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 (**A grade**)

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

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

22/8/2011-date 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 techniques, 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.

ONGOING RESEARCH GRANTS

4 national and International funded projects

COMPLETED RESEARCH GRANTS

6 national and International funded projects

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. (completed)
3. Studies on molecular basis of dystonia and movement disorders.
4. Studies on genetics of skeletal dysplasia.

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 (Selected from 34)

1. 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.

2. **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.

3. 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. **.**

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

5. 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.

6. 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.

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

8. **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.

9. 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.

10. 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.

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

12. 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.

13. 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.

14. 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.

15. 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*** 263:730-4.

16. **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]** Genetic causes of moderate to severe hearing loss point to modifiers. ***Clin Genet*** DOI:10.1111/cge.12856

17. Iqbal M, Muhammad N, Ali SA, Kostjukovits S, Makitie O, **Naz S** The Finnish founder mutation c.70 A>G in *RMRP* causes cartilage-hair hypoplasia in a Pakistani family. ***Clin Dysmorph*** DOI: 10.1097/MCD.0000000000000155

EDITED BOOK WITH 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: 5
Mphil degrees awarded: 6
PhD students under supervision: 5
Mphil students under supervision: 2