Google Scholar

Yanli Wang

M.D., Ph.D., FACMG, FAAP

Board-certified pediatrician and clinical geneticist with expertise in rare disease diagnosis, genomic medicine, and computational genomics. Research focuses on interpretation of noncoding variation and RNA regulatory mechanisms underlying rare and common diseases.

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CURRENT POSITION

Attending Physician, Division of Human Genetics - Clinical Genetics and Genomics

Children's Hospital of Philadelphia Jul 2025-curr

T32 Research Fellow (NIH/NIGMS 5T32GM008638), Division of Human Genetics - Clinical Genetics and Genomics

Children's Hospital of Philadelphia Jul 2025-curr

CERTIFICATION & LICENSURE

American Board of Medical Genetics and Genomics - Clinical Genetics

2025-curr

American Board of Pediatrics

2025-curr

Pennsylvania Medical License

2025-curr

Epic - Physician Builder Advanced

Children's Hospital of Philadelphia 2022-curr

EDUCATION & TRAINING

Residency, Combined Pediatric and Genetics

Children's Hospital of Philadelphia 2021.06-2025.06 Philadelphia, PA

M.D./Ph.D. Bioinformatics and Genomics

Penn State College of Medicine 2010.08-2020.12 Hershey, PA

- PhD Thesis: "Identification and Visualization of Regulatory Elements and 3D Genome Structure"
- Doctoral advisor: Feng Yue, PhD

B.Sc. Computer Science & Biological Sciences

University of Pittsburgh 2006.08-2009.12 Pittsburgh, PA

- Graduated *summa cum laude*

PROFESSIONAL AND LEADERSHIP EXPERIENCES

Member, American College of Medical Genetics and Genomics

2026-curr

Member, American Society of Human Genetics

2026-curr

Co-leader, NERD (New Epic Resident Development) Squad

Children's Hospital of Philadelphia 2021-2023

- Interfaces between residents, residency program and Epic (electronic health record) leadership
- Teaches Epic to new interns, helps current residents leverage the EMR's functionality.

RESEARCH HIGHLIGHTS & INTERESTS

Brain splicing QTL mapping reveals RNA processing mechanisms associated with autism spectrum disorder

- Generated brain-specific splicing quantitative trait locus (sQTL) maps using the MAJIQTL framework and GTEx transcriptomic datasets.
- Integrated sQTL maps with autism spectrum disorder (ASD) genome-wide association studies through colocalization and splicing transcriptome-wide association analyses.
- Identified candidate ASD-associated loci with evidence for RNA-processing mechanisms underlying disease susceptibility.
- Role: Lead computational analyst.

Decoding the function of non-coding genetic variants through the transcriptome

- Developing computational approaches to identify pathogenic non-coding variants affecting RNA splicing, RNA structure, and translational regulation.
- Integrating transcriptomic, genomic, and functional datasets to improve interpretation of exome/genome-negative rare disease cases.
- Applying machine learning approaches to model transcript-level consequences of genetic variation.
- Role: Lead computational analyst.

Constructing the 3D Genome Browser

- Integrated genomic, epigenomic, and chromatin conformation datasets to facilitate the functional annotation of genomic elements, giving insight into the function of non-coding elements and effects of non-coding genetic variants.
- Role: Lead computational architect, first-author
- The browser has over 640+ citations as of June 2026; the original publication received **High Attention Score** by Altmetric.

Exploring the Epigenomics and 3D Genome Structure of Zebrafish

- Contributed to large-scale functional annotation of the zebrafish genome through analysis of transcriptomic, epigenomic, and chromatin conformation datasets.
- Performed computational analyses of RNA-seq, ATAC-seq, ChIP-seq, DNA methylation, and Hi-C datasets.
- Role: Original lead analyst (graduated before project completion).

PEER-REVIEWED PUBLICATIONS

- Yang, H., Luan, Y., Liu, T., Lee, H.J., Fang, L., **Wang, Y.**, ...14 authors...Gerhard, G.S., Hardison, R.C., Wang, T., Cheng, K.C. & Yue, F. A map of cis-regulatory elements and 3D genome structures in zebrafish. Nature. 2020, Dec; 588(7837): 337-343.

- Zhang, J., Lee, D., Dhiman, V., Jiang, P., Xu, J....41 authors...**Wang, Y.**, ...6 authors...Cheng, C., Yue, F., Liu, X.S., White, K.P. & Gerstein, M. An integrative ENCODE resource for cancer genomics. Nature Communications. 2020, Jul; 11(1): 3696.

- **Wang, Y.**, Song, F., Zhang, B., Zhang, L., Xu, J., Kuang, D., Li, D., Choudhary M.N.K., Li, Y., Hu, M., Hardison, R., Wang, T., & Yue, F. The 3D Genome Browser: a web-based browser for visualizing 3D genome organization and long-range chromatin interactions. Genome Biology. 2018, Oct; 19(1): 151.

- Dixon, J.R., Xu, J., Dileep, V., Zhan, Y., Song, F....4 authors...**Wang, Y.**, ...22 authors...Ay, F., Noble, W.S., Dekker, J., Gilbert, D.M., & Yue, F. Integrative detection and analysis of structural variation in cancer genomes. Nature Genetics. 2018, Oct; 50(10): 1388-1398.

- Panganiban, R.P., **Wang, Y.**, Howrylak, J., Chinchilli, V.M., Craig, T.J., August, A., & Ishmael, F.T. Circulating MicroRNAs as Biomarkers in Allergic Rhinitis and Asthma. Journal of Allergy and Clinical Immunology. 2016, Mar; 137(5): 1423-32.

- Perez-Andreu, V., Roberts, K.G., Xu, H., Smith, C., Zhang, H., Yang, W., …20 authors… Yue, F., **Wang, Y.**, Pui, C.H., Jeha, S., Relling, M.V., Evans, W.E., Gerhard, D.S., Loh, M.L., Willman, C.L., Hunger, S.P., Mullighan, C.G., & Yang, J.J. A genome-wide association study of susceptibility to acute lymphoblastic leukemia in adolescents and young adults. Blood. 2015, Jan; 125(4): 680-6.

- Pope, B.D., Ryba, T., Dileep, V., Yue, F., Wu, W., Denas, O., Vera, D.L., **Wang, Y.**, ...6 authors... Snyder, M., Stamatoyannopoulos, J.A., Taylor, J., Hardison, R.C., Kahveci, T., Ren B., & Gilbert, D.M. Topologically-associating domains are stable units of replication-timing regulation. Nature. 2014, Nov; 515(7527): 402-5.

- Yue, F., Cheng, Y., Breschi, A., Vierstra, J., Wu, W., Ryba, T., Sandstrom, R., Ma, Z., Davis, C., Pope, B.D., Shen, Y., ...103 authors…, **Wang, Y.**, …20 authors… Gilbert, D.M., Hardison, R.C., Beer, A.M., Ren., B. A comparative encyclopedia of DNA elements in the mouse genome. Nature. 2014, Nov; 515(7527): 355-64.

- Ananda, G., Hile, S.E., Breski, A., **Wang, Y.**, Kelkar, Y., Makova, K.D., & Eckert, K.A. Microsatellite Interruptions Stabilize Primate Genomes and Exist as Population-Specific Single Nucleotide Polymorphisms within Individual Human Genomes. PLoS Genetics. 2014 Jul 17;10(7):e1004498.

- Kuppuswamy, U., Ananthasubramanian, S., **Wang, Y.**, Balakrishnan, N., & Ganapathiraju, M.K. Predicting gene ontology annotations of orphan GWAS genes using protein-protein interactions. Algorithms for Molecular Biology. 2014, 9(1):10.

CONFERENCES AND ABSTRACTS

- **Wang, Y.**, Wang, D, Gazzara, M., Pasaniuc, B., Barash, Y. Brain splicing QTL mapping reveals RNA processing mechanisms associated with autism spectrum disorder. Poster Presentation. RNADay@Penn 2026. Philadelphia, PA, USA.
- **Wang, Y.**, Mayk, C, Conlin, L.K., Bayram, Y., Spinner N.B., Montaño, C. P346: Pallister-Killian syndrome due to hexasomy 12p mosaicism found in diaphragmatic fibroblasts. Poster Presentation. ACMG Annual Clinical Genetics Meeting 2026. Baltimore, MD, USA.
- **Wang, Y.**, Wright, R., Medne, L., Keena, B., Zackai, E., Skraban, C., Reynoso Santos, F. P452: Male non-lethal FLNA phenotypes: Medical and counseling challenges. Poster Presentation. ACMG Annual Clinical Genetics Meeting 2025. Los Angeles, CA, USA.
- **Wang, Y.**, Zhang, B., Zhang, L., Wang, T. & Yue, F. Visualizing three-dimensional organization and long-range interactions of the mammalian genome with the 3D Genome Browser. Oral Presentation. RECOMB/ISCB RSG DREAM Conference 2015, Philadelphia, PA, USA.
- **Wang, Y.**, Zhang, B., Zhang, L., Wang, T. & Yue, F. Visualizing three-dimensional organization and long-range interactions of the mammalian genome with the 3D Genome Browser. Poster Presentation. ISMB/ECCB 2015, Dublin, Ireland.
- **Wang, Y.** & Yue, F. ENCODE element browser and the 3D genome browser. Oral Presentation. ENCODE 2015: Research Applications and Users Meeting, Potomac, MD, USA.

TECHNICAL EXPERTISE

- Computational Genomics: TWAS, QTL mapping, transcriptomics
- Programming: Python, R, Bash, Perl
- High-Performance Computing: Linux/Unix, Slurm
- Clinical Genomics: Variant interpretation