# Alex S. Nord

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## Professional Experience

### Postdoctoral Fellow, Genomics Division, Lawrence Berkeley National Laboratory 9/2011-Present

- Advisors: Axel Visel, Len Pennacchio, Edward Rubin
- My research focus is how mammalian enhancers contribute to development, tissue function, and human disease using a combination of computational and experimental methods, including genomic approaches, such as ChIP-seq, as well as genome engineering and mouse models. I am particularly interested in how the genetic control of expression of critical genes is achieved in normal brain development and how perturbation to these regulatory systems contributes to neurodevelopmental disorders.
- More generally, my research interest is the control of gene expression, the evolution of gene regulatory systems, and how mutations that are outside of coding regions or perturb gene dosage contribute to disease. I am additionally interested in using results and approaches from functional genomics to inform clinical care, such as sequence-based molecular diagnosis and pharmacogenomics, functional screening of putative causal mutations, and characterization of disease via epigenetic and transcriptional landscapes.

### Ph.D., Department of Genome Sciences, University of Washington 2007-2011

- Advisor: Mary-Claire King
- Methods for identifying copy number variation from next generation sequence data.
- Impact of rare variation in human disease, focusing on cancer and neurodevelopmental disorders.

### Biostatistician, Department Of Medical Genetics, University of Washington 2005-2007

- **Supervisor:** Gail Jarvik. **Description:** Analysis of the role of genetic variation in cardiovascular disease and in neurodevelopment in children following surgery to repair cardiac defects in infancy.

### Research Consultant, UC San Francisco and UC Los Angeles 2004-2005

- **Supervisors:** Thomas Ferrin, Patricia Babbitt, Bruce Conklin (UCSF), and Stephen Young (UCLA). **Description:** Design of the International Gene Trap Consortium website and computational analysis pipeline (www.genetrap.org). Consultant to BayGenomics gene trap project, responsibility for educational outreach.

### Research Associate, The Wellcome Trust Sanger Institute (Cambridge, UK) 2003-2004

- **Supervisor:** William Skarnes. **Description:** Trained and oversaw stem cell technicians in a large acale gene trapping effort. Developed database and informatics support for high-throughput gene trapping effort.

### Research Assistant, UC Berkeley 2001-2003

- **Supervisor**: William Skarnes. **Description**: Mouse embryonic stem cell technician generating gene trap lines.

## Education

**Ph.D., Department of Genome Sciences, University of Washington, Seattle, WA** **2007-2011**

Advisor, Mary-Claire King. Committee members: Evan Eichler, Jack McClellan, Josh Akey, Marshall Horwitz. Dissertation Title: Copy number variation and complex human disease.

**B.A., Carleton College, Northfield, MN** **1997-2001**

Bachelor of Arts in Biology, *Magna cum laude*. Senior thesis (received honors in major): "Invasions in the context of community ecology: community dynamics and prediction of ecological impact."

## Academic and Professional Honors, Membership Groups, and Funding

- **2013-Present**: NIH/NIGMS NRSA F32 Postdoctoral Fellowship (GM105202)
- **2013**: Invited Visiting Instructor, Bioinformatics Short Course (July 29-31, 2013): ChIP-seq Analysis Using Next Generation Sequencing, Iowa Institute of Human Genetics, University of Iowa
- **2012**: American Society of Human Genetics/Charles J. Epstein Trainee Award for Excellence in Human Genetics Research- Semifinalist
- **2012**: Invited Visiting Instructor, Bioinformatics Short Course (August 1-3, 2012): Mutation Detection Using Massively Parallel Sequencing, Iowa Institute of Human Genetics, University of Iowa
- **2009-2011**: Predoctoral Trainee, Biostatistics, Epidemiologic and Bioinformatic Training in Environmental Health, University of Washington (5T32ES015459).
- **2007-2009**: Predoctoral Trainee, Training program in genetics general and medical, Genome Sciences, University of Washington (5T32GM007735)
- **2001**: Distinction in Thesis and Major for Biology, *Magna Cum Laude*, Carleton College.
- **2000:** NSF/REU Grant Recipient, Graduate College of Marine Studies, University of Delaware

## Teaching Experience

- **2012 & 2013:** Invited Visiting Instructor, Bioinformatics Short Course, Iowa Institute of Human Genetics: I developed and delivered lectures in current topics in genetics and in bioinformatics. Course attendees included undergraduate students, graduate students, postdocs, faculty, and clinicians.
- **2011:** Teaching Assistant, Human Genetics (Genome 351, University of Washington)
- **2010:** Teaching Assistant, Introductory Genetics (Genome 371, University of Washington)

## Publications

**Research papers:**

***Most relevant to the current application:***

1. **Nord AS**, Blow MJ, Attanasio C, Akiyama JA, Holt A, Hosseini R, Phouanenavong S, Plajzer-Frick I, Shoukry M, Afzal V, Rubenstein JLR, Rubin EM, Pennacchio LA, Visel A. Rapid and Pervasive Changes in Genome-Wide Enhancer Usage During Mammalian Development. *Cell.* 2013. Dec 19;155(7): 1521-31

2. Attanasio C, **Nord AS**, Zhu Y, Blow MJ, Li Z, Liberton DK, Morrison H, Plajzer-Frick I, Holt A, Hosseini R, Phouanenavong S, Akiyama JA, Shoukry M, Afzal V, Rubin EM, FitzPatrick DR, Ren B, Hallgrímsson B, Pennacchio LA, Visel A. Fine tuning of craniofacial morphology by distant-acting enhancers. *Science*. 2013. Oct 25;342(6157):1241006. PMID: 24159046.

3. Visel A, Taher L, Girgis H, May D, Golonzhka O, Hoch RV, McKinsey GL, Pattabiraman K, Silberberg SN, Blow MJ, Hansen DV, **Nord AS**, Akiyama JA, Holt A, Hosseini R, Phouanenavong S, Plajzer-Frick I, Shoukry M, Afzal V, Kaplan T, Kriegstein AR, Rubin EM, Ovcharenko I, Pennacchio LA, Rubenstein JL. A high-resolution enhancer atlas of the developing telencephalon. *Cell*. 2013 Feb 14;152(4):895-908. PMCID: PMC3660042.

4. **Nord AS**, Lee M, King MC, Walsh T. Accurate and exact CNV identification from targeted high-throughput sequence data. *BMC Genomics*. 2011 Apr 12;12:184. PMCID: PMC3088570.

5. **Nord AS**, Roeb W, Dickel DE, Walsh T, Kusenda M, O'Connor KL, Malhotra D, McCarthy SE, Stray SM, Taylor SM, Sebat J; STAART Psychopharmacology Network, King B, King MC, McClellan JM. Reduced transcript expression of genes affected by inherited and de novo CNVs in autism. *Eur J Hum Genet.* 2011 Jun;19(6):727-31. PMCID: PMC3110052.

6. Walsh T, Pierce SB, Lenz DR, Brownstein Z, Dagan-Rosenfeld O, Shahin H, Roeb W, McCarthy S, **Nord AS**, Gordon CR, Ben-Neriah Z, Sebat J, Kanaan M, Lee MK, Frydman M, King MC, Avraham KB. Genomic duplication and overexpression of TJP2/ZO-2 leads to altered expression of apoptosis genes in progressive nonsyndromic hearing loss DFNA51. *Am J Hum Genet.* 2010 Jul 9;87(1):101-9. PMCID: PMC2896780.

7. Gaynor JW, **Nord AS**, Wernovsky G, Bernbaum J, Solot CB, Burnham N, Zackai E, Heagerty PJ, Clancy RR, Nicolson SC, Jarvik GP, Gerdes M. Apolipoprotein E genotype modifies the risk of behavior problems after infant cardiac surgery. *Pediatrics.* 2009 Jul;124(1):241-50. PMCID: PMC2840402.

8. Walsh T, McClellan JM, McCarthy SE, Addington AM, Pierce SB, Cooper GM, **Nord AS**, Kusenda M, Malhotra D, Bhandari A, Stray SM, Rippey CF, Roccanova P, Makarov V, Lakshmi B, Findling RL, Sikich L, Stromberg T, Merriman B, Gogtay N, Butler P, Eckstrand K, Noory L, Gochman P, Long R, Chen Z, Davis S, Baker C, Eichler EE, Meltzer PS, Nelson SF, Singleton AB, Lee MK, Rapoport JL, King MC, Sebat J. Rare structural variants disrupt multiple genes in neurodevelopmental pathways in schizophrenia. *Science*. 2008 Apr 25;320(5875):539-43. PMID: 18369103.

9. **Nord AS**, Vranizan K, Tingley W, Zambon AC, Hanspers K, Fong LG, Hu Y, Bacchetti P, Ferrin TE, Babbitt PC, Doniger SW, Skarnes WC, Young SG, Conklin BR. Modeling insertional mutagenesis using gene length and expression in murine embryonic stem cells. *PLoS One.* 2007 Jul 18;2(7):e617. PMCID: PMC1910612.

10. **Nord AS**, Chang PJ, Conklin BR, Cox AV, Harper CA, Hicks GG, Huang CC, Johns SJ, Kawamoto M, Liu S, Meng EC, Morris JH, Rossant J, Ruiz P, Skarnes WC, Soriano P, Stanford WL, Stryke D, von Melchner H, Wurst W, Yamamura K, Young SG, Babbitt PC, Ferrin TE. The International Gene Trap Consortium Website: a portal to all publicly available gene trap cell lines in mouse. *Nucleic Acids Res.* 2006 Jan 1;34(Database issue):D642-8. PMCID: PMC1347459.

## ***Additional publications:***

1. Dickel, DE, Zhu Y, **Nord AS**, Wylie J, Akiyama JA, Plajzer-Frick I, Bruneau BG, Cisel A, Pennacchio LA. Function-based Identification of Mammalian Enhancers. *Nature Methods* (Minor revisions pending)

2. Pennington KP, Walsh T, Harrell M, Lee M, Pennil C, Rendi M, Thornton A, Norquist BM, Casadei S, **Nord AS**, Agnew KJ, Pritchard CC, Scroggins S, Garcia R, King MC, Swisher EM. Germline and Somatic Mutations in Homologous Recombination Genes Predict Platinum Response and Survival in Ovarian, Fallopian Tube, and Peritoneal Carcinomas. *Clin Cancer Res.* (Minor revisions pending)

3. Rippey C, Walsh T, Gulsuner S, Brodsky M, **Nord AS**, Gasperini M, Pierce S, Spurrell C, Coe BP, Krumm N, Lee MK, Sebat J, McClellan JM, King MC. Formation of chimeric genes by copy-number variation as a mutational mechanism in schizophrenia. *Am J Hum Genet.* 2013 Oct 3;93(4):697-710. PMCID: PMC3791253

4. Kodera H, Kato M, **Nord AS**, Walsh T, Lee M, Yamanaka G, Tohyama J, Nakamura K, Nakagawa E, Ikeda T, Ben-Zeev B, Lev D, Lerman-Sagie T, Straussberg R, Tanabe S, Ueda K, Amamoto M, Ohta S, Nonoda Y, Nishiyama K, Tsurusaki Y, Nakashima M, Miyake N, Hayasaka K, King MC, Matsumoto N, Saitsu H. Targeted capture and sequencing for detection of mutations causing early onset epileptic encephalopathy. *Epilepsia.* 2013 Jul;54(7):1262-9. PMID: 23662938.

5. Sanchez-Castro M, Gordon CT, Petit F, **Nord AS**, Callier P, Andrieux J, Guérin P, Pichon O, David A, Abadie V, Bonnet D, Visel A, Pennacchio LA, Amiel J, Lyonnet S, Le Caignec C. Congenital Heart Defects in Patients with Deletions Upstream of SOX9. *Hum Mutat.* 2013 Dec;34(12):1628-31. PMID: 24115316.

6. Pritchard CC, Smith C, Salipante SJ, Lee MK, Thornton AM, **Nord AS**, Gulden C, Kupfer SS, Swisher EM, Bennett RL, Novetsky AP, Jarvik GP, Olopade OI, Goodfellow PJ, King MC, Tait JF, Walsh T. ColoSeq provides comprehensive lynch and polyposis syndrome mutational analysis using massively parallel sequencing. *J Mol Diagn.* 2012 Jul;14(4):357-66. PMCID: PMC3391416.

7. Walsh T, Casadei S, Lee MK, Pennil CC, **Nord AS**, Thornton AM, Roeb W, Agnew KJ, Stray SM, Wickramanayake A, Norquist B, Pennington KP, Garcia RL, King MC, Swisher EM. Mutations in 12 genes for inherited ovarian, fallopian tube, and peritoneal carcinoma identified by massively parallel sequencing. *Proc Natl Acad Sci U S A*. 2011 Nov 1;108(44):18032-7. PMCID: PMC3207658.

8. Ronald J, Rajagopalan R, Cerrato F, **Nord AS**, Hatsukami T, Kohler T, Marcovina S, Heagerty P, Jarvik GP. Genetic variation in LPAL2, LPA, and PLG predicts plasma lipoprotein(a) level and carotid artery disease risk. *Stroke.* 2011 Jan;42(1):2-9. PMCID: PMC3020902.

9. Gaynor JW, Gerdes M, **Nord AS**, Bernbaum J, Zackai E, Wernovsky G, Clancy RR, Heagerty PJ, Solot CB, McDonald-McGinn D, Jarvik GP. Is cardiac diagnosis a predictor of neurodevelopmental outcome after cardiac surgery in infancy? *J Thorac Cardiovasc Surg.* 2010 Dec;140(6):1230-7. PMCID: PMC3278908.

10. Walsh T, Lee MK, Casadei S, Thornton AM, Stray SM, Pennil C, **Nord AS**, Mandell JB, Swisher EM, King MC. Detection of inherited mutations for breast and ovarian cancer using genomic capture and massively parallel sequencing. *Proc Natl Acad Sci U S A.* 2010 Jul 13;107(28):12629-33. PMCID: PMC2906584.

11. Shahin H, Walsh T, Rayyan AA, Lee MK, Higgins J, Dickel D, Lewis K, Thompson J, Baker C, **Nord AS**, Stray S, Gurwitz D, Avraham KB, King MC, Kanaan M. Five novel loci for inherited hearing loss mapped by SNP-based homozygosity profiles in Palestinian families. *Eur J Hum Genet.* 2010 Apr;18(4):407-13. PMCID: PMC2987250.

12. Chen J, Zimmerman RA, Jarvik GP, **Nord AS**, Clancy RR, Wernovsky G, Montenegro LM, Hartman DM, Nicolson SC, Spray TL, Gaynor JW, Ichord R. Perioperative stroke in infants undergoing open heart operations for congenital heart disease. *Ann Thorac Surg.* 2009 Sep;88(3):823-9. PMCID: PMC2840405.

13. Fuller S, **Nord AS**, Gerdes M, Wernovsky G, Jarvik GP, Bernbaum J, Zackai E, Gaynor JW. Predictors of impaired neurodevelopmental outcomes at one year of age after infant cardiac surgery. *Eur J Cardiothorac Surg.* 2009 Jul;36(1):40-7. PMID: 19394849.

14. Ober C, **Nord AS**, Thompson EE, Pan L, Tan Z, Cusanovich D, Sun Y, Nicolae R, Edelstein C, Schneider DH, Billstrand C, Pfaffinger D, Phillips N, Anderson RL, Philips B, Rajagopalan R, Hatsukami TS, Rieder MJ, Heagerty PJ, Nickerson DA, Abney M, Marcovina S, Jarvik GP, Scanu AM, Nicolae DL. Genome-wide association study of plasma lipoprotein(a) levels identifies multiple genes on chromosome 6q. *J Lipid Res.* 2009 May;50(5):798-806. PMCID: PMC2666166.

15. Goode EL, Fridley BL, Sun Z, Atkinson EJ, **Nord AS**, McDonnell SK, Jarvik GP, de Andrade M, Slager SL. Comparison of tagging single-nucleotide polymorphism methods in association analyses. *BMC Proc.* 2007;1 Suppl 1:S6. PMCID: PMC2367496.

16. Reiner AP, Wurfel MM, Lange LA, Carlson CS, **Nord AS**, Carty CL, Rieder MJ, Desmarais C, Jenny NS, Iribarren C, Walston JD, Williams OD, Nickerson DA, Jarvik GP. Polymorphisms of the IL1-receptor antagonist gene (IL1RN) are associated with multiple markers of systemic inflammation. *Arterioscler Thromb Vasc Biol.* 2008 Jul;28(7):1407-12. PMCID: PMC2748384.

17. Tabbutt S, **Nord AS**, Jarvik GP, Bernbaum J, Wernovsky G, Gerdes M, Zackai E, Clancy RR, Nicolson SC, Spray TL, Gaynor JW. Neurodevelopmental outcomes after staged palliation for hypoplastic left heart syndrome. *Pediatrics.* 2008 Mar;121(3):476-83. PMID: 18310195.

18. Zeltser I, Jarvik GP, Bernbaum J, Wernovsky G, **Nord AS**, Gerdes M, Zackai E, Clancy R, Nicolson SC, Spray TL, Gaynor JW. Genetic factors are important determinants of neurodevelopmental outcome after repair of tetralogy of Fallot. *J Thorac Cardiovasc Surg.* 2008 Jan;135(1):91-7. PMID: 18179924.

19. Crawford DC, **Nord AS**, Badzioch MD, Ranchalis J, McKinstry LA, Ahearn M, Bertucci C, Shephard C, Wong M, Rieder MJ, Schellenberg GD, Nickerson DA, Heagerty PJ, Wijsman EM, Jarvik GP. A common VLDLR polymorphism interacts with APOE genotype in the prediction of carotid artery disease risk. *J Lipid Res.* 2008 Mar;49(3):588-96. PMID: 18056683.

20. Gaynor JW, Wernovsky G, Jarvik GP, Bernbaum J, Gerdes M, Zackai E, **Nord AS**, Clancy RR, Nicolson SC, Spray TL. Patient characteristics are important determinants of neurodevelopmental outcome at one year of age after neonatal and infant cardiac surgery. *J Thorac Cardiovasc Surg.* 2007 May;133(5):1344-53, 1353.e1-3. PMCID: PMC2844117.

21. Carlson CS, Heagerty PJ, **Nord AS**, Pritchard DK, Ranchalis J, Boguch JM, Duan H, Hatsukami TS, Schwartz SM, Rieder MJ, Nickerson DA, Jarvik GP. TagSNP evaluation for the association of 42 inflammation loci and vascular disease: evidence of IL6, FGB, ALOX5, NFKBIA, and IL4R loci effects. *Hum Genet.* 2007 Mar;121(1):65-75. PMID: 17115186.

22. Skarnes WC, von Melchner H, Wurst W, Hicks G, **Nord AS**, Cox T, Young SG, Ruiz P, Soriano P, Tessier-Lavigne M, Conklin BR, Stanford WL, Rossant J; International Gene Trap Consortium. A public gene trap resource for mouse functional genomics. *Nat Genet.* 2004 Jun;36(6):543-4. PMCID: PMC2716026.

## Manuscripts in review:

1. Attanasio CA, **Nord AS** (co-first authors), Zhu Y, Blow MJ, Biddie SC, Mendenhall EM, Dixon J, Wright C, Hosseini R, Akiyama JA, Holt A, Plajzer-Frick I, Shoukry M, Afzal V, Ren B, Bernstein BE, Rubin EM, Visel A, Pennacchio LA. Tissue-specific BRG1 binding at active and repressed regulatory elements during embryogenesis. *Genome Research* (In review)

2. Wu H, **Nord AS**, Akiyama JA, Shoukry M, Afzal V, Rubin EM, Pennacchio LA, Visel A. Tissue-specific enhancer RNA expression from distant-acting *in vivo* enhancers. Submitted to *Nature Genetics*.

3. Pattabiraman K, Golonzhka O, Lindtner S, **Nord AS**, Taher L, Hoch R, Silberberg SN, Zhang D, Chen B, Zeng HK, Pennacchio LA, Puelles L, Visel A, Rubenstein JLR. Transcriptional regulation of embryonic pallial enhancers that are active in distinct protodomains presages distinct regional fate. Submitted to *Cell*.

## Relevant abstracts:

**1.** **Nord AS**, Akiyama J, Blow MJ, Holt A, Hosseini R, Phouanenavong S, Plajzer-Frick I, Shoukry M, Afzal V, Rubin EM, Rubenstein JLR, Pennacchio LP, Visel A. Developmental and Evolutionary Dynamics of Mammalian Enhancers. (Selected for Lightning Talk). Presented at ENCODE Consortium Meeting, May 30, 2013, Stanford University, Palo Alto, California.

**2.** **Nord AS**, Taher L, Akiyama J, Blow MJ, Holt A, Hosseini R, Phouanenavong S, Plajzer-Frick I, Shoukry M, Afzal V, Rubin EM, Ovcharenko I, Rubenstein JLR, Pennacchio LP, Visel A. Temporally and spatially resolved catalogues of *in vivo* forebrain enhancers; (Abstract/Program #407 – Selected for platform/oral presentation). Presented at the 62nd Annual Meeting of The American Society of Human Genetics, November 9, 2012 in San Francisco, California.

## Book chapters:

*1.* **Nord AS**, Salipante SJ, Pritchard CC. Copy number variant (CNV) detection using next-generation sequencing. In Kulkarni and Pfiefer (Eds.), *Clinical Genomics: A Guide to Clinical Next Generation Sequencing*. Elsevier/Academic Press (In press)