



Alexander Eckehart Urban

Associate Professor of Psychiatry and Behavioral Sciences (Major Laboratories and Clinical Translational Neurosciences Incubator) and of Genetics

 NIH Biosketch available Online

Bio

ACADEMIC APPOINTMENTS

- Associate Professor, Psychiatry and Behavioral Sciences
- Associate Professor, Genetics
- Member, Bio-X
- Member, Maternal & Child Health Research Institute (MCHRI)
- Member, Wu Tsai Neurosciences Institute

PROFESSIONAL EDUCATION

- PhD, Yale University , Molecular, Cellular and Developmental Biology (2007)

LINKS

- Urban Lab web site: <http://urbanlab.stanford.edu>
- Google Scholar: <http://scholar.google.com/citations?user=IGyIeMsAAAAJ&hl=en>
- NCBI myBibliography: <https://www.ncbi.nlm.nih.gov/myncbi/collections/bibliography/44935252/>

Research & Scholarship

CURRENT RESEARCH AND SCHOLARLY INTERESTS

I have over two decades worth of experience in developing and applying high-throughput and high-resolution genomics analysis tools and procedures, in particular in the context of studying genomic sequence variation in brain development and function.

I have been involved on numerous occasions in using a large-scale and high-throughput setup for genomics analyses as well as carrying out analyses over several levels of genomics and epigenomics information. This includes participation in the ENCODE and 1000 Genomes projects, for the latter as a member of both the analytical and structural variation groups.

I have experience with developing and applying state-of-the-art and emerging genomics and epigenomics technologies (array and next-generation-sequencing based) for the analysis of gene expression, genomic DNA sequence and structure, DNA methylation and chromatin modification, in human cells and human cell culture systems, including stem cell culture models. For example I was co-first author of the paper in Science (Korbel, Urban, Affourtit et al., 2007, PMID 17901297) on developing next-generation-sequencing based paired-end mapping of CNVs and SVs, an approach that is now a standard part of whole-human-genome sequencing projects such as the 1000 Genomes Project. Paired-end mapping is also a critical component of advanced RNA-Seq approaches, mapping of transposable elements and the study of long-range chromatin interactions using the HiC method.

Two main, and connected, directions of research in my laboratory are the investigation of the molecular effects of large genome variants during neuronal development using iPSC model systems and the study of the nature and effects of somatic genome variation in the brain using tissue culture models and primary tissue samples.

CLINICAL TRIALS

- Building Resilience at Schools: Emotional and Biological Assessment and Treatment of Traumatic Stress, Not Recruiting

Teaching

COURSES

2023-24

- Frontiers in Biological Research: BIOC 215, DBIO 215, GENE 215 (Aut, Win, Spr)

2022-23

- Frontiers in Biological Research: BIOC 215, DBIO 215, GENE 215 (Aut, Win, Spr)

2021-22

- Frontiers in Biological Research: BIOC 215, DBIO 215, GENE 215 (Aut, Win, Spr)

2020-21

- Frontiers in Biological Research: BIOC 215, DBIO 215, GENE 215 (Aut, Win, Spr)

STANFORD ADVISEES

Postdoctoral Faculty Sponsor

Hanmin Guo, Pingping Qu

GRADUATE AND FELLOWSHIP PROGRAM AFFILIATIONS

- Genetics (Phd Program)

Publications

PUBLICATIONS

- **Integrative analyses highlight functional regulatory variants associated with neuropsychiatric diseases.** *Nature genetics*
Guo, M. G., Reynolds, D. L., Ang, C. E., Liu, Y., Zhao, Y., Donohue, L. K., Siprashvili, Z., Yang, X., Yoo, Y., Mondal, S., Hong, A., Kain, J., Meservey, et al
2023
- **Pan-conserved segment tags identify ultra-conserved sequences across assemblies in the human pangenome.** *Cell reports methods*
Lee, H., Greer, S. U., Pavlichin, D. S., Zhou, B., Urban, A. E., Weissman, T., Ji, H. P.
2023; 3 (8): 100543
- **Mutations in human DNA methyltransferase DNMT1 induce specific genome-wide epigenomic and transcriptomic changes in neurodevelopment.** *Human molecular genetics*
Davis, K. N., Qu, P. P., Ma, S., Lin, L., Plastini, M., Dahl, N., Plazzi, G., Pizza, F., O'Hara, R., Wong, W. H., Hallmayer, J., Mignot, E., Zhang, et al
2023
- **Schizophrenia-associated somatic copy-number variants from 12,834 cases reveal recurrent NRXN1 and ABCB11 disruptions.** *Cell genomics*
Maury, E. A., Sherman, M. A., Genovese, G., Gilgenast, T. G., Kamath, T., Burris, S. J., Rajarajan, P., Flaherty, E., Akbarian, S., Chess, A., McCarroll, S. A., Loh, P. R., Phillips-Cremins, et al
2023; 3 (8): 100356
- **Evaluation of C4 gene copy number in Pediatric Acute Neuropsychiatric Syndrome** *DEVELOPMENTAL NEUROSCIENCE*
Kalinowski, A., Tian, L., Pattni, R., Ollila, H., Khan, M., Manko, C., Silverman, M., Ma, M., Columbo, L., Farhadian, B., Swedo, S., Murphy, T., Johnson, et al
2023

- **Comprehensive multi-omic profiling of somatic mutations in malformations of cortical development** *NATURE GENETICS*
Chung, C., Yang, X., Bae, T., Vong, K., Mittal, S. H., Donkels, C., Westley Phillips, H., Li, Z., Marsh, A. L., Breuss, M., Ball, L., Garcia, C., George, et al
2023; 55 (2): 209-+
- **RESOLVING THE EXACT BREAKPOINTS AND SEQUENCE REARRANGEMENTS OF LARGE NEUROPSYCHIATRIC COPY NUMBER VARIATIONS (CNVS) AT SINGLE BASE-PAIR RESOLUTION USING CRISPR-TARGETED ULTRALONG READ SEQUENCING (CTLR-SEQ)**
Zhou, B., Shin, G., Vervoort, L., Greer, S., Huang, Y., Roychowdhury, T., Pattni, R., Abyzov, A., Vermeesch, J., Ji, H., Urban, A.
ELSEVIER.2022: E88-E89
- **DETECTION OF COMPLEX STRUCTURAL GENOME VARIANTS USING ARC-SV AND THEIR ENRICHMENT INSIDE GENES OF NEURODEVELOPMENTAL PATHWAYS**
Hughes, C., Zhou, B., Arthur, J. G., Kim, T., Song, G., Palejev, D., Wong, W. H., Urban, A. E.
ELSEVIER.2022: E177-E178
- **Analysis of somatic mutations in 131 human brains reveals aging-associated hypermutability.** *Science (New York, N.Y.)*
Bae, T., Fasching, L., Wang, Y., Shin, J. H., Suvakov, M., Jang, Y., Norton, S., Dias, C., Mariani, J., Jourdon, A., Wu, F., Panda, A., Pattni, et al
2022; 377 (6605): 511-517
- **Maternal attachment insecurity, maltreatment history, and depressive symptoms are associated with broad DNA methylation signatures in infants.** *Molecular psychiatry*
Robakis, T. K., Roth, M. C., King, L. S., Humphreys, K. L., Ho, M., Zhang, X., Chen, Y., Li, T., Rasgon, N. L., Watson, K. T., Urban, A. E., Gotlib, I. H.
2022
- **Somatic mosaicism reveals clonal distributions of neocortical development** *NATURE*
Breuss, M. W., Yang, X., Schlachetzki, J. M., Antaki, D., Lana, A. J., Xu, X., Chung, C., Chai, G., Stanley, V., Song, Q., Newmeyer, T. F., An Nguyen, O'Brien, S., et al
2022; 604 (7907): 689-+
- **Hyperexcitable arousal circuits drive sleep instability during aging.** *Science (New York, N.Y.)*
Li, S. B., Damonte, V. M., Chen, C., Wang, G. X., Keschull, J. M., Yamaguchi, H., Bian, W. J., Purmann, C., Pattni, R., Urban, A. E., Mourrain, P., Kauer, J. A., Scherrer, et al
2022; 375 (6583): eabh3021
- **Increased activation product of complement 4 protein in plasma of individuals with schizophrenia.** *Translational psychiatry*
Kalinowski, A., Liliental, J., Anker, L. A., Linkovski, O., Culbertson, C., Hall, J. N., Pattni, R., Sabatti, C., Noordsy, D., Hallmayer, J. F., Mellins, E. D., Ballon, J. S., O'Hara, et al
2021; 11 (1): 486
- **Cross-platform validation of neurotransmitter release impairments in schizophrenia patient-derived NRXN1-mutant neurons.** *Proceedings of the National Academy of Sciences of the United States of America*
Pak, C., Danko, T., Mirabella, V. R., Wang, J., Liu, Y., Vangipuram, M., Grieder, S., Zhang, X., Ward, T., Huang, Y. A., Jin, K., Dexheimer, P., Bardes, et al
2021; 118 (22)
- **PRG2 and AQPEP are misexpressed in fetal membranes in placenta previa and percreta1.** *Biology of reproduction*
Zhang, E. T., Hannibal, R. L., Badillo Rivera, K. M., Song, J. H., McGowan, K., Zhu, X., Meinhardt, G., Knofler, M., Pollheimer, J., Urban, A. E., Folkins, A. K., Lyell, D. J., Baker, et al
2021
- **Comprehensive identification of somatic nucleotide variants in human brain tissue.** *Genome biology*
Wang, Y., Bae, T., Thorpe, J., Sherman, M. A., Jones, A. G., Cho, S., Daily, K., Dou, Y., Ganz, J., Galor, A., Lobon, I., Pattni, R., Rosenbluh, et al
2021; 22 (1): 92
- **Machine learning reveals bilateral distribution of somatic L1 insertions in human neurons and glia.** *Nature neuroscience*
Zhu, X. n., Zhou, B. n., Pattni, R. n., Gleason, K. n., Tan, C. n., Kalinowski, A. n., Sloan, S. n., Fiston-Lavier, A. S., Mariani, J. n., Petrov, D. n., Barres, B. A., Duncan, L. n., Abyzov, et al
2021
- **Highly sensitive and full-genome interrogation of SARS-CoV-2 using multiplexed PCR enrichment followed by next-generation sequencing.**
Li, C., Debruyne, D. N., Spencer, J., Kapoor, V., Liu, L. Y., Zhou, B., Pandey, U., Bootwalla, M., Ostrow, D., Maglinte, D. T., Ruble, D., Ryutov, A., Shen, et al
AMER ASSOC CANCER RESEARCH.2020

- **CNN-Peaks: ChIP-Seq peak detection pipeline using convolutional neural networks that imitate human visual inspection.** *Scientific reports*
Oh, D., Strattan, J. S., Hur, J. K., Bento, J., Urban, A. E., Song, G., Cherry, J. M.
2020; 10 (1): 7933
- **Low C4 Copy Number of Total C4 Gene, C4B Gene and C4BL Gene in Children with Pediatric Acute-onset Neuropsychiatric Syndrome (PANS)**
Kalinowski, A., Lee, J., Hedlin, H., Pattini, R., Ollila, H., Mignot, E., Levinson, D., Swedo, S., Murphy, T., Chan, A., Thienemann, M., Urban, A., Frankovich, et al
WILEY.2020: 254–55
- **Integrated functional genomic analyses of Klinefelter and Turner syndromes reveal global network effects of altered X chromosome dosage.** *Proceedings of the National Academy of Sciences of the United States of America*
Zhang, X., Hong, D., Ma, S., Ward, T., Ho, M., Pattni, R., Duren, Z., Stankov, A., Bade Shrestha, S., Hallmayer, J., Wong, W. H., Reiss, A. L., Urban, et al
2020
- **Epigenetic signatures of attachment insecurity and childhood adversity provide evidence for role transition in the pathogenesis of perinatal depression.** *Translational psychiatry*
Robakis, T. K., Zhang, S., Rasgon, N. L., Li, T., Wang, T., Roth, M. C., Humphreys, K. L., Gotlib, I. H., Ho, M., Khechaduri, A., Watson, K., Roat-Shumway, S., Budhan, et al
2020; 10 (1): 48
- **Complex mosaic structural variations in human fetal brains.** *Genome research*
Sekar, S. n., Tomasini, L. n., Proukakis, C. n., Bae, T. n., Manlove, L. n., Jang, Y. n., Scuderi, S. n., Zhou, B. n., Kalyva, M. n., Amiri, A. n., Mariani, J. n., Sedlazeck, F. n., Urban, et al
2020
- **Network Effects of the 15q13.3 Microdeletion on the Transcriptome and Epigenome in Human-Induced Neurons.** *Biological psychiatry*
Zhang, S. n., Zhang, X. n., Purmann, C. n., Ma, S. n., Shrestha, A. n., Davis, K. N., Ho, M. n., Huang, Y. n., Pattni, R. n., Wong, W. H., Bernstein, J. A., Hallmayer, J. n., Urban, et al
2020
- **MACHINE LEARNING REVEALS BILATERAL DISTRIBUTION OF SOMATIC L1 INSERTIONS IN HUMAN NEURONS AND GLIA**
Zhu, X., Zhou, B., Pattni, R., Gleason, K., Tan, C., Kalinowski, A., Sloan, S., Fiston-Lavier, A., Mariani, J., Vogel, H., Moran, J., Vaccarino, F., Tamminga, et al
ELSEVIER.2019: S68
- **CELL TYPE SPECIFIC MULTI-LEVEL ANALYSIS OF NEUROPSYCHIATRIC GENOMIC SEQUENCE ELEMENTS**
Urban, A.
ELSEVIER.2019: S47
- **APPROACHES TO TRANSCRIPTOME ANALYSIS OF HUMAN INDUCED NEURONS IN CO-CULTURE WITH MURINE GLIA TO MODEL FUNCTIONAL SYNAPSES**
Purmann, C., Zhang, X., Pak, C., Huang, Y., Pattni, R., Grieder, S., Wernig, M., Levinson, D., Aronow, B., Sudhof, T., Urban, A.
ELSEVIER.2019: S172–S173
- **SMALL-SCALE SOMATIC STRUCTURAL VARIANTS IN FETAL AND ADULT HUMAN BRAINS**
Zhou, B., Arthur, J., Pattni, R., Zhu, X., Wong, W., Urban, A.
ELSEVIER.2019: S163–S164
- **INTEGRATED ANALYSIS OF GENE EXPRESSION, DNA METHYLATION AND CHROMATIN ACCESSIBILITY IN A HUMAN IPSC-TO-INDUCED-NEURON MODEL OF THE 15Q13.3 MICRODELETION**
Zhang, S., Zhang, X., Ma, S., Purmann, C., Davis, K., Wong, W., Bernstein, J., Hallmayer, J., Urban, A.
ELSEVIER.2019: S105
- **COMPARATIVE FUNCTIONAL GENOMICS ANALYSES OF THE 16P11.2 DELETION AND DUPLICATION CNVS IN A HUMAN IPSC-TO-INDUCED NEURON MODEL**
Zhang, X., Thomas, W., Leung, L., Zhou, B., Muench, K., Plastini, M., Pattni, R., Ho, S., Ho, M., Huang, Y., Hallmayer, J., Mourrain, P., Palmer, et al
ELSEVIER.2019: S66
- **Metabolism of cholesterol and progesterone is differentially regulated in primary trophoblastic subtypes and might be disturbed in recurrent miscarriages.** *Journal of lipid research*
Vondra, S., Kunihs, V., Eberhart, T., Eigner, K., Bauer, R., Haslinger, P., Haider, S., Windsperger, K., Klambauer, G., Schutz, B., Mikula, M., Zhu, X., Urban, et al
2019

- **Haplotype-resolved and integrated genome analysis of the cancer cell line HepG2** *NUCLEIC ACIDS RESEARCH*
Zhou, B., Ho, S. S., Greer, S. U., Spies, N., Bell, J. M., Zhang, X., Zhu, X., Arthur, J. G., Byeon, S., Pattni, R., Saha, I., Huang, Y., Song, et al
2019; 47 (8): 3846–61
- **The NASA Twins Study: A multidimensional analysis of a year-long human spaceflight** *SCIENCE*
Garrett-Bakelman, F. E., Darshi, M., Green, S. J., Gur, R. C., Lin, L., Macias, B. R., McKenna, M. J., Meydan, C., Mishra, T., Nasrini, J., Piening, B. D., Rizzardi, L. F., Sharma, et al
2019; 364 (6436): 144+
- **Allele-specific binding of RNA-binding proteins reveals functional genetic variants in the RNA** *NATURE COMMUNICATIONS*
Yang, E., Bahn, J., Hsiao, E., Tan, B., Sun, Y., Fu, T., Zhou, B., Van Nostrand, E. L., Pratt, G. A., Freese, P., Wei, X., Quinones-Valdez, G., Urban, et al
2019; 10
- **Allele-specific binding of RNA-binding proteins reveals functional genetic variants in the RNA.** *Nature communications*
Yang, E., Bahn, J. H., Hsiao, E. Y., Tan, B. X., Sun, Y., Fu, T., Zhou, B., Van Nostrand, E. L., Pratt, G. A., Freese, P., Wei, X., Quinones-Valdez, G., Urban, et al
2019; 10 (1): 1338
- **Haplotype-resolved and integrated genome analysis of the cancer cell line HepG2.** *Nucleic acids research*
Zhou, B., Ho, S. S., Greer, S. U., Spies, N., Bell, J. M., Zhang, X., Zhu, X., Arthur, J. G., Byeon, S., Pattni, R., Saha, I., Huang, Y., Song, et al
2019
- **Comprehensive, integrated, and phased whole-genome analysis of the primary ENCODE cell line K562** *GENOME RESEARCH*
Zhou, B., Ho, S. S., Greer, S. U., Zhu, X., Bell, J. M., Arthur, J. G., Spies, N., Zhang, X., Byeon, S., Pattni, R., Ben-Efraim, N., Haney, M. S., Haraksingh, et al
2019; 29 (3): 472–84
- **Comprehensive, integrated, and phased whole-genome analysis of the primary ENCODE cell line K562.** *Genome research*
Zhou, B., Ho, S. S., Greer, S. U., Zhu, X., Bell, J. M., Arthur, J. G., Spies, N., Zhang, X., Byeon, S., Pattni, R., Ben-Efraim, N., Haney, M. S., Haraksingh, et al
2019
- **The NASA Twins Study: A multidimensional analysis of a year-long human spaceflight.** *Science (New York, N.Y.)*
Garrett-Bakelman, F. E., Darshi, M. n., Green, S. J., Gur, R. C., Lin, L. n., Macias, B. R., McKenna, M. J., Meydan, C. n., Mishra, T. n., Nasrini, J. n., Piening, B. D., Rizzardi, L. F., Sharma, et al
2019; 364 (6436)
- **ANALYZING THE MOLECULAR NETWORK EFFECTS OF LARGE NEUROPSYCHIATRIC CNVS WITH IPSC BASED NEURONAL TISSUE CULTURE MODELS**
Purmann, C., Ma, S., Zhang, S., Ward, T., Huang, E., Pattni, R., Hallmayer, J., Wong, W., Urban, A.
ELSEVIER.2019: 1060
- **MACHINE LEARNING ANALYSIS OF ULTRA-DEEP WHOLE-GENOME SEQUENCING IN HUMAN BRAIN REVEALS SOMATIC GENOMIC RETROTRANSPOSITION IN GLIA AS WELL AS IN NEURONS**
Urban, A., Zhu, X., Zhou, B., Sloan, S., Pattni, R., Fiston-Lavier, A., Snyder, M., Petrov, D., Abyzov, A., Vaccarino, F., Barres, B., Vogel, H., Tamminga, et al
ELSEVIER.2019: 1240
- **PROMISES AND CHALLENGES OF USING HUMAN IPSC MODELS TO VALIDATE RARE AND COMMON GENETIC VARIANTS OF NEUROPSYCHIATRIC DISORDERS**
Duan, J., Urban, A., Gejman, P.
ELSEVIER.2019: 1059–60
- **Structural variation in the sequencing era.** *Nature reviews. Genetics*
Ho, S. S., Urban, A. E., Mills, R. E.
2019
- **Approaches and Methods for Variant Analysis in the Genome of a Single Cell** *BIOMARKERS OF HUMAN AGING*
Abyzov, A., Vaccarino, F. M., Urban, A. E., Sarangi, V., Moskalev, A.
2019; 10: 203–28
- **Neural signatures of sleep in zebrafish.** *Nature*
Leung, L. C., Wang, G. X., Madelaine, R. n., Skariah, G. n., Kawakami, K. n., Deisseroth, K. n., Urban, A. E., Mourrain, P. n.
2019; 571 (7764): 198–204

- **Extensive and deep sequencing of the Venter/HuRef genome for developing and benchmarking genome analysis tools** *SCIENTIFIC DATA*
Zhou, B., Arthur, J. G., Ho, S. S., Pattni, R., Huang, Y., Wong, W. H., Urban, A. E.
2018; 5
- **Extensive and deep sequencing of the Venter/HuRef genome for developing and benchmarking genome analysis tools.** *Scientific data*
Zhou, B., Arthur, J. G., Ho, S. S., Pattni, R., Huang, Y., Wong, W. H., Urban, A. E.
2018; 5: 180261
- **Local and global chromatin interactions are altered by large genomic deletions associated with human brain development** *NATURE COMMUNICATIONS*
Zhang, X., Zhang, Y., Zhu, X., Purmann, C., Haney, M. S., Ward, T., Khechaduri, A., Yao, J., Weissman, S. M., Urban, A. E.
2018; 9
- **Whole-genome sequencing analysis of CNV using low-coverage and paired-end strategies is efficient and outperforms array-based CNV analysis** *JOURNAL OF MEDICAL GENETICS*
Zhou, B., Ho, S. S., Zhang, X., Pattni, R., Haraksingh, R. R., Urban, A. E.
2018; 55 (11): 735-743
- **Genome amplification and cellular senescence are hallmarks of human placenta development.** *PLoS genetics*
Velicky, P., Meinhardt, G., Plessl, K., Vondra, S., Weiss, T., Haslinger, P., Lendl, T., Aumayr, K., Mairhofer, M., Zhu, X., Schutz, B., Hannibal, R. L., Lindau, et al
2018; 14 (10): e1007698
- **Genome amplification and cellular senescence are hallmarks of human placenta development** *PLOS GENETICS*
Velicky, P., Meinhardt, G., Plessl, K., Vondra, S., Weiss, T., Haslinger, P., Lendl, T., Aumayr, K., Mairhofer, M., Zhu, X., Schuetz, B., Hannibal, R. L., Lindau, et al
2018; 14 (10)
- **1q21.1 microduplication: large verbal-nonverbal performance discrepancy and ddPCR assays of HYDIN/HYDIN2 copy number** *NPJ GENOMIC MEDICINE*
Xavier, J., Zhou, B., Bilan, F., Zhang, X., Gilbert-Dussardier, B., Viaux-Savelon, S., Pattni, R., Ho, S. S., Cohen, D., Levinson, D. F., Urbana, A. E., Laurent-Levinson, C.
2018; 3: 24
- **Whole-genome sequencing analysis of CNV using low-coverage and paired-end strategies is efficient and outperforms array-based CNV analysis.** *Journal of medical genetics*
Zhou, B., Ho, S. S., Zhang, X., Pattni, R., Haraksingh, R. R., Urban, A. E.
2018
- **ANALYZING THE MOLECULAR EFFECTS OF LARGE NEUROPSYCHIATRIC CNVS WITH IPSC BASED NEURONAL TISSUE CULTURE MODELS**
Urban, A.
OXFORD UNIV PRESS.2018: S33
- **Different mutational rates and mechanisms in human cells at pregastrulation and neurogenesis** *SCIENCE*
Bae, T., Tomasini, L., Mariani, J., Zhou, B., Roychowdhury, T., Franjic, D., Pletikos, M., Pattni, R., Chen, B., Venturini, E., Riley-Gillis, B., Sestan, N., Urban, et al
2018; 359 (6375): 550+
- **Detection and Quantification of Mosaic Genomic DNA Variation in Primary Somatic Tissues Using ddPCR: Analysis of Mosaic Transposable-Element Insertions, Copy-Number Variants, and Single-Nucleotide Variants.** *Methods in molecular biology (Clifton, N.J.)*
Zhou, B. n., Haney, M. S., Zhu, X. n., Pattni, R. n., Abyzov, A. n., Urban, A. E.
2018; 1768: 173–90
- **Detection and Quantification of Mosaic Genomic DNA Variation in Primary Somatic Tissues Using ddPCR: Analysis of Mosaic Transposable-Element Insertions, Copy-Number Variants, and Single-Nucleotide Variants** *DIGITAL PCR: METHODS AND PROTOCOLS*
Zhou, B., Haney, M. S., Zhu, X., Pattni, R., Abyzov, A., Urban, A. E., KarlinNeumann, G., Bizouarn, F.
2018; 1768: 173-190
- **Local and global chromatin interactions are altered by large genomic deletions associated with human brain development.** *Nature communications*
Zhang, X., Zhang, Y., Zhu, X., Purmann, C., Haney, M. S., Ward, T., Khechaduri, A., Yao, J., Weissman, S. M., Urban, A. E.
2018; 9 (1): 5356

- **Challenges and recommendations for epigenomics in precision health** *NATURE BIOTECHNOLOGY*
Carter, A. C., Chang, H. Y., Church, G., Dombkowski, A., Ecker, J. R., Gil, E., Giresi, P. G., Greely, H., Greenleaf, W. J., Hacohen, N., He, C., Hill, D., Ko, et al
2017; 35 (12): 1128–32
- **META-ANALYSIS OF WHOLE BLOOD GENE EXPRESSION IN MAJOR DEPRESSION: IDENTIFYING COHERENT GENE NETWORKS**
Mostafavi, S., Jansen, R., Battle, A., Zhu, X., Shi, J., Montgomery, S., Urban, A., Weissman, M., Potash, J., van Grootheest, G., Smit, J., Sullivan, P., Levinson, et al
ELSEVIER SCIENCE BV.2017: S288–S289
- **COMPREHENSIVE COMPARATIVE PERFORMANCE ANALYSIS OF HIGH-DENSITY OLIGOMER ARRAY PLATFORMS FOR THE DETECTION AND ANALYSIS OF HUMAN GENOMIC COPY NUMBER VARIATION**
Haraksingh, R., Abyzov, A., Urban, A.
ELSEVIER SCIENCE BV.2017: S301–S302
- **Attachment Insecurity and DNA Methylation in Risk for Postpartum Depression**
Robakis, T., On, B., Budhan, V., Crowe, S., Williams, K., Rasgon, N., Urban, A.
ELSEVIER SCIENCE INC.2017: S298
- **Intersection of diverse neuronal genomes and neuropsychiatric disease: The Brain Somatic Mosaicism Network** *SCIENCE*
McConnell, M. J., Moran, J. V., Abyzov, A., Akbarian, S., Bae, T., Cortes-Ciriano, I., Erwin, J. A., Fasching, L., Flasch, D. A., Freed, D., Ganz, J., Jaffe, A. E., Kwan, et al
2017; 356 (6336): 395-?
- **Characterizing regression in Phelan McDermid Syndrome (22q13 deletion syndrome).** *Journal of psychiatric research*
Reiersen, G., Bernstein, J., Froehlich-Santino, W., Urban, A., Purmann, C., Berquist, S., Jordan, J., O'Hara, R., Hallmayer, J.
2017; 91: 139-144
- **One thousand somatic SNVs per skin fibroblast cell set baseline of mosaic mutational load with patterns that suggest proliferative origin.** *Genome research*
Abyzov, A., Tomasini, L., Zhou, B., Vasmatzis, N., Coppola, G., Amenduni, M., Pattni, R., Wilson, M., Gerstein, M., Weissman, S., Urban, A. E., Vaccarino, F. M.
2017
- **Comprehensive performance comparison of high-resolution array platforms for genome-wide Copy Number Variation (CNV) analysis in humans.** *BMC genomics*
Haraksingh, R. R., Abyzov, A. n., Urban, A. E.
2017; 18 (1): 321
- **Principles and Approaches for Discovery and Validation of Somatic Mosaicism in the Human Brain** *GENOMIC MOSAICISM IN NEURONS AND OTHER CELL TYPES*
Abyzov, A., Urban, A. E., Vaccarino, F. M., Frade, J. M., Gage, F. H.
2017; 131: 3–24
- **Identification of Human Neuronal Protein Complexes Reveals Biochemical Activities and Convergent Mechanisms of Action in Autism Spectrum Disorders.** *Cell systems*
Li, J., Ma, Z., Shi, M., Malty, R. H., Aoki, H., Minic, Z., Phanse, S., Jin, K., Wall, D. P., Zhang, Z., Urban, A. E., Hallmayer, J., Babu, et al
2015; 1 (5): 361-374
- **Identification of Human Neuronal Protein Complexes Reveals Biochemical Activities and Convergent Mechanisms of Action in Autism Spectrum Disorders** *CELL SYSTEMS*
Li, J., Ma, Z., Shi, M., Malty, R. H., Aoki, H., Minic, Z., Phanse, S., Jin, K., Wall, D. P., Zhang, Z., Urban, A. E., Hallmayer, J., Babu, et al
2015; 1 (5): 361-374
- **46,XY disorders of sex development and congenital diaphragmatic hernia: A case with dysmorphic facies, truncus arteriosus, bifid thymus, gut malrotation, rhizomelia, and adactyly** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
Esplin, E. D., Chaib, H., Haney, M., Martin, B., Homeyer, M., Urban, A. E., Bernstein, J. A.
2015; 167A (6): 1360-1364
- **Using iPSCs and genomics to catch CNVs in the act.** *Nature genetics*
Urban, A. E., Purmann, C.
2015; 47 (2): 100-101
- **Type I interferon signaling genes in recurrent major depression: increased expression detected by whole-blood RNA sequencing.** *Molecular psychiatry*

- Mostafavi, S., Battle, A., Zhu, X., Potash, J. B., Weissman, M. M., Shi, J., Beckman, K., Haudenschild, C., McCormick, C., Mei, R., Geroff, M. J., Gindes, H., Adams, et al
2014; 19 (12): 1267-1274
- **Characterizing the genetic basis of transcriptome diversity through RNA-sequencing of 922 individuals** *GENOME RESEARCH*
Battle, A., Mostafavi, S., Zhu, X., Potash, J. B., Weissman, M. M., McCormick, C., Haudenschild, C. D., Beckman, K. B., Shi, J., Mei, R., Urban, A. E., Montgomery, S. B., Levinson, et al
2014; 24 (1): 14-24
 - **Integrated systems analysis reveals a molecular network underlying autism spectrum disorders.** *Molecular systems biology*
Li, J., Shi, M., Ma, Z., Zhao, S., Euskirchen, G., Ziskin, J., Urban, A., Hallmayer, J., Snyder, M.
2014; 10 (12): 774-?
 - **Integrated systems analysis reveals a molecular network underlying autism spectrum disorders.** *Molecular systems biology*
Li, J., Shi, M., Ma, Z., Zhao, S., Euskirchen, G., Ziskin, J., Urban, A., Hallmayer, J., Snyder, M.
2014; 10: 774-?
 - **Child Development and Structural Variation in the Human Genome** *CHILD DEVELOPMENT*
Zhang, Y., Haraksingh, R., Grubert, F., Abyzov, A., Gerstein, M., Weissman, S., Urban, A. E.
2013; 84 (1): 34-48
 - **Enriching DNA Sequences with Nucleotide Variation by Thymidine Glycosylases Combined with Suppression PCR** *PCR TECHNOLOGY: CURRENT INNOVATIONS, 3RD EDITION*
Pan, X., Urban, A. E., Weissman, S. M., Nolan, T., Bustin, S. A.
2013: 285-297
 - **Normalizing RNA-Sequencing Data by Modeling Hidden Covariates with Prior Knowledge.** *PloS one*
Mostafavi, S., Battle, A., Zhu, X., Urban, A. E., Levinson, D., Montgomery, S. B., Koller, D.
2013; 8 (7)
 - **Normalizing RNA-sequencing data by modeling hidden covariates with prior knowledge.** *PloS one*
Mostafavi, S., Battle, A., Zhu, X., Urban, A. E., Levinson, D., Montgomery, S. B., Koller, D.
2013; 8 (7)
 - **Somatic copy number mosaicism in human skin revealed by induced pluripotent stem cells** *NATURE*
Abyzov, A., Mariani, J., Palejev, D., Zhang, Y., Haney, M. S., Tomasini, L., Ferrandino, A. F., Belmaker, L. A., Szekely, A., Wilson, M., Kocabas, A., Calixto, N. E., Grigorenko, et al
2012; 492 (7429): 438-?
 - **Copy Number Variation detection from 1000 Genomes project exon capture sequencing data** *BMC BIOINFORMATICS*
Wu, J., Grzeda, K. R., Stewart, C., Grubert, F., Urban, A. E., Snyder, M. P., Marth, G. T.
2012; 13
 - **Extensive genetic variation in somatic human tissues** *PROCEEDINGS OF THE NATIONAL ACADEMY OF SCIENCES OF THE UNITED STATES OF AMERICA*
O'Huallachain, M., Karczewski, K. J., Weissman, S. M., Urban, A. E., Snyder, M. P.
2012; 109 (44): 18018-18023
 - **A Role of Genomic Copy Number Variation in the Complex Behavioral Phenotype of Alcohol Dependence: A Commentary** *ALCOHOLISM-CLINICAL AND EXPERIMENTAL RESEARCH*
Urban, A. E.
2012; 36 (9): 1483-1486
 - **Mutations in DNMT1 cause autosomal dominant cerebellar ataxia, deafness and narcolepsy** *HUMAN MOLECULAR GENETICS*
Winkelmann, J., Lin, L., Schormair, B., Kornum, B. R., Faraco, J., Plazzi, G., Melberg, A., Cornelio, F., Urban, A. E., Pizza, F., Poli, F., Grubert, F., Wieland, et al
2012; 21 (10): 2205-2210
 - **Genome-Wide Mapping of Copy Number Variation in Humans: Comparative Analysis of High Resolution Array Platforms** *PLOS ONE*
Haraksingh, R. R., Abyzov, A., Gerstein, M., Urban, A. E., Snyder, M.
2011; 6 (11)

- **A Comprehensive Map of Mobile Element Insertion Polymorphisms in Humans** *PLOS GENETICS*
Stewart, C., Kural, D., Stroemberg, M. P., Walker, J. A., Konkel, M. K., Stuetz, A. M., Urban, A. E., Grubert, F., Lam, H. Y., Lee, W., Busby, M., Indap, A. R., Garrison, et al
2011; 7 (8)
- **Identification of genomic indels and structural variations using split reads** *BMC GENOMICS*
Zhang, Z. D., Du, J., Lam, H., Abyzov, A., Urban, A. E., Snyder, M., Gerstein, M.
2011; 12
- **Demographic history and rare allele sharing among human populations** *PROCEEDINGS OF THE NATIONAL ACADEMY OF SCIENCES OF THE UNITED STATES OF AMERICA*
Gravel, S., Henn, B. M., Gutenkunst, R. N., Indap, A. R., Marth, G. T., Clark, A. G., Yu, F., Gibbs, R. A., Bustamante, C. D.
2011; 108 (29): 11983-11988
- **CNVnator: An approach to discover, genotype, and characterize typical and atypical CNVs from family and population genome sequencing** *GENOME RESEARCH*
Abyzov, A., Urban, A. E., Snyder, M., Gerstein, M.
2011; 21 (6): 974-984
- **The promise of stem cell research for neuropsychiatric disorders** *JOURNAL OF CHILD PSYCHOLOGY AND PSYCHIATRY*
Vaccarino, F. M., Urban, A. E., Stevens, H. E., Szekely, A., Abyzov, A., Grigorenko, E. L., Gerstein, M., Weissman, S.
2011; 52 (4): 504-516
- **Mapping copy number variation by population-scale genome sequencing** *NATURE*
Mills, R. E., Walter, K., Stewart, C., Handsaker, R. E., Chen, K., Alkan, C., Abyzov, A., Yoon, S. C., Ye, K., Cheetham, R. K., Chinwalla, A., Conrad, D. F., Fu, et al
2011; 470 (7332): 59-65
- **The functional spectrum of low-frequency coding variation** *GENOME BIOLOGY*
Marth, G. T., Yu, F., Indap, A. R., Garimella, K., Gravel, S., Leong, W. F., Tyler-Smith, C., Bainbridge, M., Blackwell, T., Zheng-Bradley, X., Chen, Y., Challis, D., Clarke, et al
2011; 12 (9)
- **A map of human genome variation from population-scale sequencing** *NATURE*
Altshuler, D., Durbin, R. M., Abecasis, G. R., Bentley, D. R., Chakravarti, A., Clark, A. G., Collins, F. S., De La Vega, F. M., Donnelly, P., Egholm, M., Flicek, P., Gabriel, S. B., Gibbs, et al
2010; 467 (7319): 1061-1073
- **Behavior, Brain, and Genome in Genomic Disorders: Finding the Correspondences** *JOURNAL OF DEVELOPMENTAL AND BEHAVIORAL PEDIATRICS*
Grigorenko, E. L., Urban, A. E., Mencl, E.
2010; 31 (7): 602-609
- **Variation in Transcription Factor Binding Among Humans** *SCIENCE*
Kasowski, M., Grubert, F., Heffelfinger, C., Hariharan, M., Asabere, A., Waszak, S. M., Habegger, L., Rozowsky, J., Shi, M., Urban, A. E., Hong, M., Karczewski, K. J., Huber, et al
2010; 328 (5975): 232-235
- **EBNA1 regulates cellular gene expression by binding cellular promoters** *PROCEEDINGS OF THE NATIONAL ACADEMY OF SCIENCES OF THE UNITED STATES OF AMERICA*
Canaan, A., Haviv, I., Urban, A. E., Schulz, V. P., Hartman, S., Zhang, Z., Palejev, D., Deisseroth, A. B., Lacy, J., Snyder, M., Gerstein, M., Weissman, S. M.
2009; 106 (52): 22421-22426
- **The genetic architecture of Down syndrome phenotypes revealed by high-resolution analysis of human segmental trisomies** *PROCEEDINGS OF THE NATIONAL ACADEMY OF SCIENCES OF THE UNITED STATES OF AMERICA*
Korbel, J. O., Tirosh-Wagner, T., Urban, A. E., Chen, X., Kasowski, M., Dai, L., Grubert, F., Erdman, C., Gao, M. C., Lange, K., Sobel, E. M., Barlow, G. M., Aylsworth, et al
2009; 106 (29): 12031-12036
- **Distinct Genomic Aberrations Associated with ERG Rearranged Prostate Cancer** *GENES CHROMOSOMES & CANCER*
Demichelis, F., Setlur, S. R., Beroukhi, R., Perner, S., Korbel, J. O., LaFargue, C. J., Pflueger, D., Pina, C., Hofer, M. D., Sboner, A., Svensson, M. A., Rickman, D. S., Urban, et al

2009; 48 (4): 366-380

- **Analysis of copy number variants and segmental duplications in the human genome: Evidence for a change in the process of formation in recent evolutionary history** *GENOME RESEARCH*
Kim, P. M., Lam, H. Y., Urban, A. E., Korbelt, J. O., Affourtit, J., Grubert, F., Chen, X., Weissman, S., Snyder, M., Gerstein, M. B.
2008; 18 (12): 1865-1874
- **High-Resolution Copy-Number Variation Map Reflects Human Olfactory Receptor Diversity and Evolution** *PLOS GENETICS*
Hasin, Y., Olender, T., Khen, M., Gonzaga-Jauregui, C., Kim, P. M., Urban, A. E., Snyder, M., Gerstein, M. B., Lancet, D., Korbelt, J. O.
2008; 4 (11)
- **A procedure for highly specific, sensitive, and unbiased whole-genome amplification** *PROCEEDINGS OF THE NATIONAL ACADEMY OF SCIENCES OF THE UNITED STATES OF AMERICA*
Pan, X., Urban, A. E., Palejev, D., Schulz, V., Grubert, F., Hu, Y., Snyder, M., Weissman, S. M.
2008; 105 (40): 15499-15504
- **MEDME: An experimental and analytical methodology for the estimation of DNA methylation levels based on microarray derived MeDIP-enrichment** *GENOME RESEARCH*
Pelizzola, M., Koga, Y., Urban, A. E., Krauthammer, M., Weissman, S., Halaban, R., Molinaro, A. M.
2008; 18 (10): 1652-1659
- **The current excitement about copy-number variation: how it relates to gene duplications and protein families** *CURRENT OPINION IN STRUCTURAL BIOLOGY*
Korbelt, J. O., Kim, P. M., Chen, X., Urban, A. E., Weissman, S., Snyder, M., Gerstein, M. B.
2008; 18 (3): 366-374
- **Systematic analysis of transcribed loci in ENCODE regions using RACE sequencing reveals extensive transcription in the human genome** *GENOME BIOLOGY*
QianWu, J., Du, J., Rozowsky, J., Zhang, Z., Urban, A. E., Euskirchen, G., Weissman, S., Gerstein, M., Snyder, M.
2008; 9 (1)
- **Paired-end mapping reveals extensive structural variation in the human genome** *SCIENCE*
Korbelt, J. O., Urban, A. E., Affourtit, J. P., Godwin, B., Grubert, F., Simons, J. F., Kim, P. M., Palejev, D., Carriero, N. J., Du, L., Taillon, B. E., Chen, Z., Tanzer, et al
2007; 318 (5849): 420-426
- **Identification and analysis of functional elements in 1% of the human genome by the ENCODE pilot project** *NATURE*
Birney, E., Stamatoyannopoulos, J. A., Dutta, A., Guigo, R., Gingeras, T. R., Margulies, E. H., Weng, Z., Snyder, M., Dermitzakis, E. T., Stamatoyannopoulos, J. A., Thurman, R. E., Kuehn, M. S., Taylor, et al
2007; 447 (7146): 799-816
- **Systematic prediction and validation of breakpoints associated with copy-number variants in the human genome** *PROCEEDINGS OF THE NATIONAL ACADEMY OF SCIENCES OF THE UNITED STATES OF AMERICA*
Korbelt, J. O., Urban, A. E., Grubert, F., Du, J., Royce, T. E., Starr, P., Zhong, G., Emanuel, B. S., Weissman, S. M., Snyder, M., Gerstein, M. B.
2007; 104 (24): 10110-10115
- **Assessing the performance of different high-density tiling microarray strategies for mapping transcribed regions of the human genome** *GENOME RESEARCH*
Emanuelsson, O., Nagalakshmi, U., Zheng, D., Rozowsky, J. S., Urban, A. E., Du, J., Lian, Z., Stolc, V., Weissman, S., Snyder, M., Gerstein, M. B.
2007; 17 (6): 886-897
- **High-resolution mapping of DNA copy alterations in human chromosome 22 using high-density tiling oligonucleotide arrays** *PROCEEDINGS OF THE NATIONAL ACADEMY OF SCIENCES OF THE UNITED STATES OF AMERICA*
Urban, A. E., Korbelt, J. O., Selzer, R., Richmond, T., Hacker, A., Popescu, G. V., Clubbells, J. F., Green, R., Emanuel, B. S., Gerstein, M. B., Weissman, S. M., Snyder, M.
2006; 103 (12): 4534-4539
- **Global identification of human transcribed sequences with genome tiling arrays** *SCIENCE*
Bertone, P., Stolc, V., Royce, T. E., Rozowsky, J. S., Urban, A. E., Zhu, X. W., Rinn, J. L., Tongprasit, W., Samanta, M., Weissman, S., Gerstein, M., Snyder, M.
2004; 306 (5705): 2242-2246

- **The ENCODE (ENCyclopedia of DNA elements) Project** *SCIENCE*

Feingold, E. A., Good, P. J., Guyer, M. S., Kamholz, S., Liefer, L., Wetterstrand, K., Collins, F. S., Gingeras, T. R., Kampa, D., Sekinger, E. A., Cheng, J., Hirsch, H., Ghosh, et al
2004; 306 (5696): 636-640

- **Identification of novel functional elements in the human genome** *67th Cold Spring Harbor Symposium on Quantitative Biology*

Lian, Z., Euskirchen, G., Rinn, J., Martone, R., Bertone, P., Hartman, S., Royce, T., Nelson, K., Sayward, F., Luscombe, N., Yang, J., Li, J. L., Miller, et al
COLD SPRING HARBOR LAB PRESS, PUBLICATIONS DEPT.2003: 317-322