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Picture

Name	Dr. Rafiullah	
Designation	Assistant Professor	
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Qualification

Year	Degree/Certificate	Name of the Institute/ University	Field of study
	Post Doctorate	NA	
2017	PhD	Ruprecht Karl University of Heidelberg, Germany	Human Genetics
2009	MS/ Mphil	BUITEMS, Quetta - Pakistan	Biotechnology – Human Genetics
2005	Graduation	University of Balochistan, Quetta - Pakistan	Zoology, Botany, Chemistry

Publications in HEC Recognized journals

S. No	Title of Paper	Name of Journal	National/ International	Publication date
1	Identification and In Silico Analysis of a Homozygous Nonsense Variant in TGM1	Medicina (Kaunas)	International	2023
2	An expansion of phenotype: novel homozygous variant in the MED17	Journal of Neurogenetics	International	2022
3	Controversy on the management of patients carrying RET p.V804M mutation	Endocrine	International	2021
4	Prenatal exome sequencing and chromosomal microarray analysis in fetal structural anomalies in a highly consanguineous population reveals a propensity of ciliopathy genes causing multisystem phenotypes	Human Genetics	International	2021
5	Lessons Learned from Large-Scale, First-Tier Clinical Exome Sequencing in a Highly Consanguineous Population	American Journal of Human Genetics	International	2019
6	Sex Hormones Regulate SHANK Expression	Frontiers in Molecular Neuroscience	International	2018

7	Foxp1 expression is essential for sex-specific murine neonatal ultrasonic vocalization	Human Molecular Genetics	International	2017
8	A novel homozygous ARL13B variant in patients with Joubert syndrome impairs its guanine nucleotide-exchange factor activity.	European Journal of Human Genetics	International	2017
9	Homozygous missense mutation in the LMAN2L gene segregates with intellectual disability in a large consanguineous Pakistani family	Journal of Medical Genetics	International	2016
10	Mutations in the genes for thyroglobulin and thyroid peroxidase cause thyroid dyshormonogenesis and autosomal-recessive intellectual disability	Journal of Human Genetics	International	2016
11	Novel mutations of different genes in familial benign thyroid nodules and cancer	European journal of endocrinology (Submitted)	International	2022

Paper Presented

S. No	Title of Paper	Name of Conference	National/ International	Date
1	A novel ARL13B mutation impairs its guanine nucleotide-exchange factor activity in patients with Joubert syndrome	German Genetics Society – Bochum	International	March 2017
2	Gender-dimorphic expression of Foxp1 in the developing mouse brain and its impact on sex-specific communication	German Genetics Society – Lübeck	International	March 2016
3	Homozygous missense mutation in the LMAN2L gene segregates with intellectual disability in a large consanguineous Pakistani family	German Genetics Society – Lübeck	International	March 2016

Books Authored/ Edited

S. No	Name of book	Publisher	ISBN
	NA		

Work Experience

S. No	From (year)	To (year)	Name of the Institution/ Organization	Position held
1	2018	2021	King Faisal Specialist Hospital & Research Center International Holding, Riyadh, Saudi Arabia	Scientist
2	2021	Current	BUITEMS	Assistant Professor

Area of specialization			Human Genetics	
Research Interest			Human Genetics – Developmental disorders	
Future Research Plans				
HEC Approved supervisor			Yes	
If Yes, provide HEC URL			https://www.hec.gov.pk/english/scholarshipsgrants/ASA/Pages/APS-EPORTAL.aspx	
Research grants/ Projects			Submitted, waiting for decision	
Additional Information				