PERSONAL DATA

Name: Sadaf Naz Phone: 042-99231819
Email: naz.sbs@pu.edu.pk

Expertise Field: Molecular Biology/Human Genetics

EDUCATION

B.Sc. in Zoology, Botany, Chemistry
Kinnaird College for women (KC), Lahore, Pakistan, 1993

M.Sc. in Biochemistry/Molecular Biology
Quaid-i-Azam University (QAU), Islamabad, Pakistan, 1996

Ph.D. in Molecular Biology
Center of Excellence in Molecular Biology (CEMB), University of the Punjab (PU), Lahore, Pakistan, 2001
Thesis: "Study of Nonsyndromic Recessive Deafness by Linkage Analysis"

TRAINING/COURSES

Working with **embryonic stem cells** and generation of **transgenic mice**: "Molecular Embryology of the Mouse" (June 5-25 2002), Cold Spring Harbor Laboratories, New York, **USA**.

"Use of **bioinformatics** in genomic research" (August 19th to September 2nd 2006), COMSTECH Secretariat, Islamabad, **Pakistan**.

"**Next Generation Sequencing**" (27h June 2011 to 16th July 2011), DNA sequencing Center, Brigham Young University, Provo, Utah, **USA**.

"52nd Annual Short Course on **Medical and Experimental Mammalian Genetics**" (17th July 2011 to 29th July 2011), The Jackson Laboratory, Bar Harbor, Maine, **USA**.

POSITIONS

31/8/2019-to date Professor, School of Biological Sciences, University of the Punjab, **Pakistan**

22/8/2011-30/8/2019 Associate Professor, School of Biological Sciences, University of the Punjab, **Pakistan**

22/9/2005-2011 Assistant Professor, School of Biological Sciences, University of the Punjab, Lahore, **Pakistan**

10/2001-8/2005 Postdoctoral fellow, National Institute on Deafness and other Communication Disorders, NIH, Rockville, MD, **USA**

AWARDS and DISTINCTIONS

1996 **First** position in **M.Sc.** Biochemistry/Molecular Biology group, Quaid-i-Azam University, Islamabad, **Pakistan**.

2003 Fellows Award for Research Excellence (**FARE**) National Institutes of Health, **USA**.

2006, **2007**, **2011**, **2013-2016** Research Productivity **Award**, Pakistan Council for Science and Technology, Ministry of Science and Technology, Islamabad, **Pakistan**.

2012 Performance Evaluation **Award**, University of the Punjab, Lahore, **Pakistan**

PROFESSIONAL EXPERTISE

Human linkage analyses, gene mapping, Gene identification by positional candidate gene and next generation sequencing analyses, Molecular Biology Recombinant DNA technology, recombinant protein expression and isolation. Transgenics, mice breeding and evaluation of hearing of mice by Auditory Brainstem Response. Cell culture and immunocytochemistry.

PROFESSIONAL ASSOCIATIONS

2002 to date Member of American Society of Human Genetics, USA.
Life membership, Pakistan Society for Biochemistry and Molecular Biology.

RESEARCH GRANTS AND AWARDS

NATIONAL

2007-2010 Higher Education Commission, **Pakistan**, Grant 836, Rs. 4,198,896 (**Principal Investigator**). Molecular Characterization of Dystonia and Wolfram Syndrome in Pakistan.

2009-2012 Higher Education Commission, **Pakistan**, Grant 1262, Rs. 4,103,154 (**Co-Principal Investigator**). Genetic & Molecular Characterization of Oculocutaneous Albinism (OCA) and Related Syndrome in Pakistan.

2015-2018 Higher Education Commission, Pakistan, Grant 2877, Rs. 3,165,202 (**Principal Investigator**) Genetic studies of neurological movement disorders and related syndromes.

2016-2019 Higher Education Commission, Pakistan, Grant 4352, Rs.1,846,284 (**Co-Principal Investigator**) Genetic analysis of disease causing genes for glaucoma in Pakistani population.

2016-2019 Higher Education Commission, Pakistan, Grant 3288, Rs. 3,592,600 (**Principal Investigator**) Genetics of recessively inherited stable or progressive hearing loss in Pakistan.

2017-2018 University of the Punjab, Pakistan, Grant 105, Rs. 200, 000, (**Principal Investigator**) Identification of genes for skeletal dysplasia syndromes in families from Pakistan.

INTERNATIONAL

2006-2009 Brigham Young University, Provo, Utah, **USA**, MEG, US$20,000 (**Co-Principal Investigator**). Study of cleft Lip with or without cleft palate. ($5000 for work in Pakistan)

2007-2012, no cost extension till 2013 Fogarty International Center and National Institute on Deafness and other Communication Disorders (NIDCD), National Institutes of Health (NIH), **USA**, Grant R01TW007608, US$270,000 (**Principal Investigator**). Genetic basis of moderate to severe hearing loss in Pakistan.

2010 Deustche Forschungsgemeinschaft, DFG, **Germany**, Funding for the initiation and enhancement of bilateral co-operation with University of Lübeck, €4200. (**Principal Investigator**). Molecular characterization of the phenotype in a family with unique dystonia syndrome from Pakistan.

2015-2016 Growing Stronger, **USA** and Koshish Foundation, **USA** (**Principal Investigator**) US$21,000. Genetics of dwarfism and skeletal dysplasia.

2015-2018 Deustche Forschungsgemeinschaft, **Germany**, LO 1555/8-1, (**Co-Investigator**), €152,240. Molecular characterization of complex movement disorders with predominant dystonic features in consanguineous families.

2016-2017 University of Minnesota, **USA** (**Principal Investigator**) $4100. Molecular characterization of Psychotic disorders in Pakistan.

Travel award
2017 American Society of Human Genetics, Developing Countries award. $2500 (for travelling to USA to present research work in the annual ASHG 2017 meeting)

RESEARCH
1) Current Research (**22nd September 2005 to present**) School of Biological Sciences (SBS), University of the Punjab (PU), Lahore, Pakistan.

1. Genetics of nonsyndromic moderate to severe hearing loss in Pakistan.
2. Genetics of Cleft Lip with or without Cleft palate (CL/P) in Pakistan.
3. Studies on molecular basis of neurological and movement disorders.
4. Studies on genetics of skeletal dysplasia.
5. Studies on psychiatric disorders

2) Research and Postdoctoral fellowship (9th April 2001-31st July 2005) Laboratory of Molecular Genetics, National Institute on Deafness and other Communication Disorders (NIDCD), National Institutes of Health (NIH), USA. Mentor: Dr. Edward R Wilcox, Dr. Thomas B Friedman.

1. Genetics of severe to profound deafness
2. Functional studies in mice for a deafness gene
3. Protein expression, affinity purification, immunocytochemistry

3) Research for Ph.D. (1996 –2001) CEMB, Pakistan, (November 1998-April 1999) LMG, NIDCD, NIH, USA, Supervisor: Dr. Sheikh Riazuddin, Mentor: Dr. Edward R Wilcox.

Genetics of severe to profound deafness in Pakistan

PUBLICATIONS (Impact Factor 258.881, Citations 2015)

1. Riazuddin S, Castelein CM, Ahmed ZM, Lalwani AK, Mastroianni MA, **Naz S**, Smith TN, Liburd NA, Friedman TB, Griffith AJ, Riazuddin S, Wilcox ER (2000). Dominant modifier DFNM1 suppresses recessive deafness DFNB26. ***Nat Genet*** 26:431-434.

2. Wilcox ER, Burton QL, **Naz S**, Riazuddin S, Smith TN, Ploplis B, Belyantseva I, Ben-Yosef T, Liburd NA, Morell RJ, Kachar B, Wu DK, Griffith AJ, Riazuddin S, Friedman TB (2001). Mutations in the gene encoding tight junction claudin-14 cause autosomal recessive deafness DFNB29. ***Cell*** 104:165-172.

3. Ben-Yosef T, Wattenhofer M, Riazuddin S, Ahmed ZM, Scott HS, Kudoh J, Shibuya K, Antonarakis SE, Bonne-Tamir B, Radhakrishna U, **Naz S**, Ahmed Z, Riazuddin S, Pandya A, Nance WE, Wilcox ER, Friedman TB, Morell RJ (2001). Novel mutations of TMPRSS3 in four DFNB8/B10 families segregating congenital autosomal recessive deafness. ***J Med Genet*** 38: 396-400.

4. Liburd N, Ghosh M, Riazuddin S, **Naz S**, Khan S, Ahmed Z, Riazuddin S, Liang Y, Menon PS, Smith T, Smith AC, Chen KS, Lupski JR, Wilcox ER, Potocki L, Friedman TB (2001). Novel mutations of MYO15A associated with profound deafness in consanguineous families and moderately severe hearing loss in a patient with Smith-Magenis syndrome. ***Hum Genet*** 109:535-541.

5. Kurima K, Peters LM, Yang Y, Riazuddin S, Ahmed ZM, **Naz S**, Arnaud D, Drury S, Mo J, Makishima T, Ghosh M, Menon PS, Deshmukh D, Oddoux C, Ostrer H, Khan S, Riazuddin S, Deininger PL, Hampton LL, Sullivan SL, Battey JF Jr, Keats BJ, Wilcox ER, Friedman TB, Griffith AJ (2002). Dominant and recessive deafness caused by mutations of a novel gene, TMC1, required for cochlear hair-cell function. ***Nat Genet*** 30:277-284.

6. **Naz S**, Giguere CM, Kohrman DC, Mitchem KL, Riazuddin S, Morell R, Ramesh A, Srisailpaatahy S, Deshmukh D, Riazuddin S, Griffith AJ, Friedman TB, Smith RJH, Wilcox ER (2002). Hearing loss in *DFNB6* families associated with mutations in a novel gene, *TMIE.* ***Am J Hum Genet*** 71:632-636.

7. Park HJ, Shaukat S, Liu XZ, Hahn SH, **Naz S**, Ghosh M, Kim HN, Moon SK, Abe S, Tukamoto K, Riazuddin S, Kabra M, Erdenetungalag R, Radnaabazar J, Khan S, Pandya A, Usami SI, Nance WE, Wilcox ER, Riazuddin S, Griffith AJ (2003). Origins and frequencies of SLC26A4 (PDS) mutations in east and south Asians: global implications for the epidemiology of deafness. ***J Med Genet*** 40:242-248.

8. **Naz S**, Alasti F, Mowjoodi A, Riazuddin S, Sanati MH, Friedman TB, Griffith AJ, Wilcox ER, Riazuddin S (2003). Distinctive audiometric profile associated with *DFNB21* alleles of *TECTA*. ***J Med Genet*** 40:360-363.

9. **Naz S**, Griffith AJ, Riazuddin S, Hampton LL, Battey JF, Khan SN, Riazuddin S, Wilcox ER, Friedman TB (2004). Mutations of ESPN cause autosomal recessive deafness and vestibular dysfunction. ***J Med Genet*** 41:591-595.

10. Belyantseva IA, Boger ET, **Naz S**, Frolenkov GI, Sellers JR, Ahmed ZM, Griffith AJ, Friedman TB (2005). Myosin-XVa is required for tip localization of whirlin and differential elongation of hair-cell stereocilia. ***Nat Cell Biol*** 7:148-156.

11. Riazuddin S, Khan SN, Ahmed ZM, Ghosh M, Caution K, Nazli S, Kabra M, Zafar AU, Chen K, **Naz S**, Antonellis A, Pavan WJ, Green ED, Wilcox ER, Friedman PL, Morell RJ, Riazuddin S, Friedman TB (2006). Mutations in TRIOBP,

encoding a putative cytoskeletal organizing protein, are associated with nonsyndromic recessive deafness. ***Am J Hum Genet*** 78:137-143.

12. Malik S , Kakar N, Hasnain S, Ahmad J, Wilcox ER, **Naz S** (2010). Epidemiology of Van der Woude Syndrome from mutational analyses in affected patients from Pakistan. ***Clin Genet*** 78: 247-256.

13. Bashir R, Fatima A, **Naz S** (2010). A frameshift Mutation in *SANS* results in atypical Usher syndrome. ***Clin Genet*** 78:601-603.

14. Bashir R, Fatima A, **Naz S** [Naz S, **Co-first author**] (2010). Mutations in *CLDN14* are associated with different hearing thresholds*.* ***J Hum Genet*** 55:767-770.

15. Arif B, Grünewald A, Fatima A, Ramirez A , Ali A, Brüggemann N, Würfel J, Rolfs A, Lohmann K, Malik A, Klein C, **Naz S** (2011). An unusual neurological syndrome of crawling gait, dystonia, pyramidal signs and limited speech. ***Mov Disord*** 26:2279-2283.

16. Bashir R, Fatima A, **Naz S** (2012). Prioritized sequencing of the second exon of *MYO15A* reveals a new mutation segregating in a Pakistani family with moderate to severe hearing loss. ***Eur J Med Genet*** 55:99-102.

17. Imtiaz A, **Naz S** (2012) A rapid and cost-effective protocol for screening known genes for autosomal recessive deafness. ***Pak J Zool*** 44:641-647.

18. Mujtaba G, Bukhari I, Fatima A, **Naz S** (2012). A p.C343S missense mutation in PJVK causes progressive hearing loss. ***Gene*** 504:98-101.

19. **Naz S**, Fatima A. [Naz S, **corresponding author**] (2013). Amplification of GC-rich DNA for high throughput family based genetic studies. ***Mol Biotechnol*** 53:345-350.

20. Arif B, Kumar KR, Seibler P, Franke F, Fatima A, Winkler S, Nürnberg G, Thiele H, Nürnberg P, Jamil AZ, Brüggemann, A, Abbas G, Klein C, **Naz S**, Lohmann K (2013). A novel OPA3 mutation revealed by exome sequencing: An example of reverse phenotyping ***JAMA Neurol*** 70:783-787.

21. Bashir R, Imtiaz A, Fatima A, Alam A, **Naz S** (2013). c.42_52del11 mutation in *TPRN* and progressive hearing loss in a family from Pakistan. ***Biochem Genet*** 51:350-357.

22. Khan MR, Bashir R, **Naz S** (2013). SLC26A4 mutations in patients with moderate to severe hearing loss. ***Biochem Genet*** 51:514-523.

23. Bukhari I, Mujtaba G, **Naz S** (2013). Contribution of GJB2 mutations to hearing loss in Hazara division of Pakistan. ***Biochem Genet*** 51:524-529.

24. Doss S, Lohmann K, Seibler P Arns B, Klopstock T, Zühlke C, Freimann K, Winkler S, Lohnau T, Drungowski M, Nürnberg P, Wiegers K, Lohmann E, **Naz S**, Kasten M, Bohner G, Ramirez A, Endres M, Klein C (2014). Recessive

dystonia-ataxia syndrome in a Turkish family caused by a *COX20* (*FAM36A*) mutation ***J Neurol*** 261:207-212.

25. Malik S, Wilcox ER, **Naz S** (2014) Novel lip pit phenotypes and mutations of IRF6 in Van der Woude Syndrome patients from Pakistan. ***Clin Genet*** 85:487-491.

26. Imtiaz A, Kohrman DC, **Naz S** (2014) A frameshift mutation in GRXCR2 causes recessively inherited hearing loss ***Hum Mutat*** 35:618-624.

27. Bashir R, Sanai M, Azeem A, Altaf I, Saleem F, **Naz S** (2014) Contribution of *GLC3A* locus to primary congenital glaucoma in Pakistani population. ***Pak J Med Sci*** 30:1341-1345.

28. 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. ***Eur Arch Otorhinolaryngol*** 272:2071-2075.

29. Mujtaba G, Schultz JM, Imtiaz A, Morell RJ, Friedman TB, **Naz S** (2015). A mutation of MET, encoding hepatocyte growth factor receptor, is associated with human DFNB97 hearing loss. ***J Med Genet*** 52:548-552.

30. Bashir R, Tahir H, Yousaf K, Naz S, **Naz S** (2015). Homozygous p.G61E mutation in a consanguineous Pakistani family with co-existence of Juvenile-onset open angle and primary congenital glaucoma. ***Gene*** 570:295–298.

31. Imtiaz A, Maqsood A, Rehman AU, Morell RJ, Holt JR, Friedman TB, **Naz S** (2016) Recessive mutations of *TMC1* associated with moderate to severe hearing loss ***Neurogenet*** 17:115-123.

32. Lohmann K, Schlicht F, Svetel M, Hinrichs F, Zittel S, Graf J. Lohnau T, Schmidt A, Mir P, Krause P, Lang AE, Jabusch HC, Wolters A, Kamm C, Zeuner KE, Altenmüller E **Naz S**, Chung SJ, Kostic VS, Münchau S, Kühn AA, Brüggemann N, Klein C (2016). The role of mutations in COL6A3 in isolated dystonia. ***J Neurol*** 263:730-734.

33. Iqbal M, Muhammad N, Ali SA, Kostjukovits S, Makitie O, **Naz S** (2017) The Finnish founder mutation c.70 A>G in *RMRP* causes cartilage-hair hypoplasia in a Pakistani family. ***Clin Dysmorph*** 26:121-123.

34. **Naz S**, Imtiaz A, Mujtaba G, Maqsood A, Bashir R, Bukhari I, Khan MR, Ramzan M, Fatima A, Rehman AU, Iqbal M, Chaudhry T, Lund M, Brewer CC, Morell JR, Friedman TB **[Naz S corresponding author]** (2017) Genetic causes of moderate to severe hearing loss point to modifiers. ***Clin Genet*** 91:589-598.

35. Tariq H, **Naz S** (2017) *TFG* associated hereditary spastic paraplegia: an addition to the phenotypic spectrum. ***Neurogenet*** 18:105-109.

36. Tariq H, Mukhtar S, **Naz S** (2017). A novel mutation in *ALS2* associated with severe and progressive infantile onset of spastic paralysis. ***J Neurogenet*** 31: 26-29.

37. Manzoor H, Bukhari I, Wajid M, Zhang Y, Zhang H, Brüggemann N, Klein C, Shi Q, **Naz S** (2017) A novel *APTX* variant and ataxia with oculomotor apraxia type 1. ***J Clin Neurol*** 13:303-305.

38. Imtiaz A, Belyantseva IA, Beirl AJ, Fenollar-Ferrer C, Bashir R, Bouzid A, Shaukat U, Bukhari I, Azaiez H, Booth KT, Kahrizi K, Maqsood A, Wilson EA, Fitzgerald TS, Tlili A, Olszewski R, Lund M, Chaudhry T, Rehman AU, Starost MF, Waryah AM, Hoa M, Dong L, Morell RJ, Smith, RJH, Riazuddin S, Masmoudi S, Kindt K, **Naz S**, Friedman TB **[Naz S and Friedman TB, equal contribution]** (2018) CDC14A phosphatase is essential for hearing and male fertility in mouse and human ***Hum Mol Genet*** 27:780-798

39. Ain UN, Makitie, O, **Naz S** (2018) Autosomal recessive chondrodysplasia with severe short stature caused by a biallelic *COL10A1* variant. ***J Med Genet*** 55:403-407

40. Manzoor H, Brüggemann N, Hinrichs F, Hussain, HMJ, Wajid M, Bäumer T, Münchau A, **Naz S**, Lohmann K **[Naz S and Lohmann K, corresponding authors]** (2018) Novel homozygous variants in ATCAY, MCOLN1, and SACS in complex neurological disorders. ***Park Rel Dis*** 51:91-95

41. Bashir R, Yousaf K, Tahir H, Sanai M, Qayyum S, Naz S, **Naz S** (2018). Clinical variability of *CYP1B1* gene variants in Pakistani Primary Congenital Glaucoma families. ***J Pak Med Asso*** 68:1205-1211

42. Tariq H, Imran R, **Naz S** (2018) A novel homozygous missense mutation within SETX gene causing AOA2 ***J Clin Neurol*** 14:498-504

43. Ramzan M, Idrees H, Mujtaba G, Sobreira N, Witmer D, Baylor-Hopkins Center for Mendelian Genomics, **Naz S** (2019) Bi-allelic Pro291Leu variant in *KCNQ4* leads to early onset non-syndromic hearing loss. ***Gene*** 705:109-112

44. Ain NU, Iqbal M, Valta H, Emerling CA, Ahmed S, Makitie O, **Naz S** Novel variants in natriuretic peptide receptor 2 in unrelated patients with acromesomelic dysplasia type maroteaux ***Eur J Med Genet*** In press

45. Aslam F, **Naz S** Ataxia and dysarthria due to an ABCA2 variant: Extension of the phenotypic spectrum ***Park Rel Dis*** In press

46. Tariq H, Butt JU, Houlden H, **Naz S** Are some *C19orf12* variants monoallelic for neurological disorders? ***Park Rel Dis*** In press

ABSTRACTS

12 abstracts published in national and international meetings' proceedings

EDITED BOOK and written CHAPTER

Naz S (2012). Genetics of nonsyndromic recessively inherited moderate to severe and progressive deafness in humans in *"**Hearing Loss***" ed **Naz S**, Intech, Croatia. Chapter 12, pp 247-274. (ISBN 979-953-307-271-4)

RESEARCH SUPERVISION

PhD degrees awarded: 7
MPhil degrees awarded: 11 (One as co-supervisor)
PhD students under supervision: 5
MPhil students under supervision: 1