

Curriculum Vitae

Name: Dr. Atta Ur Rehman
Born: August 16, 1985
Nationality: Pakistani
Address: Department of Zoology, Hazara University Mansehra, KP.
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Education

- 2017 – 2020 PhD in Life Sciences
Title: Autozygome-based analysis of Pakistani families with monogenic conditions
Director: Prof. Andrea Superti-Furga, Co-Director: Prof. Carlo Rivolta
Faculty of Biology and Medicine, University of Lausanne, Switzerland
- 2009 – 2011 MPhil in Human Genetics
Title: Gene diversity and clinal analysis at ABO and Rh loci in Bajaur and Mohmand Agencies
Director: Prof. Sajid Malik
Faculty of Biological Sciences, Quaid-i-Azam University Islamabad, Pakistan
- 2005 – 2008 MSc in Zoology
Department of Zoology, University of Peshawar, Pakistan
- 2003 – 2005 BSc in Biological Sciences
University of Peshawar, Pakistan
- 2001 – 2003 Higher Secondary School Certificate (Pre-Medical)
Board of Intermediate and Secondary Education, Peshawar
- 2000 – 2001 Secondary School Certificate (Science)
Board of Intermediate and Secondary Education, Peshawar

Experience

- 2012 – Present Lecturer
Department of Zoology, Faculty of Science
Hazara University Mansehra, Pakistan
- 2009 – 2012 Lecturer
Higher Education Department
Government of Khyber Pakhtunkhwa, Pakistan

International Awards

The Swiss Government Excellence Scholarships for Foreign Scholars
(Awarded by the Swiss Federal Commission for Scholarships FCS)

Research Awards

Title: Exploring the genetic landscape of inherited retinal dystrophies in Khyber Pakhtunkhwa using whole exome sequencing approach

Funded by: Higher Education Commission of Pakistan under NRPU-2021 call

Technical Skills

- Next Generation Sequencing (NGS)
Library preparation, Quality control, and Platform optimization
- Alignment, Visualization, and Bioinformatic analysis of
Whole genome or exome sequencing data, Genes panel data as well as high-density genotype array data
- Sanger Sequencing
Primer Designing and Optimization
Polymerase Chain Reaction – Optimization and troubleshooting
Sequencing Reaction, and Sample purification
Handling of Genetic Analyzer, and Sanger results interpretation
- DNA – Extraction, Quantification, and Quality Assessment
- Gel electrophorèses

Analytical Skills

- Data access and retrieval using genome browsers – NCBI, UCSC, Ensembl etc.
- Autozygosity Mapping – using online tools such as Homozygosity Mapper, AutoMap.
- Linkage analysis – using high-density genotype data in consanguineous families
- Allele frequency calculations – using the Genome Aggregation Database (gnomAD), Exome Aggregation Consortium (ExAC), and Exome Variant Server (EVS) etc.
- Pathogenicity scoring of known variants – using the Human Gene Mutation Database (HGMD), ClinVar, and VarSome.
- In silico predictions of pathogenicity for missense variants – SIFT, PolyPhen-2, CADD, LRT, PROVEAN, Mutation Assessor, Mutation Taster and Others.
- In silico predictions of pathogenicity for splice site variants – using Human Splicing Finder, MaxEntScan, NNSPLICE etc.
- Good practice of using numerous other online tools and public databases especially for evolutionary conservation scoring, tissue expression analysis, gene network analysis, gene knock out models etc.

Publications

1. Rehman, A. U., Rashid, A., Hussain, Z., Shah, K (2022). A novel homozygous missense variant p.D339N in the PKLR gene correlates with pyruvate kinase deficiency in a Pakistani family: a case report. *Journal of Medical Case Reports*. (in press).
2. Ullah, Z., Hassan, Z., Rehman, A.U (2022).The adverse effects of industrial effluents on water quality and fish fauna of river Kabul. *JAPS, Journal of Animal and Plant Sciences* (accepted).
3. Rehman, A. U., Sepahi, N., Bedoni, N., Ravesh, Z., Salmaninejad, A., Cancellieri, F., ... & Rivolta, C. (2021). Whole exome sequencing in 17 consanguineous Iranian pedigrees expands the mutational spectrum of inherited retinal dystrophies. *Scientific Reports*, 11(1), 1-9.
4. Rehman, A. U., Anwar, I., Rashid, A., & Malik, S. (2021). Frequencies of ABO and Rh (D) Blood Group Phenotypes in Pashtuns of North-Western Pakistan: A population undergoing huge demographic changes. *International Journal of Immunogenetics*, 48(4), 336-339.
5. Rehman, A. U., Rashid, A., & Anwar, I. (2021). Single Cell RNA Sequencing (scRNA-Seq) as an Emerging Technology in Cancer Research: Applications of scRNA-seq in cancer research. *Proceedings of the Pakistan Academy of Sciences: B. Life and Environmental Sciences*, 58(3), 19-28.
6. Khan, R. A., Ullah, Z., Zaman, I. U., Khan, M. S., Mahmood, S., Akhtar, N., ... & Hussain, S. S. (2021). Population distribution and habitat analysis of Rufous treepie (*Dendrocitta vagabunda*) in Abbottabad, Pakistan. *Brazilian Journal of Biology*, 83.
7. Abdullah Muhammad, Ali Ijaz, Haleem Syed Kashif, Rehman Atta Ur, Qayyum S, Niaz Z, Ahmed S, Khan I, Khattak M. N. K, Sultana N, Tauseef Isfahan. Molecular and biochemical characterization of *Echinococcus* spp. in hydatid cyst fluid collected from human and livestock in Northern Khyber Pakhtunkhwa and Gilgit Baltistan. *Journal of Animal and Plant Sciences* 2021, 31 (5), 00-00.
8. Rehman, A. U.; Peter, V. G.; Quinodoz, M.; Rashid, A.; Khan, S. A.; Superti-Furga, A.; Rivolta, C., Exploring the genetic landscape of inherited retinal diseases in North-Western Pakistan reveals a high degree of autozygosity and prevalent founder mutations. *Investigative Ophthalmology & Visual Science* 2020, 61 (7), 2385-2385.
9. Rehman, A. U.; Peter, V. G.; Quinodoz, M.; Dawood, M.; Rivolta, C., Whole-exome sequencing in a consanguineous Pakistani family identifies a mutational hotspot in the COL7A1 gene, causing recessive dystrophic epidermolysis bullosa. *Clin Dysmorphol* 2020, 29 (2), 86-89.
10. Ravesh Z, S. A., Sepahi N, Rehman A. U, Bedoni N, Pasdar A, Quinodoz M, Peter VG, Mojarrad M, Asad AG, Piran M, Genetic Testing Identifies Known and Unknown Mutations in Iranian Families with Hereditary Retinal Diseases. *Investigative Ophthalmology & Visual Science* 2020, 10 (61(7)), 2384-2384.
11. Poulter, J. A.; Gravett, M.; Taylor, R. L.; Fujinami, K.; De Zaeytijd, J.; Bellingham, J.; Rehman, A. U.; Hayashi, T.; Kondo, M.; Rehman, A., New variants and in silico analyses in GRK1 associated Oguchi disease. *Human Mutation*. 2020 Nov 30.
12. Panagiotou, E. S.; Papatomas, T.; Nikopoulos, K.; Koukoura, S.; Quinodoz, M.; Rehman, A. U.; Giannopoulos, T.; Rivolta, C.; Konstas, A. G., Management of Full-Thickness Macular Hole in A Genetically Confirmed Case with Usher Syndrome. *Ophthalmol Ther* 2020, 9 (3), 677-684.

13. Nikopoulos, K.; Cisarova, K.; Quinodoz, M.; Koskiniemi-Kuendig, H.; Miyake, N.; Farinelli, P.; Rehman, A. U.; Khan, M. I.; Prunotto, A.; Akiyama, M.; Kamatani, Y.; Terao, C.; Miya, F.; Ikeda, Y.; Ueno, S.; Fuse, N.; Murakami, A.; Wada, Y.; Terasaki, H.; Sonoda, K. H.; Ishibashi, T.; Kubo, M.; Cremers, F. P. M.; Kutalik, Z.; Matsumoto, N.; Nishiguchi, K. M.; Nakazawa, T.; Rivolta, C., A frequent variant in the Japanese population determines quasi-Mendelian inheritance of rare retinal ciliopathy. *Nat Commun* 2019, 10 (1), 2884.
14. Rehman, A. U.; Peter, V. G.; Quinodoz, M.; Rashid, A.; Khan, S. A.; Superti-Furga, A.; Rivolta, C., Exploring the Genetic Landscape of Retinal Diseases in North-Western Pakistan Reveals a High Degree of Autozygosity and a Prevalent Founder Mutation in ABCA4. *Genes (Basel)* 2019, 11 (1).
15. Anwar, I.; Hussain, S.; Rehman, A. U.; Hussain, M., Genetic variation among the major Pakistani populations based on 15 autosomal STR markers. *International journal of legal medicine* 2019, 133 (4), 1037-1038.
16. Subhan, F.; Shahzad, R.; Tauseef, I.; Haleem, K. S.; Rehman, A. U.; Mahmood, S.; Lee, I. J., Isolation, identification, and pathological effects of beach sand bacterial extract on human skin keratinocytes in vitro. *PeerJ* 2018, 6, e4245.
17. Rehman, A. U.; Ngo, Q. A.; Martin-Merida, I.; Garcia-Sandoval, B.; Ayuso, C.; Rivolta, C., A rare chromosomal rearrangement in the X-linked opsin gene array is associated with retinal degeneration. *Investigative Ophthalmology & Visual Science* 2018, 59 (9), 5407-5407.
18. Rahman, H. U.; Rehman, A. U., Cutaneous leishmaniasis in Dir Lower District, North-West Pakistan: epidemiology and treatment response. *Journal of Pakistan Association of Dermatology* 2018, 27 (4), 353-362.
19. Khan, N.; Tabassum, S.; Ahmad, M.; Norouz, F.; Ahmad, A.; Ghayyur, S.; Rehman, A. U.; Khan, M. F., Effects of sub-lethal concentration of cypermethrin on histopathological and hematological profile of rohu (*Labeo rohita*) during acute toxicity. *Int. J. Agric. Biol* 2018, 20, 601-608.
20. Ali, H.; Tauseef, I.; Haleem, S.; Ullah, I.; Shah, A.; Khattak, M.; Mahmood, S.; Khan, M.; Rashid, A.; Rehman, A. U., Prevalence of gastrointestinal nematodes in Equines of Bajaur and Mohmand Agencies, North-West Pakistan. *JAPS, Journal of Animal and Plant Sciences* 2018, 28 (3), 695-701.
21. Rehman, A. U.; Sher, A.; Malik, S., Analysis of Genetic Differentiation at ABO and Rh Loci among the Pashtun Populations Inhabiting Lower Khyber Pakhtunkhwa, Pakistan. *Pakistan Journal of Zoology* 2017, 49 (1).
22. Tufail, M.; Rehman, A. U.; Malik, S., Determinants of consanguinity and inbreeding coefficient in the multiethnic population of Mardan, Khyber Pakhtunkhwa, Pakistan. *Asian Biomedicine* 2017, 11 (6), 451-460.
23. Saeed, A.; Rehman, A. U.; Ahmed, S.; Awais, M.; Mahmood, T.; Subhan, F.; Mahmood, S., Evaluation of Mortality Rate of Three Captive Pheasant Species at Dhodial Pheasantry, Mansehra, Pakistan. *Journal homepage: www. wesca. net* 2017, 12 (2).
24. Mahmood, S.; Rehman, A. U.; Khan, M.; Lawal, R.; Hanotte, O., Phenotypic diversity among indigenous cockfighting (Aseel) chickens from Pakistan. *The J Anim Plant Sci* 2017, 27, 1126-1132.
25. Khattak, M.; Iltaf, M.; Rehman, A. U.; Malik, S.; Zahid, M., Prevalence, socio-demographic determinants and risk factors of toxoplasmosis: case-control study in a rural community of Mardan district, northern Pakistan. *The J Anim Plant Sci* 2017, 27 (2), 617-626.

26. Khan, M. F.; Khattak, M. N. K.; He, D.; Rehman, A. U.; Chen, Y., Mitochondrial genome sequence and gene organization of Kunar Snow trout (*Schizothorax labiatus*) with phylogenetic consideration. *Gene Reports* 2017, 7, 64-73.
27. Ud-Din, A.; Rauf, M.; Ghafoor, S.; Khattak, M.; Hameed, M.; Rehman, A. U.; Shah, H., Efficient use of artificial micro-RNA to downregulate the expression of genes at the post-transcriptional level in *Arabidopsis thaliana*. *Genetics and Molecular Research: GMR* 2016, 15 (2).
28. Rehman, A. U.; Malik, S., Evaluation of Tribal Diversity of Pashtuns of Bajaur Agency, North-West Pakistan, on the Basis of Allelic Polymorphisms at ABO and Rh Loci. *Pakistan Journal of Zoology* 2016, 48 (3).
29. Rehman, A. U.; Ahmad, I.; Zaman, M.; Malik, S., Transition in consanguinity in Dir Lower district, a victim of war, natural disaster and population displacement, in north-west Pakistan—a response to Sthanadar et al.(2015). *Journal of biosocial science* 2016, 48 (3), 421-426.
30. Rashid, A.; Khattak, M.; Khan, M.; Ayaz, S.; Rehman, A. U., Gastrointestinal helminthoses: prevalence and associated risk factors in small ruminants of district kohat, pakistan. *JAPS, Journal of Animal and Plant Sciences* 2016, 26 (4), 956-962.
31. Khan, J.; Rashid, A.; Khan, K.; Malik, S.; Rehman, A. U., Distribution of ABO and Rh (D) Allelic Polymorphisms North Waziristan Agency,(Federally Administered Tribal Areas), Pakistan. *The Anthropologist* 2016, 23 (3), 423-428.
32. Ahmad, I.; Rehman, A. U.; Malik, S., Determinants of Consanguinity and Inbreeding Coefficient F in Dir Lower District, North-West Pakistan: A Multivariate Approach. *Iran J Public Health* 2016, 45 (4), 537-9.
33. Ahmad, B.; Rehman, A. U.; Malik, S., Consanguinity and Inbreeding Coefficient in Tribal Pashtuns Inhabiting the Turbulent and War-Affected Territory of Bajaur Agency, North-West Pakistan. *J Biosoc Sci* 2016, 48 (1), 113-28.
34. Rehman, A. U.; Rashid, A.; Malik, S., Genetic diversity at ABO and Rh (D) loci in the tribal groups of Mohmand Agency (Federally Administered Tribal Areas), Pakistan. *The Anthropologist* 2015, 19 (3), 679-683.
35. Saira, S.; Khattak, R.; Rehman, A. U.; Khan, A.; Khattak, M., Prevalence of goiter and Iodine status in 6-12 years school children and pregnant women of district Charsadda, Pakistan. *Acta Endocrinologica (1841-0987)* 2014, 10 (1).
36. Rehman, A. U.; Wahab, Z. u.; Khattak, M. N. K.; Malik, S., ABO and Rh (D) blood groups polymorphism in four tehsils of Bajaur Agency (Federally Administered Tribal Areas), Pakistan. *The Anthropologist* 2014, 18 (1), 259-261.

Presentations

1. Rehman, A U., et al. (2020). Exploring the genetic landscape of inherited retinal diseases in North-Western Pakistan reveals a high degree of autozygosity and prevalent founder mutations. Poster presentation at ARVO Annual Meeting, held in Baltimore, Maryland, USA (May 03 - 07, 2020).
2. Ravesh, Zeinab, Rehman, A U., et al. Genetic Testing Identifies Known and Unknown Mutations in Iranian Families with Hereditary Retinal Diseases. Co-author of a poster presentation at ARVO Annual Meeting, held in Baltimore, Maryland, USA (May 03 - 07, 2020).
3. Rehman, A. U., (2019). Exome sequencing identifies a possible mutational hotspot in the COL7A1 gene underlying recessive dystrophic epidermolysis

bullosa in a consanguineous Pakistani family. Poster Presentation at the 7th Rare Diseases Summer School organized by University of Zürich, Switzerland held from July 10th to July 12th 2019.

4. Rehman, A. U., (2019). Homozygosity mapping and mutation identification in consanguineous Pakistani families with inherited retinal degenerations). Oral Presentation at Young Researcher Vision Camp, 2019 held at Castle Wildenstein Leibertingen, Germany from July 12th to 14th, 2019.
5. Rehman, A. U., (2018). A rare chromosomal rearrangement in the X-linked opsin gene array is associated with retinal degeneration. Poster presentation at ARVO Annual Meeting, held in Honolulu, Hawaii, USA (April 29 – May 03, 2018).
6. Rehman, A. U., and Malik S (2011). Pattern of genetic heterogeneity and gene diversity at ABO and Rh loci in Bajaur and Mohmand Agencies, FATA, Pakistan. Oral Presentation at 31st Pakistan Congress of Zoology (International), University of Azad Jammu & Kashmir, Muzaffarabad (Apr. 19 - 21, 2011).
7. Khan S, Rehman, A. U. (2017). Prevalence and socio-demographic correlates of consanguinity in district Mansehra, Pakistan. Co-author of a poster presentation at 37th Pakistan Congress of Zoology (International), Government College University Faisalabad (Feb. 28 – Mar. 02, 2017).
8. Ali A and Rehman, A. U. (2017). Effects of synthetic hormones on the captive breeding of silver carp and grass carp. Co-author of a poster presentation at 37th Pakistan Congress of Zoology (International), Government College University Faisalabad (Feb. 28 – Mar. 02, 2017).

Thesis Supervisions

1. Jehan Zeb Khan (2016). Genetic heterogeneity and gene diversity analysis at ABO and Rh (D) loci in population of North Waziristan Agency, FATA, Pakistan (M.Phil Thesis, Department of Zoology, Hazara University, Mansehra, Pakistan).
2. Mohammad Javaid Khan (2016). Study of consanguinity and its socio-demographic differentials in district Shangla (M.Phil Thesis, Department of Zoology, Hazara University, Mansehra, Pakistan).
3. Hidayat Ullah (2016). Consanguinity and its determinants in population of district Battagram (M.Phil Thesis, Department of Zoology, Hazara University, Mansehra, Pakistan).
4. Sidra Khan (2016). Study on the socio-biological aspects of consanguinity in district Mansehra (M.Phil Thesis, Department of Zoology, Hazara University, Mansehra, Pakistan).
5. Atta Ur Rahim (2016). Spectrum of Genetic anomalies in district Swat (M.Phil Thesis, Department of Zoology, Hazara University, Mansehra, Pakistan).
6. Hidayat Ur Rahman (2015). Epidemiology of parasitic protozoa, *Leishmania tropica* (luhe), in the human population at Dir Lower, Malakand Division, Khyber Pakhtunkhwa, Pakistan (M.Pil Thesis, Department of Zoology, Hazara University, Mansehra, Pakistan).
7. Zia Ullah (2015). Impact of Aman Garh industrial effluents on water quality and fish fauna of river Kabul in district Nowshera, Khyber Pukhtunkhwa, Pakistan (M.Phil Thesis, Department of Zoology, Hazara University, Mansehra, Pakistan).
8. Dilawar Khan (2015). Distribution of ABO and Rh (D) blood groups in Bajaur Agency, FATA, Pakistan (B.S Thesis, Department of Zoology, Hazara University, Mansehra, Pakistan).
9. Mujeeb Ur Rahman (2015). Prevalence of consanguineous marriages in district Swat, Pakistan (M.Sc Thesis, Department of Zoology, Hazara University, Mansehra, Pakistan).
10. Ijaz Ahmad (2015). Incidence of consanguinity and its impact on human health in district Dir lower, Pakistan (M.Sc Thesis, Department of Zoology, Hazara University, Mansehra, Pakistan).
11. Muhammad Tufail (2015). Determinants of consanguinity in district Mardan, Khyber Pakhtunkhawa, Pakistan (B.S Thesis, Department of Zoology, Hazara University, Mansehra, Pakistan).
12. Abdur Rashid (2014). Prevalence of gastrointestinal parasites in small ruminants of district Kohat (M.Pil Thesis, Department of Zoology, Hazara University, Mansehra, Pakistan).
13. Haseenullah (2014). Distribution of ABO and Rh blood groups in district Dir Lower (M.Sc Thesis, Department of Zoology, Hazara University, Mansehra, Pakistan).
14. Bashir Ahmad (2012). Study of consanguineous marriages in population of Bajaur Agency (M.Sc Thesis, Department of Zoology, Hazara University, Mansehra, Pakistan).