



## Nikhita Amod Gogate

- Affiliate, Department Funds
- Fellow in Genetics

### Publications

#### PUBLICATIONS

- **Resolving SLC6A1 variable expressivity with deep clinical phenotyping and Drosophila models.** *HGG advances*  
Jay, K. L., Gogate, N., Hall, P. I., Ezell, K. M., Andrews, J. C., Jangam, S. V., Pan, H., Pham, K., German, R., Gomez, V., Jellinek-Russo, E., Storch, E. A., Yamamoto, et al  
2026; 7 (1): 100541
- **GREGoR: accelerating genomics for rare diseases.** *Nature*  
Dawood, M., Heavner, B., Wheeler, M. M., Ungar, R. A., LoTempio, J., Wiel, L., Berger, S., Bernstein, J. A., Chong, J. X., Délot, E. C., Eichler, E. E., Lupski, J. R., Shojiaie, et al  
2025; 647 (8089): 331-342
- **Improving automated deep phenotyping through large language models using retrieval-augmented generation.** *Genome medicine*  
Garcia, B. T., Westerfield, L., Yelemali, P., Gogate, N., Rivera-Munoz, E. A., Du, H., Dawood, M., Jolly, A., Lupski, J. R., Posey, J. E.  
2025; 17 (1): 91
- **Domain specific phenotypic expansion associated with variants in MACF1.** *medRxiv : the preprint server for health sciences*  
Gogate, N., Jolly, A., Rosenfeld, J. A., Bahena-Carbajal, P., Bernstein, J. A., Bonner, D., Busa, T., Cristian, I., D'Souza, P., Friedman, J., Gorokhova, S., Haaf, T., Herman, et al  
2025
- **Considerations for reporting variants in novel candidate genes identified during clinical genomic testing.** *Genetics in medicine : official journal of the American College of Medical Genetics*  
Chong, J. X., Berger, S. I., Baxter, S., Smith, E., Xiao, C., Calame, D. G., Hawley, M. H., Rivera-Munoz, E. A., DiTroia, S., Bamshad, M. J., Rehm, H. L.  
2024: 101199
- **Beyond the exome: What's next in diagnostic testing for Mendelian conditions.** *American journal of human genetics*  
Wojcik, M. H., Reuter, C. M., Marwaha, S., Mahmoud, M., Duyzend, M. H., Barseghyan, H., Yuan, B., Boone, P. M., Groopman, E. E., Délot, E. C., Jain, D., Sanchis-Juan, A., Starita, et al  
2023; 110 (8): 1229-1248
- **Modeling and integration of N-glycan biomarkers in a comprehensive biomarker data model.** *Glycobiology*  
Lyman, D. F., Bell, A., Black, A., Dingerdissen, H., Cauley, E., Gogate, N., Liu, D., Joseph, A., Kahsay, R., Crichton, D. J., Mehta, A., Mazumder, R.  
2022; 32 (10): 855-870
- **Patterns of psychopathology and cognition in sex chromosome aneuploidy.** *Journal of neurodevelopmental disorders*  
Rau, S., Whitman, E. T., Schauder, K., Gogate, N., Lee, N. R., Kenworthy, L., Raznahan, A.  
2021; 13 (1): 61
- **COVID-19 biomarkers and their overlap with comorbidities in a disease biomarker data model.** *Briefings in bioinformatics*  
Gogate, N., Lyman, D., Bell, A., Cauley, E., Crandall, K. A., Joseph, A., Kahsay, R., Natale, D. A., Schriml, L. M., Sen, S., Mazumder, R.  
2021; 22 (6)

- **Influence of hydrological factors on bacterial community structure in a tropical monsoonal estuary in India.** *Environmental science and pollution research international*  
Parvathi, A., Catena, M., Jasna, V., Phadke, N., Gogate, N.  
2021; 28 (36): 50579-50592
- **OncoMX: A Knowledgebase for Exploring Cancer Biomarkers in the Context of Related Cancer and Healthy Data.** *JCO clinical cancer informatics*  
Dingerdissen, H. M., Bastian, F., Vijay-Shanker, K., Robinson-Rechavi, M., Bell, A., Gogate, N., Gupta, S., Holmes, E., Kahsay, R., Keeney, J., Kincaid, H., King, C. H., Liu, et al  
2020; 4: 210-220
- **A Targeted Next Generation Sequencing Panel for Non-syndromic Early Onset Severe Obesity and Identification of Novel Likely - Pathogenic Variants in the MC4R and LEP Genes.** *Indian journal of pediatrics*  
Khadilkar, V., Gogate, N., Gangodkar, P., Ranade, S., Lohiya, N., Ragte, T., Patil, K., Pilankar, A., Anand, S., Bapat, A., Agarwal, M., Kaushik, K., Lote-Oke, et al  
2020; 87 (2): 105-110