DR. MUZAMMIL AHMAD KHAN

Assistant Professor
Gomal Centre of Biochemistry and Biotechnology,
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PROFESSIONAL EXPERIENCE

1- Gomal University, Dera Ismail Khan. Khyber Pakhtunkhwa, Pakistan (November 2011 to Till Date)

Working as *"Assistant Professor (BPS-19),* Gomal Centre of Biochemistry and Biotechnology, Gomal University D.I.Khan*.*

Responsibilities/Accomplishments:

- Teaching Biochemistry/Molecular Biology courses to BS, MS and Ph.D students.
- Supervising BS, MS and Ph.D research students.
- Executing Research Projects.

2- Hamad Medical Corporation Doha, Qatar (November 2014 to November 2016)

Worked as **"Post Doctoral Research Scientist,** Translational Research Institute, Hamad Medical Corporation, Doha, Qatar

Responsibilities/Accomplishments:

- Lookafter the activities of Genomic core facility like Nucleic aic lab, Sequencing Lab, Microarray Lab etc.
- Establish linkages/ Collaborations within Hamad Medical Corporation and other Biomedical Institutions in Qatar.
- Writing Research Projects and Drafting manuscripts of Scientific data
- Supervising and Training the Research Assistants and Students
- Provinding Research Consultation to the Medical Doctors for designing projects

3- Department of Human Genetics, Sidra Medical and Research Centre, Doha, Qatar (March 2022 October 2022)

Worked as *"Postdoctoral Fellow"*

4- Mufti Mehmood Memorial Teaching Hospital D I Khan (Part Time) (March 2020 to August 2021)

Worked as **"RT-PCR Consultant,** D.I.Khan Public Health Laboratory, Pakistan.

Responsibilities/Accomplishments:

- Data Interpetiton and Reporting for diagonsitcic purpose

EDUCATIONAL QUALIFICATION

- **Postdoctoral Fellowship,** Translational Research Institute, Hamad Medical Corporation, Doha, Qatar
  Specialization : Human Molecular Genetics **2014-2016**

- **Ph.D.** QAU Islamabad, Pakistan (IRSIP from University of Toronto, Canada)
  Specialization : Human Molecular Genetics/ Biochemistry **2007-2011**

- **Master (M.Sc)**, Quaid-I-University, Islamabad, Pakistan.
  Specialization : Biochemistry/Molecular Biology **2005-2007**

- **Graduation (B.Sc)** Gomal University, D.I.Khan, Pakistan
  Specialization : Chemistry, Zoology, Statistics **2004**

i- Title of Ph.D. Dissertation:-

"Genetic Mapping of Genes Involved in Autosomal Recessive Mental Retardation"

ii- Projects Executed during Postdoc:-

- *Whole exome sequencing of consanguineous Pakistani families inheriting neurological disorders.*
- *Sanger DNA sequencing of EPCAM gene in patients with Tufting Enteropathy.*
- *SNP genotyping of targeted genes in HCV infected patient with liver steatosis.*
- *Development of pharmacogenetics tests for drugs used in human cardiovascular disorders.*

RESEARCH PROJECTS

I. Molecular study of inherited neurological disorders in consanguineous Pakistani families—Funded by Higher Education Commission of Pakistan Islamabad, Pakistan **(Amount Allocated PKR. 500,000).**

II. Analysis of single nucleotide polymorphisms associated with hepatic steatosis in patients with chronic hepatitis C. Funded by Medical Research Centre, Hamad Medical Corporation, Doha, Qatar **(Amount Allocated: QR. 100,000)**.

III. Genetic Mapping and Functional Characterization of Genes Involved in Autosomal Recessive Intellectual Disability. Funded by Higher Education Commission of Pakistan, Islamabad, Pakistan **(Amount Allocated: PKR. 64,25,600).**

IV. Whole exome sequencing of families with rare genetic disorders. Sponsored by 3billion Company, South Korea **(Exome sequencing of more than 100 samples).**

COLLABORATIVE RESEARCH PROJECTS/ACCOMPLISHMENTS:

a. Identification of disease genes for human neurological and ophthalmic disorders. **(Collaborator; Dr. Christian Windpassinger, Associate Professor, Institute of Human Genetics, Graz Medical University, Austria).**

b. Genetic analysis of consanguineous Pakistani families with intellectual disability. **(Collaborator: Dr. John B. Vincent, Senior Scientist, Centre for Addiction and Mental Health, Professor, Department of Psychiatry, University of Toronto, Canada).**

ACADEMIC HONORS AND SCHOLARSHIP

1. Awarded Fellowship by European Society of Human Genetics to attend the ESHG meeting 2016 at Barcelona, Spain **(May, 2016)**
2. Awarded Fellowship by European Society of Human Genetics for attending Eye Genetics Course at Bertinoro, Italy **(September, 2015)**.
3. Best Oral Presentation Award in National Conference on Trends in Biochemistry and Molecular Biology, organized by Quaid-I-University, Islamabad, Pakistan **(February 2012)**.
4. Received Student Talent Forming Scholarship from HEC of Pakistan two times during M.Sc **(2005-2007).**
5. Selected in talented student scholarship from Quaid-I-Azam University Islamabad during M.Phil **(2008).**
6. Indigenous scholarship for PhD from Higher Education Commission of Pakistan **(2008-2011).**
7. IRSIP scholarship from Higher Education commission (HEC) of Pakistan **(Feb 2010-July 2010).**

GUEST LECTURES

- Diagnosis of SARS-CoV-2; a Virus that causes COVID-19. Delivered at **Khushal Khan Khattak University, Karak (February 2021).**
- Role of Genes in Medical Genetics. Delivered **at Gomal Medical College, D.I.Khan**, Pakistan (**March, 2018**).
- Genetics and Pathophysiology of Microcephaly. Delivered at Translational Research Institute, **Hamad Medical Corporation, Doha, Qatar.**
- Pharmacogenetics in cardiovascular disorders. 3rd CT Research Forum and Echo Cardiography Workshop, 2015. **Hamad Medical Corporation, Doha, Qatar**.
- Genome-wide homozygosity mapping revealed a novel methyltransferase gene mutation associated with new form of syndromic intellectual disability. Delivered at Translational Research Institute, **Hamad Medical Corporation, Doha, Qatar**.
- Molecular Diagnostic tools in Genetic Mental Retardation at Institute of Molecular Biology and Biotechnology, **Bahauddin Zakariya, University, Multan, Pakistan.**
- NSUN2, a novel gene, associated with distal myopathy and cognitive impairment. Delivered at Institute of Human Genetics, **Graz Medical University, Austria**.

WORKSHOPS & TRAINING COURSES

- Eye Genetics Meeting at Bertinoro, Italy, organized by European School of Genetic Medicine and sponsored by European Society of Human Genetics in **September 2015**.
- Training on Affymatrix Microarray system for gene expression assays at **Hamad Medical Corporation, 2015**, Doha, Qatar.
- Training on HiSeq 2500 and NexSeq500 next generation DNA sequencer at **Sidra Biomedical and Research Institute, 2015, Doha, Qatar.**
- 3rd one day Workshop on Bioinformatics, held on **18th June 2012**; organized by Muhammad Ali Jinah University, Islamabad.
- International Workshop on Frontiers in Molecular Biology and Biotechnology, held in March 28-30 organize by COMSTECH.

MEMBERSHIP OF SCIENTIFIC AND WELFARE SOCIETIES

- Professional Society of Genetic Counselors in Asia (**2019**)
- Canadian Association of Genetic Counsellors (**2018-19**)
- European Society of Human Genetics (**2015-2017**)
- Lions Eye Club, International (Organized Free Eye Camp in the rural areas) (**2013-2014**)
- BAQI (Biochemist Association QAU Islamabad)
- NAYS (National Association of Young Scientist)

ADMINISTRATIVE EXPERIENCE AND ACADEMIC MEMBERSHIP

- Director Gomal Centre of Biochemistry and Biotechnology, Gomal University, D.I.Khan (May 2021 – March 2022)
- Member Advance Studies and Reserch Board, Gomal University, D.I.Khan, Pakistan (2020-21)
- Member Board of Faculty of Sciences at Gomal University, D.I.Khan (2020-21).
- Member Purchase Committee of Gomal University, D.I.Khan, Pakistan (2018-2019)
- Working as an Internal controller exam at Gomal Centre of Biochemistry and Biotechnology, Gomal University, D.I.Khan, Pakistan
- Member Board of Studies at Gomal Centre of Biochemistry and Biotechnology, Gomal University, D.I.Khan, Pakistan
- Member HEC curriculum development committee for Biotechnology syllabus 2013

INTERNATIONAL TRAINING / SHORT COURSES

- Certificate of training in Laboratory Biosafty, from Centre for Addiction and Mental Health University of Toronto, Toronto, Ontario, Canada.
- Short Term Training for Next Generation Sequencing at Graz Medical University, Austria.

- 3 days training on NextSeq500 for whole exome sequencing. Organized by AGBL, Dubai.

Organized Workshop:

- Organized Two days workshop on "Fundamental Techniques in Biotechnology" on **9th-10 December 2017** at Gomal Centre of Biochemistry and Biotechnology, Gomal University, D.I.Khan, Pakistan.

RESEARCH PUBLICATIONS

Papers published in different Q ranked journals

Q1: 11 Articles **Q2: 17 Articles** **Q3: 11 Articles** **Q4: 13 Articles**

1- Lukas Kaufmann, Johannes Pilic, Lisa Auinger, Anna-Lena Mayer, Jasmin Blatterer, Johann Semmler-Bruckner, Safdar Abbas, Khurram Rehman, Muhammad Ayaz, Wolfgang Graier, Roland Malli, Erwin Petek, Klaus Wagner, Ali Al Kaissi, **Muzammil Ahmad Khan**, Christian Windpassinger. Analysis of a non-lethal biallelic frameshift mutation in ZMPSTE24 reveals utilization of alternative translation initiation codons Clinical Genetics, 2023. Q1

2- Nazif Muhammad, Syeda Iqra Hussain, Zia Ur Rehman, Sher Alam Khan, Samin Jan, Niamatullah Khan, Muhammad Muzammal, Sumra Wajid Abbasi, Naseebullah Kakar, Zia Ur Rehman, **Muzammil Ahmad Khan**, Muhammad Usman Mirza, Noor Muhammad, Saadullah Khan, Naveed Wasif. Autosomal recessive variants c.953A>C and c.97-1G>C in NSUN2 causing intellectual disability: a molecular dynamics simulation study of loss-of-function mechanisms. Frontiers in Neurology, 2023. Q2

3- Safeer Ahmad, Muhammad Zeeshan Ali, Muhammad Muzammal, Amjad Ullah Khan, Muhammad Ikram, Mari Muurinen, Shabir Hussain, Petra Loid, **Muzammil Ahmad Khan**, Outi Mäkitie. Identification of GLI1 and KIAA0825 variants in two families with postaxial polydactyly. Genes 2023, 14(4), 869. Q2

4- Safeer Ahmad, Muhammad Zeehsan Ali, Sumra Wajid Abbasi, Safdar Abbas, Iftikhar Ahmad, Shakil Abbas, Muhammad Shoaib, Mubarak Ziab, Ikhlak Ahmed, Khalid Fakhro, **Muzammil Ahmad Khan**, Ammira Akil. A GHRHR founder mutation causes isolated growth hormone deficiency type IV in a consanguineous Pakistani family. Frontiers in Endocrinology, 2023 Q1

5- Ao Ma, Jianteng Zhou, Haider Ali, Tanveer Abbas , Imtiaz Ali, Zubair Muhammad, Sobia Dil, Jing Chen, Xiongheng Huang, Hui Ma, Daren Zhao, Beibei Zhang, Yuanwei Zhang, Wasim Shah, Basit Shah, Ghulam Murtaza, Furhan Iqbal, **Muzammil Ahmad Khan**, Asad Khan, Qing Li, Bo Xu, Limin Wu, Huan Zhang, Qinghua Shi. Loss-of-function Mutations in CFAP57 Cause Multiple Morphological Abnormalities of the Flagella in Humans and Mice. JCI Insight, 2023;8(3):e166869, 2023 (impact factor: 9.484). Q1

6- Adil U Rehman, Malaika Hamid, Sher Alam Khan, Muhammad Eisa, Wasim Ullah, Zia Ur Rehman, **Muzammil Ahmad Khan**, Sulman Basit, Noor Mohammad, Saadullah Khan, Naveed Wasif. The Expansion of the Spectrum in Stuttering Disorders to a Novel ARMC Gene Family (ARMC3). Genes 2022, 13(12), 2299. Q2

7- Muhammad Muzammal, Ahmad Firoz, Hani Mohammed Ali, Arshad Farid, **Muzammil Ahmad Khan**, Khalid Rehman Hakeem. Lumateperone interact with S-protein of Ebola Virus and TIM-1 of human cell membrane: Insights from computational studies. Applied Sciences, 2022. Q2

8- Mazhar Khan, Xei Xuefeng, Muhammad Zubair, Abbas Khan, Ranjha Khan, Jianteng Zhou, Yuanwei Zhang, Muzafar Said, Sher A. Khan, Qumar Zaman, Ghulam Murtaza, **Muzammil Ahmad Khan**, Xiaoning Hou, Huan Zhang, Bo Xu, Xiaohua Jiang, Shi Qinghua, Shun Bai. A homozygous missense variant in DND1 causes non-obstructive azoospermia in humans. Frontier in Genetics, 2022. Q2

9- Raeesa Tehreem, Iris Chen, Mudassar Raza Shah , Yumei Li, **Muzammil Ahmad Khan**, Kiran Afshan, Rui Chen, and Sabika Firasat. Exome Sequencing Identified Molecular Determinants of Retinal Dystrophies in Nine Consanguineous Pakistani Families. Genes 2022, 13, 1630. Q2

10- Erich Schaflinger, Jasmin Blatterer, Aiman Saeed Khan, Lukas Kaufmann, Lisa Auinger, Benjamin Tatrai1, Sumra Wajid Abbasi, Muhammad Zeeshan Ali, Ansar Ahmad Abbasi, Ali Al Kaissi, Erwin Petek, Klaus Wagner, **Muzammil Ahmad Khan**, Christian Windpassinger. An exceptional biallelic N-terminal frame shift mutation in ZMPSTE24 leads to non-lethal Progeria due to possible utilization of a downstream alternative start codon. Gene, 2022, 833: 146582. Q2
11- Muhammad Muzammal, Alessandro Di Cerbo, Eman M. Almusalami, Arshad Farid, **Muzammil Ahmad Khan**, Shakira Ghazanfar, Mohammed Al mohaini, Abdulkhaliq J. Alsalman, Maitham A. Al Hawaj. In silico Analysis of L2HGDH Mutations and their biological significance in disease etiology. Genes, 2022, 13, 698; 1-11. Q2
12- Muhammad Zeeshan Ali, Arshad Farid, Safeer Ahmad, Muhammad Muzammal, Mohammed Al Mohaini, Abdulkhaliq J. Alsalman, Maitham A. Al Hawaj, Yousef N. Alhashem, Abdulmonem A. Alsaleh, Eman M. Almusalami, Mahpara Maryam, **Muzammil Ahmad Khan**. In Silico Analysis Identified Putative Pathogenic Missense nsSNPs in Human SLITRK1 Gene. Genes, 2022, 13: 672. Q2
13- Iftikhar Ahmed, Muhammad Muzammal, Sumra Wajid Abbasi, **Muzammil Ahmad Khan** , Asif Mir. Whole Exome Analysis in Consanguineous Pakistani Families Determined ROR2 and RPTN as Novel Candidate Genes to be involved in Autosomal Recessive Non-Syndromic Intellectual Disability. International Journal of Computational Intelligence in Control, 2021, 13 (2): 163-171.
14- Sadam Hussain, Amjad Nawaz, Malaika Hamid, Waseem Ullah, Iqbal Nawaz Khan, Mehak Afshan, Adil Rehman, Hamid Nawaz, Julia Halswick, Sohail Ahmad, Muhammad Muzammal, Noor Muhammad1, Abid Jan, Saadullah Khan, Christian Windpassinger, **Muzammil Ahmad Khan**. Mutation screening of multiple Pakistani MCPH families revealed novel and recurrent protein-truncating mutations of ASPM. Biotechnology and Applied Biochemistry, 2021. Q3
15- Muhammad Muzammal, Muhammad Zeeshan Ali, Beatrice Brugger, Jasmin Brugger, Safeer Ahmad, Sundas Taj, Syed Khizar Shah, Saadullah Khan, Christian Enzinger, Erwin Petek, Klaus Wagner, **Muzammil Ahmad Khan**, Christian Windpassinger. A novel protein truncating mutation in L2HGDH causes L-2-hydroxyglutaric aciduria in a consanguineous Pakistani family. Metabolic Brain Disease 2021. Q2
16- Hadia Gul, Abdul Haleem Shah, Ricardo Harripaul, Sumra Wajid Abbasi, Muhammad Faheem, Muhammad Zubair, Muhammad Muzammal, Saadullah Khan, John B Vincent, **Muzammil Ahmad Khan**. Homozygosity mapping coupled with whole exome sequencing and protein modelling identified a novel missense mutation in GUCY2D in a consanguineous Pakistani family with Leber congenital amaurosis. J Genet, 2021; 100: 57. Q4
17- Hadia Gul, Abdul Haleem Shah, Ricardo Harripaul, Anna Mikhailov, Ejaz Ullah Khan, Wasim Shah, Nisar Ahmad, John B Vincent, **Muzammil Ahmad Khan**. Mutation Analysis of a Pakistani Oculocutaneous Albinism Family Identifies a Novel Splice Site Defect in OCA2 gene. Pak J Zool, 2020. Q4
18- Muhammad Hanif, **Muzammil Ahmad Khan**, Muhammad Ramzan, Abdul rafey, Ali Zaman, Imran Aziz, Amanullah, Adnan Amin. Inhibitive efficacy of Nymphoides indica rhizome extract on a-glucosidae and cross-link formation of advanced glycation end products. Journal of Traditional Chinese Medicine, 2021; 41(3) 376-380. Q3
19- Safdar Abbas, Beatrice Brugger, Muhammad Zubair, Sana Gul, Jasmin Blatterer, Khurram Rehman, Benjamin Tatrai, **Muzammil Ahmad Khan**, Christian Windpassinger. Exome-wide mapping in spastic paraplegia family from Pakistan found a deletion mutation in cytochrome B5 domain of Fatty Acid 2-Hydroxylase gene. Neurological Research, 2020 (Impact Factor: 2.401). Q2
20- Kamal Hassan , Gulab Sher, Eman Hamid, Khalid Abou Hazima, Hatim Abdelrahman, Fatma Al-Medaihki, Wesam Al-Masri, Jisha Sankar, Rana Shawish, Mahlah Daryaee, **Muzammil Ahmad Khan**, Zafar Nawaz. Outcome Associated with EPCAM Founder Mutation c.499dup in Qatar. European Journal of Medical Genetics, 2020, 63 (10) 104023 (Impact Factor: 2.368). Q2
21- Muhammad Zeeshan Ali, Jasmin Blatterer, **Muzammil Ahmad Khan**, Erich Schaflinger, Erwin Petek, Safeer Ahmad, Ejazullah Khan, Christian Windpassinger. Identification of a novel protein truncating mutation p.Asp98* in XPC associated with Xeroderma Pigmentosum in a Consanguineous Pakistani family. Mol Genet and Genomics Med, 2020 8(2) e1060 (Impact Factor: 2.448). Q3
22- Khan S, Khan AK, Hamid M, Nazif M, Abbas M, Khan SA, Khan B, **Khan MA**, Jan A, Khattak B, Ullah W, Muhammad N. Association of sequence variants in FZD6 encoding the Wnt receptor frizzled 6 with autosomal recessive Nail Dysplasia (NDNC-10) in Pashtun families. J Pak Med Assoc, 2020 (Impact Factor: 0.718). Q4
23- Muhammad Muzammal, Muhammad Zubair, Sophie Bierbaumer, Jasmin Blatterer, Aisha Gul, Safdar Abbas, Muhammad Badar, Ansar Ahmad Abbasi, **Muzammil Ahmad Khan**, Christian Windpassinger. Exome sequence analysis in consanguineous Pakistani families inheriting Bardet Biedle syndrome determined

founder effect of mutation c.299delC (p.Ser100Leufs*24) in BBS9 gene. Mol Genet Genomic Med, 2019, doi: 10.1002/mgg3.834 (Impact Factor: 2.448). Q3

24- Gul H, Shah AH, Harripaul R, Mikhailov A, Khan E, Khan F, Zubair M, Ali MZ, Shah AH, Salman S, Khan S, Vincent JB, **Khan MA**. Genetic studies of multiple consanguineous Pakistani families segregating oculocutaneous albinism identified novel and reported mutations. Ann Hum Genet, 2019 doi: 10.1111/ahg.12307 (Impact Factor: 1.529). Q3

25- Ahmed J, Windpassinger C, Salim M, Wiener M, Petek E, Schaflinger E, Taj S, Hussain S, Abbas S, Abbas M, Younis I, Muhammad N, Khan S, **Khan MA**. Genetic study of Khyber-Pukhtunkhwa resident Pakistani families presenting microcephaly with intellectual disability. JPMA, 2019 69 (12)1812-1816: (Impact Factor: 0.718). Q4

26- Ahmad B, **Khan MA**, Muhammad N, Basit S, Qasim I, Khan S. Pakistan Genetic Mutation Database (PGMD); a centralized Pakistani mutome data source. European Journal of Medical Genetics, 2017 (Impact Factor: 2.004). Q2

27- Abbasi AA, Bläsius K, Hu H, Latif Z, Picker-Minh S, Khan MN, Farooq S, **Khan MA**, Kaindl AM. Identification of a novel homozygous TRAPPC9 gene mutation causing non-syndromic intellectual disability, speech disorder, and secondary microcephaly. Amr J Med Genet: Part B, 2017 (Impact Factor: 3.016). Q1

28- **Khan MA**, Windpassinger C, Ali MZ, Zubair M, Gul H, Abbas S, Khan S, Badar M, Mohammad RM, Nawaz Z. Molecular genetic analysis of consanguineous families with primary microcephaly identified pathogenic variants in the ASPM gene. J Genet, 2017 96(2)383-387 (Impact Factor: 1.09). Q4

29- Khan AK, Muhammad N, Aziz A, Khan SA, Shah K, Nasir A**, Khan MA**, Khan S. A novel mutation in homeobox DNA binding domain of HOXC13 gene underlies pure hair and nail ectodermal dysplasia (ECTD9) in a Pakistani family. BMC Med Genet, 2017 18(1)42 (Impact factor: 1.913). Q3

30- Shahzad M, Yousaf S, Waryah YM, Gul H, Kausar T, Tariq N, Mehmood U, Ali M, **Khan MA,** Waryah AM, Shaikh RS, Riazuddin S, Ahmed ZM, University of Washington Center for Mendelian Genomics. Molecular Outcomes, Clinical Consequences, and genetic diagnosis of Oculocutaneous Albinism in Pakistani families. Sci Rep, 2017 17: 44185 (Impact factor: 4.122). Q1

31- Iskandarani A, Bhat AA, Siveen K, Prabhu K, Kuttikrishnan S, **Khan MA**, Krishnankutty R, Kulinski M, Nasr R, Mohammad R, Uddin S. Bortezomib-mediated downregulation of S-phase kinase protein-2 (SKP2) causes apoptotic cell death in chronic myelogenous leukemia cells. J Transl Med, 2016 14(1):69 (Impact factor: 4.197). Q1

32- Ullah E, Saqib MAN, Sajid S, Shah N, Zubair M, **Khan MA**, Ahmed I, Ali G, Dutta AK, Danda S, Lao R, Tang PL, Kwok P, Ansar M, Slavotinek A. Genetic Analysis of Consanguineous Families Presenting with Congenital Ocular Defects. Exp Eye Resear, 2016 146: 163-71 (Impact Factor: 3.152). Q1

33- **Khan MA,** Mohan S, Zubair M, Windpassinger C. Homozygosity mapping identified a novel protein truncating mutation (p. Ser100Leufs*24) of BBS9 gene in a consanguineous Pakistani family segregating Bardet Biedl Syndrome. BMC Med Genet, 2016 17:10 (doi:10.1186/s12881-016-0271-9) (Impact Factor: 1.913). Q3

34- **Khan MA,** Rupp V, Orpinell M, Hussain MS, Altmüller J, Steinmetz MO, Enzinger C, Thiele H, Höhne W, Nürnberg G, Baig SM, Ansar M, Nürnberg P, Vincent JB, Speicher MR, Gönczy P, Windpassinger C. A missense mutation in the PISA domain of HsSAS-6 causes autosomal recessive primary microcephaly in a large consanguineous Pakistani family. HMG 2014; 15;23(22):5940-5949, doi: 10.1093/hmg/ddu318 (Impact factor: 4.902). Q1

35- **Khan MA,** Rupp V, Khan MA, Khan MP, Ansar M, Windpassinger C. Genetic analysis of a consanguineous Pakistani family with Leber Congenital Amaurosis identified a novel mutation in GUCY2D gene. J Genet 2014; 93(2) 527-530. (Impact factor: 0.995). Q4

36- **Khan MA,** Rafiq MA, Noor A, Hussain S, Flores JV, Rupp V, Vincent AK, Malli R, Ali G, Khan FS, Ishak GE, Doherty D, Weksberg R, Ayub M, Windpassinger C,Ibrahim S, Frye M, Ansar M, Vincent JB. Mutation in NSUN2, which encodes an RNA methyltransferase, causes autosomal-recessive intellectual disability. Am J Hum Genet 2012; 90(5):856-863 (Impact factor: 9.025). Q1

37- Rafiq MA, Kuss AW, Puettmann L, Noor A, Ramiah A, Ali G, Hu H, Kerio NA, Xiang Y, Garshasbi M, **Khan MA**, Ishak GE, Weksberg R, Ullmann R, Tzschach A, Kahrizi K, Mahmood K, Naeem F, Ayub M, Moremen KW, Vincent JB, Ropers HH, Ansar M, Najmabadi H. Mutations in the alpha 1,2-mannosidase gene, MAN1B1, cause autosomal recessive intellectual disability. Am J Hum Genet 2011; 89: 176-182 (Impact factor: 9.025). Q1

38- **Khan MA,** Rafiq MA, Noor A, Ali N, Ali G, Vincent JB, Ansar M. A novel deletion mutation in the TUSC3 gene in a consanguineous Pakistani family with autosomal recessive nonsyndromic intellectual disability. BMC Med Genet 2011; 12: 56 (Impact factor: 1.913). Q3

39- Rafiq MA, Ansar M, Marshall CR, Noor A, Shaheen N, Mowjoodi A, **Khan MA**, Ali G, Amin-ud-Din M, Feuk L, Vincent JB, Scherer SW. Mapping of three novel loci for non-syndromic autosomal recessive mental retardation (NS-ARMR) in consanguineous families from Pakistan. Clin Genet 2010; 78: 478-483 (Impact factor: 3.512). Q1

40- Safeer Ahmad, Muhammad Zeeshan Ali, Muhammad Muzammal, Fayyaz Ahmad Mir, **Muzammil Ahmad Khan.** The molecular genetics of human appendicular skeleton. Molecular Genetics and Genomics, 2022. Q2

41- Muhammad Muzammal, Safeer Ahmad, Muhammad Zeeshan Ali, **Muzammil Ahmad Khan**. Alopecia-Mental Retardation Syndrome (APMR); Molecular Genetics of a Rare Neuro-Dermal Disorder. Annals of Human Genetics, 2021. Q3

42- Muhammad Muzammal, Muhammad Zeeshan Ali, Safeer Ahmad, Shawana Huma, Rizwan, Sohail Ahmad, Ansar Ahmad Abbasi, Saadullah Khan and **Muzammil Ahmad Khan**. The molecular genetics of UV-sensitive syndrome; a rare dermal anomaly. J Pak Med Assoc, 2021. Q4

43- **Khan MA,** Khan S, Windpassinger C, Nawaz Z, Mohammad RM. The molecular genetics of autosomal recessive non-syndromic intellectual disability: a mutational continuum and future recommendations. Ann Hum Genet 2016 8; 342-368 (Impact factor: 1.529). Q3

44- Khan S, Basit S, **Khan MA,** Muhammad N, Ahmad W. Genetics of human isolated acromesomelic dysplasia. Eur J Med Genet, 2016 59(4): 198-203 (Impact Factor: 2.004). Q2

45- Khan SA, Muhammad N, **Khan MA,** Kamal A, Rehman ZU, Khan S. Genetics of Human Bardet-Biedl Syndrome, an Updates. Clin Genet, 2016 (doi: 10.1111/cge) (Impact Factor: 3.512). Q2

Book Chapters (Total: 01)

1. Muhammad Akhlaq, Zaheer-Ud-Din Babar, Mahvish Ajaz, **Muzammil Ahmad Khan**, Erkan Kilinc, Muhammad Adeel, Muhammad Badar, Asif Nawaz and Aamir Jalil. Covid-19 Pandemic and Coronaviruses from Discovery to Treatment: A tale of Two Decades of 21st Century'' published in Springer Book ''Modeling, Control and Drug Development for COVID-19 Outbreak Prevention'', 2021.

THESIS SUPERVISED AT M.PHIL LEVEL

1. Genetic and Functional Characterization of Pakistani Families Segregating Intellectual Disability. Aiman Nayab, **Session 2019-21.**
2. Molecular Characterization of Pakistani Families Suffering from Congenital Blindness using Positional Cloning Approach and Structural Modelling. Sana Fatima, **Session 2019-21**.
3. Exome Sequence Analysis of a Pashtoon Origin Family Inheriting Autosomal Recessive Non-Syndromic Intellectual Disability. Rizwan, **Session 2017-19**
4. Molecular Mapping and Computational Analysis of Exome Data Variants in Bardet-Biedl Syndrome Patients from Consanguineous Pakistani Families. Shawana Huma, Session **2017-19.**
5. Genetic Analysis of a New Form of Syndromic Intellectual Disability in a Consanguineous Family From District D.I.Khan. Sana Saleem Jan, **Session 2017-19.**
6. Molecular Genetic Studies of a Pakistani Origin Family Segregating Autosomal Recessive Ichthyosis. Aiman Saeed Khan, **Session 2016-18**
7. Genetic Diagnosis of a Saraiki origin Pakistani family inheriting autosomal recessive intellectual disability. Sundas Taj, **Session 2016-2018.**

8. Analysis of AGE's (advanced glycation end products) and protein cross-link inhibitory effects by Nymphoides indica roots extracts. Muhammad Hanif, **Session 2016-2018.**
9. Genetic Analysis of a Saraiki Language Family inheriting Postaxial polydactyly. Safeer Ahmad, **Session 2016-2018**.
10. Homozygosity Mapping and Candidate gene analysis in a Pakistani Family with USHER Syndrome. Farman Ullah, **Session 2015-2017.**
11. Mutation Screening of rare MCPH variants in KPK Origin Families Segregating Primary Microcephaly. Saddam Hussain, **Session 2015-2017**.
12. Genetic analysis of Saraiki origin Pakistani families inheriting autosomal recessive primary microcephaly. Muhammad Zeeshan Ali, **Session 2015-2017**.
13. Molecular Genetic Studies of Pakistani Families Segregating Muscular Dystrophies. Safdar Abbas, **Session 2015-2017**
14. Molecular Screening of OCA genes in D.I.Khan origin Pakistani families. Anam Rashid, **Session 2013-2015**.
15. Molecular analysis of Saraiki ethnic families segregating autosomal recessive Oculocutaneous Albinism. Kashmala Samad, **Session 2013-2015**.
16. Genetic mapping of known MCPH loci in Saraiki origin families of Pakistan. Aiman Hina, **Session 2013-2015**.

THESIS SUPERVISED AT Ph.D LEVEL

- The Molecular Genetic Study of Pakistani Families Inheriting Autosomal Recessive Intellectual Disability. Mr. Muhammad Muzammil **(Ph.D. Session 2016-19)**

THESIS CO- SUPERVISED AT Ph.D LEVEL

- Genetic Dissection of Pakistan families segregating autosomal recessive albinism. Miss Hadia Gul **(Ph.D. Session 2013-16).**
- The Analysis of Abnormal Spectrum of Serum Proteins in Persons Exposed to X-Radiations. Mr. Khurram Rehman (**Ph.D. Session 2015-18**)

THESIS CO- SUPERVISED AT M. PHIL

- Bioinformatics analysis of BBS9 gene. Shamsa Ali, **Session 2016-2018.**
- Bioinformatics analysis of FA2H gene. Sana Gul, **Session 2016-2018.**
- Genetic Study of autosomal recessive primary microcephaly in families from Dera Ismail Khan District. Nisar Ahmed, **Session 2012-2014.**