermitzakis ET, Groop L, Parker SCJ, McCarthy MI. 2019. Genetic variant effects on gene expression in human pancreatic islets and their implications for T2D. 2020. *Nat Commun*. Sep 30;11(1):4912. PMID: 32999275.
4. Lawlor N, Márquez EJ, Orchard P, Narisu N, Thibodeau A, Kursawe R, Erdos MR, Pak E, Dutra A, Li X, Piecuch E, Shamim MS, Varshney A, Luo O, Chines PS, Kanke M, Fuchbserger C, NIH Intramural Sequencing Center, Sethupathy P, Ruan Y, Aiden EL, Collins FS, Ucar D, Parker SCJ\*, and **Stitzel ML\***. 2019. EndoC- $\beta$ H1 multiomic profiling defines gene regulatory programs intrinsic to human  $\beta$  cell identity and function. *Cell Reports*. Jan 15;26(3):788-801.e6. PMID: 30650367 (\*co-senior authors)
5. Varshney A, VanRenterghem H, Orchard P, Boyle A, **Stitzel ML**, Ucar D, and Parker SCJ. 2019. Cell specificity of human regulatory annotations and their genetic effects on gene expression. *Genetics*. Feb; 211(2):549-562. PMID: 30593493.
6. Youn A, Marquez EJ, Lawlor N, **Stitzel ML**, and Ucar D. 2019. BiFET: A Bias-free Transcription Factor Footprint Enrichment Test. *Nucleic Acids Research*. Jan 25;47(2):e11. PMID: 30428075.

7. Thibodeau A, Uyar A, Khetan S, **Stitzel ML**, and Ucar D. 2018. A neural network based model effectively predicts enhancers from clinical ATAC-seq samples. *Sci Rep*. Oct 30;8(1):16048. PMID: 30375457.
  8. Khetan S, Kursawe R, Youn A, Lawlor N, Marquez E, Ucar D\*, and **Stitzel ML\***. 2018. Chromatin accessibility profiling uncovers genetic- and T2D disease state-associated changes in cis-regulatory element use in human islets. *Diabetes*. Sep 4. PMID: 30181159 (\*co-corresponding)
  9. Wu Z, Zhang Y, **Stitzel ML**, and Wu H. 2018. Two-phase differential expression analysis for single cell RNA-seq. *Bioinformatics*. Apr 24. PMID: 29688282
  10. Kycia I, Wolford BN, Huyghe J, Fuchsberger C, Vadlamudi S, Kursawe R, Uyar A, Khetan S, Bolisetty M, Mathur A, Kuusisto J, Laakso M, Ucar D, Mohlke KL, Boehnke M, Collins FS, Parker SCJ, and **Stitzel ML**. 2018. A common functional variant in a type 2 diabetes locus potentiates activity of an evolutionarily conserved islet stretch enhancer and increases islet *C2CD4A* expression. *Am J Hum Genet*. Apr 5;102(4):620-635 PMID: 29625024
  11. Flannick J, Fuchsberger C, Mahajan A, Teslovich TM, Agarwala V, Gaulton KJ...**Stitzel, ML** [42<sup>nd</sup> author] ... Altshuler D, Burt NP, Florez JC, Boehnke M, McCarthy MI. 2017. Sequence data and association statistics from 12,940 type 2 diabetes cases and controls. *Sci Data*. Dec 19; 4:170179. PMCID: PMC5735917
  12. Lawlor N, Youn A, Kursawe R, Ucar D\*, and **Stitzel ML\***. 2017. Alpha TC1 and Beta-TC-6 genomic profiling reveals their utility for modeling molecular genetics of islet (dys)function and type 2 diabetes. *Scientific Reports*. Sep 20;7(1):11959 PMCID: PMC5607285 (\*co-corresponding)
  13. Roman TS, Cannon ME, Vadlamudi S, Buchkovich ML, Wolford BN, Welch RP, Morken MA, Kwon GJ, Varshney A, Kursawe R, Wu Y, Jackson AU; National Institutes of Health Intramural Sequencing Center (NISC) Comparative Sequencing Program, Erdos MR, Kuusisto J, Laakso M, Scott LJ, Boehnke M, Collins FS, Parker SCJ, **Stitzel ML**, Mohlke KL. 2017. A Type 2 Diabetes-Associated Functional Regulatory Variant in a Pancreatic Islet Enhancer at the *ADCY5* Locus. *Diabetes*. Sep;66(9):2521-2530. PMID: 28684635
  14. Ucar D, Márquez EJ, Chung CH, Marches R, Rossi RJ, Uyar A, Wu TC, George J, **Stitzel ML**, Palucka AK, Kuchel GA, Banchereau J. 2017. The chromatin accessibility signature of human immune aging stems from CD8<sup>+</sup> T cells. 2017 Oct 2;214(10):3123-3144. PMID: 28904110
  15. Varshney A\*, Scott LJ\*, Welch RP\*, Erdos MR\*, Chines PS, Narisu N, Albanus RD'O, Orchard P, Wolford BN, Kursawe R, Vadlamudi S, Cannon ME, Didion J, Hensley J, Kirilusha A, NISC Comparative Sequencing Program, Bonnycastle LL, Taylor DL, Watanabe R, Mohlke K, Boehnke M\*, Collins FS\*, Parker SCJ\*, and **Stitzel ML\***. 2017. Genetic regulatory signatures underlying islet gene expression and type 2 diabetes. *Proc Natl Acad Sci USA*. Feb 28;114(9):2301-2306. PMID28193859 \*Equal contribution
  16. Lawlor N, George J, Bolisetty M, Kursawe R, Sun L, V S, Kycia I, Robson P, **Stitzel ML**. 2017. Single cell transcriptomes identify human islet cell signatures and reveal cell-type-specific expression changes in type 2 diabetes. *Genome Res*. Feb; 27(2):208-222. PMID: 27864352
- EMBL-EBI deemed these human islet experiments/datasets of high quality and selected them for inclusion into its Single Cell Expression Atlas.**
17. Fuchsberger C\*, Flannick J\*, Teslovich TM\*, ...**Stitzel ML** [42<sup>nd</sup> author] ...Boehnke †, Altshuler D†, McCarthy MI†. 2016. The genetic architecture of type 2 diabetes. *Nature*. Aug 4;536(7614):41-7. PMID: 27398621

18. Thibodeau A, Márquez EJ, Luo O, Ruan Y, Menghi F, Shin DG, **Stitzel ML**, Vera-Licona P, Ucar D. 2016. QuIN: A web server for Querying and visualizing Chromatin Interaction Networks. *PLoS Comput Biol*. Jun 23;12(6): e1004809. PMID: 27336171.
19. Zubek J, **Stitzel ML**, Ucar D, Plewczynski DM. 2016. Computational inference of H3K4me3 and H3K27ac domain length. *PeerJ*. Mar 14;4: e1750. PMID: 26989607
20. Majithia AR, Flannick J, Shahinian P, Guo M, Bray MA, Fontanillas P, Gabriel SB; GoT2D Consortium.; NHGRI JHS/FHS Allelic Spectrum Project.; SIGMA T2D Consortium, T2D-GENES Consortium, Rosen ED, Altshuler D. 2014. Rare variants in PPARG with decreased activity in adipocyte differentiation are associated with increased risk of type 2 diabetes. *Proc Natl Acad Sci U S A*. Sep 9;111(36):13127-32. PMID: 25157153.
21. Wang SR, Agarwala V, Flannick J, Chiang CW, Altshuler D; GoT2D Consortium, Hirschhorn JN. 2014. *Am J Hum Genet*. May 1;94(5):710-20. PMID: 24768551
22. 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.

**This publication was awarded the C.W. Cotterman Award, judged by the AJHG editorial board to represent an outstanding scientific contribution to the field of human genetics in 2014.**

*Period prior to 2013*

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