

**Progress since last Committee Meeting - October 2, 2017****Awards**

- ASHG Reviewer's Choice Abstract Award 2016 & 2017
- Awarded & Renewed F31 Ruth L. Kirschstein NRSA 9/2016-9/2018
- FASEB MARC Travel Award to ASHG 2014 & 2015
- Genetics and Regulation Training Grant 2013-2016

**Manuscripts in progress**

- Submitting: Mozaffari, S. V., Stein, M. M., Magnaye, K. M., Nicolae, D. L., and Ober, C. (2018). Gene Expression and Methylation of Imprinted Genes in the Hutterites
- Revising: Mozaffari, S. V., DeCara, J. M., Shah, S. J., Lang, R. M., Nicolae, D. L., and Ober, C. (2017). Parent of Origin Effects on Quantitative Phenotypes in a Founder Population. *bioRxiv*
- Submitted: Shiquan, S., Zhu, J., Mozaffari, S. V., Ober, C., Chen, M., and Zhou, X. (2018). Heritability Estimation and Differential Analysis with Generalized Linear Mixed Models in RNA Sequencing and Bisulfite Sequencing Studies

**Published**

- Hart, M. W., Stover, D. A., Guerra, V., Mozaffari, S. V., Ober, C., Mugal, C. F., and Kaj, I. (2018). Positive selection on human gamete-recognition genes. *PeerJ*, 6(1):e4259–35
- Igartua, C., Mozaffari, S. V., Nicolae, D. L., and Ober, C. (2017). Rare non-coding variants are associated with plasma lipid traits in a founder population. *Scientific Reports*, 7(1):16415
- Gamazon, E. R., Wheeler, H. E., Shah, K. P., Mozaffari, S. V., Aquino-Michaels, K., Carroll, R. J., Eyler, A. E., Denny, J. C., Nicolae, D. L., Cox, N. J., and Im, H. K. (2015). A gene-based association method for mapping traits using reference transcriptome data. *Nature Genetics*, 47(9):1091–1098

**Oral Presentations**

- GGSB Work in Progress April 2016 & March 2017  
*Parent of Origin Effects in the Hutterites*
- Genetics of Model Organisms Club April 21, 2016  
*Parent of Origin Effects in the Hutterites*
- Human Genetics Work in Progress January 20, 2016  
*Parent of Origin Effects*
- Molecular Biosciences Retreat November 5, 2015  
Mozaffari SV, DeCara J, Shah S, Herman C, Lang R, Nicolae D, Ober C., *Parent of Origin GWAS with Cardiovascular Disease Associated Traits in the Hutterites*. 2015: Nov 5; Galena, IL.
- ASHG October 9, 2015  
Mozaffari SV, DeCara J, Shah S, Herman C, Lang R, Nicolae D, Ober C., *Parent of Origin GWAS of CVD-Associated Phenotypes in the Hutterites* (Abstract Program #310). Presented at the Annual Meeting of The American Society of Human Genetics; 2015: Oct 9; Baltimore, MD.

## Posters

- Reviewer's Choice Abstract Award:  
Mozaffari SV, Nicolae D, Ober C. Identifying Imprinted Genes and Parent of Origin Effects on Gene Expression in the Hutterites. Poster presented at the Annual Meeting of The American Society of Human Genetics Conference; 2017: Oct 17-21; Orlando, FL
- Mozaffari SV, Nicolae D, Ober C. *Opposite Allele Parent of Origin Effects on Cardiovascular and Asthma Associated Traits in the Hutterites*. Poster presented at the Gordon Research Seminar and Conference; 2017: July 8-14; Stowe, VT
- Reviewer's Choice Abstract Award:  
Mozaffari SV, Nicolae D, Ober C. Opposite Allele Parent of Origin Effects on Body Mass Index in the Hutterites. Poster presented at the Annual Meeting of The American Society of Human Genetics Conference; 2016: Oct 18-22; Vancouver, Canada
- Mozaffari SV, Gamazon E, Aquino-Michaels K, Cox NJ, Im HK. *Quantifying Context Specificity of Gene Regulation using Predicted Gene Expression Levels*. Poster presented at the Annual Meeting of The American Society of Human Genetics Conference; 2014: Oct 18-22; San Diego, CA

## Teaching Assistantship Requirements Completed

- BIOS 20187 *Fundamentals of Genetics* Winter 2018  
- Undergraduate Course: Recent developments in molecular genetics and the human genome project integrated into the structure of classical genetics.
- HGEN 47000 *Human Genetics* Fall 2015 & 2017  
- Graduate Course: Classic and modern approaches to studying cytogenetic, Mendelian, and complex human diseases. Grant proposal writing course.  
- Fall 2017: Conducting two-day computational workshop on GWAS
- MGCB 31400 (BIOS 21236) *Genetic Analysis of Model Organisms* Fall 2014  
- Graduate & Undergraduate Course: Introduction to genetic tools, experiments, and model organisms

## Additional Courses

- STAT 24500 *Statistical Theory & Methods II* Winter 2015
- HGEN 46900 *Human Variation & Disease* Spring 2015
- STAT 35500 *Statistical Genetics* Spring 2015
- myChoice Mini-Course: *Effective Writing in the Biological Sciences* Fall 2015
- HGEN 48600 *Fundamentals of Computational Biology: Models & Inference* Winter 2016
- PBHS 31831 *Genetic & Molecular Epidemiology* Spring 2016

## Additional Workshops & Conferences

- ComSciCon Chicago (Communicating Science Conference & Workshop) August 2017
- Gordon Research Seminar & Conference: Human Genetics & Genomics July 2017  
Discussion Leader for mentorship session at GRS
- Summer Institute in Statistical Genetics at the University of Washington July 2016
- Master R Developer Workshop taught by Hadley Wickham May 2015

## Extracurricular

- Museum of Science & Industry: Science Connections volunteer Fall 2014-current  
Introduce genetic concepts to guests in a fun and engaging way, incorporating hands-on activities. Assist in the Fabrication Lab helping guests design and print custom objects using 3D printers and laser cutters.
- myCHOICE Data Science Trek to San Francisco Bay Area November 2017  
Visit PhD alumni at 23andMe, Genentech, Ancestry, Illumina, Genomic Health, etc.
- UChicago Software Carpentry Helper Fall 2015, 2016, & 2017
- Expanding Your Horizons (EYH) Chicago: volunteer March 2017  
Engage, inspire, and empower young girls to pursue STEM careers at a one-day STEM symposium where 300 middle school girls participate in hands-on science, technology, engineering and math led by academic and professional women.
- myCHOICE Internship: Institute of Translational Medicine: writer Summer 2015  
Translate complex research into dynamic science stories. Share translational research stories in weekly newsletter, ITM website, and social media platforms

## Since my last committee meeting:

**AIM 1** To identify and characterize parent of origin effects on quantitative traits in the Hutterites.

Completed with preprint on bioRxiv<sup>4</sup>, working on revising and resubmitting. We are working on replicating the significant opposite effects of SNPs.

**AIM 2a** To identify and characterize parent of origin effects on gene expression in 306 Hutterites.

Manuscript draft on imprinted genes identified in LCLs completed. Includes replication of genes in whole blood gene expression and whole blood methylation.

**AIM 2b** To identify and characterize parent of origin and allele specific effects on gene expression in 306 Hutterites.

I first performed opposite effect POeQTLs (similar method as in Aim 1) on total LCL gene expression. We subsetted on SNPs with at least three individuals in at least three genotype groups (out of four if you call parent of origin) and found no significant associations (Bonferonni p-value cutoff 1e-07).

I then tested maternally inherited SNPs with the maternal gene expression and paternally inherited SNPs with paternal expression in *cis*. I'll share results at the meeting.

## References

- [1] Gamazon, E. R., Wheeler, H. E., Shah, K. P., Mozaffari, S. V., Aquino-Michaels, K., Carroll, R. J., Eyler, A. E., Denny, J. C., Nicolae, D. L., Cox, N. J., and Im, H. K. (2015). A gene-based association method for mapping traits using reference transcriptome data. *Nature Genetics*, 47(9):1091–1098.
- [2] Hart, M. W., Stover, D. A., Guerra, V., Mozaffari, S. V., Ober, C., Mugal, C. F., and Kaj, I. (2018). Positive selection on human gamete-recognition genes. *PeerJ*, 6(1):e4259–35.
- [3] Igartua, C., Mozaffari, S. V., Nicolae, D. L., and Ober, C. (2017). Rare non-coding variants are associated with plasma lipid traits in a founder population. *Scientific Reports*, 7(1):16415.
- [4] Mozaffari, S. V., DeCara, J. M., Shah, S. J., Lang, R. M., Nicolae, D. L., and Ober, C. (2017). Parent of Origin Effects on Quantitative Phenotypes in a Founder Population. *bioRxiv*.
- [5] Mozaffari, S. V., Stein, M. M., Magnaye, K. M., Nicolae, D. L., and Ober, C. (2018). Gene Expression and Methylation of Imprinted Genes in the Hutterites.
- [6] Shiquan, S., Zhu, J., Mozaffari, S. V., Ober, C., Chen, M., and Zhou, X. (2018). Heritability Estimation and Differential Analysis with Generalized Linear Mixed Models in RNA Sequencing and Bisulfite Sequencing Studies.