Dr. Tahir Khan
Associate Professor
tahir.khan@numspak.edu.pk
https://scholar.google.com/citations?user=wKUMWlUAAAAJ&hl=en

Profile

Dr. Khan is an Associate Professor in the Department of Molecular Biology and Genetics at the National University of Medical Sciences (NUMS), Pakistan. He holds a PhD in Biotechnology and has over a decade of research experience in human genetics, functional genomics, and rare disease biology. His research focuses on identifying disease-causing genetic variants and dissecting their molecular and developmental mechanisms using next-generation sequencing, transcriptome profiling, and CRISPR-based genome editing. A major component of his work involves in vivo and in vitro disease modeling, integrating genomic, cellular, and neurodevelopmental phenotyping. Dr. Khan has held research positions at Duke University (USA), Northwestern University (USA), Lurie Children's Hospital of Chicago (USA), Queen Mary University of London (UK), and Uppsala University (Sweden). He has published extensively in leading journals and has contributed to the discovery and functional characterization of several novel disease-causing genes and syndromes including eponymous Khan-Khan-Katsanis syndrome. At NUMS, he is actively involved in teaching, research mentorship, and in development of functional genomics facilities.

Research Interests

Human Genetics, Functional Genomics, Human Disease Modeling

Selected Publications

Roychaudhury A, Lee YR, Choi TI, Thomas MG, Khan TN, Yousaf H, et al. (2024) SRPK3 Is Essential for Cognitive and Ocular Development in Humans and Zebrafish, Explaining X-Linked Intellectual Disability. Annals of Neurology.

Grange LJ, Reynolds JJ, Ullah F, Isidor B, Shearer RF, Latypova X, ..., Khan TN et al. (2022) Pathogenic variants in SLF2 and SMC5 cause segmented chromosomes and mosaic variegated hyperploidy. Nature Communication.

Khan TN, Khan K, Sadeghpour A, Reynolds H, Perilla Y, McDonald MT, Gallentine WB, Baig SM, Task Force for Neonatal G, Davis EE, Katsanis N (2019) Mutations in NCAPG2 Cause a Severe Neurodevelopmental Syndrome that Expands the Phenotypic Spectrum of Condensinopathies. American Journal of Human Genetics 104: 94-111

Stankiewicz P, Khan TN, Szafranski P, Slattery L, Streff H, Vetrini F, Bernstein JA, Brown CW, Rosenfeld JA, Rednam S, Scollon S, Bergstrom KL, Parsons DW, Plon SE, Vieira MW, Quaio C, Baratela WAR, Acosta Guio JC, Armstrong R, Mehta SG et al. (2017) Haploinsufficiency of the Chromatin Remodeler BPTF Causes Syndromic Developmental and Speech Delay, Postnatal Microcephaly, and Dysmorphic Features. American journal of human genetics 101: 503-515

Ta-Shma A, Khan TN, Vivante A, Willer JR, Matak P, Jalas C, Pode-Shakked B, Salem Y, Anikster Y, Hildebrandt F, Katsanis N, Elpeleg O, Davis EE (2017) Mutations in TMEM260 Cause a Pediatric Neurodevelopmental, Cardiac, and Renal Syndrome. American journal of human genetics 100: 666-675