

JSSAHER / DBT BUILDER Project



Dr. Arshia Angural
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Education details:

▪ Ph.D. Biotechnology

School of Biotechnology, Shri Mata Vaishno Devi University, Jammu and Kashmir, India

The title of doctoral thesis is “*Identification and Characterization of Rare Human Genetic Disorders in Consanguineous Population of Jammu & Kashmir*”.

Mentors: Dr. Swarkar Sharma, Ph.D., and Prof. Vijeshwar Verma, Ph.D.

▪ M.Sc. Medical Biotechnology

Manipal Life Sciences Centre, Manipal University, Karnataka, India

The title of research project dissertation is “*Study of genetic variations in host factors and their interaction for malaria susceptibility and resistance to parasite*”.

Mentor: Dr. Saadi Abdul Vahab, Ph.D

Areas of interest: Clinical Genetics, Genetics of Rare Diseases especially Neurodegenerative Disorders, Pharmacogenetics, Genomics, Newborn Screening.

Complete list of publications with impact factor in chronological order: *h-index* = 6

- 2023
- Birla, S., **Angural, A.**, Madathumchalil, A., Shende, R. V., Shastry, S. V., Mahadevappa, M., Shambhu, S. K., Vishwanath, P., and Prashant, A. “Redefining the polypill: pros and cons in cardiovascular precision medicine.” *Frontiers in Pharmacology*. 14:1268119. (**IF** = **5.6**; doi: 10.3389/fphar.2023.1268119)
 - Spolia, A., **Angural, A.**, Sharma, V., Shipra, Razdan, S., Dhar, M. K., Mahajan, A., Verma, V., Pandita, K. K., S. Sharma, and E. Rai. “Cost-effective Whole Exome Sequencing discovers pathogenic variant causing Neurofibromatosis type 1 in a family from Jammu and Kashmir, India.” *Scientific Reports* 13: 7852. (**IF** = **4.6**; doi: 10.1038/s41598-023-34941-y)
 - **Angural, A.**, Birla, S., Srinivas, A., Prasanth, A. and Hattur, B.G. “Birth Defects: A Call for Action.” *NAM S&T Centre – Fact File* 1-4 (Document No.: STI-

FF/02/2023).

- 2022 ■ Spolia, A., Shipra, **Angural, A.**, Singh, H., Verma, V., Pandita, K.K., Sharma, S. and Rai, E., 2022. Genetic Characterisation of Pantothenate Kinase Associated Neurodegeneration (PKAN) in a Consanguineous Family from Jammu and Kashmir, India. *International Journal of Human Genetics*, 22(3), pp.225-232.
- 2021 ■ **Angural, A.**, K. Ponnusamy, D. Langeh, M. Kumari, A. Spolia, E. Rai, A. Sharma, K.K. Pandita and Sharma, S. “Missense Variation in *TPPI* Gene causes Neuronal Ceroid Lipofuscinosis Type 2 in a Family from Jammu and Kashmir-India.” *Preprints* 2021070661 (doi: 10.20944/preprints202107.0661.v1)
- 2020 ■ **Angural, A.**, A. Spolia, A. Mahajan, V. Verma, A. Sharma, P. Kumar, M. K. Dhar, K. K. Pandita, E. Rai and S. Sharma. “Review: Understanding Rare Genetic Diseases in Low Resource Regions Like Jammu and Kashmir - India.” *Frontiers in Genetics* 11: 415. (IF = 3.7; doi: 10.3389/fgene.2020.00415)
- 2019 ■ **Angural, A.**, I. Sharma, P. Pandoh, V. Sharma, A. Spolia, E. Rai, V. Singh, S. Razdan, K. K. Pandita and S. Sharma. “A case report on a novel MT-ATP6 gene variation in atypical mitochondrial Leigh syndrome associated with bilateral basal ganglia calcifications.” *Mitochondrion* 46: 209-213. (IF = 4.4; doi: 10.1016/j.mito.2018.06.005)
- 2018 ■ Sharma, V., I. Sethi, I. Sharma, G. Singh, A. Mahajan, **A. Angural**, A.J.S. Bhanwer, M. K. Dhar, K. K. Pandita, V. Singh, E. Rai and S. Sharma. “Replication of *MACF1* gene variant rs2296172 with type 2 diabetes susceptibility in the Bania population group of Punjab, India.” *International Journal of Diabetes in Developing Countries* 38: 387-390. (IF = 0.9; doi: <https://doi.org/10.1007/s13410-017-0598-6>)
- 2017 ■ **Angural, A.**, I. Singh, A. Mahajan, P. Pandoh, M. K. Dhar, S. Kaul, V. Verma, E. Rai, S. Razdan, K. K. Pandita and S. Sharma. “A variation in *PANK2* gene is causing Pantothenate kinase-associated Neurodegeneration in a family from Jammu and Kashmir-India.” *Scientific Reports* 7: 4834. (IF = 4.6; doi: 10.1038/s41598-017-05388-9)
- Rai, E., **A. Angural**, A. Sapolia, S. Razdan, K. K. Pandita, and S. Sharma. “Geographic Isolation and Endogamous Practices Provide Higher Risk of Genetic Disorders in Jammu and Kashmir.” *Canadian Journal of Biotechnology* 1 (Special), 250. (abstract publication; doi: <https://doi.org/10.24870/cjb.2017-a234>)
- Sharma, V., I. Sethi, I. Sharma, A. Mahajan, **A. Angural**, A.J.S. Bhanwer, M. K. Dhar, K. K. Pandita, V. Singh, E. Rai and S. Sharma. “*MACF1* gene variant rs2296172 is associated with type 2 diabetes susceptibility in the Bania population group of Punjab-India.” *Canadian Journal of Biotechnology* 1 (Special), 79. (abstract publication; doi: <https://doi.org/10.24870/cjb.2017-a66>)
- Sharma, V., I. Sharma, I. Sethi, A. Mahajan, G. Singh, **A. Angural**, A.J.S. Bhanwer, M.K. Dhar, V. Singh, E. Rai and S. Sharma. “Replication of Newly Identified Type 2 Diabetes Susceptible Loci in Northwest Indian Population.” *Diabetes Research and Clinical Practice* 126: 160-163. (IF = 5.1; doi:10.1016/j.diabres.2017.02.013)
- 2016 ■ Rai, E., A. Mahajan, P. Kumar, **A. Angural**, M. K. Dhar, S. Razdan, K. Thangaraj, C. A. Wise, S. Ikegawa, K. K. Pandita and S. Sharma. “Whole Exome Screening Identifies Novel and Recurrent *WISP3* Mutations Causing Progressive Pseudorheumatoid Dysplasia in Jammu and Kashmir-India.” *Scientific Reports* 6: 27684. (IF = 4.6; doi: 10.1038/srep27684)
- 2013 ■ Saadi, A. V., H. Gupta, **A. Angural**, S. K. Dhanya, S. Mony, D. Oberoi, S. C. D'Souza, R. C. Sahoo, M. H. Hande, P. M. Gopinath and K. Satyamoorthy. “Single

nucleotide polymorphisms of ADRB2 gene and their association with susceptibility for Plasmodium falciparum malaria and asthma in an Indian population.” *Infection, Genetics and Evolution* 20: 140-147. (IF = 4.4; doi: 10.1016/j.meegid.2013.08.026)

- Gupta, H., S. C. Sakharwade, A. Angural, A. Kotambail, G. K. Bhat, M. H. Hande, S. C. D'Souza, P. Rao, V. Kumari, A. V. Saadi and K. Satyamoorthy. “Evidence for genetic linkage between a polymorphism in the GNAS gene and malaria in South Indian population.” *Acta Tropica* 128(3): 571-577. (IF = 2.7; doi: 10.1016/j.actatropica.2013.08.005)

Grant details, if any:

- Awarded travel grant “International Travel Scheme” (ITS) from DST-SERB for attending and presenting research work at the American Society of Human Genetics Annual Meeting 2019, Houston, USA (15th October to 19th October, 2019).

Achievements:

- Awarded 3rd position in BIOBIZ contest in TITIKSHA (technical fest), SMVDU (2016).
- Qualified CSIR-UGC National Eligibility Test (NET) for Lectureship in Life Sciences, India (June, 2013).
- Qualified national-level examination “Graduate Aptitude Test in Engineering” (GATE) in Biotechnology, India (2013).

Memberships:

- Membership for the Indian Society of Human Genetics.
- Annual Membership for the American Society of Human Genetics (2019).