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Designation	Professor (Tenured)	
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Qualification

Year	Degree/Certificate	Name of the Institute/ University	Field of study
2007-2010	Post Doctorate	WHO-IARC, Lyon, France	Molecular Genetics
2002-2006	PhD	CEMB, University of the Punjab, Lahore	Molecular Biology
2000-2001	MPhil	CEMB, University of the Punjab, Lahore	Molecular Biology
1993-1999	Graduation (M.Sc / B.Sc)	University of the Punjab, Lahore	• Botany • Chemistry, Zoology, Botany

Publications in HEC Recognized journals

S. No	Title of Paper	Name of Journal	National/ International	Publication date
1.	Variants in KIAA0825 underlie autosomal recessive postaxial polydactyly	Human Genetics	International	2019
2.	Identification of Mutations in Gene BRCA1/2 in Breast Cancer Cases from Balochistan, Pakistan	Pakistan Journal of Zoology	International	2019
3.	Unknown mutations and genotype/phenotype correlations of autosomal recessive congenital ichthyosis in patients from Saudi Arabia and Pakistan	Molecular genetics & Genomic Medicine	International	2019
4.	XPC gene mutations in families with xeroderma pigmentosum from Pakistan; prevalent founder effect	Congenital Anomalies	International	2019
5.	Novel insertion and a previously reported nonsense variant of ALOXE3 gene lead to autosomal recessive ichthyosis in two Balochi families	Congenital Anomalies	International	2018
6.	Expanding the phenotype associated	American Journal	International	2018

	with biallelic WDR60 mutations: Siblings 7with retinal degeneration and polydactyly lacking other features of short rib thoracic dystrophies	of Medical Genetics Part A		
7.	Genetic Characterization of Serotypes A and Asia-1 Foot-and-mouth Disease Viruses in Balochistan, Pakistan, in 2011	Transboundary and Emerging Diseases	International	2017
8.	The pattern of invasive lobular carcinoma in the patients diagnosed with breast cancer from Balochistan	Indian Journal of Cancer	International	2017
9.	Identification of two novel ALS2 mutations in infantile-onset ascending hereditary spastic paraplegia	Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration	International	2016
10.	No evidence that protein truncating variants in BRIP1 are associated with breast cancer risk: implications for gene panel testing	Journal of Medical Genetics	International	2016
11.	Novel Nonsense Variants c. 58C> T (p. Q20X) and c. 256G> T (p. E85X) in the CHEK2 Gene Identified dentified in Breast Cancer Patients from Balochistan	Asian Pacific Journal of Cancer Prevention	International	2016
12.	Exome sequencing and CRISPR/Cas genome editing identify mutations of ZAK as a cause of limb defects in humans and mice	Genome Research	International	2016
13.	Genetic characterization of the Makrani people of Pakistan from mitochondrial DNA control-region data	Legal Medicine	International	2015
14.	Mutational spectrum of the TYR and SLC45A2 genes in Pakistani families with oculocutaneous albinism, and potential founder effect of missense substitution (p.Arg77Gln) of tyrosinase	Clinical and Experimental Dermatology	International	2015
15.	A recurrent de novo mutation in KCNC1 causes progressive myoclonus epilepsy	Nature Genetics	International	2015
16.	Homozygous missense and nonsense mutations in BMPR1B cause acromesomelic chondrodysplasia-type Grebe	European Journal of Human Genetics	International	2014
17.	Dopamine transporter deficiency syndrome: phenotypic spectrum from infancy to adulthood	Brain	International	2014
18.	Identification of a novel mutation (p.Ile198Thr) in gene TYR in a Pakistani family with nonsyndromic oculocutaneous albinism	Clinical and Experimental Dermatology	International	2014
19.	Missense mutations (p.H371Y, p.D438Y) in gene CHEK2 are associated with breast cancer risk in	Molecular Biology Reports	International	2014

	women of Balochistan origin			
20.	Risk factors for lung cancer in the Pakistani population	Asian Pac J Cancer Prev	International	2014
21.	Exon skipping and severe childhood-onset obesity caused by a leptin receptor mutation	Am J Med Genet A	International	2013
22.	An Alu repeat-mediated genomic GCNT2 deletion underlies congenital cataracts and adult i blood group	Human Genetics	International	2012
23.	A homozygous splice site mutation in TRAPPC9 causes intellectual disability and microcephaly	Eur J Med Genet	International	2012
24.	A missense mutation (p.G274R) in gene ASPA causes Canavan disease in a Pakistani family	Molecular Biology Reports	International	2012
25.	Detection of BRCA1/2 mutations in breast cancer patients from Thailand and Pakistan	Clinical Genetics	International	2012
26.	Novel mutations of endothelin-B receptor gene in Pakistani patients with Waardenburg syndrome	Molecular Biology Reports	International	2012
27.	Various aspects, patterns and risk factors in breast cancer patients of Balochistan	Asian Pac J Cancer Prev	International	2012
28.	Epidemiology of Van der Woude syndrome from mutational analyses in affected patients from Pakistan	Clinical Genetics	International	2012
29.	Loss-of-Function Mutations of ILDR1 Cause Autosomal Recessive Hearing Impairment DFNB42	Am J Hum Genet	International	2011
30.	Identification of a novel LCA5 mutation in a Pakistani family with Leber congenital amaurosis and cataracts	Molecular Vision	International	2011
31.	Prevalence of hepatitis C virus (HCV) genotypes in Balochistan	Molecular Biology Reports	International	2009
32.	Frequencies of PrP genotypes and their implication for breeding against scrapie susceptibility in nine Pakistani sheep breeds	Molecular Biology Reports	International	2009
33.	Genetic Variability at seven codons of the prion protein gene in famous Pakistani sheep	Journal of Genetics	International	2008
34.	A novel HSF4 gene mutation (R405X) causing autosomal recessive congenital cataracts in a large consanguineous family from Pakistan	BMC Medical Genetics	International	2008
35.	Genome wide Significant Linkage to Stuttering on Chromosome 12	Am J Hum Genet	International	2005
36.	A new locus for nonsyndromic deafness DFNB49 maps to chromosome 5q12.3-q14.1	Human Genetics	International	2005

37.	DFNB48, a new nonsyndromic recessive deafness locus maps to chromosome 15q23-q25.1	Human Genetics	International	2005
38.	PCDH15 is expressed in the neurosensory epithelium of the eye and ear and mutant alleles are responsible for both USH1F and DFNB23	Human Molecular Genetics	International	2003

Paper Presented

S. No	Title of Paper	Name of Conference	National/ International	Date
1.	Intensive course on Bioreactor operation	Institute of Biosciences, University of Putra Malaysia organized by Fermentation Technology Unit, Laboratory of Industrial Biotechnology Institute of Biosciences, and University of Putra, Malaysia	International	March 21-23, 2007
2.	Course on Introduction to Cancer Epidemiology	IARC summer school on Cancer Epidemiology Lyon, France	International	June 9-20, 2008
3.	Course on Methodological Issues in the design and analysis of gene and environment studies	IARC summer school on Cancer Epidemiology Lyon, France	International	June 23-27, 2008
4.	20th EACR meeting	Lyon, France	International	July 5-8, 2008

Books Authored/ Edited

S. No	Name of book	Publisher	ISBN

Work Experience

S. No	From (year)	To (year)	Name of the Institution/ Organization	Position held
1.	June 2016	To-date	BUITEMS, Quetta	Professor (Tenured)
2.	Sep 2011	June 2016	BUITEMS, Quetta	Associate Professor (TTS)
3.	Jan 2011	Sep 2011	BUITEMS, Quetta	Associate Professor (BPS)
4.	Sep 2006	Jan 2011	BUITEMS, Quetta	Assistant Professor (BPS)
5.	Sep 2005	Sep 2006	BUITEMS, Quetta	Lecturer (BPS)
6.	Mar 2002	Sep 2005	CEMB, University of the Punjab, Lahore	PhD Research Scholar
7.	Mar 2000	Dec 2001	CEMB, University of the Punjab, Lahore	M.Phil Research Scholar

Area of specialization	Cancer Genetics / Human Genetics / DNA forensics
Research Interest	Molecular Biology
Future Research Plans	-

HEC Approved supervisor	Ye
If Yes, provide HEC URL	http://sc.hec.gov.pk/aphds/submit.asp?supid=2091
Research grants/ Projects	1- Study of basal breast cancer cases from Pakistan (WHO-IARC Funded) 2- Identification of new genes responsible for recessive mental retardation in Pakistani population (HEC funded) 3- Grant for Repair and maintenance of Scientific equipments (HEC funded)
Additional Information	
https://scholar.google.com.pk/citations?user=Ceq6kEEAAAAJ&hl=en&oi=ao https://www.researchgate.net/profile/Jamil_Ahmad21	