ASMA ALI KHAN

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ACADEMIC QUALIFICATION

Nov 2012	**PhD**, Life and Health Sciences **University of Lorraine, FRANCE**
Jun 2009	**MS** Structural, Cellular and Molecular Biology **University of Henri Poincaré Nancy 1, FRANCE**
Jun 2006	**M.Sc.** Microbiology and Molecular Genetics **University of the Punjab, Lahore PAKISTAN**

WORK EXPERIENCE

June 2016 to date	Assistant Professor (TTS) CEMB, University of the Punjab Lahore, PAKISTAN
Feb 2013 to Feb 2016	Assistant Professor (Contract) CEMB, University of the Punjab Lahore, PAKISTAN

LIST OF PUBLICATIONS

- Ali M, Khan SY, Rodrigues TA, Francisco T, Jiao X, Qi H, Kabir F, Irum B, Rauf B, **Khan AA**, Mehmood A, Naeem MA, Assir MZ, Ali MH, Shahzad M, Abu-Amero KK, Akram SJ, Akram J, Riazuddin S, Riazuddin S, Robinson ML, Baes M, Azevedo JE, Hejtmancik JF, Riazuddin SA. ***A missense allele of PEX5 is responsible for the defective import of PTS2 cargo proteins into peroxisomes***. Hum Genet. 2021 Apr;140(4):649-666. doi: 10.1007/s00439-020-02238-z.
- Khan SY, Ali M, Lee MW, Ma Z, Biswas P, **Khan AA**, Naeem MA, Riazuddin S, Riazuddin S, Ayyagari R, Hejtmancik JF, Riazuddin SA. ***Whole genome sequencing data of multiple individuals of Pakistani descent.*** Sci Data. 2020 Oct 13;7(1):350. doi: 10.1038/s41597-020-00664-2.
- Tona R, Lopez IA, Fenollar-Ferrer C, Faridi R, Anselmi C, **Khan AA**, Shahzad M, Morell RJ, Gu S, Hoa M, Dong L, Ishiyama A, Belyantseva IA, Riazuddin S, Friedman TB. ***Mouse Models of Human Pathogenic Variants of TBC1D24 Associated with Non-Syndromic Deafness DFNB86 and DFNA65 and Syndromes Involving Deafness.*** Genes (Basel). 2020 Sep 24;11(10):1122. doi: 10.3390/genes11101122. **Impact Factor: 3.886**
- Richard EM, Polla DL, Assir MZ, Contreras M, Shahzad M, **Khan AA**, Razzaq A, Akram J, Tarar MN, Blanpied TA, Ahmed ZM, Abou Jamra R, Wieczorek D, van Bokhoven H, Riazuddin S, Riazuddin S. ***Bi-allelic Variants in METTL5 Cause Autosomal-Recessive Intellectual Disability and Microcephaly.*** Am J Hum Genet. 2019 Oct 3;105(4):869-878. doi: 10.1016/j.ajhg.2019.09.007.
- Faridi R, Tona R, Brofferio A, Hoa M, Olszewski R, Schrauwen I, Assir MZK, Bandesha AA, **Khan AA**, Rehman AU, Brewer C, Ahmed W, Leal SM, Riazuddin S, Boyden SE, Friedman TB. ***Mutational and Phenotypic Spectra of KCNE1 deficiency in Jervell and Lange-Nielsen Syndrome and Romano-Ward Syndrome.*** Hum Mutat. 2019 Feb;40(2):162-176. doi: 10.1002/humu.23689.
- Richard EM, Santos-Cortez RL, Faridi R, Rehman AU, Lee K, Shahzad M, Acharya A, **Khan AA**, Imtiaz A, Chakchouk I, Takla C, Abbe I, Rafeeq M, Liaqat K, Chaudhry T, Bamshad MJ, Nickerson DA; University of Washington Center for Mendelian Genomics, Schrauwen I, Khan SN, Morell RJ, Zafar S, Ansar M, Ahmed ZM, Ahmad W, Riazuddin S, Friedman TB, Leal SM, Riazuddin S. ***Global genetic insight contributed by consanguineous Pakistani families segregating hearing loss.*** Hum Mutat. 2019 Jan;40(1):53-72. doi: 10.1002/humu.23666.

- Riazuddin, S., Hussain, M., Razzaq, A., Iqbal, Z., Shahzad, M., Polla, D.L., Song, Y., van Beusekom, E., **Khan, A.A.**, Tomas-Roca, L., Rashid, M., Zahoor, M.Y., Wissink-Lindhout, W.M., Basra, M.A., Ansar, M., Agha, Z., van Heeswijk, K., Rasheed, F., Van de Vorst, M., Veltman, J.A., Gilissen, C., Akram, J., Kleefstra, T., Assir, M.Z., Grozeva, D., Carss, K., Raymond, F.L., O'Connor, T.D., Riazuddin, S.A., Khan, S.N., Ahmed, Z.M., de Brouwer, A.P., and van Bokhoven, H. ***Exome sequencing of Pakistani consanguineous families identifies 30 novel candidate genes for recessive intellectual disability.*** Molecular Psychiatry. 2017 Nov;22(11):1604-1614. doi: 10.1038/mp.2016.109.
- Faridi, R., Rehman, A.U., Morell, R.J., Friedman, P.L., Demain, L., Zahra, S., **Khan, A.A.**, Tohlob, D., Assir, M.Z., Beaman, G., Khan, S.N., Newman, W.G., Riazuddin, S., and Friedman, T.B. ***Mutations of SGO2 and CLDN14 collectively cause coincidental Perrault syndrome.*** Clinical genetics. 2017 Feb;91(2):328-332. doi: 10.1111/cge.12867.
- Jiao, X., Kabir, F., Irum, B., Khan, A. O., Wang, Q., Li, D., **Khan, A. A.**, Husnain, T., Akram, J., Riazuddin, S., Hejtmancik, J. F. & Riazuddin, S. A. ***A Common Ancestral Mutation in CRYBB3 Identified in Multiple Consanguineous Families with Congenital Cataracts.*** PLoS One. 2016 Jun 21;11(6):e0157005. doi: 10.1371/journal.pone.0157005.
- Jiao, X., Khan, S. Y., Irum, B., Khan, A. O., Wang, Q., Kabir, F., **Khan, A. A.**, Husnain, T., Akram, J., Riazuddin, S., Hejtmancik, J. F. & Riazuddin, S. A. ***Missense Mutations in CRYAB Are Liable for Recessive Congenital Cataracts.*** PLoS One. 2015 10(9): e0137973. doi:10.1371/journal.pone.0137973.
- Rehman, A.U., Santos-Cortez, R.L., Morell, R.J., Drummond, M.C., Ito, T., Lee, K., **Khan, A.A.**, Basra, M.A., Wasif, N., Ayub, M., Ali, R.A., Raza, S.; University of Washington Center for Mendelian Genomics, Nickerson, D.A., Shendure, J., Bamshad, M., Riazuddin, S., Billington, N., Khan, S.N., Friedman, P.L., Griffith, A.J., Ahmad, W., Riazuddin, S., Leal, S.M., Friedman, T.B. ***Mutations in TBC1D24, a Gene Associated With Epilepsy, Also Cause Nonsyndromic Deafness DFNB86.*** The American Journal of Human Genetics. 2014 Jan 2; 94(1):144-52. doi: 10.1016/j.ajhg.2013.12.004.
- Bonnet, C., **Khan, A. A***., Bresso E., Vigouroux C., Béri-Dexheimer, M., Lejczak S., Deemer B., Andrieux J., Philippe, C., Moncla A., Giurgea I., Leheup B., and Jonveaux,P. ***Extended spectrum of MBD5 mutations in neurodevelopmental disorders.*** European Journal of Human Genetics. 2013 Dec; 21(12):1457-61 doi: 10.1038/ejhg.2013.22.
- Huynh, M. T., Béri-Dexheimer, M., Bonnet, C., Bronner, M., **Khan, A. A.**, Allou, L., Philippe, C., Vigneron, J. and Jonveaux, P. ***RUNX1T1, a chromatin repression protein, is a candidate gene for autosomal dominant intellectual disability.***
  American Journal of Medical Genetics PartA, 2012 158A: 1782–1784. doi: 10.1002/ajmg.a.35386

ACHEIVEMENTS

GRANTS:

- NRPU project entitled "Identification and Characterization of Genes Responsible For Hearing Impairment In Pakistani Population" 8.4M PKR sponsored by Higher Education Commission of Pakistan *project No. 2934*

AWARDS:

- HEC Overseas Scholarships for MS/MPhil Leading to Ph.D. (FRANCE) 2008
- Higher Education Commission (HEC) Approved Supervisor 2013 to date

RESEARCH SUPERVISION

4 PhD and 12 MPhil students have successfully defended their theses