

I. PERSONAL

Name: **Ren-Hua Chung (鍾仁華), Ph.D.**

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Home Address: 11367 NW 83 Way, Doral, FL, 33178

Current Academic Rank: Assistant Professor (Research track)

Primary Department: Dr. John T. Macdonald Foundation Department
of Human Genetics

Webpage: <http://hihq.med.miami.edu/find-a-doctor/profile?id=95484&submit=true>

Citizenship: Taiwan

II. HIGHER EDUCATION

North Carolina State University Raleigh, NC	Ph.D. Bionformatics, minor in Statistics	2006
University of California at Davis Davis, CA	M.S. Computer Science	2003
National Chiao-Tung University Hsin-Chu, Taiwan	B.S. Computer Science	2000

III. EXPERIENCE

<u>Institution:</u>	<u>Rank/Status:</u>	<u>Dates:</u>
University of Miami Leonard M. Miller School of Medicine Dr. John T. Macdonald Foundation Department of Human Genetics John P. Hussman Institute for Human Genomics Center for Genetic Epidemiology and Statistical Genetics	Assistant Professor	2008 - present

Center for Human Genetics Duke University Medical Center Durham, NC	Postdoctoral Fellow	2007 - 2008
Center for Human Genetics Duke University Medical Center Durham, NC	Internship	2003 - 2006

IV. PUBLICATIONS

Book Chapters and monographs published:

Chung R-H, Martin ER. Chapter 12: Linkage disequilibrium and association analysis. Genetic analysis of complex diseases, 3rd edition. In preparation. Wiley-Liss, USA.

Chung R-H, Martin ER. Chapter 19: Single marker family-based association analysis conditional on parental information. In Press. Statistical genetics in the methods of molecular biology series, Humana Press, USA.

Refereed journals:

1. **Chung R-H**, Schmidt MA, Martin ER. CAPL: an efficient association software package using family and case-control data and accounting for population stratification. *BMC Bioinformatics*, 2011. Published Online: May 25: 12-201.
2. Hussman JP, **Chung R-H**, Griswold AJ, Jaworski JM, Salyakina D, Ma D, Konidari I, Whitehead P, Vance JM, Martin ER, Cuccaro ML, Gilbert JR, Haines JL, Pericak-Vance MA. A Noise-Reduction GWAS Analysis Implicates Altered Regulation of Neurite Outgrowth and Guidance in Autism. *Molecular Autism*, 2011 Jan 19;2(1):1.
3. **Chung R-H**, Schmidt MA, Morris RW, Martin ER. CAPL: A Novel Association Test Using Case-Control and Family Data and Accounting for Population Stratification. *Genetic Epidemiology*, 2010 34(7):747-55.
4. **Chung R-H**, Schmidt S, Martin ER, Hauser ER. Ordered subset analysis (OSA) for family-based association mapping of complex traits. *Genetic Epidemiology*, 2008 32 (7):627-37.
5. Zhang L, Martin ER, **Chung R-H**, Li Y-J, Morris RW. X-LRT: A likelihood approach to estimate genetic risks and test association with X-linked markers using a case-parents design. *Genetic Epidemiology*. 2008 32(4):370-380.
6. **Chung R-H**, Morris RW, Martin ER. Response to "XMCPDT does have correct type I error rates" by Jie Ding and Shili Lin. *The American Journal of Human Genetics*. 2008 82(2):530-531.
7. **Chung R-H**, Morris RW, Zhang L, Li Y-J, Martin ER. X-APL: An improved family-based test of association for the X chromosome. *The American Journal of Human Genetics* 2007 80(1): 59-68.
8. **Chung R-H**, Hauser ER, Martin ER. Interpretation of simultaneous linkage and family-based association tests in genome screens. *Genetic Epidemiology* 2007 31:134-142.
9. **Chung R-H**, Hauser ER, Martin ER. The APL test: Extension to general nuclear families and haplotypes and examination of its robustness. *Human Heredity* 2006 61(4):189-199.

10. Martin ER, Bronson PG, Li YJ, Wall N, **Chung R-H**, Schmechel DE, Small G, Xu PT, Bartlett J, Schnetz-Boutaud N, Haines JL, Gilbert JR, Pericak-Vance MA. Interaction between the alpha-T catenin gene (VR22) and APOE in Alzheimer's disease. *Journal of Medical Genetics* 2005 42(10):787-792.
11. **Chung R-H**, Gusfield D. Perfect phylogeny haplotyper: haplotype inferral using a tree model. *Bioinformatics*. 2003 19(6): 780-781.

Submitted:

1. **Chung RH**, Ma D, Wang K, Hedges DJ, Jaworski JM, Gilbert JR, Cuccaro ML, Wright HH, Abrahamson RK, Konidari I, Whitehead PL, Schellenberg GD, Hakonarson H, Haines JL, Pericak-Vance MA, Martin ER. An X-chromosome-wide association study in autism families identifies a novel autism gene TBL1X. *Molecular Autism*, 2011. Submitted.
2. **Chung RH**, Chen YE. A two-stage random forest-based pathway analysis method. *PLoS One*. 2011. Submitted.

In preparation:

1. **Chung RH**, Scott WK, Vance JM, Martin ER. A novel analysis method based on gene-gene interactions in pathways defined by protein-protein interaction networks. Invited article for the *Frontier in Statistical Genetics and Methodology*.
2. Park YS, Schmidt MA, Martin ER, Whitehead PW, Konidari I, Cuccaro ML, Haines JL, Pericak-Vance MA, **Chung RH**. A novel set of genetic variants associated with autism spectrum disorder (ASD) susceptibility is revealed by application of the Pathway-PDT.

Other works, publications and abstracts:

1. **Chung RH**, Gusfield D. Empirical exploration of perfect phylogeny haplotyping and haplotypers. COCOON'03 Proceedings of the 9th annual international conference on Computing and combinatorics. 2003. Page 15.

Refereed abstracts:

1. **Chung R-H**, Ma D, Wang K, Hedges DJ, Jaworski JM, Gilbert JR, Cuccaro ML, Wright HH, Abrahamson RK, Konidari I, Whitehead PL, Schellenberg GD, Hakonarson H, Haines JL, Pericak-Vance MA, Martin ER. An X-chromosome-wide association study identifies a novel autism spectrum disorder gene. 60th Annual Meeting of the American Society of Human Genetics (ASHG), Washington, DC, Nov. 2-6, 2010.
2. Schmidt M A, Kinnamon DD, Powell EH, Beecham GW, **Chung R-H**, Martin ER. Tests for association in next-generation sequence data with uncertain genotypes. 60th Annual Meeting of the American Society of Human Genetics (ASHG), Washington, DC, Nov. 2-6, 2010, (abstract accepted for platform presentation).
3. Ma DQ, Griswold AJ, Cukier H, Schmidt MA, **Chung R**, Kinnamon DD, Arpit M, Ulloa R; Jaworski J, Salyakina D, Nations LD, Konidari I, Whitehead P, Wright HH, Abramson RK, Williams SM, Menon R, Edwards EY; Martin ER, Haines JL, Gilbert JR, Cuccaro ML, Pericak-Vance MA. Evaluation of copy number variations in autism spectrum disorders. 60th Annual Meeting of the American Society of Human Genetics (ASHG), Washington, DC, Nov. 2-6, 2010.
4. **Chung R-H**, Edwards TL, Scott WK, Almonte C, Burt A, Powell EH, Beecham GW, Konidari I, Pericak-Vance MA, Haines JL, Zuchner S, Wang G, Wang L, Vance JM, Martin ER. Predictive modeling for Parkinson Disease. 59th Annual Meeting of the American Society of Human Genetics (ASHG), Honolulu, Hawaii, Oct. 20-24, 2009.

5. Martin, ER, **Chung R-H**, MA DQ, Jaworski JM, Gilbert JR, Hedges DJ, Hoffman J, Anderson A, Konidari I, Abramson RK, Wright HH, Cuccaro ML, Haines JL, Pericak-Vance MA. Family-based association study of the X chromosome reveals ASD genes. 8th Annual International Meeting for Autism Research (IMFAR), Chicago, IL., May 7-9, 2009.
6. Schmidt MA, Martin ER, **Chung R-H**. Accommodating Population Structure in the APL Test. 58th Annual Meeting of the American Society of Human Genetics (ASHG), Philadelphia, Pennsylvania, Nov. 11 – 15, 2008.
7. Martin ER, **Chung R-H**, Ma DQ, Jaworski J, Gilbert JR, Hedges D, Cuccaro ML, Pericak-Vance MA. A first look at the X chromosome in autism spectrum disorder through GWAS eyes. 58th Annual Meeting of the American Society of Human Genetics (ASHG), Philadelphia, Pennsylvania, Nov. 11 – 15, 2008.
8. **Chung R-H**, Morris RW, Schmidt MA, Martin ER. CAPL: An association test combining nuclear family and case-control data. 58th Annual Meeting of the American Society of Human Genetics (ASHG), Philadelphia, Pennsylvania, Nov. 11 – 15, 2008.
9. **Chung, R-H**, Morris RW, Zhang L, Li YJ, Martin ER. X-APL: A family-based association test for the X chromosome. National Health Research Institute, Zhunan, Taiwan, 2007 (platform).
10. **Chung R-H**, Schmidt S, Qin X, Lou X, Martin ER, Hauser ER. Ordered subset Analysis for Association Mapping. 57th Annual Meeting of the American Society of Human Genetics (ASHG) San Diego, CA, October 23-27, 2007. (platform).
11. **Chung, R-H**, Morris RW, Zhang L, Li YJ, Martin ER. X-APL: A family-based association test for the X chromosome. (2315) 56th Annual Meeting of the American Society of Human Genetics (ASHG), New Orleans, Louisiana, October 9 -13, 2006.
12. Zhang L, Morris R, **Chung R-H**, Martin ER, LI Y-J. Likelihood ratio tests of association for X-linked QTL in family-based designs. (2214) 56th Annual Meeting of the American Society of Human Genetics (ASHG), New Orleans, Louisiana, October 9 -13, 2006.
13. **Chung, R-H**, Martin, ER, Family-based association analysis in the presence of linkage (APL) with missing parental genotypes: A computer program for single-locus and haplotype tests. 55th Annual Meeting of the American Society of Human Genetics, Salt Lake City, UT, October 24-29, 2005.
14. Martin ER, Bronson P, **Chung R-H**, Xu P-T, Haines JL, Gilbert JR, Pericak-Vance MA. Association of ACE polymorphisms and Alzheimer disease. 54th Annual Meeting of the American Society of Human Genetics, Toronto, Canada, October 26-30, 2004.
15. Bronson PG, Quan H, **Chung R-H**, Bartlett J, Haines JL, Schmechel D, Small G, Xu PT, Gilbert J, Pericak-Vance MA, Martin ER. Significant association and linkage for Alzheimer disease and SNPs in the VR22 and LRRTM3 genes on chromosome 10q. 54th Annual Meeting of the American Society of Human Genetics, Toronto, Canada, October 26-30, 2004.

V. PROFESSIONAL

Funded research performed in the past five years:

<u>Present:</u> Name of Grant (Funding Agency, Number, Title)	Role in Grant (PI, CO-PI)	Effort in calendar months	Amount of Award	Duration

1. (Chung R-H) Alzheimer's Association <i>"A novel statistical method using case-control and family data"</i>	PI	2.04	\$79,006	07/01/09 to 08/31/11
2. 5RC2HG005605-02 (Martin, ER) NIH/NHGRI <i>"Statistical Methods for Next-Gen Sequencing in Disease Association Studies"</i> This project will develop new statistical approaches for analysis of next-generation sequence data which is currently being generated to study genetic variation underlying important human disease traits. Our approaches will improve statistical techniques for identifying sequence variation in individuals and for relating specific variants to disease status. Our methods have the potential to identify functionally important variants that can be used in diagnosis and prediction and as a basis for novel translational therapies for human genetic diseases.	Co-investigator	2.4	\$1,000,000	09/30/09 to 07/31/11

Pending: Name of Grant (Funding Agency, Number, Title)	Role in Grant (PI, Co-PI)	Effort in calendar months	Amount of Award	Duration
1. R03 (Chung RH) NIH <i>"A novel comprehensive pathway-based analysis of breast cancer risk and prognosis"</i> Identifying pathways associated with breast cancer can help researchers understand disease mechanism, provide therapeutic targets, and potentially improve diagnosis and prediction of breast cancer.	PI	1.2	\$153,001	4/1/12 to 3/31/14
2. R21 (Chung RH) NIH <i>"Family-Based Pathway Association Analysis for Next Generation Sequencing Data"</i> The proposed research is relevant to public health because identifying pathways associated with complex diseases can help researchers understand disease mechanism, provide therapeutic targets, and potentially improve diagnosis and prediction of disease.	PI	3.6	\$420,750	4/1/12 to 3/31/14

<p>3. U01 (Martin ER) NIH <i>"Integrated Software for Genotype Calling and Association Testing in Sequence Data"</i> The proposed research is relevant to human health because it will speed discovery of genes underlying genetic diseases by providing a comprehensive analysis package for NGS data that is accessible to the broad research community. Ultimately this will aid in diagnosis and treatment for human genetic diseases, reducing the burden of human disability</p>	Co-investigator	1.8	\$2,973,764	12/01/11 to 11/30/15
<p>4. R01 (Gilbert, JR/Martin,ER/Mash,D) NIH <i>"Transcription and Epigenetics in Alzheimers Disease"</i> The ultimate aim is to understand AD related abnormalities at the network level to both understand underlying disease etiology and potentially identify points of intervention in molecular and biochemical pathways for potential therapeutic approaches.</p>	Co-investigator	1.2 (1 st yr.) 2.4 (2 nd -3 rd yr.) 6.0 (5 th yr.)	\$3,515,341	04/01/11 to 03/31/16

Past: Name of Grant (Funding Agency, Number, Title)	Role in Grant (PI, CO-PI)	Effort in calendar months	Amount of Award	Duration
<p>1. (Chung R-H) Stanley J. Glaser Foundation Research Award <i>"A novel statistical method using case-control and family data with consideration of population stratification"</i></p>	PI	No effort requested	\$45,000	06/01/09 to 08/31/10 (no cost extension)
<p>2. R01 MH059528-07 (Hauser, ER) NIH/NIMH <i>"Software for Integrated Linkage and Association Analysis"</i> The goals of this grant are to develop and extend software for the analysis of complex genetic traits. Each newly developed method will be examined with simulation studies using SIMLA, a general software package for simulation of complex genetic diseases, also developed under this grant.</p>	Other significant contributor	.48	\$170,673	07/01/00 to 11/30/09

<p>3. R01 NS051355-03 (Martin, ER) NIH <i>“Statistical tests for association with X-linked genes”</i> The goal of this grant is to develop family-based tests of association for markers on the X chromosome. Methods for dichotomous and quantitative traits will be developed and these will be applied to Autism, Parkinson disease and cardiovascular disease.</p>	<p>Other significant contributor</p>	<p>2.76</p>	<p>\$234,707</p>	<p>04/01/05 to 03/31/10</p>
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Current research projects:

1. *Development of statistical methods for pathway analysis:* I have developed Pathway-PDT, which is a powerful family-based pathway analysis tool, and PUPPI, which accounts for gene-gene interactions based on protein-protein interaction network. Pathway-PDT identified a candidate pathway (GO: 32103; positive regulation of response to external stimulus) for Autism Spectrum Disorder that survived the stringent permutation-based multiple testing correction based on a family-based ASD GWAS dataset. PUPPI identified the Parkinson disease pathway defined in the pathway database KEGG (hsa05012) as the top hit based on a case-control Parkinson GWAS dataset.
2. *Statistical method development for next-generation sequencing data:* I am working with Dr. Eden Martin, the director of Center for Genetic Epidemiology and Statistical Genetics, for the development of statistical methods for next-generation sequencing (NGS) data. Since the NGS technique will be widely applied to generate data for disease studies, it is important to develop powerful statistical tools to analyze such data.
3. *Autism research:* I am advising a PhD student to conduct pathway analysis for autism. I am also involved in the autism next-generation sequencing project. The goal of the project is to sequence 1000 unrelated cases and 1000 unrelated controls in candidate regions to identify rare variants associated with autism.
4. *Breast cancer analysis:* I am collaborating with Dr. Jennifer Hu at the Sylvester Cancer Center at the University of Miami for breast cancer analysis. Non-synonymous SNPs (nsSNPs), including rare and common variants selected from genes in candidate pathways with functions of DNA repair, immune functions and candidate genes from previous GWAS studies, were genotyped. Methods that can accommodate both rare and common variants will be developed to analyze the dataset.
5. *Parallelization of statistical algorithms:* Statistical algorithms for disease association studies are usually computationally intense, as the datasets are large. We are applying parallel algorithms to our methods to distribute the code to a cluster of computers for efficient computations.

Editorial responsibilities:

2010 Review Editor, *Frontiers in Statistical Genetics and Methodology*, Frontiers Research Foundation, Lausanne, Switzerland

Professional and Honorary Organizations:

American Society for Human Genetics (ASHG) 2004 - present
 American Statistical Association (ASA) 2010-present

Post-doctoral Fellowships:

2007-2008 Postdoctoral Fellow, Center for Human Genetics, Duke University Medical Center, Durham, N.C

Consultant appointments:

1/27/2011 Dr. Deborah Mash, University of Miami Miller School of Medicine. Analyze Sequenom SNPs run in cocaine study.

3/15/2011 Dr. Jennifer Hu, University of Miami Miller School of Medicine. Analyze subset of Omni2.5 SNPs

for prostate cancer study of toxicity outcome

- 3/23/2011 Dr. Jennifer Hu, University of Miami Miller School of Medicine. Adjusted logistic regression analysis on SNPs in candidate pathways and genes for breast cancer
- 6/8/2011 Dr. Kathleen Egan, Moffitt Cancer Center. Pilot study of 8 samples genotyped on Illumina Omni2.5M platform.
- 6/16/2011 Dr. Xiong Li, Maine Institute for Human Genetics and Health. CNV and LOH analysis on normal and gene-modified cell clones.

Grant and Proposal reviewer:

2010 Grant reviewer, Dystonia Medical Research Foundation

Journal reviewer:

American Journal of Human Genetics
Annals of Human Genetics
American Journal of Epidemiology
Genetic Epidemiology
Heredity
International Journal of Neuroscience
Statistics in Medicine

Invited Speaker, Recent years only:

- 2011 Invited Speaker, "Pathway-PDT: a novel family-based pathway analysis method" 2011. Joint Statistical Meeting, Miami Beach conference center, Miami, FL.
- 2011 Invited Speaker, "Family-based pathway association test for GWAS" 2011 Emerging Information and Technology Conference (EITC), University of Chicago, Chicago, Ill.
- 2009 Invited Speaker, "A combined approach using case-control and family data with consideration of population stratification" 2010 Emerging Information and Technology Conference (EITC), MIT, Cambridge, MA.
- 2006 Invited Speaker, "Exploring correlation between tests of linkage and association in families". Research Conference, Center for Human Genetics, Duke University Medical Center, Durham, N.C.
- 2005 Invited Speaker, "Family-based association study in the presence of linkage". Research Conference, Center for Human Genetics, Duke University Medical Center, Durham, N.C.
- 2005 Invited Speaker, "Family-based association analysis on the X chromosome". Bioinformatics Research Center Retreat, Atlantic Beach, N.C.
- 2004 Invited Speaker, "Family-based association study in the presence of linkage". Bioinformatics Research Center Retreat, Smokey Mountain, N.C.

VI. TEACHING

Teaching Specialization:

2011 Instructor: HGG 631 Human Genetics II, Genes in Population, Spring 2011

Student Advising:

9/1/2009 - 12/1/2009 Debashis Biswas, MS, Department of Computer Science, University of Miami

7/1/2010 - Now YoSon Park, PhD student, Dr. John T. Macdonald Foundation Department of Human Genetics, University of Miami Miller School of Medicine

VII. SERVICE

Graduate Committees: (Last Two Years)

YoSon Park, PhD student, Dr. John T. Macdonald Foundation Department of Human Genetics, University of Miami Miller School of Medicine

**Administrative Responsibilities:
University of Miami**

2011 – present Division Director, Division of Statistical Genetics in the Center for Genetic Epidemiology and Statistical Genetics, University of Miami, Leonard M. Miller School of Medicine, Dr. John T. Macdonald Foundation Department of Human Genetics, John P. Hussman Institute for Human Genomics, Center for Genetic Epidemiology and Statistical Genetics.

2008- present Assistant Professor, University of Miami, Leonard M. Miller School of Medicine, Dr. John T. Macdonald Foundation Department of Human Genetics, John P. Hussman Institute for Human Genomics, Center for Genetic Epidemiology and Statistical Genetics.