Stanford



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, Wu Tsai Human Performance Alliance
- 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, Recruiting

Teaching

COURSES

2026-27

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

2025-26

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

2024-25

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

2023-24

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

STANFORD ADVISEES

Doctoral Dissertation Advisor (AC)

Ally Heesu Kim

GRADUATE AND FELLOWSHIP PROGRAM AFFILIATIONS

- Genetics (PhD Program)

Publications

PUBLICATIONS

- **Multi-platform framework for mapping somatic retrotransposition in human tissues.** *bioRxiv : the preprint server for biology*
  Wang, S., Bae, M., Wang, J., Zhao, B., Nguyen, K., Mallett, S., Switzenberg, J. A., Losh, S. J., Sexton, C. E., Miao, B., Dong, S., Zeng, X., Wang, et al
  2025

- **The Somatic Mosaicism across Human Tissues Network.** *Nature*
  Coorens, T. H., Oh, J. W., Choi, Y. A., Lim, N. S., Zhao, B., Voshall, A., Abyzov, A., Antonacci-Fulton, L., Aparicio, S., Ardlie, K. G., Bell, T. J., Bennett, J. T., Bernstein, et al
  2025; 643 (8070): 47-59

- **Cell-type specific global reprogramming of the transcriptome and epigenome in induced neurons with the 16p11.2 neuropsychiatric CNVs.** *European journal of human genetics : EJHG*
  Ward, T. R., Qu, P. P., Leung, L. C., Zhou, B., Muench, K. L., Khechaduri, A., Plastini, M. J., Charlton, C. A., Pattni, R., Ho, S., Ho, M., Huang, Y., Zhou, et al
  2025

- **Prioritizing disease-related rare variants by integrating gene expression data.** *PLoS genetics*
  Guo, H., Urban, A. E., Wong, W. H.
  2024; 20 (9): e1011412

- **Resolving the 22q11.2 deletion using CTLR-Seq reveals chromosomal rearrangement mechanisms and individual variance in breakpoints.** *Proceedings of the National Academy of Sciences of the United States of America*

Zhou, B., Purmann, C., Guo, H., Shin, G., Huang, Y., Pattni, R., Meng, Q., Greer, S. U., Roychowdhury, T., Wood, R. N., Ho, M., Dohna, H. Z., Abyzov, et al
2024; 121 (31): e2322834121

- **Synaptic pruning in schizophrenia is not classical.** *Brain, behavior, and immunity*
  Kalinowski, A., Urban, A. E.
  2024

- **Detection and analysis of complex structural variation in human genomes across populations and in brains of donors with psychiatric disorders** *Cell*
  Zhou, B., Arthur, J. G., Guo, H., et al
  2024; Published online September 30, 2024

- **Genomic data resources of the Brain Somatic Mosaicism Network for neuropsychiatric diseases.** *Scientific data*
  Garrison, M. A., Jang, Y., Bae, T., Cherskov, A., Emery, S. B., Fasching, L., Jones, A., Moldovan, J. B., Molitor, C., Pochareddy, S., Peters, M. A., Shin, J. H., Wang, et al
  2023; 10 (1): 813

- **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. P. 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. C. 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., Kebschull, 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., Hsiaol, 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)
- **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.
  edited by 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
- **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.
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  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.
  edited by 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

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