Justin L. Cotney

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ACADEMIC APPOINTMENTS:

Assistant Professor (May 2015- present)
Department of Genetics and Genome Sciences, UConn Health Center, Farmington, CT

Associate Research Scientist (May 2014 – April 2015)
Department of Genetics, Yale School of Medicine, New Haven, CT

EDUCATIONAL BACKGROUND:

Ph.D.	Genetics and Molecular Biology	Emory University, Atlanta, GA	May, 2008
B.S.	Biology	Birmingham Southern College, Birmingham, AL	May, 2002
Postdoc	Genetics	Yale School of Medicine, New Haven, CT	April, 2014
Postdoc	Molecular Biology	Yale University, New Haven, CT	May, 2009

PRINT AND ORAL SCHOLARSHIP:

Peer Reviewed Publications (* indicates equal contribution)

1. Wilderman A, Kron J, VanOudenhove J, Noonan JP, **Cotney J**. High Resolution Epigenomic Atlas of Early Human Craniofacial Development. bioRxiv. 2017. (under review)

2. Stewart TA, Liang C, **Cotney JL**, Noonan JP, Sanger TJ, Wagner GP. Evidence against tetrapod-wide digit identities and for a limited frame shift in bird wings. (under review)

3. Oran AR, Adams CM, Zhang X-Y, Gennaro VJ, Pfeiffer HK, Mellert HS, Seidel HE, Mascioli K, Kaplan J, Gaballa MR, Shen C, Rigoutsos I, King MP, **Cotney JL**, Arnold JJ, Sharma SD, Martinez-Outschoorn UE, Vakoc CR, Chodosh LA, Thompson JE, Bradner JE, Cameron CE, Shadel GS, Eischen CM, McMahon SB. Multi-focal control of mitochondrial gene expression by oncogenic MYC provides potential therapeutic targets in cancer. *Oncotarget*. Impact Journals; 2016 Aug 31;7(45):72395–414.

4. **Cotney JL**, Noonan JP. Chromatin immunoprecipitation with fixed animal tissues and preparation for high-throughput sequencing. Cold Spring Harb Protoc. 2015 Feb;2015(2):191–9.

5. Reilly SK, Yin J, Ayoub AE, Emera D, Leng J, **Cotney J**, Sarro R, Rakic P, Noonan JP. Evolutionary genomics. Evolutionary changes in promoter and enhancer activity during human corticogenesis. *Science*. 2015 Mar 6;347(6226):1155–9.

6. **Cotney J**, Muhle RA, Sanders SJ, Liu L, Willsey AJ, Niu W, Liu W, Klei L, Lei J, Yin J, Reilly SK, Tebbenkamp AT, Bichsel C, Pletikos M, Šestan N, Roeder K, State MW, Devlin B, Noonan JP.

The autism-associated chromatin modifier CHD8 regulates other autism risk genes during human neurodevelopment. *Nature Communications*. 2015;6:6404.

7. Willsey AJ, Sanders SJ, Li M, Dong S, Tebbenkamp AT, Muhle RA, Reilly SK, Lin L, Fertuzinhos S, Miller JA, Murtha MT, Bichsel C, Niu W, **Cotney J**, (16 additional authors), Noonan JP, Roeder K, Devlin B, Sestan N, State MW. 2013. Co-expression networks implicate human mid-fetal deep cortical projection neurons in the pathogenesis of autism. *Cell* 155, 997-1007.

8. **Cotney, J.***, Leng, J.*, Yin, J., Reilly, S.K., Demare, L.E., Emera, D., Ayoub, A.E., Rakic, P., and Noonan, J.P. 2013. The evolution of lineage-specific regulatory activities in the human embryonic limb. *Cell* 154, 185–196.

9. Commentary in Nature News & Views: Flicek, P. 2013 Evolutionary biology: The handiwork of tinkering. *Nature* 500: 158-159.

10. DeMare LE, Leng J, **Cotney J**, Reilly SK, Yin J, Sarro R, Noonan JP. 2013 The genomic landscape of cohesin-associated chromatin interactions. *Genome Res*. 23,1224-34.

11. Clark VE, Erson-Omay EZ, Serin A, Yin J, **Cotney J**, Ozduman K, Avşar T, Li J, Murray PB, Henegariu O, et al. 2013. Genomic analysis of non-NF2 meningiomas reveals mutations in TRAF7, KLF4, AKT1, and SMO. *Science* 339: 1077–1080.

12. Bandyopadhyay U*, **Cotney J***, Nagy M, Oh S, Leng J, Mahajan M, Mane S, Fenton WA, Noonan JP, Horwich AL. 2013. RNA-Seq Profiling of Spinal Cord Motor Neurons from a Presymptomatic SOD1 ALS Mouse. *PLoS ONE* 8: e53575.

13. **Cotney J**, Leng J, Oh S, Demare LE, Reilly SK, Gerstein MB, and Noonan JP. 2012. Chromatin state signatures associated with tissue-specific gene expression and enhancer activity in the embryonic limb. *Genome Res* 22: 1069–1080.

14. Raimundo N, Song L, Shutt TE, McKay SE, **Cotney J**, Guan M-X, Gilliland TC, Hohuan D, Santos-Sacchi J, and Shadel GS. 2012. Mitochondrial stress engages E2F1 apoptotic signaling to cause deafness. *Cell* 148: 716–726.

15. Surovtseva YV, Shutt TE, **Cotney J**, Cimen H, Chen SY, Koc EC, and Shadel GS. 2011. Mitochondrial ribosomal protein L12 selectively associates with human mitochondrial RNA polymerase to activate transcription. *Proc Natl Acad Sci USA* 108: 17921–17926.

16. Ayoub AE, Oh S, Xie Y, Leng J, **Cotney J**, Dominguez MH, Noonan JP, and Rakic P. 2011. Transcriptional programs in transient embryonic zones of the cerebral cortex defined by high-resolution mRNA sequencing. *Proc Natl Acad Sci USA* 108: 14950–14955.

17. Shutt TE, Lodeiro MF, **Cotney J**, Cameron CE, and Shadel GS. 2010. Core human mitochondrial transcription apparatus is a regulated two-component system in vitro. *Proc Natl Acad Sci USA* 107: 12133–12138.

18. **Cotney J**, McKay SE, and Shadel GS. 2009. Elucidation of separate, but collaborative functions of the rRNA methyltransferase-related human mitochondrial transcription factors B1 and B2 in mitochondrial biogenesis reveals new insight into maternally inherited deafness. *Hum Mol Genet* 18: 2670–2682.

19. **Cotney J**, Wang Z, and Shadel GS. 2007. Relative abundance of the human mitochondrial transcription system and distinct roles for h-mtTFB1 and h-mtTFB2 in mitochondrial biogenesis and gene expression. *Nucleic Acids Res* 35: 4042–4054.

20. Wang Z, **Cotney J**, and Shadel GS. 2007. Human mitochondrial ribosomal protein MRPL12 interacts directly with mitochondrial RNA polymerase to modulate mitochondrial gene expression. *J Biol Chem* 282: 12610–12618.

21. **Cotney J**, and Shadel GS. 2006. Evidence for an early gene duplication event in the evolution of the mitochondrial transcription factor B family and maintenance of rRNA methyltransferase activity in human mtTFB1 and mtTFB2. *J Mol Evol* 63: 707–717.

22. Pezzementi L, Johnson K, **Cotney J**, Barker A, and Manning E. Amino acids involved in substrate and inhibitor specificity in cholinesterase 2 from amphioxus. *Cholinesterases in the Second Millennium: Biomolecular and Pathological Aspects*, (N.C. Inestrosa and E.O. Campos, eds.), 2004. MIFAB: Santiago, Chile, pp. 223-224.

23. Pezzementi L, Johnson K, Tsigelny I, **Cotney J**, Manning E, Barker A, and Merritt S. 2003. Amino acids defining the acyl pocket of an invertebrate cholinesterase. *Comp. Biochem. Physiol. B, Biochem. Mol. Biol.* 136: 813–832.

Oral Presentations

Peer reviewed submissions

Epigenetic profiling of early human craniofacial development. 36th Annual Meeting of the Society of Craniofacial Genetics and Developmental Biology, October 2013, Boston, MA

Invited presentations

Regulation of autism risk networks by the chromatin remodeler CHD8 during human neurodevelopment. UConn Autism Workshop, December 2016, Farmington, CT

Roadmap to identifying craniofacial enhanceropathies. Institute for Systems Genomics Networking Workshop, June 2016, Farmington, CT

Unlocking the regulome to understand craniofacial disorders and skeletal development. UConn Musculoskeletal Institute Research Day, April 2016, Avon, CT

Developmental Enhancers: Roles in Human Evolution and Disease. UCSF Institute for Human Genetics. April 2014, San Francisco, CA

Developmental Enhancers: Roles in Human Evolution and Disease. JAXGM. November 2013. Farmington, CT

Developmental Enhancers: Roles in Human Evolution and Disease. 3rd Kavli Community Symposium. August 2013, Trondheim, Norway

The evolution of lineage-specific regulatory activities in the human embryonic limb. Gordon Research Seminar: Human Genetics and Genomics. July 2013. Smithfield, RI

The evolution of lineage-specific regulatory activities in the human embryonic limb. Yale Center for Genome Analysis. June 2013. New Haven, CT

The evolution of lineage-specific regulatory activities in the human embryonic limb. Illumina Northeast User Meeting. October 2012, New Haven, CT

Posters

High Resolution Epigenomic Atlas of Early Human Craniofacial Development. Biology of Genomes. May 2017. Cold Spring Harbor Laboratory, Cold Spring Harbor, NY

Roadmap to Identifying Craniofacial Enhanceropathies. Meeting of the American Society of Human Genetics. October 2016, Vancouver, Canada

Regulation of autism risk networks by the chromatin remodeler CHD8 during human neurodevelopment. Genomics of Common Diseases. September 2014. Bolger Center, Potomac, MD.

The evolution of lineage-specific regulatory activities in the human embryonic limb. Biology of Genomes. May 2013. Cold Spring Harbor Laboratory, Cold Spring Harbor, NY

Enhancers maintain a constitutive open chromatin state independent of tissue-specific activity during embryonic development. Systems Biology. March 2012. Cold Spring Harbor Laboratory, Cold Spring Harbor, NY

Chromatin state transitions identify tissue-specific regulatory elements and predict gene expression gradients during mammalian embryonic development. Biology of Genomes. May 2011. Cold Spring Harbor Laboratory, Cold Spring Harbor, NY

Control of embryonic limb development through changes in global chromatin organization and gene expression. Systems Biology. March 2010. Cold Spring Harbor Laboratory, Cold Spring Harbor, NY

GRANTS:

NIH (1R01NS105501-01)
Cellular and Molecular Mechanisms underlying Angelman syndrome
Role: Collaborator
Pending

NIH/NIGMS (5R35GM119465-02)
Unraveling mechanism of genome regulation to understand and improve human health
Role: PI
$250,000
Current

NIH/NIDCR (5R00DE024194-05)
Identification of human orofacial enhancers and their role in orofacial clefts
Role: PI
$150,018
Current

Academic Plan, Tier 1 grant
TSC2 Variant without Clinical Findings of Tuberous Sclerosis is a Risk Factor for Idopathic Autism Spectrum Disorder
Role: Collaborator
$100,000
Current

NIH/NIDCR (K99DE024194-02)
Identification of human orofacial enhancers and their role in orofacial clefts
Role: PI
$108,853
Completed

OTHER ACADEMIC ACCOMPLISHMENTS/SERVICE:

Awards and Honors
Rudolph J. Andersen Foundation Fellow. Yale University School of Medicine. (July 2010 – June 2011)

Scientific Review Panels
Peer Reviewed Medical Research Program (PRMRP) for the Department of Defense Congressionally Directed Medical Research Programs (2016)
NSF CREST Center Competition Grant Review (2015, 2016)
Medical Research Council (UK) Grant Review (2015, 2016)
Wellcome Trust and Royal Society (UK) Sir Henry Dale Fellowship Review (2015, 2016)

Journal Reviews
Guest Associate Editor, PLOS Genetics, 2016
Ad Hoc Reviewer: Nature, Nature Genetics, Cell, PNAS, Molecular Cell, Genome Research, Nucleic Acids Research, Biological Pyschiatry, PLOS Genetics, PLOS One, BioEssays

TEACHING AND TRAINING:

Courses
MEDS 6448 Foundations of Biomedical Science I (Fall 2015, Fall 2016)
DENT 5440 Integrating Biotechnology with Clinical Dentistry (Fall 2015)
HB2015 Human Genetics (Fall 2015)
MEDS 5369-F40 Advanced Genetics and Molecular Biology (Spring 2016, Spring 2017)
MEDS 5415 Craniofacial and Oral Biology (Fall 2016)
MEDS 5322 Developmental Biology (Spring 2017)

Graduate Student Training
Thesis Advisor:
Andrea Wilderman (2016 – present)

Thesis Committees:
Meagan Gross (Committee Member, 2015-2016)
Kelly Brewer (Committee Member, 2016 -)
Shubham Khetan (Committee Member, 2016 -)
Michael Chung (Committee Member, 2017 -)
Michelle Spoto (Committee Member, 2017 -)

Rotation Students:
Elizabeth Kurowski (Summer 2016)
Kirby Madden-Hennessey (Fall 2016)
Edward Vigneau (Fall 2016)