DR. MUHAMMAD UMAIR

EDUCATION:

PhD: Doctor of Philosophy (Biochemistry/Molecular Biology),
Quaid-i-Azam University, Islamabad, Pakistan.

Sandwich HEC Fellowship: Technical University Munich and Helmholtz Zentrum Neuherberg, Munich, Germany.

Dissertation Title (2013-2018):

"***Clinical and Genetic Characterization of Human Hereditary Skeletal Deformities in Consanguineous Families*** "

M.Phil: Masters of Philosophy in Biochemistry/Molecular Biology

"Characterization of Certain Human Hereditary Muscle and Skeletal Dysplasias"

(Quaid-i-Azam University, Islamabad)

M.Phil Dissertation Title (2010-2012):

M.Sc: Masters in Biochemistry/Molecular Biology

2008-2010 (Quaid-i-Azam University, Islamabad)

RESEARCH EXPERIENCE AND CURRENT POSITIONS:

Research Scientist in Medical Genomics Research Department at **King Abdullah International Medical Research Center (KAIMRC)**, Ministry of National Guard Health Affairs (MNGHA), Riyadh, Kingdom of Saudi Arabia (KSA). **[26 Oct, 2018-Present].**

Team Leader Functional Studies in the Medical Genomics Research Department at **King Abdullah International Medical Research Center (KAIMRC)**, Ministry of National Gard Health Affairs (MNGHA), Riyadh, Kingdom of Saudi Arabia (KSA). **[12 May, 2019-Present].**

Research Advisor in the Department of Life Sciences, School of Science, **University of Management and Technology (UMT)**, Lahore, Pakistan **[01 July, 2022-Till date].**

Adjunct Researcher in the Department of Life Sciences, School of Science, **University of Management and Technology (UMT)**, Lahore, Pakistan **[28 July, 2021-31 June, 2022].**

Head and Chief Technologist of Biochemistry/Molecular Biology Section**, Al-Habib Clinical Labs,** Kohat, Pakistan **[01 March 2018 -26 October, 2018].**

Teaching Assistant in Department of Biochemistry/Molecular Biology (BMB), **Quaid-i-Azam University**, Islamabad, Pakistan **[01 September 2017 -21 February, 2018].**

National Internship Programme (NIP) in Department of Biochemistry/ Molecular Biology (BMB), **Quaid-i-Azam University**, Islamabad, Pakistan **[14 April 2011 -13 April, 2012].**

RESEARCH PROFILE:

An enthusiastic and self-driven scientist with over +6 years of post-PhD experience specializing in the genetic characterization of inherited disorders. Expertise includes establishing genotype-phenotype correlations, conducting functional analyses of causal variants, and utilizing both conventional and advanced genetics/molecular biology techniques.

CORE SKILLS:

1) WES/WGS Data Analysis.
2) Functional Studies.
3) Novel Gene Discovery.
4) Genotype-Phenotype Coorelations.
5) PGT-A, PGT-M, NIPT.

SCIENTIFIC ACHIEVEMENTS

1. Most impactful achievements was the identification of NAV3 variants associated with neurodevelopmental disorder, which was recently recognized by **OMIM** as **"UMAIR-Alfadhel neurodevelopmental disorder" [OMIM# 621182]—**a testament to the originality and global significance of his work.
2. **Stanford–Elsevier World's Top 2% Scientists (2022, 2023, 2024),** Recognized among the world's top 2% most-cited scientists, based on a standardized citation metrics database developed by Stanford University and Elsevier (Ioannidis et al., PLOS Biology).
3. **Winner Gold Medal in Genetics**, 2nd Dr. Sajjad Aslam Shami **Gold Medal in Genetics**. The Applied Zoological Society of Pakistan (AZSP-2021) 12th March-2021, QAU, Islamabad, Pakistan.
4. **Winner (1st position): Best Poster Presentation** at 4th Middle East Genetic and Metabolic Academy (MEGMA) Symposium, 2023, Riyadh, KSA. [Title of poster: VWA8 causes developmental delay, microcephaly, scoliosis and responsible for skeletal development and morphogenesis in Zebrafish].
5. **Top cited Article 2022-2023: Journal-Clinical Genetics;** Genetic overview of postaxial polydactyly: Updated classification.
6. Awarded with an **International Research Support Initiative Program (IRSIP),** Fellowship (06 months) for **Technical University Munich, Germany**, Institute of Human Gnentics, and Helmholtz Zentrum Neuherberg, Munich, Germany from Higher Education Commission (HEC), Pakistan.
7. Awarded with a Competitive **Indigenous Ph.D. Scholarship** from Higher Education Commission (HEC), Pakistan (2013-2017).
8. Merit Scholarship holder throughout M.Phil (MS).

RESEARCH PROJECTS

1. Functional Characterization of Genes Associated with Neurodevelopmental disorders (NDDs). **(NRC23R/177/02) {PI}** (Funded by: KAIMRC, Riyadh, Saudi Arabia).
2. Genetic and Functional Characterization of genes associated with Polydactyly. **(NRC22R/661/12) {PI}** (Funded by: KAIMRC, Riyadh, Saudi Arabia).
3. Molecular Characterization of Inherited Hematological Disorders. **(NRR25/059/2) {PI}** (Funded by: KAIMRC, Riyadh, Saudi Arabia).
4. Genetic and Rare Diseases (GARD) programme Saudi Arabia: A Step Towards Therapeutic Genomics. **(RC19/120/R) {Co-PI}** (Funded by: KAIMRC, Riyadh, Saudi Arabia).
5. Genetic and Molecular Characterization of Genes Involved in Causing Rare Conjoined Twins. **(RC19/070/R) {Co-PI}** (Funded by: KAIMRC, Riyadh, Saudi Arabia).
6. Genodermatosis in Saudi Arabia: a long road to traverse. **(RC19/138/R) {Co-PI}** (Funded by: KAIMRC, Riyadh, Saudi Arabia).
7. Preventative Genome medicine for inherited genetic disorder in Saudi Arabia. **RC19/115/R) {Co-PI}** (Funded by: KAIMRC, Riyadh, Saudi Arabia). [NIPT, PGS, PGT-M and PGT-A].
8. Genetic and Rare Disease Registry. **(RC20/453/R) {Co-PI}** (Funded by: KAIMRC, Riyadh, Saudi Arabia).

9. Implementation of new guidelines for dietary management of propionic acidemia and methylmalonic acidemia: a prospective cohort study. **(NRC23R/145/03) {Co-PI}** (Funded by: KAIMRC, Riyadh, Saudi Arabia).

10. KAIMRC Genomic Database version 2. **(RC20/546/R) {Co-PI}** (Funded by: KAIMRC, Riyadh, Saudi Arabia).

11. Clinical, molecular, and biochemical characteristics of primary mitochondrial disorders in Saudi Arabia. **(NRC23R/521/08) {Co-PI}** (Funded by: KAIMRC, Riyadh, Saudi Arabia).

12. Natural History Study on Patients with Propionic Acidemia or Methylmalonic Acidemia. **(SRR25/001/5) {Co-PI}** (Funded by: Moderna). mRNA-RDOB-P901 Study Protocol.

13. Pilot Study Towards a ***Nationwide Newborn Genome Sequencing Program***: Saudi BabySeq. **{Co-PI}** (Funded by: ***King Salman Center for Disability Research***, Riyadh, Saudi Arabia).

14. Discovery of Genetic Diagnosis of Disable children's by using Next generation Sequencing. **{Co-PI}** (Funded by: ***King Salman Center for Disability Research***, Riyadh, Saudi Arabia).

15. Genome Profiling of Genetic Forms of Obesity in Saudi Population for Biomarkers Identification and Precision Medicine. **(NRR25/115/1) {Co-PI}** (Funded by: KAIMRC, Riyadh, Saudi Arabia).

16. Saudi Genomic Database Proposal. **(RC19/315/R) {Co-PI}** (Funded by: KAIMRC, Riyadh, Saudi Arabia). **[Completed Dec, 2020].**

17. Genetic and Clinical Characterization of Genetic Skeletal Disorders (GSDs). **(RC19/352/R) {PI}** (Funded by: KAIMRC, Riyadh, Saudi Arabia). **[Completed Feb, 2023]**.

18. Basic Characterization of Rare Diseases Novel Genes and Variants In Saudi Patients **(RC18/017/R) {Co-PI}** (Funded by: KAIMRC, Riyadh, Saudi Arabia). **[Completed 2023].**

TRAINING COURSES, CONFERENCES & WORKSHOPS

1. Presented Poster at "American College of medical genetics and Genomics" [ACMG], (18-22 March, 2025). Title of poster: Congenital myopathy and split-foot malformation: A unique case without mesoaxial polydactyly.
2. Participated as Speaker at 4th Middle East Genetic and Metabolic Academy (MEGMA) Symposium, 2023, Riyadh, KSA. [Topic: Limitations of Gene Therapy].
3. 3rd Middle East Genetic & Metabolic Academy (MEGMA) Neurogenetics Conference, AbuDhabi, November 4-6, 2022.

4. Presented Poster at "The American Society of Human Genetics" [ASHG], (25-29 Oct, 2022). Title of poster: A Novel de novo Nonsense Variant in TBX2 Cause Osteochondrodysplasia.

5. The International Parkinson and Movement Disorder Society, Participated in "Approach and Management of Sleep Problems in Parkinson's Disease" 2022, Sep 21, 2022. Awarded 1.00 AMA PRA category 1 credit(s)™.

6. The International Parkinson and Movement Disorder Society, Participated in "Animal Models for Parkinson's Disease" 2022, Sep 21, 2022. Awarded 1.00 AMA PRA category 1 credit(s)™.

7. The International Parkinson and Movement Disorder Society, Participated in "MDS-PAS Movement Disorders for Basic Scientists: From the Clinic to the Lab" 2022, Sep 21, 2022. Awarded 1.00 AMA PRA category 1 credit(s)™.

8. The International Parkinson and Movement Disorder Society, Participated in "Genetic Testing 101 Part A" 2022, Sep 21, 2022. Awarded 1.00 AMA PRA category 1 credit(s)™.

9. The International Parkinson and Movement Disorder Society, Participated in "Approach and Practical Management of Treatable Adult-onset Progressive Ataxias" 2022, Sep 21, 2022. Awarded 1.00 AMA PRA category 1 credit(s)™.

10. Participated as a speaker in KIAMRC Weekly seminar under the title "Rare Genetic diseases-dilemma". KAIMRC, November 23, 2021, Riyadh, Saudi Arabia.

11. The International Parkinson and Movement Disorder Society, Participated in MDS first Movement Disorders Clinical Practice Conference- Education 2021, Nov 18, 2021-Nov 19, 2021 (21MDCPC-EDU-001-107336).

12. Secrets for Powerful Speaking and Listening Workshop. Organized by KAIMRC, Riyadh, on 06-07 October 2021.

13. The International Parkinson and Movement Disorder Society, Participated in Educational Activity entitled MDS virtual congress 2021, Sep 17, 2021- Sep 22, 2021 (IC21V-CERT-001-107336). Awarded 34.00 AMAPRA Category 1 Credit(s)™.

14. Research Project Management, conducted by KAIMRC, Riyadh, 14-06-21-2021 to 16-06-2021 (18) training hours.

15. 1st Middle East Genetic & Metabolic Academy (MEGMA) Neurogenetics Conference [Virtual], Dubai, July 2-3, 2021.

16. "COVID-19 Contact Tracing" an online non-credit course authorized by Johns Hopkins University and offered through Coursera, july 7th, 2020.

17. Eppendorf webinar, The Pandemic: From a laboratory Prespective, July 30, 2020.

18. COVID19 awareness among staff and students of Saudi academic institutions, 18 July, 2020, Jamia Hmadard, Riyadh, Saudi Arabia.

19. ASPED Live Webinar "Telemedicine in Paediatric Diabetes during the COVID Era and Beyond". August 21, 2020. President, Arab Society for Paediatric Endocrinology & Diabetes, Abu Dhabi, United Arab Emirates.

20. Webinar, interntional E-conference ON " Biological sciences vis-à-vis Sustainable Dvelopment and Enviromantal Conservation", 03-04 July, 2020, Carrier point university, Himachal Pradesh, India.

21. E-conference on Phytopharmaceuticals (PHP 2020), August 06, 2020, Jamia hamdard, New Delhi, India.

22. 1st International virtual conference on Advances in Colloidal and polymeric Systems (ACAPS-2020), 22 August, 2020, Vellore, India.

23. 10th European Conference on Rare Diseases & Orphan Products held online on 14-15 May 2020.

24. 7th Annual Mendelian Data Analysis Workshop at Washington University, Aug 11-16, 2019, Seattle, USA.

25. Nature research Academies, Effectively Communicating Science to the Public, "Author Academy" course. King Abdullah International Medical Research Center (KAIMRC), Dec 22, 2019, Riyadh Saudi Arabia.

26. Nature research Academies, completing the "Science Communication Academy" course. King Abdullah International Medical Research Center (KAIMRC), Dec 23, 2019, Riyadh Saudi Arabia.

27. Poster Presentation at 7th Pediatrics Research Day, KASCH, Dec 05, 2019 Riyadh Saudi Arabia.

28. 10th Annual forum for Medical Research Nov 25-26, 2019 Riyadh, Saudi Arabia.

29. Research to Publication workshop, British Medical Journal (BMJ) in collaboration with University of California, San Francisco (UCSF), held in KAIMRC, Sep 05, 2019 Riyadh, Saudi Arabia.

30. Ensemble Online training course, Wellcome Trust Genome Campus, Cambridge, UK.

31. Research joint project forum workshop, King Saud bin Abdulaziz University for Health Sciences KSAU-HS, Oct 27, 2019 Riyadh, Saudi Arabia.

32. Health Intellectual Property Awareness Workshop, Vision 2030-Accelerating Saudi Arabia's Emergence as a Leading healthcare Innovator, Nov 20-21, 2019 KAIMRC, Saudi Arabia.

33. Arabic language Course, Nov 11, 2019 National Guard of health Affairs, Riyadh, Saudi Arabia.

34. Poster presentation at 1st Annual Health Research Conference "Health Research-A Step Towards Attaining SDGs 2030", Sep 24-25, 2018 Islamabad, Pakistan.

35. Intellectual property Rights, PASTIC/PSF, Ministry of science and technology, Aug 18, 2016, Islamabad, Pakistan.

36. International Workshop on Ion Beam Application, NCP, Oct 27-28, 2016 Islamabad, Pakistan.

37. World Blood Donor Day 2015 PNCA, Islamabad, Pakistan.

38. National Conference on Trends in Biochemistry and Molecular Biology, Feb 21, 2012 Islamabad, Pakistan.

39. International Conference on Applications of Molecular Biology in Medicine and Agriculture, August 20-22, 2013 Islamabad, Pakistan.

Contribution to Novel Genes Discovery

	Genes	Disease
1	***IQCE***	Autosomal recessive Post-axial Polydactyly
2	***STKLD1***	Autosomal recessive Pre-axial Polydactyly
3	***EPS15L1***	Autosomal recessive Split hand/Split Foot Malformation
4	***DLX6***	Autosomal dominant Split hand/Split Foot Malformation
5	***UGDH***	Developmental Delay and Axial Hypotonia
6	***NEK10***	Primary Ciliary Dyskinesia
7	***DLL1***	Congenital Vertebral Malformation
8	***RAP1GDS1***	Dysmorphic Feature, Intellectual Disability & Speech Delay
9	***EMC10***	Global Developmental Delay, Mild Intellectual Disability, and Speech Delay
10	***DCBDL2***	Recessive Lethal Restrictive Cardiomyopathy & Developmental Delay
11	***DACH1***	Autosomal recessive Post-axial Polydactyly
12	***VWA8***	Developmental Delay, Microcephaly, and Scoliosis
13	***PPP1R1B/DARPP-32***	Generalized Dystonia
14	***SPTBN5***	Intellectual Disability, Developmental Delay, and Seizures.
15	***SIX5***	Autosomal recessive Non-syndromic Hearing Impairment
16	***TBX2***	Osteochondrodysplasia
17	***ATP9A***	Attention Deficit Hyperactivity Disorder (ADHD)
18	***AMFR***	Autosomal recessive Spastic Paraplegia
19	***PIP5KIγ***	Neurodevelopmental Syndrome
20	***NUDT2***	Autosomal recessive Neurodevelopmental Disorder
21	***CSMD1***	Autosomal recessive Neurodevelopmental Disorder
22	***NAV3***	Autosomal recessive Neurodevelopmental Disorder

23	***HMGXB4***	Autosomal recessive Neurodevelopmental disorder
24	***INPP4A***	Neurodevelopmental disorder
25	***SPAG9***	CACD syndrome
26	***EEFSEC***	Neurodegeneration
27	***AIRIM/C1orf109***	Autosomal recessive Neurodevelopmental disorder

LIST OF PUBLICATIONS {Total Impact Factor: 2,322.774}

First Author Publications	Corresponding Author Publications	Co-Author Publications	**Total Publications**	**H-Index**	I-Index	**Total Citations**
32	**43**	**133**	**208**	**44**	**134**	**17,449**

1. **Umair M**, Palander O, Bilal M, Almuzzaini B, Alam Q, Ahmad F, Younus M, Khan A, Waqas A, Alfadhel M. Biallelic variant in DACH1, encoding Dachshund Homolog 1, defines a novel candidate locus for recessive postaxial polydactyly type A. ***Genomics*** 2021; 113(4):2495-2502. https://doi.org/10.1016/j.ygeno.2021.05.015. **(IF: 5.736).**
2. **Umair M**, Khan MF, Aldrees M, Nashabat M, Alhamoudi KM, et al. Mutated VWA8 is associated with developmental delay, microcephaly, scoliosis and play a novel role in early development and skeletal morphogenesis in Zebrafish. ***Front Cell Dev Biol*** 2021; 9:736960. doi: 10.3389/fcell.2021.736960. **(IF: 6.684).**
3. **Umair M**, Alfadhel M. Genetic Disorders Associated with Metal Metabolism. ***Cells*** 2019 8(12): 1598. https://doi.org/10.3390/cells8121598. **(IF: 6.600).**
4. **Umair M**, Ahmad F, Alam Q, Mohd Rehan, Alqosaibi AI, et al. A novel homozygous missense mutation in the zinc finger DNA binding domain of the GLI1 causes recessive post-axial polydactyly. ***Front Genet*** 2021;12:746949. doi: 10.3389/fgene.2021.746949. **(IF: 4.599).**
5. **Umair M**, Shah K, Alhaddad B, Haack TB, Graf E, et al. Exome Sequencing Revealed a Splice Site Variant in the IQCE Gene Underlying Post-axial Polydactyly type A Restricted to Lower Limb. ***Eur J Hum Genet*** 2017; 25(8): 960-965. **(IF: 4.246).**
6. **Umair M**, Ullah A, Ahmad F, Shah K, Abbas S, Ahmad W. First direct evidence of involvement of a homozygous loss-of-function variant in the EPS15L1 gene underlying split-hand/split-foot malformation. ***Clin Genet*** 2018; 93(3):699-702. **(IF: 4.442).**
7. **Umair M**, Eckstein G, Wieland T, Rudolph G, Strom T, et al. Homozygous XYLT2 variants as a cause of spondyloocular syndrome. ***Clin Genet*** 2018; 93(4):913-918. **(IF: 4.442).**
8. **Umair M**, Hassan A, Jan A, Ahmad F, Imran M, Samman MI, Basit S, Ahmad W. Homozygous sequence variants in the FKBP10 gene underlie osteogenesis imperfecta in consanguineous families. ***J Hum Genet*** 2016; 61(3); 207-213. **(IF: 3.172).**
9. **Umair M**, Alhaddad B, Rafique A, Jan A, Haack TB, Graf E, Ullah A, Ahmad F, Strom TM, Meitinger T, Ahmad W. Exome Sequencing Revealed a Novel Homozygous Splice Site Variant in WNT1 Gene Underlying Osteogenesis Imperfecta Type 3. ***Pediatric Research*** 2017; 82(5):753-758. **(IF: 3.756).**

10. **Umair M**, Ahmad F, Bilal M, Ahmad W and Alfadhel M. Clinical Genetics of Polydactyly: An Updated Review. ***Front Genet*** 2018: 9:447. doi: 10.3389/fgene.2018.00447. **(IF: 4.599).**
11. **Umair M**, Rafique A, Ullah A, Ahmad F, Ali RH, et al. Novel Homozygous Sequence Variants in the GDF5 Gene Underlie Acromesomelic Dysplasia type-Grebe (AMDG) in Consanguineous Families. ***Congenit Anom (Kyoto)*** 2016; 57(2):45-51. **(IF: 1.409).**
12. **Umair M**, Ballow M, Asiri A, Alyafee Y, Tuwaijriet AA, et al. EMC10 Homozygous Variant Identified in a Family with Global Developmental Delay, Mild Intellectual Disability, and Speech Delay. ***Clin Genet*** 2020; 98(6):555-561. doi: 10.1111/cge.13842. **(IF: 4.442).**
13. **Umair M,** Seidel H, Ahmed I, Ullah A, Haack TB, Alhaddad B, Jan A, Rafique A, Strom TM, Ahmad F, Meitinger T, Ahmad W. Ellis-van Creveld syndrome and profound deafness resulted by sequence variants in the EVC/EVC2 and TMC1 genes. ***J Gent*** 2017; 96(6):1005-1014. **(IF: 1.166).**
14. **Umair M**, Bilal M, Ali RH, Alhaddad B, Ahmad F, Abdullah, et al. Whole exome sequencing revealed a nonsense mutation in STKLD1 causing non-syndromic pre-axial polydactyly type A affecting only upper limb. ***Clin Genet*** 2019; 96(2): 134-139. doi: 10.1111/cge.13547. **(IF: 4.442).**
15. **Umair M**, Wasif N, Albalawi AM, Ramzan K, Alfadhel M, Ahmad W, Basit S. Exome Sequencing Revealed a Novel Loss-of-Function Variant in the GLI3 Transcriptional Activator 2 Domain Underlies Nonsyndromic Postaxial Polydactyly. ***Mol Genet & Genomic Med*** 2019; 7(7): e00627. doi: 10.1002/mgg3.627. **(IF: 2.183).**
16. **Umair M**, Ahmad F, Bilal M, Abbas S. Syndactyly genes and classification: A Mini-review. ***J Biochem Clin Gene*** 2018 1(1):34–47. **(IF:0.00).**
17. **Umair M**, Ahmad F, Bilal M, Arshad M. Frontonasal dysplasia: A review. ***J Biochem Clin Gene*** 2018 1(2):2–14. **(IF:0.00).**
18. **Umair M**, Khan A, Amin W, Abbas S, Younus M, Alfadhel M, Ahmad F. Biallelic Missense Mutation in the ECEL1 Underlies Distal Arthrogryposis Type 5 (DA5D). ***Front in Pedriatics*** 2019; 7:343. doi: 10.3389/fped.2019.00343. **(IF: 3.418).**
19. **Umair M**, Hayat A. Non-syndromic Split Hand-Foot Malformation (SHFM): Recent Classification. ***Mol Syndromol*** 2019;10: 243-254. https://doi.org/10.1159/000502784 **(IF: 1.631).**
20. **Umair M**, Mahmood RT, Inam M, Waqas A, Wazir I. Sero-prevalence of hepatitis B, hepatitis C, human immunodeficiency virus, syphilis and malaria in blood donors of Mirpur, Azad Jammu Kashmir, Pakistan. ***JPHBS*** 2012; p.110-114; ISSN 2305-8668. **(IF: 0.00).**
21. **Umair M**, Ahmad F, Ullah A. Whole Exome Sequencing as a Diagnostic Tool for Genetic Disorders in Pakistan. ***PJMR*** 2018;57(2);90-91. **(IF: 0.00).**
22. **Umair M**, Ahamd F, Bilal M, Asiri A, Younus Y, Khan A. A Comprehensive review of genetic skeletal disorders reported from Pakistan: A brief commentary. ***Meta Gene*** 2019; (20); 1-7; 100559. doi.org/10.1016/j.mgene.2019.100559. **(IF: 0.00).**

23. **Umair M**, Alfadhel M. Prevention of hemoglobinopathies in Saudi Arabia: efficacy of national premarital screening and the feasibility of preimplantation genetic diagnosis. ***Biochem Clin Gene*** 2020;3(2): 94–99. doi.org/10.24911/JBCGenetics/183-1595487640. **(IF: 0.00).**

24. Ullah A, **Umair M**, Yousaf M, Khan SA, Din N, et al. Sequence Variants in four Genes Underlying Bardet-Biedl Syndrome (BBS) in Consanguineous Families. ***Mol Vision*** 2017; 23:482-494. ***Equal first author contribution***. **(IF: 2.367).**

25. **Umair M**, Kharfy TA, Sajjad S, Alfadhel M. FIG4-associated Yunis-Varon Syndrome: Identification of a novel missense variant. ***Mol Syndrom*** 2021; 12(6): 386-392. doi: 10.1159/000516971. **(IF: 1.631).**

26. Ahmad F, Ahmed I, Alam Q, Ahmad T, Khan A, Ahmad I, Bilal M, Hayat A, Khan A, Waqas A, Rafeeqi MM, Sainj ZM, **Umair M.** Variants in the PNPLA1 Gene in Families with Autosomal Recessive Congenital Ichthyosis Reveal Clinical Significance. ***Mol Syndromology*** 2021; 12(6): 351–361. doi:10.1159/000516943. ***[Corresponding author]*. (IF: 1.631).**

27. Hayat A, Hussain S, Bilal M, Kausar M, Almuzzaini B, Abbas S, Tanveer A, Khan A, Siddiqi S, Foo JN, Ahmad F, Khan F, Khan B, Anees M, Mäkitie O, Alfadhel M, Ahmad W, **Umair M**. Biallelic variants in four genes underlying recessive osteogenesis imperfect. ***Eur J Med Gent*** 2020; 63(8):103954. doi: 10.1016/j.ejmg.2020.103954.***[Corresponding author]*** **(IF: 2.708).**

28. Nayab A, Alam Q, Alzahrani OR, Khan R, Sarfaraz S, Albaz AA, Rafeeq MM, Sain ZM, Waqas A, **Umair M**. Targeted exome sequencing identified a novel frameshift variant in the PGAM2 gene causing glycogen storage disease type X. ***Eur J Med Gent*** 2021. 64(9): 104283. https://doi.org/10.1016/j.ejmg.2021.104283 **[Corresponding author] (IF: 2.708).**

29. Younus M, Ahmad F, Malik E, Bilal M, Kauser M, Abbas S, Shaheen S, Alfadheld M, **Umair M**. SGCD Homozygous Nonsense Mutation (p.Arg97∗) Causing Limb-Girdle Muscular Dystrophy Type 2F (LGMD2F) in a Consanguineous Family, a Case Report. ***Front Genet*** 2019; 9:727. doi: 10.3389/fgene.2018.00727. **[*Corresponding author]* (IF: 4.599).**

30. Ullah A, Lin Z, Younus M, Shafiq S, Khan S, Rasheed M, Mahmood A, Alqosaibi AI, Alshehri MA, Khan A, **Umair M**. Homozygous missense variant in POPDC3 causes recessive limb girdle muscular dystrophy type 26. ***J Gene Med*** 2022; 24(4):e3412. doi: 10.1002/jgm.3412. **[Corresponding author] (IF: 4.565).**

31. Alfadhel M, **Umair M**, Almuzzaini B, Asiri A, Alhamoudi KM, Al-Owain M. Identification of TTC26 splice variant associated with new complex ciliopathy syndrome leading to Biliary, Renal, Neurological and Skeletal, manifestations (BRENS) syndrome. ***Mol Syndromol*** 2021;12(3): 133-140. doi: 10.1159/000513829. **(IF: 1.631).**

32. Alyafee Y, Al Tuwaijri A, Alam Q, **Umair M**, Haddad S. Next Generation Sequencing Based Non-invasive Prenatal Testing (NIPT): First Report From Saudi Arabia. ***Front Genet*** 2021; 12: 630787. doi: 10.3389/fgene.2021.630787. **(IF: 4.599)**.

33. Asiri A, Alwadaani D, **Umair M**, Alhamoudi KM, et al. Pancytopenia, Recurrent Infection, Poor Wound Healing, Heterotopia of the Brain Probably Associated with a Candidate Novel

de Novo *CDC42* Gene Defect: Expanding the Molecular and Phenotypic Spectrum. ***Genes (Basel)*** 2021, 12, 294. doi.org/10.3390/genes12020294. **(IF: 4.096).**

34. Alyafee Y, Alam Q, Altuwaijri A, **Umair M**, Haddad S, et al. Next-Generation Sequencing-Based Pre-Implantation Genetic Testing for Aneuploidy (PGT-A): First Report from Saudi Arabia. ***Genes (Basel)*** 2021;12(4):461. doi: 10.3390/genes12040461. **(IF: 4.096).**

35. Alfadhel M, Almuqbil M, Mutairi FA, **Umair M**, et al. The Leukodystrophy Spectrum in Saudi Arabia: Epidemiological, Clinical, Radiological and Genetic Data. ***Front Pediatr*** 2021; 9: 633385. doi: 10.3389/fped.2021.633385. **(IF: 3.418).**

36. Hayat A, **Umair M**, Abbas S, Rauf A, Ahmad F, et al. Identification of a novel biallelic missense variant in the KIAA0825 underlies postaxial polydactyly type A. ***Genomics*** 2020; 112(4):2729-2733. doi: 10.1016/j.ygeno.2020.03.006. **(IF: 5.736)**.

37. Liaqat K, Hussain S, Bilal M, Nasir A, Acharya A, Ali RH, Nawaz S, **Umair M**, Schrauwen I, et al. Further evidence of involvement of TMEM132E in autosomal recessive nonsyndromic hearing impairment. ***J Hum Genet*** *2020*; 65(2): 187-192. doi: 10.1038/s10038-019-0691-4. **(IF: 3.172).**

38. Khan A, Wang R, Han S, **Umair M**, Abbas S, et al. Homozygous missense variant in the TTN gene causing autosomal recessive limb-girdle muscular dystrophy type 10. ***BMC Med Genet*** 2019; 20(1):166. doi: 10.1186/s12881-019-0895-7. **(IF: 2.103).**

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Books

1. **Umair M,** Rafeeq M, Alam Q. (2024). Rare Genetic Disorders: Advancements in Diagnosis and Treatment (1st Ed pp. XIII, 275). **SPRINGER Nature,** April 13, 2024. ISBN: 978-981-99-9322-2. https://doi.org/10.1007/978-981-99-9323-9.
2. Alam Q, Rafeeq M, **Umair M.** (2025). Innovations and Implications in Molecular Diagnostics (1st Ed). **Taylor & Francis,** 10 September 2025. ISBN: 9781003489177. https://doi.org/10.1201/9781003489177.

Book Chapters

1. **Umair M,** Alfadhel M. EMC10-Related Neurodevelopmental Disorder. 2023 Jun 15. In: Adam MP, Mirzaa GM, Pagon RA, Wallace SE, Bean LJH, Gripp KW, Amemiya A, editors. ***GeneReviews®*** [Internet]. Seattle (WA): University of Washington, Seattle; 1993–2023. PMID: 37319253.
2. Madhu NR, Biswas G, Paul S, Adhikari S, Sarkar B, Rafeeq MM, **Umair M**. Challenges and Future Opportunities in Rare Genetic Disorders: A Comprehensive Review. In: **Umair, M**., Rafeeq, M., Alam, Q. (eds) Rare Genetic Disorders. ***Springer***, **(2024).** https://doi.org/10.1007/978-981-99-9323-9_9.
3. Alkurbi, M.O., Alghamdi, S., Aslam, A., **Umair, M**. Targeting Apicoplasts in Plasmodium falciparum, Origin and Pathways. In: Drug Targets for Plasmodium Falciparum: Historic to Future Perspectives. ***Springer***, Singapore (2024). https://doi.org/10.1007/978-981-19-4484-0_5.
4. **Umair M**, Alfadhel M. Homocystinuria due to Deficiency of N(5,10)-Methylenetetrahydrofolate Reductase Activity. 2025 May 29. In: Adam MP, Feldman J, Mirzaa GM, Pagon RA, Wallace SE, Amemiya A, editors. ***GeneReviews®*** [Internet]. Seattle (WA): University of Washington, Seattle; 1993–2025. Bookshelf ID: NBK615089.
5. Nayab A, Waqas A, Jelani M, **Umair M**. Polymerase chain reaction and its variants in molecular diagnostics. In: Alam Q, Rafeeq M, **Umair M** (eds) Innovations and Implications in Molecular Diagnostics (1st Ed). **Taylor & Francis (2025). ISBN: 9781003489177.**
6. Rafeeq M, **Umair M,** Alam Q, Thani AB. Polymerase chain reaction and its variants in molecular diagnostics. In: Alam Q, Rafeeq M, **Umair M** (eds) Innovations and Implications in Molecular Diagnostics (1st Ed). **Taylor & Francis (2025). ISBN: 9781003489177.**
7. Tamboli QY, Debasmita D, **Umair M**, Bushnak I, Panda J, Nayak D, Mohanta YK. Nanomedicines for neurological disorders: Recent advances. In: Nanomedicine and Nutrigenomics: Pioneering the Future of Health. Advances in Biotechnology and Bioengineering. ***Elsevier***; 2026. p. 143–168. doi:10.1016/B978-0-443-26761-1.00010-7

Patents and Designs

Certificate of Registration for UK Design

Design Title: Device for Identification of Genetic Rare Disorders in Children

Design Number: 6396874

Grant Date: 16 October 2024

Registration Date: 11 October 2024

Inventors: Dr. Alaa Hamed Habib, Dr. Farhan Ahmad Khan, Dr. Misbahuddin Rafeeq, Dr. Muhammad Barkaat Hussain, Dr. Qamre Alam, **Dr. Muhammad Umair**

International Design Classification:

Version: 14-2023

Class: 10 - Clocks and Watches and Other Measuring Instruments, Checking and Signaling Instruments

Subclass: 05 - Instruments, Apparatus, and Devices for Checking, Security, or Testing

Description: A specialized device designed for the identification and early detection of rare genetic disorders in children.

Link: https://www.registered-design.service.gov.uk/find

PhD STUDENTS UNDER Co-SUPERVISION

1. **Mr. Zaheer Ahmed, Thesis title:** *Mapping of Genes Involved in Syndromic and Non-syndromic Limb Anomalies. 2021-TILL DATE.*
2. **Mr. Afzal Rafique, Thesis title:** *Genetic and Molecular Analysis of Congenital Skeletal Anomalies. 2021-TILL DATE.*

MS/M.Phil STUDENTS UNDER Co-SUPERVISION

1. **Mr. Muhammad Touseef Akbar, Thesis title:** Histopathological alterations in gills and muscles of Oreochromis niloticus collected from selected sites of Indus River, Punjab, Pakistan. **[UOE, Multan, Pakistan] 21-UE-06342, MSF2100801: 2021-2023.**
2. **Ms. Iqra Farooq, Thesis title:** Epidemiological and associated risk factors of MODY in the population of district Multan. **[UOE, Multan, Pakistan] MSF2100813: 2021-2023.**
3. **Mr. Imran Malik, Thesis title:** Prevalence of common risk factors of cardiovascular disease among patients at Nishtar Hospital, Multan. **[UOE, Multan, Pakistan]. 2021-2023.**
4. **Ms. Sadia Malik, Thesis title:** ADAMTS19 Association with genetic Heart-Diseases. **[UOE, Multan, Pakistan]. 2021-2023.**

Mawhiba Program, KAIMRC, Riyadh, KSA

1. **Investigating Disease-causing Genetic Mutations in Heritable Diseases (Mr. Abdulaziz Abdullah Almansour), Mawhiba Program 2023.**
2. **Clinical, Molecular, and Functional Characterization of Rare Genetic Disorders (Mr. Marowan Alzeraa), Mawhiba Program 2023.**
3. **Genetic and Functional characterization of Neurodevelopmental Disorders (NDDs) (Ms. Amina Othman), Mawhiba Program 2024.**
4. **Genetic and Functional Insights into Neurodevelopmental Disorders: Advancing Precision Medicine (Mr. Azad Moshrif), Mawhiba Program 2025.**

MEMBERSHIPSMS

1. **Member of The Global Parkinson's Genetics Program (GP2):** Underrepresented Populations Working Group. **[https://gp2.org/working-groups/underrepresented-populations-working-group/].**
2. **Member of the "Undiagnosed Disease Network International" (UNDI), representing Kingdom of Saudi Arabia.** [https://www.udninternational.org/schede-14-members].
3. **American Society of Human Genetics [2000-2025] (https://www.ashg.org/).**
4. **European Society of Human Genetics [2000-2025] (https://www.eshg.org/home).**
5. **Scientifc Board Member for Middle East Genetic and Metabolic Academy (MEGMA).** [https://www.megma.org/scientific-board].
6. **Member of International Parkinson and Movement Disorder Society (https://www.movementdisorders.org/),** Member ID: 107336, 01-10-2021 till 31-12-2024.
7. **Member of International Parkinson and Movement Disorder Society; he Middle East Working Group (MEWG) (https://www.movementdisorders.org/MDS-AOS/Middle-East-Working-Group.htm).**
8. **Membership of American Society of Human Genetics (ASHG), 2020 till date.**

EDITORIAL OF SCIENTIFIC AND MEDICAL JOURNALS

1. ***Emerging Editorial Board member of The Journal of Gene Medicine,*** ISSN:1521-2254,[https://onlinelibrary.wiley.com/page/journal/15212254/homepage/emerging_editors], **[4.565 IF].**
2. ***Guest Editor: Special issue; Frontiers in Medicine; Section: Translational Medicine; Neurodegenerative Disorders and Co-Morbidity:*** From Bench to Bedside 2024. *https://www.frontiersin.org/research-topics/62298/neurodegenerative-disorders-and-co-morbidity-from-bench-to-bedside/overview* ***[4.772 IF].***
3. ***Guest Editor: Special issue; Frontiers in Medicine;*** *Section: Toxicogenomics: "The Evolution in Toxicogenomics: 2023". https://www.frontiersin.org/research-topics/53786/the-evolution-in-toxicogenomics-2023.* ***[4.772 IF].***
4. ***Guest Editor: Genes [MDPI], Special Issue "Functional Studies of Genetic Variants Involved in Human Genetic Diseases".*** *https://www.mdpi.com/journal/genes/special_issues/72JTFF6M9C.* ***[4.1 IF].***
5. ***Guest Editor: Special issue; Frontiers in Genetics;*** *Section: Translational Medicine: "Rising Stars in Translational Medicine: 2022".* ***[3.50 IF].***
6. **Associate Editor:** ***Molecular Syndromology***, e-ISSN: 1661-8777 (Online), [https://www.karger.com/Journal/Home/247640], **[1.631 IF].**
7. **Editor-in-Chief:** ***International Journal of Biochemistry & Physiology (IJBP)***, ISSN: 2577-4360 [https://medwinpublishers.com/IJBP/editorial-board.php].
8. **Editor**: ***American Journal of Pediatrics (AJP)***: ISSN Online: 2472-0909 [https://www.sciencepublishinggroup.com/home/index].
9. **Editor:** ***Journal of Biochemical and Clinical Genetics***, Online ISSN: 1234-1234, [https://jbcgenetics.com/].

REVIEWER OF SCIENTIFIC AND MEDICAL JOURNALS

1. Frontiers in Genetics [4.599 IF].
2. Frontiers in Pediatrics [3.418 IF].
3. Frontiers in Medicine [5.019 IF].
4. Clinical Genetics [ISSN: 1399-0004] [4.438 IF].
5. American Journal of Medical Genetics: Part A [ISSN:1552-4833] [2.802 IF].
6. Molecular Genetics & Genomic Medicine
7. Congenital Anomalies [ISSN:1741-4520] [1.409 IF].
8. Journal of Cellular and Molecular Medicine [ISSN: 1582-4934] [5.310 IF].
9. Brain Sciences [ISSN: 2076-3425] [3.394 IF].
10. COJ Biomedical Science & Research.
11. Journal of Biochemical and Clinical Genetics [ISSN: 1658-807X].
12. Gene Reports [ISSN: 2452-0144; Elsevier].
13. International Journal of Biological Macromolecules [ISSN: 0141-8130; Elsevier].

RESEARCH PROFILES

- Research Gate: https://www.researchgate.net/profile/Muhammad-Umair-58
- KAIMRC Profile: https://kaimrc.ksau-hs.edu.sa/en/Pages/Scientists'-Google-Scholar-Profiles.aspx
- ORCID: https://orcid.org/0000-0001-6729-9746
- Google Scholar: https://scholar.google.com/citations?hl=en&user=J6XYIAUAAAAJ

HOBBIES

1. Travelling
2. Photography