



# BUITEMS

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<b>Name</b>	<b>Dr. Abdul Wali Tareen</b>	
<b>Designation</b>	Tenured Professor	
<b>Department</b>	Biotechnology	
<b>Faculty</b>	Life Sciences & Informatics	
<b>E-mail address</b>	Official	abdul.wali@buitms.edu.pk
<b>Telephone Number</b>	Office Extension	+92(0)81-111-717-111 (625)

## Educational Qualification

Year	Degree/Certificate	Name of the Institute/ University	Field of study
2013	Post-Doctorate	University of Bonn, Bonn, Germany	Human Molecular Genetics
2008	M.Phil leading to PhD	Quaid-i-Azam University, Islamabad	Human Molecular Genetics
2004	M.Sc	Quaid-i-Azam University, Islamabad	Biochemistry and Molecular Biology

## Awards and Fellowships

2017	Best University Teacher Award by Higher Education Commission, Pakistan
2017	SATHA Innovation Award by South Asian Triple Helix Association
2016	Young Development Leader Award by Government of Pakistan
2011	Postdoc fellowship awarded by Alexander von Humboldt Foundation, Germany
2004	PhD fellowship awarded by Higher Education Commission, Pakistan

## Work Experience

S. No	From	To	Name of the Institution/ Organization	Position held
1	July 2020	Present	BUITEMS, Quetta, Pakistan	Tenured Professor
2	Jan 2019	Mar 2019	Qatar Biomedical Institute (QBRI), Doha, Qatar	Guest Researcher
3	June 2016	June 2020	BUITEMS, Quetta, Pakistan	Associate Professor
4	Sep 2011	Aug 2013	Institute of Human Genetics, University of Bonn, Germany	Postdoc Fellow
5	Oct 2010	Nov 2010	Max Plank Institute for Molecular Genetics, Berlin, Germany	Guest Researcher
6	Mar 2010	June 2016	BUITEMS, Quetta, Pakistan	Assistant Professor
7	Sep 2008	Feb 2010	Kohat University of Science Technology (KUST), Kohat, Pakistan	Assistant Professor

## Research Projects

Year	Project Title	PI/Co-PI	Amount Allocated	Funding Agency
2019-2022	Genetic Analysis and Functional Studies of Inherited Skin Disorders from Balochistan	Principle Investigator	PKR. 7.8 million	Higher Education Commission, Pakistan
2020-2021	Vascular-Endothelial Growth Factor (VEGF) Gene Polymorphism in Diabetes Mellitus and Diabetic Retinopathy Patients of Balochistan	Co-Principle Investigator	PKR. 2.9 million	ORIC-BUITEMS and Government of Balochistan
2018-2019	Identification of mutations in TYR gene causing albinism in patients of Balochistan	Co-Principle Investigator	PKR. 0.486 million	Higher Education Commission, Pakistan
2017-2018	Identification of Disease Causing Genes in Xeroderma Pigmentosum Patients	Principle Investigator	PKR. 0.163 million	ORIC-BUITEMS
2016-2017	Identification of genes responsible for autosomal recessive monogenic disorders in the Pakistani population	Co-Principle Investigator	PKR. 0.453 million	Higher Education Commission, Pakistan
2016-2017	Mapping of Candidate Genes in Nail Dysplasia from Balochistan	Principle Investigator	PKR. 0.196 million	ORIC-BUITEMS
2015-2016	Sequencing of candidate genes in families suffering from split-hand/foot malformation	Principle Investigator	PKR. 0.195 million	ORIC-BUITEMS
2014-2015	Genetic linkage studies in autosomal recessive hypohidrotic ectodermal dysplasia families	Principle Investigator	PKR. 0.169 million	ORIC-BUITEMS
2014-2015	Identification of Genes in Pakistani Families with Dowling-Degos Disease	Principle Investigator	Euro. 6,500	Humboldt Foundation, Germany
2011-2012	Mapping of Candidate Genes in Families with Primary Microcephaly	Principle Investigator	PKR. 0.10 million	ORIC-BUITEMS

## Research Articles Published in HEC Recognized Journals

S. No	Title of Paper	Name of Journal	National/ International	Publication Year
1	A TMC8 splice variant causes epidermodysplasia verruciformis in a Pakistani family.	<b>Clinical &amp; Experimental Dermatology</b> 48(4):434-437	International	2023
2	Identification and <i>In Silico</i> Analysis of a Homozygous Nonsense Variant in TGM1 Gene Segregating with Congenital Ichthyosis in a Consanguineous Family.	<b>Medicina</b> (Kaunas) 59(1):103	International	2023

		doi: 10.3390/medicina5 9010103		
3	An expansion of phenotype: novel homozygous variant in the MED17 identified in patients with progressive microcephaly and global developmental delay.	<b>Journal of Neurogenetics</b> 36(4):108-114	International	2022
4	NPHP3 splice acceptor site variant is associated with infantile nephronophthisis and asphyxiating thoracic dystrophy; A rare combination.	<b>European Journal of Medical Genetics</b> <a href="http://doi.org/10.1016/j.ejmg.2022.104578">http://doi: 10.1016/j.ejmg.2022.104578</a>	International	2022
5	ADAMTS1, MPDZ, MVD, and SEZ6: Candidate genes for autosomal recessive nonsyndromic hearing impairment	<b>European Journal of Human Genetics</b> 30(1):22-33	International	2022
6	Identification of a Novel Homozygous Missense (c.443A>T:p.N148I) Mutation in BBS2 in a Kashmiri Family with Bardet-Biedl Syndrome.	<b>BioMed Research International;</b> <a href="http://doi.org/10.1155/2021/6626015">http://doi.org/10.1155/2021/6626015</a>	International	2021
7	Novel frameshift mutations in XPC gene underlie xeroderma pigmentosum in Pakistani families	<b>Indian Journal of Dermatology;</b> 66(2): 220-222	International	2021
8	Sequencing and Characterization of Mitochondrial Protein Coding Genes for Schizothorax niger (Cypriniformes: Cyprinidae) with Phylogenetic Consideration	<b>BioMed Research International;</b> <a href="http://doi.org/10.1155/2020/5980135">http://doi.org/10.1155/2020/5980135</a>	International	2020
9	Apparent Missense Variant in COL7A1 Causes a Severe Form of Recessive Dystrophic Epidermolysis Bullosa via Effects on Splicing	<b>Acta Dermato-Venereologica;</b> <a href="http://doi.org/10.2340/00015555-3634">http://doi.org/10.2340/00015555-3634</a>	International	2020
10	Novel missense alteration in LRP4 gene underlies Cenani-Lenz syndactyly syndrome in a consanguineous family	<b>The Journal of Gene Medicine;</b> <a href="http://doi.org/10.1002/jgm.3143">http://doi.org/10.1002/jgm.3143</a>	International	2020
11	Whole exome sequencing identifies a nonsense mutation in the gene UVSSA in two consanguineous pedigrees from Pakistan	<b>Journal of Dermatological Science;</b> 95: 113-118	International	2019
12	Biallelic mutations in the LPAR6 gene causing autosomal recessive wooly hair/hypotrichosis phenotype in five Pakistani families	<b>International Journal of Dermatology;</b> 58: 946-952	International	2019
13	Novel insertion and a previously reported nonsense variant of ALOXE3 gene lead to autosomal recessive ichthyosis in two Balochi families	<b>Congenital Anomalies;</b> 59: 179-180	International	2019

14	XPC gene mutations in families with xeroderma pigmentosum from Pakistan; prevalent founder effect	<b>Congenital Anomalies;</b> 59: 18-21	International	2019
15	Homozygous sequence variants in the WNT10B gene underlie split hand/foot malformation	<b>Genetics and Molecular Biology;</b> 41: 1-8	International	2018
16	A rare case of three-way complex variant translocation in chronic myeloid leukemia t(6;9;22)(p21;q34;q11): A case report	<b>Biomedical Reports</b> 7(4): 377-379	International	2017
17	Genetic analysis of Xp22.3 micro-deletions in seventeen families segregating isolated form of X-linked ichthyosis	<b>Journal of Dermatological Sciences;</b> 80: 214-217	International	2015
18	Familial Primary Localized Cutaneous Amyloidosis results from either Dominant or Recessive Mutations in OSMR	<b>Acta-Dermato-Venereologica;</b> 95: 1005-1007	International	2015
19	Mutations in the gene phospholipase C, delta-1 (PLCD1) underlying hereditary leukonychia.	<b>European Journal of Dermatology;</b> 22: 736-739	International	2012
20	Alterations of the CIB2 calcium- and integrin-binding protein cause Usher syndrome type 1J and nonsyndromic deafness DFNB48.	<b>Nature Genetics;</b> 44:1265-1271	International	2012
21	Mutations in the LPAR6 and LIPH genes underlie autosomal recessive hypotrichosis/woolly hair in 17 consanguineous families from Pakistan.	<b>Clinical &amp; Experimental Dermatology;</b> 36: 652-654	International	2011
22	Digenic inheritance of an autosomal recessive hypotrichosis in two consanguineous pedigrees.	<b>Clinical Genetics;</b> 79: 273-81	International	2010
23	Mutation Analysis of the ASPM Gene in 18 Pakistani Families with Autosomal Recessive Primary Microcephaly.	<b>Journal of Child Neurology;</b> 25: 715-720	International	2009
24	Recurrent mutations in functionally-related EDA and EDAR genes underlie X-linked isolated hypodontia and autosomal recessive hypohidrotic ectodermal dysplasia.	<b>Archive Dermatological Research;</b> 301: 625-629	International	2009
25	Mutations in P2RY5 gene underlie autosomal recessive hypotrichosis in thirteen Pakistani families.	<b>British Journal of Dermatology;</b> 160: 1006-1010	International	2009
26	Novel missense mutations in lipase H (LIPH) gene causing autosomal recessive hypotrichosis (LAH2).	<b>Journal of Dermatological Sciences;</b> 54: 12-16	International	2009
27	Novel mutations in G protein-coupled receptor gene (P2RY5) in families with autosomal recessive hypotrichosis (LAH3).	<b>Human Genetics;</b> 123: 515-519	International	2008
28	Localization of a Novel Autosomal Recessive Hypotrichosis Locus (LAH3) to chromosome 13q14.11-q21.32.	<b>Clinical Genetics;</b> 72: 23-29	International	2007

29	Mapping of a Gene for Alopecia with Mental Retardation Syndrome (APMR3) on Chromosome 18q11.2-q12.2.	<b>Annals of Human Genetics;</b> 71: 1-8	International	2007
30	Ectodermal dysplasia of hair and nail type: mapping of a novel locus to chromosome 17p12-q21.2.	<b>British Journal of Dermatology;</b> 155: 1184-1190	International	2006
31	A novel locus for alopecia with mental retardation syndrome (APMR2) maps to chromosome 3q26.2-q26.31.	<b>Clinical Genetics;</b> 70: 233-239	International	2006
32	Atrichia with papular lesions resulting from a novel insertion mutation in the human hairless gene.	<b>Clinical &amp; Experimental Dermatology;</b> 31: 695-698	International	2006
<b>Area of Research Interest</b>		Major areas of research interest are mapping, identification, cloning and functional characterization of genes responsible for human hereditary disorders, and Cancer Genetics		
<b>Research Profile</b>		<a href="https://www.scopus.com/authid/detail.uri?authorId=14044434700">https://www.scopus.com/authid/detail.uri?authorId=14044434700</a>		
		<a href="https://www.webofscience.com/wos/author/record/3492280">https://www.webofscience.com/wos/author/record/3492280</a>		
		<a href="https://orcid.org/0000-0001-8818-5369">https://orcid.org/0000-0001-8818-5369</a>		
		<a href="https://www.researchgate.net/profile/Abdul-Wali-5">https://www.researchgate.net/profile/Abdul-Wali-5</a>		
		<a href="https://scholar.google.com/citations?user=zFXYCGkAAAAJ&amp;hl=en&amp;oi=ao">https://scholar.google.com/citations?user=zFXYCGkAAAAJ&amp;hl=en&amp;oi=ao</a>		
<b>Expertise</b>		Human Molecular Genetics, Molecular Biology, Cancer Genetics		
<b>HEC Approved supervisor</b>		Yes		
<b>If Yes, provide HEC URL</b>		<a href="http://sc.hec.gov.pk/aphds/">http://sc.hec.gov.pk/aphds/</a>		