

CURRICULUM VITAE

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EDUCATION AND TRAINING

1986, H.S. Lansing High School
Lansing, New York

1990, B.A. Cornell University (Biological Sciences)
Ithaca, New York

January-
June 1989 University of Sussex
Falmer, England

1997, Ph.D. University of Michigan Medical School (Human Genetics)
Department of Human Genetics
Ann Arbor, Michigan

1997-1998 Post-doctoral research in the laboratory of Dr. Jeffrey Innis
Department of Human Genetics
University of Michigan
Co-mentor: Zhi Chen, Assistant Professor
Department of Internal Medicine

1998-2002 Postdoctoral research in the laboratory of Dr. David Kingsley
Department of Developmental Biology
Stanford University

PROFESSIONAL EMPLOYMENT

Academic Appointments

- 2002-2010 Assistant Professor
 Center for Human Genetics Research
 Department of Molecular Physiology and Biophysics
 and Department of Pediatrics
 Vanderbilt University School of Medicine
- 2010-Present Research Assistant Professor
 Center for Human Genetics Research
 Department of Molecular Physiology and Biophysics
 and Department of Pediatrics
 Vanderbilt University School of Medicine

Other Professional Positions

- 1987-1990 Research technician for Dr. Richard T. Roush
 Department of Entomology
 Cornell University
- 1991 Research technician for Dr. Maja Suter
 Department of Pathology
 Cornell College of Veterinary Medicine and
 Dr. Elizabeth B. Keller
 Section of Biochemistry
 Cornell University

PROFESSIONAL ORGANIZATIONS

Societies

- Member American Society of Human Genetics
Member American Society for Bone and Mineral Research

PROFESSIONAL ACTIVITIES: INTRAMURAL

Affiliations: Vanderbilt Interdepartmental Programs

- Investigator Center for Human Genetics Research
Member Developmental Biology Program
Member Vanderbilt Center for Stem Cell Biology

- 2003 Organizer, 4th Annual Genetics Symposium
Sponsored by the Vanderbilt-Meharry Alliance and Center for
Genetics and Health Policy
Theme: *Mouse Models of Complex Disease*
Invited Speakers: Dr. Joe Nadeau and Dr. Oliver Smithies
Meharry Medical College
Nashville, TN
- 2003-2004 Center for Human Genetics Research Core Oversight Board
Vanderbilt University Medical Center, Nashville, TN
- 2003-2006 Faculty Director
DNA Resources Core
Vanderbilt Center for Human Genetics Research
Nashville, TN
- 2004-Present Oversight Committee
Vanderbilt Ph.D. Program in Human Genetics
- 2008-2010 Faculty Search Committee
Vanderbilt Center for Human Genetics Research
- 2008-2015 Scientific Co-Director
Vanderbilt Transgenic Mouse / ES Cell Shared Resource
<https://labnodes.vanderbilt.edu/tmescsr>
- 2010-2015 Operator/BAC Recombineering Service Provider
Vanderbilt Transgenic Mouse / ES Cell Shared Resource
- 2012-Present Associate Director of Education, Vanderbilt Ph.D. Program in Human
Genetics

PROFESSIONAL ACTIVITIES: EXTRAMURAL

- 2003 Chair, Comparative Genomics Session
Advances in Genome Biology and Technology Annual Meeting
February 4-7, 2004
Marco Island, FL
- 2008 Chair, Musculoskeletal Disease Session
American Society for Human Genetics Annual Meeting
Nov. 11-15, 2008
Philadelphia, PA
- 2010 “Meet-the-Professor” session, American Society for Bone and Mineral
Research Annual Meeting

Curriculum Vitae: DOUGLAS P. MORTLOCK

Oct. 15-19, 2010

Toronto, Canada

2014-2016 Member, American Society for Human Genetics Program Committee

2015 Co-Chair, ASHG Workgroup on Genomic Editing

Ad Hoc Journal Article Reviews

American Journal of Human Genetics, Annals of Human Genetics, BMC Biology, BMC Biotechnology, Computational and Structural Biotechnology Journal, Current Molecular Medicine, Development, Developmental Biology, FEBS Letters, Genesis, Genome Research, Genomics, Molecular and Cellular Biology, Molecular and Cellular Biochemistry, Molecular Biology Reports, Nucleic Acids Research, PeerJ Computational Science, PLOS Genetics

Grant Reviewing

2007 National Science Foundation

2009 National Centre for the Replacement, Refinement and Reduction of Animals in Research (NC3Rs) (United Kingdom)

2009 Vanderbilt Diabetes Research and Training Center (DRTC)

2010 NIH, electronic grant reviewer for ARRA applications

2012 Ad hoc member, NIH Developmental Biology Subcommittee Study Section (CHHD-C)

2013 & 2014 NIH Special Emphasis Panel ZRG1-GGG- Q (PAR Panel: Casual Variants for Autoimmune and Musculoskeletal Diseases)

Conference Abstract Reviewer

2007, 2008, 2014, 2015 American Society of Human Genetics Annual Meeting

HONORS AND AWARDS

1986 Lansing High School Valedictorian Award

Fall 1986,
Spring 1987,
Spring 1988,
& Fall 1988
Dean's List, Cornell University School of Arts and Sciences

1990 Golden Key National Honor Society

May 1990 B.A. with Distinction in All Subjects
Cornell University

- Sept 1992 Howard Kramer Fellowship Award
Department of Human Genetics
University of Michigan
- Sept 1991-
June 1994 NIH Genetics Training Grant Fellowship
- 1994-1995 Cancer Center Fellowship
University of Michigan
- 1995-1996 Research Partnership Program Award
University of Michigan
- 1997-1998 Organogenesis Postdoctoral Fellowship
University of Michigan Center
- 1999-2001 National Research Service Award Postdoctoral Fellowship

TEACHING ACTIVITIES

Vanderbilt intramural teaching contributions

MPB 340: Human Genetics	
Spring 2002	Lectures: (3 hours). Topic: Animal models of human disease, lectures I and II
Spring, Fall 2004	Evaluated student literature discussions (3 hours) Lectures: (1.5 hours). Topic: Introduction to genome browsers
Fall 2005	Lectures: (4 hours)
Fall 2006	Lectures: (4 hours)
Fall 2007	Lectures: (8 hours)
HGEN I / MPB 340/8340: Human Molecular Genetics	
Fall 2008	Course co-organizer with Dr. Marshall Summar. Lectures: (10 hours)
Fall 2009	Course co-organizer with Dr. Marshall Summar. Lectures: (7 hours)
Fall 2010, 2011, 2012, 2013	Course co-organizer with Dr. Shirley Russell. Lectures: (16 hours)
Fall 2014	Course organizer. Lectures: (21 hours).
Fall 2015	Course organizer. Lectures: (32 hours).

HGEN / MPB 385: Fundamentals of Genetic Analysis	
Spring 2012, 2013, 2014	Course organizer. Discussions: (28 hours).
Human Genetics Course, MSCI program (Masters of Science in Clinical Investigation)	
Fall 2006, 2007, 2008, 2009, 2010	Lectures: (3.5-7.5 hours per year)
IGP 300A / Bioregulation I (First year course for IGP program)	
Fall 2002	Lecture: (1 hour) Topic: Using online databases for accessing mouse/human genomic information
Fall 2004	Mouse Developmental Genetics section Lectures: (5 hours). Topics: Introduction to Genetics in the Mouse; Stem Cells & Chimeras; Experimental Manipulation of the Genome; Genetics of Embryonic Patterning Organized a 1-hour "flextime" session (student journal club presentations)
MPB 324: Tutorials in Physiology (seminar course for second-year MPB program graduate students)	
Fall 2003, Spring 2004	Co-supervisor with Drs. Anne Kenworthy and Chao-Lan Yu Organizer, Spring 2004 Contact hours: (14)
Molecular Developmental Biology (CDB 341)	
Spring 2004	Lecturer (1.5 hours) Topic: Mouse embryology and gastrulation
Spring 2006	Instructor, 1-credit module. Topic : Long-range gene regulation in development Contact hours: (12)

Genetics of Model Organisms (MPB 349)	
Spring 2005	Lectures: (2 hours) Topics: Introduction to Mouse Genomics, Genome Browsers and Comparative Genomics
Spring 2009; Spring 2011	Lectures: (3 hours) Topics: Comparative sequence analysis; Evolution of vertebrate genomes; Analysis of regulatory elements.

Extramural teaching contributions: Undergraduate or graduate level

Introductory Genetics Laboratory Course for Undergraduates	
Fall 1993	Teaching Assistant University of Michigan
Molecular Genetics (Human Genetics 541)	
1996, 1997	Guest Lecturer, Dept. of Human Genetics University of Michigan
Genetics and Risk Evaluation Workshop (a continuing education short course for clinicians and counselors)	
July 1997	Division of Hematology and Oncology Department of Internal Medicine University of Michigan Medical School
Project Lab Course (an upper-level course for undergraduates; Dr. Leilani Miller, director)	
Nov. 1999	Guest Lecturer Department of Biology Santa Clara University
Introductory Biology (Dr. Ben Jorge, section leader)	
Oct. 14, 2015	Guest Discussion Participant: Gene Editing Belmont University

Outreach / high school teaching contributions

School for Science and Math at Vanderbilt (On-site program for regional high school students)	
Fall 2007, 2010, 2011,	Lecture and presentation: 1 hour

2012, 2013, 2014	Topic: "Blue genes"
Supervised student visits to Center for Human Genetics laboratories	
Nov. 2002, Feb. 2005	"Research Explorers" high school student evenings. Topic: Scientific research and hands-on lab activities
April, 2012	Supervised Visit to CHGR laboratories for MTSU Masters in Science and Professional Science student group.

Advisory and Supervisory Responsibilities

Postdoctoral Trainees, Mortlock Laboratory

2008-2014 Steven Pregizer, Ph.D.

2014-present. Benjamin Jorge, Ph.D.

Postdoctoral Mentorship Committees

2009-Present Alencia Woodard-Grice
Postdoctoral Research Fellow
Clinical Pharmacology (Division: Medicine)

2010-2013 Anthony J. Baucum
Postdoctoral Research Fellow
Department of Molecular Physiology and Biophysics
Primary Mentor: Roger Colbran, Ph.D.

Doctoral Thesis Students, Mortlock Laboratory

2003-2007 Ronald L. Chandler, Pre-doctoral Student
Department of Molecular Physiology & Biophysics
Doctoral exam completed April 6, 2007

2003-2008 Kelly J. Chandler, Pre-doctoral Student
Department of Molecular Physiology & Biophysics
Doctoral exam completed March 10, 2008

2005-2010 Nykolaus Reed, Pre-doctoral Student
Department of Microbiology (Meharry Medical College)
Doctoral exam completed April 6, 2010

2007-2012 Dawn E. Clendenning, Pre-doctoral student
Human Genetics Ph.D. Program
Doctoral exam completed March 30, 2012

Masters Thesis Students, Mortlock Laboratory

Curriculum Vitae: DOUGLAS P. MORTLOCK

2008-2013 Eva Broeckelmann
 Interdisciplinary degree in Human Genetics
 Masters' thesis completed Spring, 2013

Pre-Doctoral Rotation Students, Mortlock laboratory

Ronald Chandler; Kelly Chandler; Karen McFarland; Nykolaus Reed; Sabrina Mitchell; Jami Day; Jennifer Corpening; Rachel Ostroff; Dawn Foehr; Weiguang Wang; Eva Broeckelmann

Initiative for Minority Student Development Students, Mortlock laboratory

Fall 2004-Spring 2005 Jessica Witherspoon

Vanderbilt Summer Science Academy Students, Mortlock laboratory

Summer 2006 Eva Broeckelmann

Undergraduate researchers, Mortlock laboratory

Spring 2011 Sam Linton

High school volunteer/observers, Mortlock laboratory

Aug 2013 - Aug 2014 Christina Quigley

Chaired Doctoral Thesis Committees

Graduation	Student	Program	Mentor
2005	Angela Eeds	Molecular Physiology & Biophysics	Marshal Summar, M.D.
2007	Kylee Spencer	Human Genetics	Jonathan L. Haines, Ph.D.
2008	Will Bush	Human Genetics	Marylyn Ritchie, Ph.D.
2010	Marcia Schilling	Molecular Physiology & Biophysics	Richard O'Brien, Ph.D.
2010	Sabrina Mitchell	Human Genetics	Marshall Summar, M.D.
2010	Lauren Walters	Human Genetics	Michelle Southard-Smith, Ph.D.
In progress	David Rinker	Human Genetics	Lawrence Zwiebel, Ph.D.
In progress	Nuri Kodaman	Human Genetics	Scott Williams, Ph.D. and Melinda Aldrich, Ph.D.
In progress	Alex Fish	Human Genetics	Will Bush, Ph.D. and Tony Capra,

			Ph.D.
In progress	Corrine Simonti	Human Genetics	Tony Capra, Ph.D.

Other Doctoral Thesis Committees

2005	Jake McCauley	Molecular Physiology & Biophysics	James S. Sutcliffe, Ph.D
2005	Shannon Kenealy	Molecular Physiology & Biophysics	Jonathan L. Haines, Ph.D
2007	Elizabeth Tweedie	Molecular Physiology & Biophysics	Maureen Gannon, Ph.D
2007	Sharon (Xueying) Liang	Human Genetics	Jonathan L. Haines, Ph.D.
2007	Alli Antar	Molecular Physiology & Biophysics	Alyssa Hasty, Ph.D.
2008	Jeffrey Raum	Molecular Physiology & Biophysics	Roland Stein, Ph.D.
2009	Christina Koo Speirs	Biological Sciences	Lilianna Solnica-Krezel, Ph.D.
2009	Blairanne Williams	Neuroscience	Aaron Bowman, Ph.D.
2010	Justin Layer	Molecular Physiology & Biophysics	Tony Weil, Ph.D.
2010	Meaghan Neill	Human Genetics	Marshall Summar, M.D.
2010	Na Lian	Pharmacology	Xiangli Yang, Ph.D.
2011	Michelle Guney	Molecular Physiology & Biophysics	Maureen Gannon, Ph.D.
2011	Weiguang Wang	Pharmacology	Xiangli Yang, Ph.D.
2012	Megnan Tian	Pharmacology	Robert Macdonald, M.D., Ph.D.
2013	Jeffrey Bennett, MSTP student	Pharmacology	Dan Roden, MD

In progress	Nicole Halstead	Cell and Developmental Biology	Chris Wright, Ph.D.
2013	Olivia Veatch	Human Genetics	Jonathan L. Haines, Ph.D.
2014	Melissa Musser, MSTP student	Human Genetics	Michelle Southard-Smith, Ph.D.
2014	Nicole Hawkins	Neuroscience	Jennifer Kearney, Ph.D.
2014	Ben Jorge	Neuroscience	Jennifer Kearney, Ph.D.
In progress	Rebecca Levinson	Human Genetics	David Samuels, Ph.D.
In progress	Kristen Syring	Molecular Physiology & Biophysics	Richard O'Brien, Ph.D.

Master's Thesis Committees

2006	Jared Boustead	Molecular Physiology & Biophysics
2006	Brent Anderson	Master of Laboratory Investigation program

RESEARCH FUNDING**Active Funding**

1R01HL114751-01 (Mortlock) 08/23/2012-06/30/2016

Multi-PI grant with Melissa Rogers, UMDNJ.

Administrative PI: Mortlock

NIH/NHLBI

BMP2 Gene Regulation in Calcific Aortic Valve Disease

The goal of this project is to determine critical transcriptional and post-transcriptional regulatory elements that control BMP2 gene expression in a mouse model of calcific aortic valve disease (CAVD).

2R01 ES16931-06A1 (Bowman) 5/7/2014-5/28/2019

NIH/NIEHS

Gene-neurotoxicant Interactions in Huntington Disease

Dr. Mortlock will help oversee the Bowman lab's efforts to do CRISPR-mediated mutagenesis of Huntingtin and key interacting genes, and study their effects on metal toxicity in neuronal cells.

1R01CA188214-01 (Zheng)

09/01/2014-08/31/2019

NIH/NCI

Colorectal cancer risk loci: GWAS, fine-mapping, and functional analysis

Dr. Mortlock's role in this project is to assist with functional analysis of GWAS variants in cell culture, including design of CRISPR-mediated mutagenesis in relevant cell types.

5R01 EY020894 (Kuchtey)

09/01/2010-08/31/2014

NIH/NEI

Identifying a disease gene causing primary open angle glaucoma

The goal of this project is to use CRISPR to engineer null and specific missense mutations into the mouse Adamts10 gene to study the role of this gene in glaucoma.

Completed Funding

5R01 AR049529-04 (Tiller/Mortlock) *

09/01/2002-08/31/2006 *

NIH/NIAMS

The Role of Sedlin in Maintaining Cartilage Integrity

PI from 09/01/02-10/01/05: George Tiller

PI from 10/01/05-08/31/07: Douglas Mortlock

The goal of this project was to characterize the role of the Sedlin gene product in cartilage integrity by analyzing its structure, in vivo expression, interactions with other proteins, effects on expression of other genes, and Sedlin deficiency in animal models.

* (Dr. George Tiller was original P.I. of this grant from 09/01/02 to 11/2/05. Dr. Mortlock then became P.I. of the grant at Dr. Tiller's request.)

5R01 AI044924-05 (P.I: Aune)

07/01/2004- 06/30/2008

NIH/NIAID

IFN-Gamma Gene Regulation in T Cells and NK Cells

The primary goal of this project is to characterize regulatory processes at the DNA level that control restricted IFN-Gamma expression in T cell lineages. The grant supports Dr. Mortlock's role in designing transgenes to identify IFN-Gamma cis-regulatory elements.

3R01HD047880-01S1 (Mortlock) 09/01/2005-08/31/2007
Supplement to 1 R01 HD47880-01

Diversity supplement to fund stipend of Nykolaus Reed, a Predoctoral student in the Mortlock lab.

1R03 NS051695-01A1 (Mortlock) 1/1/2006-12/31/2008
NIH/NIINDS

A Conserved Sequence Approach for MS Association Studies

The goal of this project was to identify genetic variants on chromosome 1q43 associated with multiple sclerosis.

CHGR 2007 Pilot Grant (Mortlock) 11/01/2007-6/30/2008

Vanderbilt Center for Human Genetics Research

Chromatin status of BMP gene deserts during osteoblast differentiation

The goal of this pilot grant is to demonstrate our ability to perform high-density ChIP-on-chip assays that will interrogate the distribution of histone modifications across the *Bmp2* and *Bmp4* genes in osteoblasts.

1R01 HD047880-01 (Mortlock) 01/17/2005-09/29/2010
NIH/NICHHD

Gdf6 Gene Expression and Evolution in Vertebrates

The goal of this project is to compare expression of the Gdf6 gene during skeletal development in mouse and zebrafish, and compare functional roles of conserved Gdf6 cis-regulatory sequences across species, by performing transgenic reporter assays in both systems.

2R01 NS032830-15 (Haines) 09/30/2009- 08/31/2012
NIH/ NINDS

The Genetic Epidemiology of Multiple Sclerosis

The primary goal of this project is to identify the genes underlying multiple sclerosis using a combination of locational and candidate gene approaches. The grant supports the genotyping and statistical analysis involved in the project.

1R21 DA027002-01 (O'Brien) 09/30/2009-08/31/2012

NIH/NIDA

Characterization of Effects of G6PC2 Gene Variants on Transcription and Splicing

The goal of this project is to characterize effects of G6PC2 gene variants on its function and relation to beta cell regulation.

1R01CA136924-01A1 (USC; Coetzee) 02/23/2010-12/30/2012

NIH/NCI

Genomic Enhancers at 8q24 and Prostate Cancer

The goal of this project is to identify distant cis-enhancers at the C-MYC gene region and determine functional effects on C-MYC expression of genetic variants that are associated with human cancers.

5P30 CA068485-13 (Pietenpol) 9/28/2004-08/31/2015

National Cancer Institute

Cancer Center Support Grant

Dr. Mortlock provides scientific guidance to the Core and its users regarding transgene vector design and construction, for purposes of pronuclear injection or gene targeting. Dr. Mortlock has specific expertise in engineering BACs (Bacterial Artificial Chromosomes) for construction of transgene vectors, and the impact of gene structure issues on vector design. Dr. Mortlock also provides oversight and approval for incoming pronuclear injection projects.

3R01 MH095044-03S1 (Blakely) 5/1/2012-4/30/2017

NIMH

Pre-synaptic regulation of *C. elegans* dopamine transporter

The goal of this project is to identify novel and conserved regulators of Dopamine signaling by studying vertebrate homologs of the worm DA transporter. Dr. Mortlock will supervise the CRISPR-mediated mutagenesis of the *Mblac1* gene in mouse embryos.

5R01 MH86530-01A1 (Sarter/Blakely) 3/1/2014-2/28/2015

NIMH

Choline transporter capacity limits motivated behavior on mice, rats and humans

The goal of this project is to perform interdisciplinary research on the regulation

and function of the high-affinity choline transporter (CHT). Dr. Mortlock will supervise the CRISPR-mediated genomic editing of the *ChT* gene in mouse embryos.

3R01 ES10563-13S1 (Aschner/Bowman) 3/4/2014-10/31/2014

NIH/NIEHS

Mechanisms of Manganese Neurotoxicity

Dr. Mortlock will help oversee the Bowman lab's efforts to do CRISPR-mediated mutagenesis of key genes involved in metal toxicity, transport and response in neuronal cells.

LABORATORIES REQUESTING TRANSGENIC MICE GENERATED IN THE MORTLOCK LAB

Bmp2 BAC lines

Dr. Mark deCaestecker, Vanderbilt University, Nashville, TN

Dr. Louis Gerstenfeld, Boston University, Boston, MA

Dr. Stephen Harris, University of Texas Health Science Center, San Antonio, TX

Dr. Richard P. Harvey, Victor Chang Cardiac Research Institute, Sydney, Australia

Dr. Deneen Wellik, University of Michigan, Ann Arbor, MI

Dr. Frolian Granero-Molto, University of Navarra, Pamplona, Spain

Dr. Florent Eleferiou, Vanderbilt University, Nashville, TN

Dr. Jon Shoenecker, Vanderbilt University, Nashville, TN

Bmp4 BAC lines

Dr. Mark deCaestecker, Vanderbilt University, Nashville, TN

Dr. Motoko Yanagita, Kyoto University, Kyoto, Japan

BIBLIOGRAPHY

Peer-Reviewed Research Papers

1. ffrench-Constant RH, Roush RT, **Mortlock D**, and Dively GP. Isolation of dieldrin resistance from field populations of *Drosophila melanogaster* (Diptera: Drosophilidae). *Journal of Economic Entomology* 83(5):1733-1737 (1990).
2. ffrench-Constant RH, **Mortlock DP**, Shaffer CD, MacIntyre RJ, and Roush RT. Molecular cloning and transformation of cyclodiene resistance in *Drosophila*: an invertebrate gamma-aminobutyric acid subtype A receptor locus. *Proceedings of the National Academy of Sciences* 88:7209-7213 (1991).

3. **Mortlock D**, Keller EB, Ziegra CJ, and Suter MJ. High efficiency transfection of monkey kidney COS-1 cells. *Journal of Tissue Culture Methods* 15:176-180 (1993).
4. Bespalova IN, Farjo Q, **Mortlock DP**, Jackson AU, Meisler MH, Swaroop A, and Burmeister M. Mapping of the neural retina leucine zipper gene, Nrl, to mouse chromosome 14. *Mammalian Genome* 4:618-620 (1993).
5. Innis JW, Darling SM, Kazen-Gillespie K, Post LC, **Mortlock DP**, and Yang T. Orientation of the Hoxa complex and placement of the Hd locus distal to Hoxa2 on mouse Chromosome 6. *Mammalian Genome* 7:216-217 (1996).
6. **Mortlock DP**, Nelson MR, and Innis JW. An efficient method for isolating putative promoters and 5'-transcribed sequences from large genomic clones. *Genome Research* 6:327-335 (1996).
7. **Mortlock DP**, Post LC, and Innis JW. The molecular basis of hypodactyly (Hd): a deletion in Hoxa13 leads to arrest of digital arch formation. *Nature Genetics* 13:284-289 (1996).
8. **Mortlock DP** and Innis JW. Mutation in HOXA13 in Hand-Foot-Genital syndrome. *Nature Genetics* 15:179-180 (1997).
9. Innis JW and **Mortlock DP**. Limb development: Molecular dysmorphology is at hand! *Clinical Genetics* 53:337-348 (1998).
10. **Mortlock DP**, Sateesh P, and Innis JW. Evolution of N-terminal sequences of the vertebrate Hoxa13 protein. *Mammalian Genome* 11:151-158 (2000).
11. Goodman FR, Bachelli C, Brady AF, Brueton LA, Fryns J, **Mortlock DP**, Innis JW, Holmes LB, Donnerfeld AE, Feingold M, Beemer FA, Hennekam RCM, and Scambler PJ. Novel HOXA13 mutations and the phenotypic spectrum of Hand-Foot-Genital Syndrome. *American Journal of Human Genetics* 67:192-202 (2000).
12. Innis JW, Goodman FR, Bacchelli C, Williams TM, **Mortlock DP**, Sateesh P, Scambler PJ, McKinnon W, and Guttmacher AE. A HOXA13 allele with a missense mutation in the homeobox and a dinucleotide deletion in the promoter underlies Guttmacher syndrome. *Human Mutation* 19(5):573-4 (2002).
13. **Mortlock DP**, Guenther C, and Kingsley DM. A general approach for identifying distant regulatory elements applied to the Gdf6 gene. *Genome Research* 13(9):2069-81 (2003).
14. Innis JW, **Mortlock D**, Chen Z, Ludwig M, Williams ME, Williams TH, Doyle CD, Shao Z, Glynn M, Mikulic D, Mundlos S, and Utsch B. Polyalanine expansion in HOXA13: Three new affected families and the molecular consequences in a mouse model. *Human Molecular Genetics* 13(22):2841-51 (2004).
15. **Mortlock DP**, Portnoy ME, Chandler RL, NISC Comparative Sequence Program, and Green ED. Comparative sequence analysis of the Gdf6 locus reveals a

- duplicon-mediated chromosomal rearrangement in rodents and rapidly diverging coding and regulatory sequences. *Genomics* 84(5):814-823 (2004).
16. Portnoy ME, McDermott KJ, Antonellis A, Margulies EH, Prasad AB, NISC (NIH Intramural Sequencing Core), Kingsley DM, Green ED, and **Mortlock DP**. Detection of potential Gdf6 regulatory elements by multi-species sequence comparisons and identification of a skeletal joint enhancer. *Genomics* 86(3):295-305 (2005).
 17. Deal KD, Cantrell VA, Chandler RL, Saunders TS, **Mortlock DP**, and Southard-Smith M. Distant regulatory elements in a Sox10betaGEO BAC transgene are required for expression of Sox10 in the enteric nervous system and other neural-crest derived tissues. *Developmental Dynamics* 235(5):1413-1432 (2006).
 18. Eeds AM, **Mortlock DP**, Wade-Martins R, and Summar ML. Assessing the functional characteristics of synonymous and non-synonymous mutation candidates by use of large DNA constructs. *Am. J. Hum. Genet.* 80(4):740-750 (2007).
 19. Chandler RL, Chandler KJ, McFarland KA, and **Mortlock DP**. Bmp2 transcription in differentiating osteoblasts is regulated by a distant 3' enhancer located 156.3 kilobases from the promoter. *Mol. Cell. Biol.* 27(8):2934-2951 (2007).
 20. Spagnoli A, O'Rear L, Chandler RL, **Mortlock DP**, Granero-Molto F, Gorska AE, Longobardi L, Chytli A, Shimer K, and Moses HL. TGF-beta signaling is essential for joint morphogenesis. *J. Cell. Biol.* 177(6):1105-1117 (2007).
 21. McCauley JL, Kenealy SJ, Margulies EH, Schnetz-Boutaud NS, Gregory SG, Hauser SL, Oksenberg JR, Pericak-Vance MA, Haines JL, and **Mortlock DP**. SNPs in Multi-Species Conserved Sequences (MCS) as useful markers in association studies: a practical approach. *BMC Genomics* 8:266 (2007).
 22. Chandler KJ, Chandler RL, Broeckelmann E, Hou Y, Southard-Smith EM, and **Mortlock DP**. Relevance of BAC transgene copy number in mice: Transgene copy number variation across multiple transgenic lines and correlations with transgene integrity and expression. *Mammalian Genome* 18(10):693-708 (2007).
 23. Boyle S, Misfeldt A, Chandler KJ, Deal KK, Southard-Smith EM, **Mortlock DP**, Baldwin HS, and Mark de Caestecker. Fate mapping using Cited1-CreERT2 mice demonstrates that the cap mesenchyme contains self-renewing progenitor cells and gives rise exclusively to nephronic epithelia. *Developmental Biology* 313(1):234-245 (2008). [PMC2699557](#)
 24. Luppen CA, Chandler RL, Noh T, **Mortlock DP**, and Baruch Frenkel. BMP-2 versus BMP-4 expression and activity in glucocorticoid-arrested MC3T3-E1 osteoblasts: smad signaling, not alkaline phosphatase activity, predicts rescue of mineralization. *Growth Factors* 26(4):226-37 (2008). [PMC3760374](#)

25. Chandler KJ, Chandler RL, and **Mortlock DP**. Identification of an ancient *Bmp4* mesoderm enhancer located 49.7 kilobases from the promoter. *Developmental Biology* 327:2, p 590-602 (2009). [PMC2846791](#)
26. Granero-Moltó F, Weis JA, Miga MI, Landis B, Myers TJ, O'Rear L, Longobardi L, Jansen ED, **Mortlock DP**, Spagnoli A. Regenerative Effects of Transplanted Mesenchymal Stem Cells in Fracture Healing. *Stem Cells*. 2009 Aug; 27(8):1887-98. [PMC3426453](#)
27. McCauley JL, Zuvich RL, Bradford Y, Kenealy SJ, Schnetz-Boutaud N, Gregory SG, Hauser SL, Oksenberg JR, **Mortlock DP**, Pericak-Vance MA, Haines JL. Follow-up examination of linkage and association to chromosome 1q43 in multiple sclerosis. *Genes and Immunity*. 2009 Oct; 10(7):624-30. [PMC2765552](#)
28. Pregizer S and **Mortlock DP**. Long-range regulation of BMP family genes. *Cytokine and Growth Factor Reviews*. 2009 Oct-Dec; 20(5-6):509-15. [PMC2787762](#)
29. Anderson L, Lowery JW, Frank DB, Jones M, **Mortlock DP**, Chandler R, de Caestecker M. Opposing effects of Bmp2 and Bmp4 in hypoxic pulmonary hypertension. *American Journal of Physiology – Regulatory, Integrative, and Comparative Physiology*. 2010 Mar; 298(3):R833-42. [PMC2838658](#)
30. Jiang S, Chandler RL, Fritz DT, **Mortlock DP**, Rogers MB. Repressive BMP2 gene regulatory elements near the BMP2 promoter. *Biochemical and Biophysical Research Communications*. 2010 Feb 5; 392(2):124-8. [PMC2822113](#)
31. Reed NP and **Mortlock DP**. Identification of a distant cis-regulatory element controlling pharyngeal arch-specific expression of zebrafish *gdf6a/radar*. *Dev Dyn*. 2010 Apr; 239(4):1047-60. [PMC3110066](#)
32. Collins P, Chang S, Henderson M, Soutto M, Davis G, McLeod A, Townsend M, Glimcher L, **Mortlock D**, Aune T. Distal regions of the human IFNG locus direct cell-type specific expression. *J Immunol*. 2010 Aug 1; 185(3):1492-501. [PMC2923829](#)
33. Kruithof BP, Fritz DT, Liu Y, Garsetti DE, Frank DB, Pregizer SK, Gaussin V, **Mortlock DP**, Rogers MB. An autonomous BMP2 regulatory element in mesenchymal cells. *J Cell Biochem*. 2011 Feb;112(2):666-74. [PMC4198144](#)
34. Zuvich RL, Bush WS, McCauley JL, Beecham AH, De Jager PL, the IMSGC, Ivins AJ, Compston A, Hafler DA, Hauser SL, Sawcer SJ, Pericak-Vance MA, **Mortlock DP**, Barcellos LF, Haines JL. Interrogating the complex role of chromosome 16p13.13 in multiple sclerosis susceptibility: Independent genetic signals in CIITA-CLEC16A-SOCS1 gene complex. *Human Molecular Genetics*. 2011 Sep 1;20(17):3517-24. [PMC3153306](#)
35. Mastsubara H, Hogan DE, Morgan EF, **Mortlock DP**, Einhorn TA, Gerstenfeld

- LC. Vascular tissues are a primary source of BMP2 expression during bone formation induced by distraction osteogenesis. *Bone*. 2012 Jul;51(1):168-80.
36. Clendenning DE, **Mortlock DP**. The BMP Ligand Gdf6 Prevents Differentiation of Coronal Suture Mesenchyme in Early Cranial Development. *PLOS One*. **2012**;7(5):e36789. Epub 2012 May 31. [PMC3365063](#)
37. Ainoya K, Moriguchi T, Ohmori S, Souma T, Takai J, Morita M, Chandler KJ, **Mortlock DP**, Shimizu R, Engel JD, Lim KC, Yamamoto M. UG4 Enhancer-Driven GATA-2 and Bone Morphogenetic Protein 4 Complementation Remedies the CAKUT Phenotype in Gata2 Hypomorphic Mutant Mice. *Mol Cell Biol*. 2012 Jun;32(12):2312-22. [PMC3372261](#)
38. Kamiya N, Shafer S, Oxendine I, Mortlock DP, Chandler RL, Oxburgh L, Kim HK. [Acute BMP2 upregulation following induction of ischemic osteonecrosis in immature femoral head](#). *Bone*. 2012 Dec 3;53(1):239-247. PMC in progress.
39. Marsell, R, Steen, B, Bais, MV, **Mortlock DP**, Einhorn TA, Gerstenfeld LC. [Skeletal trauma generates systemic BMP2 activation that is temporally related to the mobilization of CD73+ cells](#). *J. Orthop. Res*. 2013. Epub ahead of print. PMC in progress.
40. Samuels DC, Li C, Li B, Song Z, Torstenson E, Clay HB, Rokas A, Thornton-Wells TA, Moore JH, Hughes TM, Hoffman RD, Haines JL, Murdock DG, **Mortlock DP**, Williams SM. Recurrent tissue-specific mtDNA mutations are common in humans. *PLOS Genetics*, 2013. Nov. 9(11)e1003929. PMC3820769
41. Swineheart IT, Schlientz AJ, Quintanilla CA, **Mortlock DP**, and Wellik DM. Hox11 genes are required for regional patterning and integration of muscle, tendon and bone. *Development*, 2013 Nov. 140(22):4574-82. PMC3817943.
42. Yutzey KE, Demer LL, Body SC, Huggins GS, Towler D, Giachelli C, Hoffman-Bowman M, **Mortlock D**, Rogers M, Sedeghi MM, Aikawa E. Calcific aortic valve disease: A consensus summary. *Arteriosclerosis, Thrombosis and Vascular Biology*, 2014 Nov;34(11):2387-93. PMC pending.
43. Pregizer SK and **Mortlock DP**. Dynamics and Cellular Localization of Bmp2, Bmp4, and Noggin Transcription in the Postnatal Mouse Skeleton. *J. Bone Miner. Res.*, 2015 Jan;30(1):64-70. PMC pending.

Non-Peer Reviewed Papers/Chapters

1. **Mortlock DP**. Comparative bioinformatics for mouse and human genes: getting started. *Current Protocols in Human Genetics*, Wiley & Sons, 2004.

2. **Pregizer, S and Mortlock DP.** Identifying functional annotations for noncoding genomic sequences. *Current Protocols in Human Genetics*, Wiley & Sons, 2012.

Abstracts

Slide Presentations by Mortlock or Mortlock lab members

1. **Mortlock DP** and Innis JW. Identification of genes within large genomic regions using promoter capture. Slide presentation, 9th International Mouse Genome Conference, November 12-15, 1995, Ann Arbor, MI.
2. **Mortlock DP**, Post LC, and Innis JW. The molecular basis of Hypodactyly (Hd): a deletion in *Hoxa13* leads to arrest of digital arch formation. Slide Presentation, University of Michigan Medical School Department of Pediatrics Annual Research Symposium, September 13, 1996, Ann Arbor, MI.
3. **Mortlock DP** and Innis JW. Reversion to an ancestral uterine phenotype by mutation in the human *HOXA13* gene. Slide presentation, Breakthrough Research Session, Annual Meeting of the American Society of Human Genetics, October 29- November 3, 1996, San Francisco, CA.
4. **Mortlock DP**, Schoor MS, and Kingsley DM. Defining cis-acting regulatory elements by transgenic BAC scanning and comparative sequencing: examples from the GDF gene family. Slide presentation, Genome Sequencing and Biology Meeting, May 9-13 2001, Cold Spring Harbor Laboratory, Cold Spring Harbor, N.Y.
5. **Mortlock DP**, Guenther C, Schoor MS, and Kingsley DM. Defining cis-acting regulatory elements by transgenic BAC scanning and comparative sequencing: examples from the GDF/BMP gene family. Slide presentation, Annual Meeting of the American Society of Human Genetics, October 12-16 2001, San Diego, CA.
6. **Mortlock DP**, Guenther C, and Kingsley DM. Identifying long-range cis-acting regulatory elements of the *Gdf6* gene using BAC deletion analysis and comparative sequencing. Slide presentation, Advances in Genome Biology and Technology Meeting, February 5-8, 2003, Marco Island, FL.
7. Chandler RL, McDermott KJ, McFarland K, and **Mortlock DP**. Functional analysis of a 395-kilobase region surrounding the mouse *Bmp2* gene: in vivo BAC transgenes and comparative analysis. Slide presentation, Annual Meeting of the American Society for Bone and Mineral Research, October 1-5, 2004, Seattle, WA.
8. **Mortlock DP**, Chandler RL, McDermott KJ, and Ramirez L. Gene deserts flanking the *Gdf6* and *Bmp2* genes contain numerous conserved, long-range regulatory enhancers that function during embryonic development. Slide presentation, Annual Meeting of the American Society of Human Genetics, October 26-30, 2004, Toronto, Canada.

9. Chandler RL and **Mortlock DP**. Functional analysis of a distant 3' Bmp2 cis-regulatory region. 2006 Southeast Regional Society for Developmental Biology Meeting, April 7-9, 2006, Nashville, TN.
10. Chandler KJ, Chandler RL, and **Mortlock DP**. Mapping long-range enhancers of Bmp4 and exploring the role of long-range evolutionarily conserved regions flanking Bmp4. Slide presentation, 20th Annual International Mammalian Genome Conference, November 12-15, 2006, Charleston, S.C.
11. Zhao M, Chandler RL, Liu J, Mundy GR, and **Mortlock DP**. Molecular mechanisms for BMP-2 transcription by the Hedgehog pathway. Slide presentation, Annual Meeting of the American Society for Bone and Mineral Research, September 16-19, 2007, Honolulu, HI.
12. **Mortlock DP**. Long-range regulatory elements control expression of Bmp2 and Bmp4. 7th International Conference on Bone Morphogenetic Proteins, July 9-13, 2008, Lake Tahoe, California.
13. Clendenning, D and **Mortlock DP**. Long-range regulatory elements control expression of Bmp2 and Bmp4. 8th International Conference on Bone Morphogenetic Proteins, Sept. 15-18, 2010, Leuven, Belgium.

INVITED PRESENTATIONS

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|----------------|--|
| Nov 15, 2001 | Genetics Interest Group Seminar Series
Program in Human Genetics
Vanderbilt University Medical School
Nashville, Tennessee |
| May 13, 2002 | Joint Seminar Series for the Eric Green and Shawn Burgess
laboratories
National Human Genome Research Institute
Bethesda, Maryland |
| April 17, 2003 | Distant DNA elements that control Gdf6 expression in developing
skeletal joints Rheumatology Seminar Series
Vanderbilt University School of Medicine
Nashville, Tennessee |
| Sept 12, 2005 | Mapping long-range regulatory landscapes around BMP genes
Department of Human Genetics Seminar Series
University of Michigan
Ann Arbor, Michigan |

Curriculum Vitae: DOUGLAS P. MORTLOCK

- Sept 13, 2005 Molecular regulation of synovial joint development
Rheumatology Division Seminar
University of Michigan
Ann Arbor, Michigan
- Oct 28, 2005 Long-range elements that regulate the Gdf6 and Bmp2 genes
Session on Long-Range Regulatory Elements
American Society for Human Genetics Annual Meeting
Salt Lake City, Utah
- Feb 17, 2006 Long-range regulatory sequences control Bmp2 expression in
developing kidney, bone and other tissues
Renal Research Group
Vanderbilt University School of Medicine
Nashville, Tennessee
- May 23, 2006 Mapping long-range regulatory landscapes around BMP genes
Integrative Genomics Seminar Series
Vanderbilt University School of Medicine
Nashville, Tennessee
- June 20, 2006 Mapping long-range regulatory landscapes around BMP genes
Regeneron Pharmaceuticals
Tarrytown, New York
- Feb 1, 2007 Mapping long-range regulatory elements around BMP family genes
Department of Molecular Physiology and Biophysics
Vanderbilt University School of Medicine
Nashville, Tennessee
- Feb 13, 2007 Mapping long-range regulatory elements that control gene
expression
Neurogenomics Postdoctoral Training Program
Vanderbilt University School of Medicine
Nashville, Tennessee
- Sept 21, 2007 Mapping long-range regulatory elements around BMP family genes
Institute for Genetic Medicine
University of Southern California
Los Angeles, California
- April 8, 2008 Mapping long-range regulatory elements around BMP family genes
Department of Animal Science, Cornell University
Ithaca, NY

Curriculum Vitae: DOUGLAS P. MORTLOCK

- June 27, 2008 Mapping long-range regulatory elements around BMP family genes
J. Wesley Pike lab, Department of Biochemistry, University of
Wisconsin
Madison, WI
- July 17, 2008 Mapping long-range regulatory elements around BMP family genes
Miami Institute for Human Genomics
University of Miami Miller School of Medicine
Miami, FL
- Oct 9, 2008 Mapping long-range regulatory elements around BMP family genes
Department of Biochemistry and Molecular Biology
University of Medicine and Dentistry of New Jersey
Newark, NJ
- Nov 21, 2008 Mapping long-range regulatory elements around BMP family genes
Department of Developmental Biology
Harvard School of Dental Medicine
Boston, MA
- Feb 5, 2009 Epigenetic landscapes around BMP genes
Genetics Interest Group
Center for Human Genetics Research, VUMC
Nashville, TN
- March 26, 2009 Mapping long-range regulatory elements around BMP family genes
Vanderbilt Center for Stem Cell Biology, VUMC
Nashville, TN
- May 14, 2009 Mapping long-range regulatory elements around BMP family genes
University of Alabama Birmingham School of Dentistry
Birmingham, AL
- Sept 6, 2009 Mapping long-range regulatory elements around BMP family genes
1st International BMP Workshop
Berlin, Germany
- March 24, 2010 Mapping long-range regulatory elements around BMP family genes.
Center of Excellence in Field Biology, Austin Peay State
University
Clarksville, TN
- May 19, 2010 Mapping long-range regulatory elements around BMP family genes.
University of Illinois
Champaign-Urbana, IL
- Aug. 13, 2013 ENCODE data: Making sense of noncoding genome annotations.
Vanderbilt Epidemiology Center, VUMC
Nashville, TN

Curriculum Vitae: DOUGLAS P. MORTLOCK

- Nov. 19, 2013 Using bioinformatics in the quest to map regulatory structure around BMP genes.
Trevecca Nazarene University
Nashville, TN
- Aug. 21, 2014 What is CRISPR/Cas, and what can we do with it?
Vanderbilt Vascular Biology Center How-To series, VUMC
Nashville, TN
- Sep. 5, 2014 What is CRISPR/Cas, and what can we do with it?
Vanderbilt Epidemiology Center, Functional Genomics Journal Club, VUMC
Nashville, TN
- April 28, 2015 CRISPR/Cas gene targeting: What can CRISPR do for you?
Vanderbilt Digestive Disease Research Center (VDDRC) retreat
Nashville, TN