CURRICULUM VITAE

Name: **SHAHEEN N. KHAN**
(Maiden Name: Shaheen S. Siddiqui)

Designation: Professor

Postal Address: Office: Centre of Excellence in Molecular Biology, 87-West Canal Bank Road, Thokar Niaz Baig, Lahore-53700, Pakistan.
(Tel: 042-5293141-46 ext 119)
Email: snkhan50@yahoo.com

Res: **77**-J, EME Housing Society, Multan Road, Lahore, Pakistan.
(Tel:042-37512077 & 37513192)
Email: snkhan50@yahoo.com

Date of Birth: August 18, 1950
Nationality: Pakistani
Marital Status: Married, 2 Children

PRESENT POSITION:

July 2006 - todate: Professor, Centre of Excellence in Molecular Biology, 87-West Canal Bank Road, Thokar Niaz Baig, Lahore-53700, Pakistan.

ADMINISTRATION:

- Incharge MoST Section of the Centre. —– Nov. 2009
- Member Board of Studies. —– Till todate
- Member Advisory Committee of the Centre. —– Till todate
- Member Purchase Committee
- Member Journal Club

ACADEMIC QUALIFICATIONS:

1995 - 00: Ph.D in Molecular Biology, University of the Punjab, Lahore.
Thesis Title: *Molecular Characterization of β-thalassemia in Pakistan.*

1969 - 70: M.Sc in Zoology (Physiology), University of Karachi.
Thesis Title: *Morphogenesis of erythrocytes in Rana Cyanophlyctis*.

1967 - 68: B.Sc., University of Karachi.

1963 – 66: SSC & HSC, Karachi Board.

INTERNATIONAL COURSES / WORKSHOPS ATTENDED:

- Autism Diagnostic Observation Schedule (ADOS) workshop conducted by Dr. Catherine Lord at Los Angeles, California. Jan. 24-25, 2002.
- Training in Molecular Diagnosis of α and β-thalassemia at the Institute of Clinical and Biological Research, Microcitemie Hospital, University of Cagliari, Sardinia, Italy. Sept. 1998- Aug. 1999.
- Microbiology course at the Toronto Institute of Medical Technology, Toronto, Canada. Feb. 1981.
- Programming Language "Cobol", and Introduction to Data Processing Computer Courses from George Brown College of Applied Arts and Technology, Toronto, Canada. In June & January 1980.

NATIONAL COURSES / WORKSHOPS ATTENDED:

- Occupational Health and Safety course conducted by M/S Bench Mark Certification, Australia at National University of Science and Technology (NUST) Aug. 3, 2000.
- Understanding ISO 9000: version 2000 course conducted by M/S Bench Mark Certification, Australia at National University of Science and Technology (NUST). Aug. 2, 2000.
- Molecular Biology Courses at the National Centre of Excellence in Molecular Biology, University of the Punjab, Lahore, Pakistan. 1995.
- Computer Courses in DOS, HG & WORD at the Centre of Excellence in Molecular Biology, Lahore, Pakistan. 1992.

RESEARCH EXPERIENCE:

April 2003 – todate

- Studies on the regeneration potential of Stem cells.
- Studies on the syndromic and non syndromic recessive deafness.
- DNA typing for Forensic uses (till 2009).

June 2000 - March 2003:

- Studies on the syndromic and non syndromic recessive deafness.
- Studies on polycystic kidney disease and autism.
- Studies on α and β-thalassemia.

July 1990 - May 2000:

- Studies on α and β-thalassemia.
- Studies on the syndromic and non syndromic recessive deafness.
- Studies on the genetic basis of stuttering.

1987 - 1990:

- Screening for Inborn errors of metabolism and deficiency of Pseudocholinesterase.

1981 - 1986:

- Overall supervision of clinical pathology laboratory, including Hematology, Biochemistry and Microbiology.

1974 - 1981:

- Randomized clinical trials for Breast cancer, Hodgkin's disease and Leukemia.

EMPLOYMENT RECORD:

June 10, 2009 - Oct 05, 2009	Acting Director Centre of Excellence in Molecular Biology , 87-West Canal Bank Road, Thokar Niaz Baig, Lahore-53700, Pakistan.
July 2006 - todate	Professor Centre of Excellence in Molecular Biology , 87-West Canal Bank Road, Thokar Niaz Baig, Lahore-53700, Pakistan.
April 2003 - June 2006	Principal Research Officer Centre for Applied Molecular Biology,
June 2000 - March 2003	Senior Research Officer Centre for Applied Molecular Biology.
July 1990 - May 2000	Research Officer Centre for Applied Molecular Biology.
1987-1990	Research Officer National Health Research Complex, Shaikh Zayed Hospital, Lahore-53700, Pakistan.
1981-1986	Supervisor M.D. Pathology Laboratories, Sharfabad, Karachi-5 Pakistan.
1974-1981	Research Assistant Ontario Cancer Research Institute, Princess Margaret Hospital, Toronto, Ontario, Canada.

SCHOLARSHIPS AND AWARDS:

2007	**Civil Award "Pride of Performance"**
2004	**Presidential Award "Izaz-e-Kamal"**
2004	**Mohtarma Fatima Jinnah Gold Medal Award**
1997	**First prize** for Scientific data recording at the National Centre of Excellence in Molecular Biology given by Rich Roberts (Nobel Laureates)
1965 - 70:	**Merit Scholarship** from F.Sc to M.Sc and **First position in M.Sc**

PRINCIPAL OR CO-INVESTIGATOR IN RESEARCH PROJECTS:

- Identification of the genetic factors responsible for the variable phenotype of thalassemia in Pakistan sponsored by University Grants Commission (UGC).
- Study of non syndromic recessive hereditary hearing impairment (UGC)
- Screening for hearing impairment in rural Sindh (UGC)
- Genetic and Molecular Basis of Recessive Deafness sponsored by Higher Education Commission of Pakistan.
- Received Research Grant of Rs 1.0 million from PSF for productive scientists.
- Studies on the genetic & molecular basis of hearing & vision impairment sponsored by WHO-COMSTECH [No. RAB&GH 06-07-24].
- Molecular Genetics of Gaucher's Disease in Pakistan sponsored by WHO-COMSTECH [No. RAB&GH 10-11/05].

PRINCIPAL INVESTIGATOR IN DEVELOPMENT PROJECTS:

- Molecular basis of recessive hereditary hearing impairment in Balochistan and NWFP PC-I sponsored by MoST.

IMPORTANT CONTRIBUTIONS / ACHIEVEMENTS:

<u>Human Resources Development:</u>

- Trained 49 M.Phil students. 29 students have completed their Ph D. two Ph.D students have submitted their thesis & six Ph.D students are at various stages of completing their research.
- Trained two lecturers form BUITMS University, Quetta under the HEC training programme for technical/scientific staff & researchers.
- Trained DCP students of Shaikh Zayed Hospital and students from other institutes for special biochemical tests

Laboratory Research:

- Identified five rare inherited metabolic disorders.
- Molecularly characterized 22 β-thalassemia mutations having different frequencies among the major ethnic groups in Pakistan including three rare (unreported so far in Asian population) mutations. Identified three β-globin chain variants HbS, HbE & HbD Punjab. Identified three α-thalassemia mutations with significant ethnic differences between Pashtoon, Balochi and Sindhi along with a rare α_2 globin chain variant Hb Sallanches. Developed procedures for prenatal diagnosis of β-thalassemia.
- Identified factors like α-gene deletions and Xmn1 polymorphism responsible for the milder phenotype in thalassemia intermedia patients.
- Identified novel mutations causing Pendred syndrome (deafness with goiter) and Usher syndrome (deafness with night vision problem) & non syndromic deafness.
- Identified 21 new recessive deafness/ vision loci/ genes.
- Demonstrated the regenerative and repair abilities of bone-marrow stromal cells in animal models of diabetes, infarcted heart, fibrotic liver and Ischemic kidney.
- Ongoing collaboration with LRBT hospital for the repair of damaged eye by limbal stem cell for the restoration of vision.
- Construction of bioengineered skin and its repair potential is already demonstrated in rat burn model. Now in collaboration with AIMC/Jinnah Hospital the generation of plasma based bioengineered human skin substitute containing dermal and epidermal cells is in process.

Application of Laboratory Research:

- Procedures developed for prenatal diagnosis of β-thalassemia are being used by gynecologists.
- Procedures developed for limbal stem cell culture to reconstruct damaged ocular surface. In collaboration with LRBT ophthalmologists the transplanted cells improved patients vision.

SCIENTIFIC SYMPOSIA/MEETINGS ATTENDED:

- International Symposium on "Biotechnology Applications in New Emerging Fields". December 21-23, 2010 at Allama Iqbal Medical College, Lahore. Sponsored by ISESCO, AIMBN, CEMB and AIMC.
- Symposium on "Future Trends in Molecular Biological Research & Its Applications in Agriculture & Health". March 25-27, 2009.
- Mini-Workshop on CEMB Vision 2015 presented Progress in Medical Molecular Biology at National Centre of Excellence in Molecular Biology. August 16, 2008.
- A mini symposium on New Vistas of Plant & Medical Molecular Biology Research at National Centre of Excellence in Molecular Biology, Lahore. July 21, 2008.
- Second National Conference on Health Biotechnology at Pakistan Academy of Sciences. May 27-28, 2008.
- Symposium on Molecular Biological Research "Repair Potential of Stem Cell in damaged heart". March 26, 2008.
- National Bioforum 2008 at National Centre of Excellence in Molecular Biology. March 24-28, 2008.

- International symposium on Stem Cells jointly organized by NCEMB, HEC & COMSTECH Jan. 8-20, 2007.
- DNA for Justice a mini workshop organized twin Centres of Molecular Biology, University of the Punjab, Lahore, Dec.16, 2006.
- Lahore Bioforum, jointly organized by twin Centres of Molecular Biology, University of the Punjab, Lahore, Pakistan & PISELL CONSULTING S.L. Barcelona, Spain. March 13-15, 2006.
- BINASIA-Pakistan National workshop, organized by National Centre of Excellence in Molecular Biology, Lahore. March 11-12, 2006.
- 4th International Symposium on Genetic Engineering and Biotechnology organized by Centre for Molecular Genetics University of Karachi, Dec. 4-8, 2005.
- 18th FAOBMB Symposium on Genomics and Proteomics in Health and Agriculture organized by School of Biological Sciences, University of the Punjab, Nov. 20-23, 2005.
- First National Conference on Health Biotechnology organized by National Commission on Biotechnology at University of Health Sciences, Lahore Jan. 27-28, 2005.
- A Symposium/Workshop on the Application of Molecular Biological Research in Agriculture and Health at the National Centre of Excellence in Molecular Biology, Lahore. March 27-31, 2004.
- 17th International Biennial Conference of Pakistan Association & Scientific Conference of Paediatric Association of SAARC Countries organized by Pakistan Paediatric Association Punjab at Pearl Continental, Lahore. Feb.19-22, 2004.
- A Seminar on Development of Research Grant Proposals Jointly Organized by Higher Education Commission and Punjab University at the National Centre of Excellence in Molecular Biology, Lahore. Oct. 21, 2003
- A Seminar on Autism. Karachi. Sep.18, 2002.
- A workshop on the Autism Diagnostic Observation Schedule (ADOS) Los Angeles CA USA. Jan 24-25, 2002.
- International Atomic Energy Agency (IAEA) Research Coordination Meeting on Genotype/Phenotype in Genetic Disorders. Tunis, Tunisia. February 28 to March 2, 2001.
- Third International Biennial Conference of Pakistan Society for Microbiology, Jointly organized by Pakistan Society for Microbiology and the National Centre of Excellence in Molecular Biology at Pearl Continental, Lahore. March 28-30, 2000.
- A mini Symposium on Molecular Biological Approaches to Diagnosis and Prevention of Genetic Diseases, Jointly organized by Lahore Chapter of Pakistan Academy of Sciences and the National Centre of Excellence in Molecular Biology, Lahore, Pakistan. Feb. 4, 2000.
- International Atomic Energy Agency (IAEA) Research Coordination Meeting on Genotype/Phenotype in Genetic Disorders. Cape Town, South Africa. March 15-19, 1999.
- International Atomic Energy Agency (IAEA) Research Coordination Meeting on Diagnosis of Genetic Disorders using Radio-Nuclide Based Molecular Methods. Nicosia, Cyprus. April 6-9, 1998.

- Fifth International Symposium-workshop on the Application of Molecular Biology Research in Agriculture, Health and Environment, organized and held at the National Centre of Excellence in Molecular Biology, Lahore, Pakistan. Oct. 14-15, 1997.
- Inaugural International Conference on Genetics, organized by Pakistan Genetical Society at the National Agricultural Research Centre (NARC), Islamabad, Pakistan. Nov. 26-28, 1996.
- Fourth International Symposium-workshop on the Application of Molecular Biology Research in Agriculture, Health and Environment, organized and held at the National Centre of Excellence in Molecular Biology, Lahore, Pakistan. April 08-11, 1995.
- Third International Symposium-workshop on Application of DNA Technology to Agriculture and Health, organized and held at the National Centre of Excellence in Molecular Biology, Lahore, Pakistan. Oct. 24-29, 1992.
- SAARC Symposium-workshop on Biological Control of Agriculturally Important Plant pests organized and held at National Centre of Excellence in Molecular Biology, Lahore, Pakistan. Dec. 16-18, 1991.

ABSTRACTS:

1. **Shaheen N. Khan.** Stem Cell as a Source of Regenerative Medicine organized by ISESCO, AIMBN, CEMB and AIMC. December 21-23, 2010 at Allama Iqbal Medical College, Lahore.

2. **Shaheen N. Khan.** Overview of Medical Molecular Biology Research at CEMB at Symposium on Future Trends in Molecular Biological Research and Its Applications in Agriculture and Health organized by Centre of Excellence in Molecular Biology, March 25-28, 2009.

3. **Shaheen N. Khan.** Genetic Basis of Retinitis Pigmentosa in consanguineous Pakistani families in Second National Conference on Health Biotechnology organized by Pakistan Academy of Sciences, May 27-28, 2008.

4. **Shaheen N.Khan.** Repair Potential of Stem Cell in damaged Heart at Symposium on Molecular Biological Research organized by Centre of Excellence in Molecular Biology, March 26, 2008.

5. **Shaheen N. Khan**, Sabiha Nazli, Jamil Ahmed, Khushnooda Ramzan, Rehan, Sadiq, Saima Riazuddin, Zubair M. Ahmed & Sheikh Riazuddin. Genetic and molecular basis of hearing impairment at 4th International Symposium on Genetic Engineering and Biotechnology organized by Centre for Molecular Genetics University of Karachi, Dec. 4-8, 2005.

6. **Shaheen N. Khan,** Sabiha Nazli, Khushnooda Ramzan, Rehan Sadiq, Jamiil Ahmad, Saba Tasneem, Saeeda Kalsoom & Sheikh Riazuddin. Genetic studies of hereditary hearing impairment at 18th FAOBMB Symposium on Genomics and Proteomics in Health and Agriculture organized by School of Biological Sciences, University of the Punjab, Nov. 20-23, 2005.

7. **Shaheen N. Khan**, Sabiha Nazli, Rehan, Sadiq, Khushnooda Ramzan, Jamil Ahmed, Saima Riazuddin, Zubair M. Ahmed & Sheikh Riazuddin. Molecular studies of hereditary hearing impairment at the First National Conference on Health Biotechnology organized by National Commission on Biotechnology at University of Health Sciences, Lahore Jan. 27-28, 2005.

8. **Khan, S.N**., Ramzan, K., Sadiq, R., Ahmed, J., Nazli, S and Riazuddin, S. Genetics of hereditary hearing impairment in Pakistan at 17th International Biennial Conference of Pakistan Association & Scientific Conference of Paediatric Association of SAARC Countries organized by Pakistan Paediatric Association Punjab. Feb.19-22, 2004.

9. Ahmad J., Ahmed ZM., Riazuddin S., Nazli S., Shaikh RS., Ramzan K., Awais M.,
Khan SN., Riazuddin S. Genotype-phenotype correlation in DFNB23 and USH1F at 2nd International Symposium on Biotechnology organized by Institute of Biotechnology & Genetic Engineering University of Sindh, Jamshoro and Nuclear Institute of Agriculture (NIA) Tando Jam, Pakistan. Jan. 19-21, 2004.

10. Aslam, M; **Khan, S.N** and Riazuddin, S. Establishment of pre-symptomatic DNA based early diagnostic procedure for Autosomal Dominant Polycystic Kidney Disease (ADPKD) in Pakistan. 7th International conference of Pakistan Society For Biochemistry and Molecular Biology on "Trends in Biochemistry and Molecular Biology", Lahore. April 2-5,2003.

11. **Khan, S. N**. and Riazuddin, S. Molecular genetics of α-thalassemia and Thalassemia intermediate in Pakistan. International Atomic Energy Agency (IAEA) Research Coordination Meeting on Genotype/Phenotype in Genetic Disorders. Tunis, Tunisia. February 28 to March 2, 2001.

12. **Khan, S. N**. and Riazuddin, S. Molecular genetics of thalassemia in Pakistan. Third International Biennial Conference of Pakistan Society for Microbiology, Lahore, Pakistan. March 28-30, 2000.

13. **Khan, S. N**. and Riazuddin, S. Molecular diagnosis and prevention of α and β-thalassemia in Pakistan. A mini Symposium on Molecular Biological Approaches to Diagnosis and Prevention of Genetic Diseases, Jointly organized by Lahore Chapter of Pakistan Academy of Sciences and the National Centre of Excellence in Molecular Biology, Lahore, Pakistan. Feb. 4, 2000.

14. **Khan, S. N.** and Riazuddin, S. Identification of α-thalassemia in the different ethnic groups of Pakistan. International Atomic Energy Agency (IAEA) Research Coordination Meeting on Genotype/Phenotype in Genetic Disorders. Cape Town, South Africa. March 15-19, 1999.

15. **Khan, S. N**. and Riazuddin, S. Molecular characterization and prenatal diagnosis for the prevention of β-thalassemia in Pakistan. International Atomic Energy Agency (IAEA) Research Coordination Meeting on Diagnosis of Genetic Disorders using Radio-Nuclide Based Molecular Methods. Nicosia, Cyprus. April 6-9, 1998.

PROFESSIONAL AND ACADEMIC RECORD:

I started my research carrier in 1970 with M.Sc thesis work on the morphogenesis of erythrocytes in Rana cyanophlyctis, comparing the changes in normal with thyroxin and radiation induced metamorphosis. As a result of excellent work, I published six papers
(1-6) and got first position in M.Sc. I received merit scholarships from Matric to M.Sc. From 1974 to 1981, I worked on breast cancer and lymphomas at the Ontario Cancer Research Institute, Toronto, Canada. After passing a course in Microbiology at Toronto Institute of Medical Technology, I joined a clinical pathology laboratory in Karachi in 1981. In 1987 I joined National Health Research Complex, Shaikh Zayed Hospital, Lahore where I worked on inherited metabolic disorders and published six papers (7-12).

In 1990, I joined CAMB to start a new career in molecular biology. My research interests were focused on determining the molecular and genetic basis of inherited disorders. The first candidate was **thalassemia**, which is the most prevalent hemoglobin disorder in Pakistan. I have published in Pakistan the first paper on the molecular genetic diagnosis of β-thalassemia (13). I have molecularly characterized α and β-thalassemia and reported
22 β-thalassemia mutations including three rare (unreported so far in Asian population) (14) and three α-thalassemia mutations with variable frequencies among six major ethnic/linguistic groups in Pakistan (25). In addition three β-globin chain hemoglobin variants and one rare α2 -globin chain variant Hb Sallanches (15 & 16) were also identified. I have reported molecular factors like α-gene deletions and Xmn1 polymorphism causing milder phenotypes in thalassemia patients (25). Participated in an IAEA sponsored multi-centre study to define further the molecular basis of β-thalassemia in Thailand, Pakistan, Sri Lanka, Mauritius, Syria, Cyprus and India and to develop a simple molecular diagnostic strategy by ARMS-PCR (18). On the basis of above results, developed procedures for prenatal diagnosis. The above success and encouragement from Professor S. Riazuddin, other genetic diseases like polycystic kidney disease and hereditary hearing and vision impairment were also included in the programme.

Hereditary Hearing & Vision Impairment As member of Dr. Riazuddin's group, we have identified 35 reported non syndromic recessive deafness loci besides Usher and Pendred syndromes in Pakistani population. Seven reported deafness & vision impairment genes were sequenced in a number of families and new or reported mutations have been identified (20, 22, 24, 38, 40, 46, 48, 49 and 52). Identified 11 new recessive deafness loci namely DFNB36 (27), DFNB37 (26), DFNB48 (29), DFNB49 (28), DFNB51 (30), DFNB56 (unpublished), DFNB67 (32), DFNB63 (34), DFNB72 (36), USH1H (39) & DFNB79 (45) and 16 new genes namely CDH23 (17), PCDH15 (19), TMC1 (21), MYO6 (26), ESPN (27), TRIOBP (31), TMHS (32), Tric (33), RDX (35), LTBP2 (41), LTBP2 (42), HGF (43), C9orf75(45), MSRB3 (54), ILDR1 (55) and GIPC3 (57).

Stem Cell & Regenerative Medicine: As a consequence of input during the last six years two main projects have been started. a) Corneal stem cell therapy for visual deficiencies. The main objective of this programme is to regenerate damaged corneal epithelium using limbal stem cells. We are using amniotic membrane to transplant cultured autologous limbal stem cells into the defective cornea to reverse visual impairment. b) Repair and regenerative potential of stem cells. In this programme mouse models has been developed to demonstrate the regenerative and repair abilities of bone-

marrow stromal cells in Diabetes, infarcted heart, fibrotic liver and ischemic kidney. Diabetic mouse models have also been used to study the effect of diabetes on heart and kidney.

My research work has been published in 65 papers with an impact factor of 304.783. I have Participated/attended 29 national/ international scientific symposia/meetings to present my research findings. I am Principal/ co- Investigator of several research projects funded by national and international funding agencies. Forty nine M.Phil & 29 Ph.D. students have completed their research in genetic diseases and stem cell and regenerative medicine programme. Three Ph.D students have submitted their thesis & six Ph.D students are at different stages of completing their research. I have received **Mohtarma Fatima Jinnah Gold Medal** in March 2004, Presidential award **Izaz-e-Kamal** 2004 and Civil award **Pride of Performance** 2007. I have received **Research Productivity Allowance** from 2003-2008.

LIST OF PUBLICATIONS

		Impact Factor
1.	Ahmed M., **Siddiqui, S.S**. 1976. The relationship between capillaryarea and red cell eccentricity at metamorphosis in Rana cyanophlyctis. Zool. Anz., Jena 197(5/6), S.332-334.	N/A
2.	Ahmed M., **Siddiqui, S.S**. 1976. The relationship between the red cell forms and surface area during thyroxin induced metamorphosis in Rana cyanophlyctis. Acta Physiol Latinoam., 26: 447-452.	N/A
3.	Ahmed M., **Siddiqui, S.S.** 1977. The relationship between the red cell forms and surface area during normal metamorphosis in Rana cyanophlyctis. Agr. Pak. 28(3): 203-211.	N/A
4.	Ahmed M., **Siddiqui, S.S**. 1977. The relationship between capillary area and red cell eccentricity at radiation induced metamorphosis in Rana cyanophlyctis. Zool. Anz., Jena 198:1/2, S 103-108.	N/A
5.	Ahmed M., **Siddiqui, S.S**. 1979. Physiology of circulation in the architecture of anura. Z. mikrosk-anat. Forsch., Leipzig. 93: 1,S. 161-168.	N/A
6.	Ahmed M., **Siddiqui, S.S**. 1979. Blood cell surface area at radiation induced metamorphosis in Rana cyanophlyctis tadpoles. Natural Sciences 1: 3, 129-136.	N/A
7.	Rauf, A., **Khan, S. N**., Khan, N.R. 1988. A case of low Pseudocholinesterase level. Pakistan Journal of Medical Research. 27: 1, 61-63.	N/A
8.	Khan, N.R., Maqbool, S., **Khan, S.N**., Mohydin, M.A.Z. 1988. A case of Mucopolysaccharidoses, Hurlers's disease. Pakistan Journal of Medical Research. 27:2, 137-142.	N/A
9.	Khan, N.R., Maqbool, S., **Khan, S.N**., Mohydin, M.A.Z. 1988. A case of Homocystinuria. Pakistan Journal of Medical Research. 27:4, 306-308.	N/A
10.	Khan, N.R., **Khan, S.N**., Maqbool S. 1990. A case of Alkaptonuria. Pakistan Journal of Medical Research. 29:1, 51-52.	N/A
11.	Khan, N.R., **Khan S.N**. 1991. Serum Cholinesterase levels in a sample village population. Pakistan Journal of Medical Research.30:3, 143-146.	N/A
`		
12.	Khan, N.R., **Khan, S.N**., Maqbool S. 1993. Screening for Inherited Metabolic Disorders at Shaikh Zayed Hospital. Pakistan Journal of Medical Research.32:3, 180-184.	N/A
13.	**Khan**, **S.N.,** Zafar, A.U. and Riazuddin, S. 1995. Molecular Genetic Diagnosis of β-thalassemia in Pakistan. J. Pak. Med. Assoc. 45: 66-70.	N/A
14.	**Khan, S.N.** and Riazuddin ,S. 1998. Molecular characterization of β-thalassemia in Pakistan. Hemoglobin 22(4):333-345.	1.274

15. **Khan, S.N.,** Riazuddin, S. and Galanello, R. 2000. Identification of Three Rare β-thalassemia Mutations in the Pakistani Population. Hemoglobin 24(1):15-22. 1.274

16. **Khan, S.N.,** Butt F.I.; Riazuddin, S. and Galanello, R. 2000. A rare α2-globin chain variant Hb Sallanches in a Pakistani family with three homozygous patients. Hemoglobin 24(1):31-35. 1.274

17. Bork, J. M., Peters, L. M., Riazuddin, S., Bernstein, S.L., Ahmed, Z. M., Ness, S. L., Polomeno, R., Ramesh, A., Schloss, M., Srisailpathy, C. R. S., Wayne, S., Bellman, S., Desmukh, D., Ahmed, Z., **Khan, SN. N**., et al. 2001. Usher syndrome ID and non-syndromic recessive deafness DFNB12 are caused by allelic mutations of the novel cadherin-like gene CDH23. Am. J. Hum. Genet. 68: 26-37. 12.303

18. Old, J., **Khan, SN.**, Verma, I., Fucharoen, S., Kleanthous, M., Iaonnou, P., Kotea, N., Fisher, C., Riazuddin, S., Saxena, R., Winichagoon, P., Kyriacov, K., Quobaili, FA., and Khan, B. 2001. A multi-centre study in order to define further the molecular basis of β-thalassemia in Thailand, Pakistan, Sri Lanka, Mauritius, Syria and India and to develop a simple molecular diagnostic strategy by ARMS-PCR. Hemoglobin 25(4): 397-407. 1.274

19. Ahmed, Z. M., Riazuddin, S., Bernstein, S. L., Ahmad, Z., **Khan, SN.**, Griffith, A. J., Morell, R. J., Friendman, T. B., Riazuddin, S. and Wilcox, E. 2001. Mutations of Protocadherin Gene PCDH15 Cause Usher Syndrome Type IF. A. J. Hum. Gene. 69:25-34. 12.303

20. Liburd. N., Ghosh. M., Riazuddin, S., Naz, S. **Khan**, **SN.**, Ahmed, Z., Riazuddin, S., Liang, Y., Menon, PSN., Smith, T., Smith, ACM., Chen, KS., Lupski, JR., Wilcox, ER., Potocki, L and Friedman, TB. 2001. Novel mutations of MYO15A associated with profound deafness in consanguineous families and moderately-serve hearing loss in a patient with Smith Magenis syndrome Hum. Genet. 109: 535-541. 4.529

21. Kurima, K., Yang, Y., Riazuddin, S., Ahmed, Z., Naz, S., Mo, J., Makishima, T., Ghosh, M., Menon, P.S.N., Deshmukh, D., Oddoux, C., Ostrer, H., **Khan, SN.**, Riazuddin, S., Hampton, L.L., Battey, J. F. Jr., Wilcox, E.R., Friedman, T.B., Griffith, A.J. (2002). Dominant and recessive deafness caused by mutations of a novel gene TMC1, required for cochlear hair-cell function. Nature Genetics, 30: 277-284. 31.434

22. Astuto, LM., Bork, JM., Weston, MD., Askew, JW., Fields, RR., Orten, DJ., Ohliger, SJ., Riazuddin, S., Morell, RJ., **Khan, SN.,** Riazuddin, S., Kremer, H., Van Hauwe, P., Moller, CG., Cremers, CWRJ., Ayuso, C., Heckenlively, JR., Rohrschneider, K., Spandau, U., Greenberg, J., Ramesar, R., Reardon, W., Bitoun, P., Millan, J., Legge, R., Friedman, TB., Kimberling, WJ. 2002. CDH23 Mutation and Phenotype Heterogeneity: A Profile of 107 Diverse families with Usher Syndrome and Non-Syndromic Deafness. Am. J. Hum. Genet. 71: 262-275. 12.303

23.	Bork, J.M., Morell, R.J., **Khan, SN**, Riazuddin, S., Wilcox, E.R., Friedman, T.B., Griffith, A.J. 2002. Clinical Presentation of DFNB 12 and Usher Syndrome Type 1D. Adv. Otorhinolaryngol. 61, 145-152.	0.346
24.	Park, H. J., Shaukat.S., Liu, X. Z., Hahn, S., Naz, S., Ghosh, M., Kim, H. N., Moon, S. K., Abe, S., Kukamoto, K., Riazuddin, S., Kabra, M., Erdenetungalag, R., Radanaabazar, J., **Khan, SN**., Pandya, A., Usmi, SI., Nance, WE., Wilcox, ER., Riazuddin, S., and Griffith, AJ. 2003 Origin and frequencies of SLC26A4 mutations in East and South Asians: Global implications for the epidemiology of deafness. J Med. Genetics. 40: 242-246.	5.751
25.	**Khan S.N**., Hasan, F., Sollaino. C., Perseu, L., Riazuddin, S. 2003. Molecular characterization of α-thalassemia in Pakistan. Hemoglobin 27(3): 161-166.	1.274
26.	Ahmed, Z. M., Morell, R. J., Riazuddin, S., Gropman, A., Shaukat, S., Ahmad, M. M., Mohiddin, S. A., Fananapazir, L., Caruso, R. C., Husnain, T., **Khan, S. N.**, Riazuddin, S., Griffith, A. J., Friedman, T. B., Wilcox, E. R. 2003. Mutations of *MYO6* are associated with recessive deafness *DFNB37*;. Am. J. Hum. Genet. 72: 1315-1322.	12.303
27.	Naz S., Griffith A.J., Riazuddin S., Hampton L.L., Battey J.F., **Khan S.N.,** Riazuddin S., Wilcox E.R., and Friedman T. B., 2004. Mutations of ESPN cause Vestibular Dysfunction and Autosomal Recessive Deafness. J. Med. Genet. 41:591-595.	5.751
28.	Ramzan K., Shaikh R. S., Ahmad J., **Khan S N.,** Riazuddin S., Ahmed Z M., Friedman T B., Wilcox E R., Riazuddin S. 2005. A new locus for Nonsyndromic deafness DFNB49 maps to chromosome 5q12.3-q14.1. Hum Genet. 116: 17-22.	4.529
29.	Ahmad J., **Khan S N.,** Khan S. Y., Ramzan K., Riazuddin S., Ahmed Z M., Wilcox E R., Friedman T B., Riazuddin S. 2005. DFNB48, a new Nonsyndromic recessive deafness locus maps to chromosome 15q23-q25.1. Hum Genet. 116:407-412.	4.529
30.	Shaikh R. S., Ramzan K., Nazli, S., Sattar S., **Khan S N.,** Riazuddin S., Ahmed Z M., Friedman T B., Wilcox E R., Riazuddin S. 2005. A new locus for nonsyndromic deafness DFNB51 maps to chromosome 11p13-p12. Am. J. Med. Genet. A. 138A:392-395.	2.404
31.	Riazuddin S., **Khan S N.,** Ahmed Z M., Ghosh M., Caution K., Nazli S., Kabra M., Zafar AU., Chen K., Naz S., Antonellis A., Paban WJ., Green ED., Wilcox ER., Friedman PL., Morell RJ., Riazuddin S., Friedman TB. 2005. Mutations in TRIOBP, which encodes a putative cytoskeletal-organizing protein, are associated with nonsyndromic recessive deafness. Am. J. Hum. Genet. 78: 137–143.	12.303
32.	Shabbir MI, Ahmed ZM, Khan SY, Riazuddin S, Waryah AM, **Khan SN**, Camps RD, Ghosh M, Kabra M, Belyantseva IA, Friedman TB, Riazuddin S. 2006. Mutations of human TMHS cause recessively inherited non-syndromic hearing loss. J Med Genet. 43(8): 634-40.	5.751

33.	Riazuddin S, Ahmed ZM, Fanning AS, Lagziel A, Kitajiri S, Ramzan K, **Khan SN**, Chattaraj P, Friedman PL, Anderson JM, Belyantseva IA, Forge A, Riazuddin S, Friedman TB. 2006. Tricellulin Is a Tight-Junction Protein Necessary for Hearing. Am. J. Hum. Genet., 79:1040-1051.	12.303
34.	Khan SY, Riazuddin S, Tariq M, Anwar S, Shabbir MI, Riazuddin SA, **Khan SN**, Husnain T, Ahmed ZM, Friedman TB, Riazuddin S. 2007. Autosomal recessive nonsyndromic deafness locus DFNB63 at chromosome 11q13.2-q13.3. Hum Genet. 120 (6): 789-93.	4.529
35.	Khan SY, Ahmed ZM, Shabbir MI, Kitajiri S, Kalsoom S, Tasneem S, Shayiq S, Ramesh A, Srisailpathy S, **Khan SN**, Smith RJ, Riazuddin S, Friedman TB, Riazuddin S. 2007. Mutations of the RDX gene cause nonsyndromic hearing loss at the DFNB24 locus. Hum. Mutat, 28 (5): 417-23.	7.033
36.	Ain Q., Nazli S., Riazuddin S., Jaleel A.U., Riazuddin S.A., Zafar A.U., **Khan SN,** Husnain T., Griffith A.J, Ahmad Z.M. Friedman T.B, Riazuddin S. (2007). The autosomal recessive nonsyndromic deafness locus DFNB72 is located on chromosome 19p13.3. Hum Genet. 122(5): 445-50	4.529
37.	Verma I.C., Kleanthous M., Saxena R., Fucharoen S., Winichagoon P., Raizuddin S., **Khan S.N.,** Akbari M.T., Izadyar M., Kotea N., Old J.M., Ioannou P.A., and Khan B. (2007). Multicenter Study of the Molecular Basis of Thalassemia Intermedia in Different Ethnic Populations. Hemoglobin, 31 (4): 439-52.	1.274
38.	Riazuddin, S., Nazli, S., Ahmed, Z.M., Yang, Y., Zulfiqar, F., Shaikh, R.S., Zafar, A.U., **Khan, S.N.,** Sabar, M.F., Javid, F.T., Wilcox, E.R., Ekaterini, T., Boger, E.T., Sellers, J.R., Belyantseva, I.A., Riazuddin S. and Friedman T. (2007). Mutation Spectrum of MYO7A and Evaluation of a Novel Nonsyndromic Deafness DFNB2 Allele with Residual Function. Hum Mutat. 29(4): 502-11	7.033
39	Ahmed ZM, Riazuddin S, **Khan SN**, Friedman PL, Riazuddin S., Friedman T. (2008). USH1H, a novel locus for type I Ushersyndrome, Maps to chromosome 15q22-23. Clin Genet 75(1): 86-91	3.304
40	Firasat S., Riazuddin SA., **Khan SN.,** Riazuddin S. (2008). Novel CYP1B1 mutations in consanguineous Pakistani families with Primary congenital glaucoma. Mol. Vision, 14: 2002-2009.	2.464
41	Khan M., Manzoor S., Mohsin S., **Khan SN.,** Ahmed FJ. (2009). IGF-1 and G-CSF complement each other in BMSC migration towards infarcted myocardium in a novel in vitro model. Cell Biol. Int. 33(6):650-657	1.619
42	Ali M., Mckibbin M., Booth A., Parry DA., Jain P., Riazuddin SA., Hejtmancik JF., **Khan SN.,** Firasat S., Shires M., Gilmour DF., Towns K., Murphy AL., Azmanov D., Tournev I., Cherninkova S., Jafri H., Raashid Y., Toomes C., Craig J., Mackey DA., Kalaydjieva L., Riazuddin S., Inglehearn CF. (2009) Null mutations in LTBP2 cause primary congenital glaucoma. Am J Hum. Genet. 84 (5):664-671	12.303

43	Schultz TM., **Khan SN.,** Ahmed ZM., Riazuddin S., Waraya AA. Chhatre D., Starost MF., Ploplis B., Buckley S., Velasquez D., Kabra M., Lee K.,. Hassan MJ., Ali G., Ansar M., Ghosh M., Wilcox ER., Ahmad W., Merlino G., Leal SM., Riazuddin S., Friedman TB., and Morell RJ. (2009). Noncoding Mutations of HGF are associated with nonsyndromic hearing loss, DFNB39. Am. J. Hum Genet. 85, 25-39.	12.303
44	Khan M, Mohsin S, **Khan SN** and Riazuddin S. (2009). Repair of senescent infarcted myocardium by mesenchymal stem cells is dependent on the age of donor mice. J. Cell Mol. Med. December 11, 2009 [Epub ahead of print]	5.900
45	Rehman A.U., Morell R.J., Khan S.Y., Belyantseva I.A., Boger E.T., Shahzad M., Ahmed Z.M., Riazuddin S., **Khan S.N.,** Riazuddin S., and Friedman T.B. (2010) Targeted Capture and Next-Generation Sequencing Identifies C9orf75, Encoding TAPERIN, as the mutated gene in nonsyndromic deafness DFNB79. Am J Hum Genet. 86, 378-388.	12.303
46	Kaul H, S. Riazuddin SA, Yasmeen A, Mohsin S, Khan M, Nasir IA, **Khan SN,** Husnain T, Akram J, Hejtmancik FJ, Riazuddin S. (2010). A new locus for autosomal recessive congenital cataract identified in a Pakistani family. Mol. Vis 16:240-245.	2.464
47	Kaul H, S. Riazuddin SA, Shahid M, Kousar S, Butt NH, Zafar AU, **Khan SN**, Husnain T, Akram J, Hejtmancik FJ, Riazuddin S. (2010). Autosomal recessive congenital cataracts are associated with *EPHA2* in consanguineous Pakistani family. Mol. Vis 16:511-517.	2.464
48	Yasmeen A, S. Riazuddin SA, Kaul H, Mohsin S, Khan M, Qazi ZA, Nasir IA, Zafar AU, **Khan SN**, Husnain T, Akram J, Hejtmancik FJ, Riazuddin S. (2010). Autosomal recessive congenital cataract in consanguineous Pakistani families is associated with mutations in *GALK1*. Mol. Vis 16:682-688.	2.464
49	Riazuddin SA, Iqbal M, Wang Y, Masuda T, Chen Y, Bowne S, Sullivan LS, Waseem NH, Bhattacharya S, Daiger SP, Zhang K, **Khan SN,** Riazuddin S, Hejtmanic FJ, Sieving PA, Zack DJ and Katsanis N. (2010). A splice site mutations in a Retina-specific Exon of BBS8 cause syndromic Retinitis Pigmentosa. Am. J. Hum. Genet. 86: 1-8.	12.303
50	Khan M, Akhtar S, Mohsin S, **Khan SN,** and Riazuddin S. (2010). Growth factor preconditioning increases the function of diabetes-impaired mesenchymal stem cells. Stem Cells Dev. 20(1): 67-75.	4.146
51	Khan, M., Mohsin, S, **Khan, S.N**, Riazuddin, S. (2010) Lin-c-kit^{+} BM-derived stem cells repair Infarcted Heart. JSRM 6(1): 15-25.	------
52	Shehzadi A, Riazuddin SA., Ali S., Li D.,Sabar MF., Khan SN., Sieving, PA., Hejtmancik JF., Riazuddin S., (2010) Nonsense mutation of MERTK causes autosomal recessive retinitis pigmentosa in a consanguineous Pakistani family. Br J Ophthalmol. 94(8):1094-9.	2.680

53	Sabir N., Riazuddin S.A., Khul H., Iqbal F., Nasir IA., Zafar AU., Qazi ZA., Butt NH., **Khan SN**., Husnain T., Hejtmancik JF and Riazuddin S (2010). Mapping of a novel locus associated with autosomal recessive congenital cataract to chromosome 8P. Mol. Vision.16:2911-2915.	2.464
54	Sabir N., Riazuddin SA., Butt T., Iqbal F., Nasir IA, Zafar AU., Qazi ZA., Butt NH., **Khan SN**., Husnain T., Hejtmanick JF and Riazuddin S (2010). Mapping of a new locus associated with autosomal recessive congenital cataract at chromosome 3q. Mol. Vision.16:2634-2638.	2.464
55	Ali RA., Rehman AU., **Khan SN**., Husnain T., Riazuddin S., Friedman TB., Ahmed ZM and Riazuddin S (2011). DFNB86, a novel autosomal recesive non-syndromic deafness locus on chromosome 16p13.3. Clin Genet. Letter to the Editor p1-3.	
56	Mohsin S, Shams S, Nasir GA, Khan M, Awan SJ, **Khan SN,** and Riazuddin S. (2011). Enhanced hepatic differentiation of mesenchymal stem cells after pretreatment with injured liver tissue. Differentiation. 81:42-48.	3.310
57	Ahmed ZM, Yousaf R, Lee BC, **Khan SN,** Lee S, Lee K, Husnain T, Rehman AU, Bonneux S, Ansar M, Ahmad W, Leal SM, Gladyshev VN, Belyantseva IA, Camp GV, Riazuddin S, Friedman TB and Riazuddin S (2011). Functional null mutations of MSRB3 encoding methionine sulfoxide reductase are associated with human deafness DFNB 74. AM J Hum Genet. 88(1): 19-29.	12.303
58	Borck G, Rehman AU, Lee K, Pogoda HM, Kakar N, von Ameln S, Grillet N, Hildebrand MS, Ahmed ZM, Nürnberg G, Ansar M, Basit S, Javed Q, Morell RJ, Nasreen N, Shearer AE, Ahmad A, Kahrizi K, Shaikh RS, Ali RA, **Khan SN,** Goebel I, Meyer NC, Kimberling WJ, Webster JA, Stephan DA, Schiller MR, Bahlo M, Najmabadi H, Gillespie PG, Nürnberg P, Wollnik B, Riazuddin S, Smith RJ, Ahmad W, Müller U, Hammerschmidt M, Friedman TB, Riazuddin S, Leal SM, Ahmad J, Kubisch C (2011). Loss of function mutations of ILDRI cause autosomal recessive hearing impairment DFNB42. Am j Hum Genet. 88(2):127-137.	12.303
59	Riazuddin S, Ahmed ZM, Hegde RS, **Khan SN,** Nasir I, Shaukat U, Riazuddin S, Butman JA, Griffith AJ, Friedman TB and Choi BY (2011). Variable expressivity of FGF3 mutations associated with deafness and LAMM syndrome BMC Med Genet; 12: 21.	2.56
60	Rehman AU, Gul K, Morell RJ, Lee K, Ahmed ZM, Riazuddin S, Ali RA, Shahzad M, Ateeq-ul-Jaleel, Andrade PB, **Khan SN**, Khan S, Brewer CC, Ahmad W, Leal SM, Riazuddin S, and Friedman TB (2011). Mutations of GIPC3 cause nonsyndromic hearing loss DFNB72 but not DFNB81 that also maps to chromosome 19p. Hum Genet. 130: 759-765.	4.529

61 Choudhery MS., Khan M., Mahmood R., Mehmood A., **Khan SN** and Riazuddin S (2012). Bone marrow derived mesenchymal stem cells from aged mice have reduced wound healing, angiogenesis, proliferation and anti apoptosis capabilities. Cell Biol Int. CB120110183. 1.800

62 Choudhery MS., Khan M., Mahmood R., Mohsin S., Akhtar S., Ali F., **Khan SN** and Riazuddin S (2012). Mesenchymal stem cells conditioned with glucose depletion augments their ability to repair infarcted myocardium. J. Cell. Mol. Med 20 (10): 1-12. 5.288

63 Ali G., Mohsin S., Khan M., Nasir GA., Shams S., **Khan SN** and Riazuddin S (2012). Nitric oxide augments mesenchymal stem cell ability to repair liver fibrosis. J. Transl. Med. Doi:10.1186/1479-5876-10-75 3.500

64 Naeem MA., Chavali VRM., Ali S., Iqbal M., Riazuddin S., **Khan SN**., Husnain T., Sieving PA., Ayyagari R., Riazuddin S., Hejtmancik JF and Riazuddin SA (2012). GNAT1 associated with autosoamal recessive congenital stationary night blindness. Investigative ophthalmology & visual science. 53(3):1353- 1361 3.466

65 Jaworek TJ., Bhatti R., Latiej N., Khan SN., Riazuddin S and Ahmed ZM (2012). USH 1K, a novel locus for type 1 Usher syndrome, maps to chromosome 10p11.21-q21.1. J. Hum Genet. 00,1-5 4.529

Total Impact Factor = 304.783