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Education

- Doctor of Philosophy (2009-2015) School of Biological Sciences (SOBS), University of the Punjab, Lahore, Pakistan under the guidance of Dr. Sadaf Naz and Dr. Thomas B. Friedman.
- Master of Science (2007-2009) Department of Microbiology and Molecular Genetics, University of the Punjab, Lahore, Pakistan.

Research Experience

- Post doctorate fellow (April 2015 – August 2019) Laboratory of Molecular Genetics, National Institute on Deafness and Other Communication Disorders, NIH, Bethesda, MD. I continued my pre-doctorate research of the human deafness gene *CDC14A*. I developed several mouse and zebrafish models using commercially available IMRC Embryonic Stem cell clones and CRIPR/Cas9 mediated genome editing. I characterized phenotypes of these mice and zebrafish mutants and studied the underlying histopathologies using light and scanning electron microscopy. I conducted a yeast-two-hybrid screen to identify interacting partners of CDC14A in the inner ear. Now, I am in the process of validating interacting partners/ substrates of this protein by immunohistochemistry, 2D gel electrophoresis, and comparative phosho-proteomics (LC/MS). In addition to my CDC14A project, I am also involved in the studies of *Triobp*, *Tprn*, *Tbc1d24* mouse models and transcriptomics.

- Doctor of Philosophy Thesis Research (*Genetic Studies of Severe Degree of Hearing loss:* January 2011-April 2013) Laboratory of Dr. Sadaf Naz, School of Biological Sciences, University of the Punjab, Lahore, Pakistan. Identified and enrolled large consanguineous families. Used the techniques of homozygosity mapping, linkage analysis (SNP and STR genotyping), Sanger and massively parallel sequencing and identified contributions of known and novel genes in the etiology of moderate to severe hearing loss. Studied effect of mutations on mRNA and protein expression/stability.

- Pre-doctoral Fellow (April 2013 – April 2015) Laboratory of Molecular Genetics, National Institute on Deafness and Other Communication Disorders, NIH. Bethesda, MD. Identified novel genes that cause hearing loss by massively parallel sequencing and SNP genotyping. Selected one of these genes (*CDC14A*, Cell Division Cycle gene 14A) for further studies and studied alternative splice isoforms of this gene in mouse and human tissues. Studied the localization pattern of CDC14A in mice and zebrafish inner ear through Immunohistochemistry and gene gun transfections. Worked on creating mouse and zebrafish models of human deafness caused by mutations of *CDC14A*.

- Enzymology Research project (November 2009 – April 2010) Laboratory of Dr. Muzaffar Iqbal (Calzyme, USA), School of Biological Sciences, Lahore. Worked on the isolation, purification, crystallization and characterization of LDH, MDH from bovine organs and ascorbate oxidase and citrate synthase from the cucumber peels and oranges, respectively.

- Major Research project in the M.Sc (2007 – 2009)

    - Identification and characterization of Bacteriocin producing bacteria from different dairy items
    - Characterization of metal resistance of microbes present in industrial effluents
    - Identification of new antibiotic producing strains of bacteria and fungi from soil

Publications

Nadeem M.S, Nissar A, Shahid S, **Imtiaz A,** Mahfooz M, Asghar M.T, Shakoori A.R (2011)" Purification and characterization of lactate dehydrogenase from the heart ventricles of river buffalo (Bubalus Bubalis)" Pakistan Journal of Zoology, 43 (2): 315-319.

Imtiaz A, Naz S (2012) "A rapid and cost-effective protocol for screening known genes for autosomal recessive deafness" Pakistan Journal of Zoology, 44 (3): 641-647.

Bashir R, **Imtiaz A*,** Fatima A, Alam A, Naz S (2013) "The c.42_52del11 mutation in *TPRN* and progressive hearing loss in a family from Pakistan". Biochemical Genetics, 51 (5-6): 350-357. **[* Co-first author]**

Imtiaz A, Kohrman D.C, Naz S (2014) "A Frameshift Mutation in *GRXCR2* Causes Recessively Inherited Hearing Loss" Human Mutation, 2014 35(5):618-24.

Salman M, Bashir R, **Imtiaz A**, Maqsood A, Mujtaba G, Iqbal M, Naz S (2015) "Mutations of *GJB2* Encoding Connexin 26 Contribute to nonsyndromic Moderate and Severe Hearing Loss in Pakistan". European Archives of Otorhinolaryngology, 2015 272(8):2071-5.

Mujtaba G, Schultz J.M, **Imtiaz A**, Morell R.J, Friedman T.B, Naz S (2015) A mutation of *MET*, encoding hepatocyte growth factor receptor, is associated with human *DFNB97* hearing loss. Journal of Medical Genetics, 2015 52(8):548-52

Imtiaz A, Maqsood A, Rehman A.U, Morell R.J, Holt J.R, Friedman T.B, Naz S (2016) Recessive mutations of TMC1 associated with moderate to severe hearing loss. Neurogenetics, 2016 17(2):115-23

Rehman A.U, Bird J.E, Faridi R, Shahzad M, Shah S, Lee K, Khan S.N, **Imtiaz A**, Ahmed Z.M, Riazuddin S, Santos-Cortez R.L, Ahmad W, Leal S.M, Riazuddin S, Friedman T.B. "Mutational Spectrum of *MYO15A* and Molecular Mechanisms Underlying Human Deafness DFNB3" Human Mutation, 2016 Oct;37(10):991-1003.

Naz S, **Imtiaz A**, Mujtaba G, Maqsood A, Bashir R, Bukhari I, Khan M.R, Ramzan M, Fatima A, Rehman A.U, Iqbal M, Chaudhry T, Lund M, Brewer C.C, Morell R.J, Friedman T.B (2017). Genetic causes of moderate to severe hearing loss point to modifiers. Clinical Genetics, 2017 91(4):589-598**. [* Co-first author]**

Imtiaz A, Belyantseva I.A, Beirl A.J, Fenollar-Ferrer C, Bashir R, Bukhari I, Bouzid A, Shaukat U, Azaiez H, Booth K.T, Kahrizi K, Najmabadi H, Maqsood A, Wilson E.A, Fitzgerald T.S, Tlili A, Olszewski R, Lund M, Chaudhry T, Rehman A.U, Starost M.F, Waryah A.M, Hoa M, Dong L, Morell R.J, Smith R.J.H, Riazuddin S, Masmoudi S, Kindt K, Naz S, Friedman T.B (2018). CDC14A Phosphatase is Essential for Hearing and Male Fertility in Mouse and Human. Human Molecular Genetics, 27(5):780-798.

Richard EM, Santos-Cortez RLP, Faridi R, Rehman AU, Lee K, Shahzad M, Acharya A, Khan AA, **Imtiaz A**, Chakchouk I, Takla C, Abbe I, Rafeeq M, Liaqat K, Chaudhry T, Bamshad MJ, Nickerson DA; University of Washington Center for Mendelian Genomics, Schrauwen I, Khan SN, Morell RJ, Zafar S, Ansar M, Ahmed ZM,

Ahmad W, Riazuddin S, Friedman TB, Leal SM, Riazuddin S (2019). Global genetic insight contributed by consanguineous Pakistani families segregating hearing loss. Human Mutation, 2019 Jan;40(1):53-72.

Katsuno T, Belyantseva I.A, Cartagena-Rivera A.X, Ohta K, Crump S.M, Petralia R.S, Ono K, Tona R, **Imtiaz A**, Rehman A, Kiyonari H, Kaneko M, Wang Y.X, Abe T, Ikeya M, Fenollar-Ferrer C, Riordan G.P, Wilson E, Fitzgerald T.S, Segawa K, Omori K, Ito J, Frolenkov G.I, Friedman T.B, Kitajiri S (2019). TRIOBP-5 sculpts stereocilia rootlets and stiffens supporting cells for hearing. Accepted for publication, JCI Insights, 2019 June: 4(12) doi: 10.1172/jci.insight.128561

Podium Presentations

Imtiaz A, Naz S**.** Identification of genes involved in moderate to severe hearing loss. 32ND Pakistan Congress of Zoology, March 2012.

Katsuno T, Belyantseva I.A, Petralia R.S, Wang Y, Ohta K, Ono K, Ikeya M, Riordan G.P, Duda J, Wilson E.A, Fitzgerald T.S, Rehman A.U, **Imtiaz A**, Ito J, Friedman T.B, Kitajiri S. The development and maintenance of hair cell stereocilia rootlets by isoform specific functions of TRIOBP. 2017 Hannover 54th Workshop on Inner Ear Biology and Symposium, Hannover, Germany.

Imtiaz A, Belyantseva I.A, Beirl A.J, Fenollar-Ferrer C, Bashir R, Bukhari I, Bouzid A, Shaukat U, Azaiez H, Booth K.T, Kahrizi K, Najmabadi H, Maqsood A, Wilson E.A, Fitzgerald T.S, Tlili A, Olszewski R, Lund M, Chaudhry T, Rehman A.U, Starost M.F, Waryah A.M, Hoa M, Dong L, Morell R.J, Smith R.J.H, Riazuddin S, Masmoudi S, Kindt K, Naz S, Friedman T.B. Mutations of CDC14A are associated with nonsyndromic deafness DFNB32 or HIIMS, Hearing Impairment Infertile Male Syndrome. American Society of human genetics, 2017 Annual Meeting, Orlando, FL, USA.

Imtiaz A, Belyantseva I.A, Beirl A.J, Fenollar-Ferrer C, Dong L, Kindt K.S, and Friedman T.B. CDC14A phosphatase is essential for hearing and male fertility in mouse and human. C-CEBH and NIDCD Joint Meeting October 9, 2018, Maryland, USA.

Poster presentations

Nadeem M.S, Nissar A, Shahid S, **Imtiaz A,** Mahfooz M, Asghar M.T, Shakoori A.R (2011)" Purification and characterization of lactate dehydrogenase from the heart ventricles of river buffalo (Bubalus Bubalis)" 30TH Pakistan Congress of Zoology, March 2010.

Imtiaz A, Bashir R, Mubtaja G, Maqsood A, Bukhari I, Rehman A, Morell R.J, Friedman T.B, Naz S. Alleles of the reported deafness genes are major contributors to the etiology of moderate to severe hearing loss in Pakistani population. October 18-22 2014, 64th Annual meeting of American Society of Human Genetics.

Imtiaz A, Bashir R, Mubtaja G, Maqsood A, Bukhari I, Rehman A, Morell R.J, Friedman T.B, Naz S. Alleles of the reported deafness genes are major contributors to the etiology of moderate to severe hearing loss in Pakistani population. January 13, 2015, NIH Graduate Student Research Symposium.

Chaudhry T, Lund M, Naz S, Friedman T.B, **Imtiaz A**. Mutations of Genes Causing Moderate to Severe Hearing Loss. August 6, 2015. Summer student poster day. NIH, Bethesda.

Lund M, Williams C, Masood A, Belyantseva I.A, Naz S, Friedman T.B. Ayesha Imtiaz. Deafness-causing gene encodes a protein colocalizing with tubulin. August 6, 2016. Summer student poster day. NIH, Bethesda.

Belyantseva I.A, Katsuno T, Riordan G, Rehman A.U, **Imtiaz A**, Morozko E, Duda J, Wilson E.A, Petralia R, Fitzgerald T, Friedman T.B, Kitajiri S. TRIOBP5 Deficient Mouse is Deaf but Hair Cells Develop Stereocilia Rootlets. 40TH Annual MidWinter Meeting 2017, Association of Research in Otolaryngology, Baltimore, Maryland, USA.

Imtiaz A, Belyantseva I.A, Beirl A.J, Fenollar-Ferrer C, Bashir R, Bukhari I, Bouzid A, Shaukat U, Azaiez H, Booth K.T, Najmabadi H, Rehman A.U, Hoa M, Dong L, Morell R.J, Smith R.J.H, Riazuddin S, Masmoudi S, Kindt K, Naz S, Friedman T.B. Human and mouse CDC14A phosphatase activity is essential for hearing and male fertility**.** 41st Annual MidWinter Meeting 2018, Association of Research in Otolaryngology, San Diego, California, USA.

Teaching Experience

- Lecturer of biology and chemistry (2004-2007) in different private organizations (Paksitan Academy of Sciences, Imran Science Academy, Rana Academy, Iqbal Town, Lahore).
- Mentoring of summer interns (June-July 2015, June-August 2016), NIDCD/NIH, Bethesda.
- Mentored NIH pre-doctorate visiting fellows Azra Maqsood, Memoona Ramzan and Rabia Faridi.
- Mentoring of NIH Post baccalaureate IRTA, Taimur Chaudary and Wenqian Chen from August 2018 to August 2019.

Honors

- President scientific, social work and debating societies at school and college (2000-2007).
- Roll of Honor, Samanabad College, Lahore, Pakistan (2007).
- Merit Scholarship, M.Sc. University of the Punjab, Lahore, Pakistan (2007-2009).
- Roll of Honor, M.Sc. University of the Punjab, Lahore, Pakistan (2009).
- Competitive HEC Indigenous PhD scholarship (2012-2014)
- Highest CGPA in M.Phil leading to Ph.D course work, School of Biological Sciences, Lahore (2011).
- NIDCD Director's Award for Educating children in the community about hearing, presented November 2018.