

Curriculum vitae



Zafar Ali, PhD

Assistant professor

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Education

PhD (Biotechnology)	National Institute for Biotechnology & Genetic Engineering (NIBGE), Faisalabad, Pakistan (2013-2018) PhD research conducted at Department of Immunology, Genetics and Pathology, Uppsala Biomedical Centre, Uppsala University, Sweden, from September 2014 to September 2016. PhD thesis title: Elucidating the Genetic Basis of Neurological Disorders in Pakistani Population
M.Phil (Molecular Biology & Biotechnology)	Institute of Biochemistry and Biotechnology, UVAS Lahore (2008-2010) M.Phil thesis title: Molecular Investigation of Mental Retardation Locus (MRT1)/Gene PRSS12 by LinkageAnalysis
BS Hons (Biotechnology)	University of Malakand, Pakistan (2003-2008)

Professional Experience

- **Assistant professor** at Centre for Biotechnology and Microbiology, University of Swat, Swat, Pakistan, May 3rd 2021- Present
- **Postdoctoral Researcher** at Department of Cellular and Molecular Medicine, Medical Genetics Program, University of Copenhagen, Blegdamsvej 3B, 2200 København, from 01-06-2020 to 30-04-2021.
- **Sr. Researcher/Postdoc fellow** at Human Molecular Genetics Laboratory, National Institute for Biotechnology and Genetic Engineering (NIBGE), Faisalabad, Pakistan from 01-07-2019 to 31-03-2020
- **Researcher** at Human Molecular Genetics Laboratory, National Institute for Biotechnology and Genetic Engineering (NIBGE), Faisalabad, Pakistan from 31-05-2012 to 30-06-2019.

- **Guest Researcher** at Department of Immunology, Genetics and Pathology, Uppsala Biomedical Centre, Uppsala University, Sweden, from September 2014 to September 2016.

Professional Interests

I am interested in elucidating the molecular genetic basis of human abnormal phenotypes (both simple and complex) and understanding their patho-mechanisms both at cellular and at organismal level in model systems such as zebrafish.

Skills and Expertise

Human molecular genetics, Whole-exome sequencing, SNP array genotyping, Homozygosity mapping, Sanger sequencing, Gene cloning and expression, Using cultured cell and Zebrafish as animal model to study human diseases, Bioinformatics tools for mutation analysis, phenotypic investigations of families with inherited disorders.

Awards/Fellowships

- **September 2014 - September 2015:** Awarded fellowship (guest PhD) from Department of Immunology, Genetics and Pathology, Uppsala Biomedical Centre, Uppsala University, Sweden (120000 SEK).
- **February 2016 - September 2016:** Higher Education Commission of Pakistan's IRSIP scholarship for sandwich PhD at Department of Immunology, Genetics and Pathology

Mentoring/Training Activities

- Training of two MS students one each in 2017 and 2018 at NIBGE
- Training of two PhD student (2018-2019) at NIBGE

Editor/ Reviewer (Journals)

- Acta Neurologica Belgica (ISSN: 0300-9009), Springer Nature (Italy)

Publications (Total I.F = 77.047)

1. E.U.H. Makhdoom, S.S. Waseem, M. Iqbal, U. Abdullah, G. Hussain, M. Asif, B. Budde, W. Höhne, S. Tinschert, S.M. Saadi, H. Yousaf, **Zafar Ali**, A. Fatima, E. Kaygusuz, A. Khan, M. Jameel, S. Khan, M. Tariq, I. Anjum, J. Altmüller, H. Thiele, S. Höning, S.M. Baig, P. Nürnberg, M.S. Hussain, "Modifier Genes in Microcephaly: A Report on WDR62, CEP63, RAD50 and PCNT Variants Exacerbating Disease Caused by Biallelic Mutations of ASPM and CENPJ", Genes, vol. 12, no. 5, pp. 731, 2021. <https://doi.org/10.3390/genes12050731>
I.F = 3.759
2. S. Faryal, M. Farooq, U. Abdullah, **Zafar Ali**, S. M. Saadi, F. Ullah, K. Khan, Y. Sarwar, M. Sher, A. A. Chopra, N. Tommerup, S. M. Baig, "A GDF5 frameshift mutation segregating with Grebe type chondrodysplasia and Brachydactyly Type C+ in a 6 generations family: Clinical report and mini review", European Journal of Medical Genetics, vol. 64, no. 7, p. 104226, (2021).
I.F = 2.368
3. A. Fatima, J. Hoeber, J. Schuster, E. Koshimizu, C. Maya-Gonzalez, B. Keren, C. Mignot, T. Akram, **Zafar Ali**, S. Miyatake, J. Tanigawa, T. Koike, M. Kato, Y. Murakami, U. Abdullah,

- M. A. Ali, R. Fadoul, L. Laan, C. Castillejo-López, M. Liik, Z. Jin, B. Birnir, N. Matsumoto, S. M. Baig, J. Klar, N. Dahl, "Monoallelic and bi-allelic variants in NCDN cause neurodevelopmental delay, intellectual disability, and epilepsy," *The American Journal of Human Genetics*, vol. 108, no. 4, pp. 739-748, (2021). **I.F = 10.502**
4. S. Rasool, J. M. Baig, A. Moawia, I. Ahmad, M. Iqbal, S. S. Waseem, M. Asif, U. Abdullah, E. U. H. Makhdoom, E. Kaygusuz, M. Zakaria, S. Ramzan, S. u. Haque, A. Mir, I. Anjum, M. Fiaz, **Zafar Ali**, M.Tariq, N. Saba, W. Hussain, B. Budde, S. Irshad, A. A. Noegel, S. Höning, S. M. Baig, P. Nürnberg, M. S. Hussain, "An update of pathogenic variants in ASPM, WDR62, CDK5RAP2, STIL, CENPJ, and CEP135 underlying autosomal recessive primary microcephaly in 32 consanguineous families from Pakistan", *Molecular genetics & genomic medicine*, vol. 8, no. 9, pp. e1408, (2020). **I.F = 1.995**
 5. S. Zulfiqar, M. Tariq, **Zafar Ali**, A. Fatima, J. Kla2, U. Abdullah, A. Ali, S. Ramzan, S. He, J. Zhang, A. Khan, S. Shah, S. Khan, E. H. Makhdoom, J. Schuster, N. Dahl, S. M. Baig. Whole Exome Sequencing Identifies Novel Mutation Underlying Hereditary Spastic Paraparesis (HSP) in Consanguineous Pakistani Families. *Journal of Clinical Neuroscience*, doi: 10.1016/j.jocn.2019.06.039, vol. 67, pp. 19-23, (2019). **I.F= 1.76**
 6. M. Sher, M. Farooq, U. Abdullah, **Zafar Ali**, S. Faryal, M. Zakaria, F. Ullah, H. Bukhari, R.S. Møller, N. Tommerup, S. M. Baig,. A novel in-frame Mutation in CLN3 Leads to Juvenile Neuronal Ceroid Lipofuscinosis in a large Pakistani family. *International Journal of Neurosciences*, vol. 129, no. 9, pp. 890-895, (2019). **I.F= 1.848**
 7. M. Zakaria, A. Fatima, J. Klar, J. Wikstrom, U. Abdullah , **Zafar Ali**, A. Talia, M. Tariq, H. Ahmad, J. Schuster, S. M. Baig, N. Dahl,. Primary microcephaly, primordial dwarfism and brachydactyly in adult cases with bi-allelic skipping of RTTN exon 42. *Human Mutation*, doi:10.1002/humu.23755, vol. 40, no. 7, pp. 899-903, (2019). **I.F= 4.453**
 8. S. A. Shah, M. E. Babar, J. Ahmad and **Zafar Ali**, Linkage Analysis of Autosomal Recessive Non-syndromic Mental Retardation Locus in Pakistani Families. *International Journal of Human Genetics*, DOI: 10.31901/24566330.2019/19.1-3.731, Vol. 19, no. 3, pp 152-157 (2019). **I.F= 0.172**
 9. S.M. Baig, A. Fatima, M. Tariq, T.N. Khan, **Zafar Ali**, M. Faheem, H. Mahmood, P. Killela, M. Waitkus, Y. He, F. Zhao, S. Wang, Y. Jiao, H. Yan, Hereditary brain tumor with a homozygous germline mutation in PMS2: pedigree analysis and prenatal screening in a family with constitutional mismatch repair deficiency (CMMRD) syndrome. *Familial cancer*, Nov 26, (2018). **I.F= 2.209**
 10. **Zafar Ali**, S. Zulfiqar, J. Klar, F. Ullah, A. Khan, U. Abdullah, S.M. Baig, J. Wikström, N. Dahl, Homozygous GRID2 missense mutation predicts a shift in the D-serine binding domain of GluD2 in a case with generalized brain atrophy and unusual clinical features, *BMC Medical Genetics*, doi. 10.1186/s12881-017-0504-6, vol. 18, no. 1. pp 144, (2017). **I.F= 1.740**
 11. J. Klar*, **Zafar Ali***, M. Farooq, K. Khan, J. Wikström, M. Iqbal, S. Zulfiqar, S. Faryal, S.M. Baig, N. Dahl. A missense variant in ITPR1 provides evidence for autosomal recessive SCA29 with asymptomatic cerebellar hypoplasia in carriers, *European Journal of Human Genetics*, vol.25, no. 7, pp. 848-853, (2017). **I.F= 3.657**
(* = Joint first author)
 12. A. Moawia, R. Shaheen, S. Rasool, S. S. Waseem, N. Ewida, B. Budde, A. Kawalia, S. Motameny, K. Khan, A. Fatima, M. Jameel, F. Ullah, T. Akram, **Zafar Ali**, U. Abdullah, S.

Irshad, W. Hohne, A. A. Noegel, M. Al-Owain, K. Hortnagel, P. Stobe, S. M. Baig, P. Nurnberg, F. S. Alkuraya, A. Hahn, and M. S. Hussain, "Mutations of KIF14 cause primary microcephaly by impairing cytokinesis," *Annals of Neurology*, doi: 10.1002/ana.25044, vol. 82, no. 4, pp 562-577, Sep 11 (2017). **I.F= 9.496**

13. **Zafar Ali**, J. Klar, M. Jameel, K. Khan, A. Fatima, R. Raininko, S. M. Baig, N. Dahl, "Novel SACS mutations associated with intellectual disability, epilepsy and widespread supratentorial abnormalities," *Journal of the Neurological Sciences*, vol. 371, pp. 105-111, Dec 15 (2016). **I.F= 3.115**

14. C. A. Martin, I. Ahmad, A. Klingseisen, M. S. Hussain, L. S. Bicknell, A. Leitch, G. Nurnberg, M. R. Toliat, J. E. Murray, D. Hunt, F. Khan, **Zafar Ali**, S. Tinschert, J. Ding, C. Keith, M. E. Harley, P. Heyn, R. Muller, I. Hoffmann, V. C. Daire, H. Dollfus, L. Dupuis, A. Bashamboo, K. McElreavey, A. Kariminejad, R. Mendoza-Londono,..... S. M. Baig, P. Nürnberg, A. P. Jackson, Mutations in PLK4, encoding a master regulator of centriole biogenesis, cause microcephaly, growth failure and retinopathy. *Nature Genetics*, vol. 46, no. 12, pp 1283-1292. (2014). **I.F= 27.605**

15. T. N. Khan, J. Klar, **Zafar Ali**, F. Khan, S.M. Baig, N. Dahl, Cenani-Lenz syndrome restricted to limb and kidney anomalies associated with a novel LRP4 missense mutation. *European Journal of Medical Genetics*, vol. 56, no. 7, pp 371-374, (2013). **I.F= 2.368**

16. **Zafar Ali**, M. E. Babar, J. Ahmad, S. A. Shah, The study of gene GJB2/DFNB1 causing deafness in humans by linkage analysis from district Peshawar. *Indian Journal of Human Genetic*, Doi. 10.4103/0971-6866.100771. Vol. 18 no. 2 pp 217-221, (2012).

17. **Zafar Ali**, M. E. Babar, J. Ahmad, M. Zubair Yousaf, M. Asif, S. A. Shah, Molecular investigation of mental retardation locus gene PRSS12 by linkage analysis. *Indian Journal of Human Genetic*, Vol. 17, no. 2, pp 65--69, (2011).

Conference papers/ abstract

1. M. Iqbal, S.M. Baig, U. Abdullah, E.UH. Makhdoom, **Zafar Ali**, S. Khan, M. Jameel, A.A. Noegel, P. Nuernberg, M. Osmond, M.S. Hussain "Mutations of PCDHGC4 encoding protocadherin gamma-C4 cause primary microcephaly and intellectual disability", vol. 28, pp. 399-401, 2020. (53rd European Society of Human Genetics (ESHG) Conference).
2. A. Fatima, J. Schuster, J. Hoeber, J. Klar, L. Laan, R. Fadoul, **Zafar Ali**, et al. "Neurochondrin missense variant associated with autosomal recessive intellectual disability and epilepsy." In *European Journal of Human Genetics*, vol. 27, pp. 1397-1398, 2019. (52nd European Society of Human Genetics (ESHG) Conference).
3. AIA Khayyat, SM Baig, U Abdullah, EU Haq, **Zafar Ali**, NA Malik, M Tariq, B Budde, AA Noegel, P Nuernberg, MS Hussain, "Mutation in C1orf131, encoding a novel nucleolar protein, causes intellectual disability in a large Pakistani family", vol. 27, pp. 1392-1393, 2019. (52nd European Society of Human Genetics (ESHG) Conference).

References

1. **Dr. Shahid Mahmood Baig, PhD**
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