Name: Sadaf Naz **Position**: Acting Director General/Professor
Gender: Female **Phone:** (+92) 4299231819 **Email address:** naz.sbs@pu.edu.pk

EDUCATION

Bachelor of Science (BSc)
Kinnaird College (KC), Lahore, Pakistan [1991-1993]
Fields of study: Botany, Zoology, Chemistry

Master of Science (MSc)
Quaid-i-Azam University (QAU), Islamabad, Pakistan [1994-1996]
Fields of study: Biochemistry, Molecular Biology

Doctor of Philosophy (PhD)
Center of Excellence in Molecular Biology (CEMB),
University of the Punjab (PU), Lahore, Pakistan [1996-2001]
Field of study: Molecular Biology

TRAINING
"Molecular Embryology of the Mouse." Course for embryonic stem cells and generation of transgenic mice: *Cold Spring Harbor Laboratories, New York, **USA*** [5th June to 25th June 2002]

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

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

CONTINUING EDUCATION
"52nd Annual Short Course on Medical and Experimental Mammalian Genetics" *The Jackson Laboratory, Bar Harbor, Maine, **USA*** [17th July 2011 to 29th July 2011]

Genetics in your clinic: what you can and should do now" Virtual symposium: *American Society of Human Genetics, **USA*** [22nd March 2016]

Advancing Precision Medicine, *BioTechniques, **USA*** [13th–15th November 2019]

POSITIONS

Acting Director General
*School of Biological Sciences, University of the Punjab, **Pakistan*** [21/3/2022-to date]

Professor
*School of Biological Sciences, University of the Punjab, **Pakistan*** [31/8/2018-To date]

Associate Professor
*School of Biological Sciences, University of the Punjab, **Pakistan*** [22/8/2011-30/8/2018]

Assistant Professor
*School of Biological Sciences, University of the Punjab, **Pakistan*** [22/9/2005-21/8/2011]

Postdoctoral Fellow & Researcher
Postdoctoral fellow, *National Institute on Deafness and other Communication Disorders (NIDCD), National Institutes of Health (NIH), Rockville, MD, **USA*** [1/10/2001-31/7/2005 & 9/4/2001-30/9/2001, respectively]

AWARDS and DISTINCTIONS

Distinction in Education

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

Awards for Research

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

Pakistan Council for Science and Technology, Ministry of Science and Technology, Islamabad, ***Pakistan*** Research Productivity Award [2006, 2007, 2011, 2013, 2014, 2015, 2016-discontinued later]

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

American Society of Human Genetics, **USA** Developing Countries award [2017]

PROFESSIONAL ASSOCIATIONS

Member, *American Society of Human Genetics,* ***USA*** [2002 to date]

Life Member, *Pakistan Society for Biochemistry and Molecular Biology,* ***Pakistan*** [2010 to date]

Member, *Organization for women in Science for the Developing World (OWSD), Trieste,* ***Italy*** [2019 to date]

TECHNICAL SKILLS

Skilled in human linkage analyses, gene mapping by microsatellites and SNPs, gene identification by positional candidate gene and massively parallel sequencing analyses, Molecular Biology techniques, recombinant DNA technology, recombinant protein expression and isolation. Familiar with transgenics, mice breeding and evaluation of hearing of mice by Auditory Brainstem Response. Proficient in many aspects of cell culture and immunocytochemistry.

PERSONAL STATEMENT

My passion in science is driven by a deep desire to enhance my knowledge. I have a great interest in identification of variants which cause inherited genetic disorders in humans. I was the first scientist in 1996 to initiate systematic genetic characterizations of recessive deafness in Pakistan as part of my PhD studies, which I extended during my postdoctoral research in USA. Back in Pakistan as an independent researcher, I have focused my work to recessively inherited, moderate to severe hearing loss. This shift from profound deafness enabled discovery of three new genes, variants of which cause progressive or moderate to severe hearing loss. It also delineated aetiology of nonsyndromic recessively inherited moderate to severe deafness and pointed to a major role of modifiers which can reduce severity of hearing loss. I have obtained training with the technical and analytical aspects of massively parallel sequencing and have applied it successfully for gene discovery in other disorders as well. As a teacher, I love to instruct students about latest findings in Genomics and help them with both their educational and personal problems.

RESEARCH OVERVIEW

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

1. Genetic characterization of **moderate to severe hearing loss**: This study has so far established genetic underpinnings of hearing loss in 130 consanguineous families. It has identified founder variants, pinpointed a major role of modifiers in this disorder and has also revealed a role of three previously undiscovered genes in hearing loss or its syndromes. (19 publications, others in preparation)

2. Molecular characterization of short stature and **skeletal dysplasias**: This ongoing research has revealed the cause of short stature or skeletal dysplasias in 50 families. Variants of four genes were established for the first time as cause of recessively inherited skeletal dysplasias. (10 publications, others submitted and in preparation)

3. Studies on molecular basis of **movement disorders:** Among the most significant findings in this ongoing project are the discovery of *ABCA2* variants related to ataxia in humans and a helpful marker for its diagnosis as a clinical indicator. (12 publications, others submitted and in preparation)

4. Genetics of **neurodevelopmental disorders** including epilepsy: In this relatively newer project, *TTC5* variants were established, while work on others is ongoing. (1 publication, others in preparation)

5. Molecular characterization of **psychotic disorders**: This paradigm shifting research has so far identified three genes which are candidates for causing psychosis in patients and could delineate new avenues for treatment. (2 manuscripts under review, others in preparation)

Research and postdoctoral fellowship *Laboratory of Molecular Genetics, National Institute on Deafness and other Communication Disorders (NIDCD), National Institutes of Health (NIH),* ***USA***. [9th April 2001 to 31st July 2005] Mentors: Dr. Edward R Wilcox, Dr. Thomas B Friedman.

1. Cloning of **deafness causing** genes: Four novel genes were identified for profound deafness while genotype-phenotype correlations were revealed for three known deafness genes. [7 publications]

2. Transgenic **mouse model** for hearing loss: A knock-in mouse model was successfully generated and partially characterized as a model for *DFNA28*, a human disorder characterized by progressive hearing loss. [Manuscript under preparation]

3. Functional studies for protein **localizations** and **protein interactions***:* Epitopes against GRHL2, WHRN and MYO3A were produced in bacteria and injected into rabbits for antibody generation. Antisera were affinity purified and for GRHL2 were validated by Western blotting and immunostaining of eukaryotic cell lines transfected with GFP-*Grhl2*. Expression pattern of *Grhl2* in mice was also explored using RT-PCR experiments on mRNA extracted from various mouse tissues and by light and confocal microscopy on mouse inner ear sections and whole mount organ of Corti using specific antisera against GRHL2. Antisera against WHRN helped in identification of WHRN interaction with MYO15A. [1 publication, second in progress]

Research for Ph.D *CEMB, University of the Punjab,* ***Pakistan***, (1996 –2001), LMG, NIDCD, NIH, **USA**, [November 1998-April 1999] Supervisor: Dr. Sheikh Riazuddin, Mentor: Dr. Edward R Wilcox.

1. Research on **profound deafness**: Samples were ascertained from 37 families and characterized by linkage analyses and Sanger sequencing. [3 publications]

CURRENT RESEARCH GRANTS

2020-2022 National Institute on Mental Health, National Institutes of Health, USA, R21 MH120692-01A1, $340,178 (Joint Principal Investigator with Dr. Jose Pardo, University of Minnesota, USA). Mendelian Variants Associated with Psychosis

2021-2024 Pakistan Science Foundation, Pakistan, Grant PSF/Res/P-PU/Biotech (239) Rs. 20,00000 (Principal Investigator) Gene discovery for skeletal dysplasia, protein localization and in vitro variant pathogenic analyses

PREVIOUS RESEARCH GRANTS

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 3288, Rs. 3,592,600 (Principal Investigator) Genetics of recessively inherited stable or progressive hearing loss in Pakistan

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

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

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 US$21,000 (Principal Investigator) Genetics of dwarfism and skeletal dysplasia

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

RESEARCH PROFILE ID and URL

ORCID 0000-0002-1912-0235
Publications https://www.ncbi.nlm.nih.gov/myncbi/collections/mybibliography/
Webpage http://faculty.sadaf-naz.pu.edu.pk/

SUPERVISION OF RESEARCH STUDENTS

12 PhD completed and 5 in progress, 17 MPhil completed and 2 in progress

COMPLETE LIST of PUBLICATIONS (Impact factor 308.34)

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** (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#. [*corresponding author, #equal contribution] (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 [*corresponding author, #equal contribution] (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*** [*corresponding authors] (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* [*equal contribution] (2018). CDC14A phosphatase is essential for hearing and male fertility in mouse and human. ***Hum Mol Genet*** 27:780-798.

39. Ain NU, 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* [*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. Ain NU, Iqbal M, Valta H, Emerling CA, Ahmed S, Makitie O*, **Naz S*** [*corresponding authors] (2019). Novel variants in natriuretic peptide receptor 2 in unrelated patients with acromesomelic dysplasia type maroteaux. ***Eur J Med Genet*** 62: (103554) 1-7.

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

45. Aslam F, **Naz S** (2019). Ataxia and dysarthria due to an ABCA2 variant: Extension of the phenotypic spectrum. ***Park Rel Dis*** 64:328-331.

46. Tariq H, Butt JU, Houlden H*, **Naz S*** [*corresponding authors] (2019). Are some *C19orf12* variants monoallelic for neurological disorders? ***Park Rel Dis*** 65:267-269.

47. ***Naz S**, *Friedman TB [*corresponding authors] (2020). Growth factor and receptor malfunctions associated with human genetic deafness ***Clin Genet*** 97:138–155.

48. Bashir R, Irfan B, Khalid M, Naz S, Saleem F Nouman U, **Naz S** (2020). Association of hepatocyte growth factor (HGF) gene polymorphisms with primary angle closure glaucoma from Lahore, Pakistani population. ***J Pak Med Assoc*** 70:208-212.

49. Shaukat M, Ishaq M, Muhammad N, **Naz S** (2020). RIN2 and BBS7 variants as cause of a coincidental syndrome. ***Eur J Med Genet*** 63: (103755) 1-4.

50. Yasin S, Mustafa S, Ayesha A, Latif M, Hassan M, Faisal M, Makitie O, Iqbal F*, **Naz S*** [*corresponding authors] (2020). A novel homozygous missense variant in MATN3 causes spondylo-epimetaphyseal dysplasia Matrilin 3 type in a consanguineous family. ***Eur J Med Genet*** 63: (103958) 1-5.

51. Arif B, Rasheed A, Kumar KR, Fatima A, Ali G, Wohler E, Sobriera N, Lohmann K, **Naz S** (2020). A novel homozygous KY variant causing a complex neurological disorder. ***Eur J Med Genet*** 63: (104031) 1-4.

52. Ramzan M, Bashir R, Salman M, Mujtaba G, Sobreira N, Witmer D, Baylor-Hopkins Center for Mendelian Genomics, **Naz S** (2020). Spectrum of genetic variants in moderate to severe sporadic hearing loss in Pakistan. ***Sci Rep*** 10: 11902 1-7

53. Yasin S, Makitie O, **Naz S** (2021). Spondylocarpotarsal synostosis syndrome due to a novel loss of function *FLNB* variant: A case report. ***BMC Muscoskel Disord*** 22:31 1-6.

54. Rasheed A, Gumus E, Zaki MS, Johnson K, Manzoor H, LaForce G, Ross D, McEvoy-Venneri J, Stanley V, Lee S, Virani A, Ben-Omran T, Gleeson JG, **Naz S***, Schaffer AE* [*corresponding authors] (2021). Bi-allelic *TTC5* variants cause delayed developmental milestones and intellectual disability. ***J Med Genet*** 58:237-246.

55. Ain NU*, Muhammad N, Dianatpour M, Baroncelli M, Iqbal M, Fard MAF, Bukhari I, Ahmed S, Hajipour M, Tabatabaie Z, Foroutan H, Nilsson O, Faghihi MA, Makitie O*, **Naz S*** [*corresponding authors] (2021). Biallelic TMEM251 variants in patients with severe skeletal dysplasia and extreme short stature. ***Hum Mutat*** 42:89-101.

56. Ain NU, Baroncelli M, Costantini A, Ishaq T, Taylan F, Nilsson O, Makitie O*, **Naz S*** [*corresponding authors] (2021). Novel form of rhizomelic skeletal dysplasia associated with a homozygous variant in *GNPNAT1*. ***J Med Genet*** 58:351–356.

57. Tariq H*, Tariq I, Bourinaris T, Houlden H*, **Naz S*** [*corresponding authors] (2021). Some pathogenic *SETX* variants are partially conserved during evolution. ***Gene*** 771**:**145360 1-5.

58. Ain NU, Fatima Z, ***Naz S**, *Makitie, O [*corresponding authors] (2021). *RAB33B* and *PCNT* variants in two Pakistani families with skeletal dysplasia and short stature. ***BMC Muscoskel Disord*** 22:630 1-6.

59. Ramzan M, Philippe C, Belyantseva IA, Nakano Y, Fenollar-Ferrer C, Tona R, Yousaf R, Basheer R, Imtiaz A, Faridi R, Munir Z, Idrees H, Salman M, Nambot S, Vitobello A, Kartti S, Zarrik O, Witmer DP, Sobreria N, Ibrahimi A, Banfi B, Moutton S, Friedman TB*, **Naz S*** [*corresponding authors] (2021).Variants of human *CLDN9* cause mild to profound hearing loss. ***Hum Mutat*** 42:1321-1335.

60. Muhammad N, Yasin S, Fatima Z, Ain NU, Faizan M, **Naz S** (2021) The c.1138G>A variant of fibroblast growth factor receptor 3 is a common cause of achondroplasia in Pakistan ***Pak J Zool*** 53: 2519-2521.

61. **Naz S** (2021). Molecular genetic landscape of hereditary hearing loss in Pakistan. ***Hum Genet*** doi: 10.1007/s00439-021-02320-0

62. Faridi R, Rea A, Fenollar-Ferrer C, O'Keefe RT, Gu S, Munir Z, Khan AA, Riazuddin S, Hoa M, **Naz S**, Newman WG, Friedman TB (2021). New insights into Perrault syndrome, a clinically and genetically heterogeneous disorder. ***Hum Genet*** doi.org/10.1007/s00439-021-02319-7

LINK TO PUBLICATIONS' ABSTRACTS

https://www.ncbi.nlm.nih.gov/myncbi/collections/mybibliography/

EDITED BOOK WITH CHAPTER

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

CONFERENCES AND SEMINARS

22 abstracts published in national and international proceedings [3 won ASHG developing Countries Awards]