

Curriculum Vitae

Name: Hilary Chenevix Martin

Date of birth: 26 July, 1988

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Education

October 2011 to present – University of Oxford: D.Phil. in Genetics under the supervision of Prof. Peter Donnelly. Intended title: “Applications of whole genome sequencing in medical and population genetics”. Intended completion: October 2015.

February 2007 to July 2011 - University of Queensland: Bachelor of Science, majoring in Genetics. Grade Point Average: 7 (out of 7). First Class Honours.

2001 to 2005 - Brisbane Girls' Grammar School. Overall Position: 1 (awarded to approximately the top 2% of school students in the state).

Awards

2015 - Finalist for the Fitch award from the Society for Molecular Biology and Evolution (to speak in the Fitch symposium at the meeting in Vienna, June 2015)

2012 - Graduate of the Year award from the University of Queensland

2011 - University Medal from the University of Queensland

2011 - Promega Student Award to present my Honours work at the Lorne Genome Conference

2009 - Maude Walker Prize, University of Queensland (awarded to the Bachelor of Science student who achieves the greatest proficiency while enrolled as a second year student)

Scholarships

2013 - Phizackerley Senior Scholarship, awarded to a D.Phil. student in the medical sciences at Balliol College, Oxford

2011-15 - Prize Studentship and Osler Award from the Nuffield Department of Medicine, University of Oxford

2011-14 - Clarendon Award, University of Oxford

2011-14 - Oxford-Australia James Fairfax Award

2010 - Tuition scholarship to attend the Summer Institute of Statistical Genetics, University of Washington

2010 – University of Queensland exchange scholarship to study for a semester at the University of California, Berkeley

2009 - Student scholarship to attend the annual conference of the Genetics Society of Australasia

Publications

1. **Martin HC**, Christ R, Hussin JG, O'Connell J, Gordon S, Mbarek H, Hottenga JJ, McAloney K, Willemsen G, Gasparini P, Pirastu N, Montgomery GW, Navarro P, Soranzo S, Toniolo D, Vitart V, Wilson JF, Marchini J, Boomsma DI, Martin NG, Donnelly P. Multi-cohort analysis of the maternal age effect on recombination. *Nature Communications* (in press).
2. **Martin HC***, Taylor JC*, ..., the WGS500 Consortium, ..., Donnelly P, McVean G. Factors influencing success of clinical genome sequencing across a broad spectrum of disorders. *Nature Genetics* (in press).
3. Kim GE, Kronengold J, Barcia G, Quraishi IH, **Martin HC**, Blair E, Taylor JC, Dulac O, Colleaux L, Nabbout R, Kaczmarek LK. Human slack potassium channel mutations increase positive cooperativity between individual channels. *Cell Rep.*, 9(5):1661-72 (2014).
4. **Martin HC***, Kim GE*, Pagnamenta AT, (25 other authors), Blair E, Donnelly P, Kaczmarek LK, Taylor JC. Clinical whole-genome sequencing in severe early-onset epilepsy reveals new genes and improves molecular diagnosis. *Human Molecular Genetics*, 23(12):3200-11 (2014).
5. Ceroni F, Simpson NH, Francks C, Baird G, Conti-Ramsden G, Clark A, Bolton PF, Hennessy ER, Donnelly P, Bentley DR, **Martin H**; IMGSAC; SLI Consortium; WGS500 Consortium, Parr J, Pagnamenta AT, Maestrini E, Bacchelli E, Fisher SE, Newbury DF. Homozygous microdeletion of exon 5 in ZNF277 in a girl with specific language impairment. *Eur J Hum Genet.*, 22(10):1165-71 (2014).
6. **Martin HC**, Wani S, Steptoe AL, Krishnan K, Nones K, Nourbakhsh E, Vlassov A, Grimmond SM & Cloonan N. Imperfect centered miRNA binding sites are common and can mediate functional repression of target mRNAs. *Genome Biology*, 15(3):R51 (2014).
7. Krishnan K, Steptoe AL, **Martin HC**, Wani S, Nones K, Waddell N, Mariasegaram M, Simpson PT, Lakhani SR, Gabrielli B, Vlassov A, Cloonan N, Grimmond SM. MicroRNA-182-5p targets a network of genes involved in DNA repair. *RNA*, 19(2):230-42 (2013).
8. Krishnan K, Steptoe AL, **Martin HC**, Pattabiraman DR, Nones K, Waddell N, Mariasegaram M, Simpson PT, Lakhani SR, Vlassov A, Grimmond SM, Cloonan N. miR-139-5p is a regulator of metastatic pathways in breast cancer. *RNA*. 19(12):1767-80 (2013).
9. **Martin HC***, Roop JI*, Schraiber JG*, Hsu TY & Brem RB. Evolution of a membrane protein regulon in *Saccharomyces*. *Molecular Biology and Evolution*, 29(7):1747-56 (2012).
10. Cloonan N, Wani S, Xu Q, Gu J, Lea K, Heater S, Barbacioru C, Steptoe AL, **Martin HC**, (16 other authors), Grimmond SM. MicroRNAs and their isomiRs function cooperatively to target common biological pathways. *Genome Biology*, 12(12):R126 (2011).
11. Idaghdour Y, Czika W, Shianna K, Lee H, Visscher PM, **Martin HC**, Miclaus K, Jadallah S, Goldstein DB, Wolfinger RD & Gibson G. Geographical genomics of human leukocyte gene expression variation in southern Morocco. *Nature Genetics*, 42(1):62-7 (2010).

Research Experience

October 2011 to present – I have worked on three major projects in my DPhil:

1. Analysis of clinical whole genome sequence data, focusing on Mendelian neurological diseases and on evaluation of different strategies for analysis and experimental design (papers #2, #3 and #4 above)
2. Meta-analysis of the maternal age effect on meiotic recombination in different human cohorts, including developing and fitting Bayesian hierarchical models (paper #1 above)
3. Analysis of whole genome sequencing data from 58 platypus samples for studies on recombination, population history and sex chromosome evolution (in progress)

July 2010 to July 2011 – I did my 4th year Honours project at the Queensland Centre for Medical Genomics, under the supervision of Prof. Sean Grimmond and Dr. Nicole Cloonan. The focus of my research was on the determination of microRNA targets from microarray and high-throughput sequencing data (papers #5, #6, #7 and #9 above).

May 2010 to July 2010 – I worked on a project with Prof. Rachel Brem (Department of Molecular and Cell Biology, University of California, Berkeley. This involved developing nonparametric statistical methods to detect polygenic regulatory evolution in yeast (paper #8 above).

January to May 2010 – I took two graduate-level courses at Berkeley that culminated in independent research projects. One was on estimating transcript-specific expression levels from RNA-seq data, and the other on identifying gene pathways that have been under selection in *Drosophila athabasca*.

January to May 2010 – I worked in Prof. Rasmus Nielsen's group in the Department of Integrative Biology at Berkeley, looking for evidence of natural selection on genes implicated in human behaviour.

September to November 2009 – I investigated the effect of genomic aberrations on *Drosophila* evolution, under the supervision of Dr. Daniel Ortiz-Barrientos (School of Biological Sciences, University of Queensland).

March to August 2009 – I did my third-year research project with Prof. Greg Gibson (School of Biological Sciences, University of Queensland), working on a study of environmental effects on human gene expression (paper #10 above).

November 2008 to February 2009 – Through the Undergraduate Research Program at Cold Spring Harbor Laboratory, I worked in Prof. Bruce Stillman's lab, which studies DNA replication.

February to November 2008 – I undertook a project in Prof. Linda Richards' lab at the Queensland Brain Institute, attempting to map the insertion site of a construct in a strain of knockout mice.

Conference Presentations

May 2015 – Biology of Genomes, Cold Spring Harbor

Talk: "Insights into recombination and sex chromosome evolution from whole-genome sequencing of the platypus"

November 2014 - American Society of Human Genetics meeting, San Diego
Poster: "Bayesian analysis of the maternal age effect on recombination in multiple cohorts."

May 2014 – Quantitative Genomics meeting, London
Talk: "Insights into platypus population history from whole-genome sequence data."

July 2013 – Society for Molecular Biology and Evolution meeting, Chicago
Poster: "An early look at platypus population genetics using whole-genome sequencing."

November 2012 – American Society of Human Genetics meeting, San Francisco
Poster: "Whole-genome sequencing identifies mutations in known and novel genes for early infantile epileptic encephalopathy."

October 2011 – International Congress of Human Genetics, Montreal
Poster: "High-throughput identification of direct miRNA targets."

April 2011 – Australasian Human Gene Mapping Meeting, Hobart
Talk: "Redefining miRNA-mRNA interactions."

February 2011 – Lorne Genome Conference
Talk: "Redefining the rules of miRNA-mRNA interactions"
Poster: "Transcriptional rewiring of a membrane protein regulon in *Saccharomyces*."

October 2009 – American Society of Human Genetics meeting, Honolulu
Poster: "Geographical genomics of expression variation in southern Morocco."

Related Academic Activities

December 2012 to May 2013 – helped organise a student conference in Quantitative Genomics at the Academy of Medical Sciences in London

2012 to 2013 – member of the students and postdocs committee at the Wellcome Trust Centre for Human Genetics; helped organise a Python course, a Careers seminar series, and social events

Computational skills

Programming in Perl, R and bash – competent

Programming in Python – basic

Familiarity with commonly used software for genetic analysis e.g. samtools, GATK, Picard, plink, vcftools

Undergraduate Course Results

Third Year:

At University of California, Berkeley (on the reciprocity program):

STAT245F Statistical Genomics

A+

MCB247 Genome Project Laboratory

A+

INTEG199 Independent Study Project (Nielsen lab)

Pass (Pass/Fail Course)

All other courses were taken at the University of Queensland. Grades range from 1=lowest to 7=highest.

SCIE3017 Research Project (Gibson Lab)	7
GNET3001 Quantitative Genetics	7
STAT3001 Mathematical Statistics	7
MATH2000 Calculus and Linear Algebra II	7

Second Year:

BIOC2012 Biochemistry: Molecular Components of Cells	7
BIOL2009 Genetics I: Molecular Genetics	7
MATH2210 Computational Biology	7
STAT2003 Probability and Statistics	7
BIOC3003 Human Molecular Genetics and Disease	7
BIOL2007 Genetics II: Organismal Genetics	7
BIOL2014 Immunology and Infectious Disease	7
STAT2004 Statistical Modeling and Analysis	7
SCIE2017 Research Project (Richards Lab)	Pass (Pass/Fail Course)

First Year:

MATH 1051 Calculus and Linear Algebra	7
PHYS1001 Mechanics and Thermal Physics I	7
CHEM1020 General Chemistry	7
BIOL1011 Genetics and Evolution	7
MATH1052 Multivariate Calculus and Ordinary Differential Equations	7
PHYS1002 Electromagnetism, Optics, Relativity and Quantum Physics I	7
BIOL1014 Molecular and Microbial Biology	7
BIOL1017 Perspectives in Science	Pass (Pass/Fail Course)

Referees:

Prof. Peter Donnelly – D.Phil. supervisor
Director, Wellcome Trust Centre for Human Genetics
Professor of Statistical Science, University of Oxford
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Prof. Gil McVean – supervised the clinical genome sequencing component of my D.Phil. work
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Prof. Sean Grimmond – Honours supervisor
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