CV

Name	Muhammad Ansar
Date of Birth	17.10.1976
Present address:	Department of Biochemistry, Faculty of Biological Sciences, Quaid-I-Azam University Islamabad. Ph. # 92-51- 90643146 E-mail: ansar@qau.edu.pk
Permanent address:	DK-743/21, Dhoke Kashmirian, Service Road, Kiyani Bazar, 46300, Rawalpindi. mansar_76@yahoo.com

EDUCATIONAL QUALIFICATION

Qualification	Year	Major Subject	Institution/ University
Post-Doc	Sep 2013-June 2015	Human Genetics	Baylor College of Medicine, USA
PhD	2000-04	Biochemistry/Molecular Biology	Quaid-I-Azam University
MSc	1997-99	Biochemistry/Molecular Biology	Quaid-I-Azam University
BSc	1995-97	Botany, Zoology, Chemistry	Punjab University

Professional Trainings & Workshops

- Received Postdoctoral training on Next Generation Sequencing (NGS) from Baylor College of Medicine, Houston, USA (September 2013 to June 2015)
- Received training on **ABI PRISM 310 DNA sequencer** conducted by Applied Biosystems at Dubai (2003).
- Received training on **CEQ 8000 DNA sequencer and Product Applications** conducted by Beckman Coulter UK, Ltd from 25th July to 28th July (2006).
- **Agilent aCGH Workshop,** Organized by Agilent and MD Anderson Cancer Center, (29th-30th April, 2015), Houston, USA

- **Computational Biology Workshop**, Organized by Sultan Qaboos University, 24th - 28th January (2009), Muscat, Oman.
- **Statistical Gene Mapping: Case-Control Association Data,** Organized by Beijing Institute of Genomics, Chinese Academy of Sciences, Beijing, 14th -19th June, (2009), China.
- **Protein Molecular Modeling and Docking Workshop** Organized by University of Arid Agriculture Rawalpindi, Pakistan, 23rd - 27th January, (2006).

Research Grants

Project Title	Status	Amount	Funding Agency	Duration
Elucidating the Genetic Etiology of Intellectual Disability in African, Asian and European Families	Principal Investigator Pakistan Side	USD 86,400/ Year (5 year = 432,000 $)	National Institute of Health, USA	Aug 2023 to July 2028 (In Progress)
Elucidation of Genetic Players Responsible for Intellectual Disability in Pakistani Families	Principal Investigator	Rs. 6352991/-	Higher Education Commission (HEC)	2018-2021 (Completed)
Molecular Genetic Studies of Inherited Neurological Disorders	Principal Investigator	Rs. 3706402/-	Higher Education Commission (HEC)	2008-2010 (Completed)
Characterization of Loci Responsible for Inherited Eye Anomalies in Families of Pakistan	Co-Principal Investigator	Rs. 4,936,800/-	Higher Education Commission (HEC)	2005-2007 (Completed)

Research Supervision

Degree	No	Status	Program
PhD Completed	09+01	Supervisor + Co supervisor	Biochemistry
PhD In Progress	04	Supervisor	Biochemistry
M.Phil Completed	88	Supervisor	Biochemistry
M.Phil In Progress	05	Supervisor	Biochemistry

Teaching

Course Title	Credit Hours	PhD/M Phil/BS
Enzymology	3	BS
Advances in Molecular Biology	3	MPhil/PhD
Gene Expression & Regulation	3	MPhil/PhD
High Throughput Sequencing Platforms & Applications	3	MPhil/PhD
Genetics	3	BS

Awards & Honors

1. AR Shakoori Gold Medal in Biological Sciences for Year 2016 by Zoological Society of Pakistan
2. Fellowship for participation in World Science Forum 2015 (WSF2015), (4th to 7th November 2015) Budapest, Hungary
3. Research Productivity Award (RPA) for years 2004, 2005 and 2006, 2010, 2011, 2012, 2013 Pakistan Council for Science and Technology, Pakistan.
4. Pakistan Academy of Sciences Gold Medal in Biochemistry (2010).
5. BioVision 2011 Fellowship Award & TWAS travel grant for participation in BioVision 2011 Life Sciences Forum, Lyon, France (26th -30th March 2011).

Membership

1. Member American Society of Human Genetics
2. Member Pakistan Academy of Sciences
3. Pakistan Society for Biochemistry and Molecular Biology (PSBMB), Pakistan

Professional Experience

1. Tenured Professor (7th Oct 2016-To date)
Department of Biochemistry, Faculty of Biological Sciences
Quiad-I-Azam University
Islamabad, Pakistan.

2. Tenured Associate Professor (21st May 2011-6th Oct 2016)
Department of Biochemistry, Faculty of Biological Sciences
Quiad-I-Azam University
Islamabad, Pakistan.

3. Assistant Professor on TTS (Aug 2007-20th May 2011)
Department of Biochemistry, Faculty of Biological Sciences
Quiad-I-Azam University
Islamabad, Pakistan.

4. Assistant Professor (1st Feb, 2005- Aug 2007)
Department of Biosciences
COMSATS Institute of Information Technology
Islamabad, Pakistan.

5. Lecturer Molecular Biology (20th Nov 2003- 31st Jan, 2005)
Department of Biosciences
COMSATS Institute of Information Technology
Islamabad, Pakistan.

6. Chairperson Biochemistry Department (August 2015 to July 2018)
Department of Biochemistry, Quaid-I-Azam University

Conferences & Workshops

- Ullah E, Lao R, Tang P, Wan E, Bardakjian T, Kwok P, Schneider A, **Ansar M**, Slavotinek A. Two missense mutations in SALL4 in a patient with microphthalmia, coloboma and optic nerve hypoplasia. **European Society of Human Genetics (ESHG2015), June 6-9, 2015, Glasgow, UK.**

- Zeinab Ravesh, Nicole Weisschuh, Bernd Wissinger, **Muhammad Ansar**. Genetic analysis of hereditary retinal dystrophies in consanguineous Pakistani families. **Meeting of Asia-Association for Research in Vision and Ophthalmology (ARVO 2015), February 16-19, 2015, Yokohama, Japan.**

- **M. Ansar**, K. Lee, RL. Santos-Cortez, E. Ullah, Z. Ravesh, MA. Saqib, X. Wang, JD. Smith, J. Shendure, MJ. Bamshad, DA. Nickerson, W. Ahmad, SM. Leal. Linkage Analysis and Gene Identification in Consanguineous Pakistani Families with Autosomal Recessive Retinal Dystrophy. **American Society of Human Genetics 64th Meeting** (18th to 22nd October, 2014)**, San Diego, California, USA**

- E. Ullah, MA. Saqib, N. Shah, S. Sajid, R. Lao, E. Wan, PL. Tang, P. Kwok, **M. Ansar**, A. Slavotinek. Mutations in ALDH1A3, FOXE3 and VSX2 cause ocular abnormalities in consanguineous Pakistani families. **American Society of Human Genetics 64th Meeting** (18th to 22nd October, 2014)**, San Diego, California, USA**

- RLP. Santos-Cortez, A. U. Rehman, M. C. Drummond, M. Shahzad, K. Lee, R. J. Morell, **M. Ansar**, A. Jan, X. Wang, A. Aziz, S. Riazuddin, J. D. Smith, G. T. Wang, Z. M. Ahmed, K. Gul, A. E. Shearer, R. J. H. Smith, J. Shendure, M. J. Bamshad, D. A. Nickerson, J. Hinnant, S. N. Khan, R. A. Fisher, W. Ahmad, K. H. Friderici, S. Riazuddin, T. B. Friedman, E. S. Wilch, SM Leal. Why Next-Generation Sequencing Studies May Fail: Challenges and Solutions for Gene Identification in the Presence of Familial Locus Heterogeneity. **American Society of Human Genetics 64th Meeting** (18th to 22nd October, 2014)**, San Diego, California, USA**

- VM Rupp, MA Khan, M. Orpinell, M. S. Hussain, J. Altmüller, M. O. Steinmetz C. Enzinger, H. Thile, W. Höhne, G. Nürnberg, S. M. Baig, **M. Ansar**, P. Nürnberg, J. B. Vincent, M. R. Speicher, P. Gönczy, C. Windpassinger. A missense mutation in the PISA domain of HsSAS-6 causes autosomal recessive primary microcephaly in a large consanguineous Pakistani family. **American Society of Human Genetics 64th Meeting (18th to 22nd October, 2014), San Diego, California, USA.**

- Saqib MAN, Ullah E, Khan FS, Venturini G, Nikopoulos K, **Ansar M**, Rivolta C. Homozygosity mapping and disease gene screening in Pakistani families with inherited retinal dystrophies. **Annual meeting of Association for Research in Vision and Ophthalmology (ARVO 2014), May 4-8, 2014, Orlando, Florida, USA**.

- Human and Molecular Genetics Retreat (9th to 10th January 2014) organized by Molecular Genetics Department, Baylor College of Medicine, USA at Moody gardens, Galveston, Texas, USA (**Poster presentation**).

- M. Rafiq, K. Mittal, I. A. Balouch, A. Noor, C. Windpassinger, A. Mikhailov, M. Aslam, M. Ayaz, A. Mir, **M. Ansar**, P. John, M. Ayub, J. B. Vincent. Autozygosity Mapping in

Pakistani Intellectual Disability Families. American Society of Human Genetics 63rd Meeting (22-26 Oct, 2013), Boston, USA

- National Symposium on Biochemical and Molecular Basis of Human Diseases, organized by University of Gujrat and Institute of Chemical and Biological Sciences, 17-18 May, 2012 **(Invited Lecture entitled "Retinal and Neurological Disorders").**

- National Conference on Trends in Biochemistry and Molecular Biology organized by Department of Biochemistry and BAQI, 21st February, 2012. (**Conference Secretary**)

- Rafiq MA, Noor A, Mittal K, Wiane E, Kaufman L, Mikhailov A, Naeem F, Van Schaftingen E, Nasir T, **Ansar M**, Ayub M, Vincent JB. Homozygosity Mapping in 7 Pakistani Intellectual Disability Families Identifies 3 New Autosomal Recessive Loci. **American Society of Human Genetics Meeting (October 11-15, 2011), Montreal, Canada.**

- Vincent JB , Khan MA, Rafiq MA, Noor A, Hussain S, Rupp V, Vincent AK, Flores J, Ishak GE, Doherty D, Weksberg R, Ayub M, Windpassinger C, Ibrahim S, **Ansar M**,. Identification of the tRNA methyltransferase gene NSUN2 as the gene for a new form of autosomal recessive intellectual disability. **American Society of Human Genetics Meeting (October 11-15, 2011), Montreal, Canada.**

- **UAAR/NCB Workshop on Biotechnology for Secondary School Teachers**, University of Arid Agriculture Rawalpindi, Pakistan, 5th - 9th January, 2006. (Invited Lecture).

- **International Thematic Workshop on the use of Bioinformatics in Genomic Research** organized by the COMSTECH and HEC at COMSTECH Secretariat, Islamabad, Pakistan, 19th – 2nd September, 2006.

List of Publications

1. Dore R, Chang CT, Declève A, Brunori G, Ludlam WG, Huang A, Movahedinia M, Damseh NS, Anwar I, Vahidi Mehrjardi MY, Ny A, Khorrami M, Kheirollahi M, Frederiksen H, Eghbal F, Mirjalili MR, Dehghani M, Karimiani EG, Oreshkov S, Alves C, Striano P, Suri M, Martinez-Agosto J, **Ansar M**, Zahid M, Akram S, Ansar M, Nelson SF; Undiagnosed Diseases Network; Antonarakis SE, Houlden H, Copmans D, Martemyanov KA, Maroofian R. ELFN1 Deficiency: the mechanistic basis and phenotypic spectrum of a neurodevelopmental disorder with epilepsy. **Genet Med. 2025 Jun 23:101506. doi: 10.1016/j.gim.2025.101506.**

2. Saeed T, Bibi N, Ahmad A, Khan S, **Ansar M**, Wasif N, Kalsoom UE. A Novel Biallelic Variant in IHH Causing Acrocapitofemoral Dysplasia in a Pakistani Family. **Mol Genet Genomic Med. 2025 Mar;13(3):e70085.**

3. Thomas HB, Demain LAM, Cabrera-Orefice A, Schrauwen I, Shamseldin HE, Rea A, Bharadwaj T, Smith TB, Oláhová M, Thompson K, He L, Kaur N, Shukla A, Abukhalid M, **Ansar M**, Rehman S, Riazuddin S, Abdulwahab F, Smith JM, Stark Z, Mancilar H, Tumer S, Esen FN, Uctepe E, Topcu V, Yesilyurt A, Afzal E, Salari M, Carroll C, Zifarelli G, Bauer P, Kor D, Bulut FD, Houlden H, Maroofian R, Carrera S, Yue WW, Munro KJ, Alkuraya FS, Jamieson P, Ahmed ZM, Leal SM, Taylor RW, Wittig I, O'Keefe RT, Newman WG. Bi-allelic variants in MRPL49 cause variable clinical presentations, including sensorineural hearing loss, leukodystrophy, and ovarian insufficiency. **Am J Hum Genet. 2025 Apr 3;112(4):952-962**.

4. Basharat R, de Bruijn SE, Zahid M, Rodenburg K, Hitti-Malin RJ, Rodríguez-Hidalgo M, Boonen EGM, Jarral A, Mahmood A, Corominas J, Khalil S, Zai JA, Ali G, Ruiz-Ederra J, Gilissen C, Cremers FPM, **Ansar M**, Panneman DM, Roosing S. Next-generation sequencing to genetically diagnose a diverse range of inherited eye disorders in 15 consanguineous families from Pakistan. **Exp Eye Res. 2024 Jul;244:109945.**

5. Khan FU, Khan H, Ullah K, Nawaz S, Abdullah, Khan MJ, Ahmed S, Ilyas M, Ali A, Ullah I, Sohail A, Hussain S, Ahmad F, Faisal, Sufyan R, Hayat A, Hanif T, Bibi F, Hayat M, Ullah R, Khan IU, Ali RH, Hasni MS, Ali H, Bilal M, Peralta S, Buchert R, Zehri Z, Hassan G, Liaqat K, Zahid M, Shah K, Mikitie O, Haack TB, Ji W, Lakhani SA, **Ansar M**, Ahmad W. Clinical and genetic investigation of 14 families with various forms of short stature syndromes. **Clin Genet. 2024 Sep;106(3):347-353.**

6. Abbas E, Fawwad A, Siddiqui IA, Afzal MS, **Ansar M**, Saqib MAN, Shahid SM. Risk Factors for the Development of Early Onset Diabetes in the Population of Sindh Province, Pakistan. **Biomedicines. 2025 May 2;13(5):1107**.

7. Malik MA, Saqib MAN, Mientjes E, Acharya A, Alam MR, Wallaard I, Schrauwen I, Bamshad MJ, Santos-Cortez RLP, Elgersma Y, Leal SM, **Ansar M**. A loss of function variant in AGPAT3 underlies intellectual disability and retinitis pigmentosa (IDRP) syndrome. **Eur J Hum Genet. 2023 Dec;31(12):1447-1454.**

8. Basharat R, Rodenburg K, Rodríguez-Hidalgo M, Jarral A, Ullah E, Corominas J, Gilissen C, Zehra ST, Hameed U, **Ansar M**, de Bruijn SE. Combined Single Gene Testing and Genome Sequencing as an Effective Diagnostic Approach for Anophthalmia and Microphthalmia Patients. **Genes (Basel). 2023 Aug 1;14(8):1573.**

9. Khan S, Umair M, Abbas S, Ali U, Zaman G, **Ansar M**, Wang R, Zhang X, Houlden H, Harlalka GV, Gul A. Overlapping neurological phenotypes in two extended consanguineous families with novel variants in the CNTNAP1 and ADGRG1 genes. **J Gene Med. 2023 May 13:e3513.**

10. Liaqat K, Hussain S, Acharya A, Nasir A, Bharadwaj T, **Ansar M**, Basit S, Schrauwen I, Ahmad W, Leal SM. Phenotype Expansion for Atypical Gaucher Disease Due to Homozygous Missense PSAP Variant in a Large Consanguineous Pakistani Family. **Genes (Basel). 2022 Apr 9;13(4):662**.

11. Bibi S, Ahmad F, Alam MR, **Ansar M**, Yeou KS, Wahedi HM. Lapachol-Induced Upregulation of Sirt1/Sirt3 is linked with Improved Skin Wound Healing in Alloxan-induced Diabetic Mice. **Iran J Pharm Res. 2021 Summer;20(3):419-430.**

12. Nawal W, Ullah A, Ullah U, Farrakh K, Ahmad F, Khan H, Ahmad GS, Khan B, **Ansar M,** Kalsoom UE, Ahmad W. Loss of Function Variants in the XPC Causes Severe Xeroderma Pigmentosum in Three Large Consanguineous Families. **Klin Padiatr. 2022 May;234(3):123-129.**

13. Levy MA, Beck DB, Metcalfe K, Douzgou S, Sithambaram S, Cottrell T, **Ansar M**, Kerkhof J, Mignot C, Nougues MC, Keren B, Moore HW, Oegema R, Giltay JC, Simon M, van Jaarsveld RH, Bos J, van Haelst M, Motazacker MM, Boon EMJ, Santen GWE, Ruivenkamp CAL, Alders M, Luperchio TR, Boukas L, Ramsey K, Narayanan V, Schaefer GB, Bonasio R, Doheny KF, Stevenson RE, Banka S, Sadikovic B, Fahrner JA. Deficiency of TET3 leads to a genome-wide DNA hypermethylation episignature in human whole blood. **NPJ Genom Med. 2021 Nov 8;6(1):92.**

14. Acharya A, Raza SI, Anwar MZ, Bharadwaj T, Liaqat K, Khokhar MAS, Everard JL, Nasir A; University of Washington Center for Mendelian Genomics, Nickerson DA, Bamshad MJ, **Ansar M**, Schrauwen I, Ahmad W, Leal SM. Wolfram-like syndrome with bicuspid aortic valve due to a homozygous missense variant in CDK13. **J Hum Genet. 2021 Oct;66(10):1009-1018.**

15. Shah K, Basit S, Ali G, Ramzan K, **Ansar M**, Ahmad W. A novel homozygous frameshift variant in the C3orf52 gene underlying isolated hair loss in a consanguineous family. **Eur J Dermatol. 2021 Jun 1;31(3):409-411.**

16. Rasheed M, Khan V, Harripaul R, Siddiqui M, Malik MA, Ullah Z, Zahid M, Vincent JB, **Ansar M**. Exome sequencing identifies novel and known mutations in families with intellectual disability. **BMC Med Genomics. 2021 Aug 27;14(1):211.**

17. Beck DB, Petracovici A, He C, Moore HW, Louie RJ, **Ansar M**, Douzgou S, Sithambaram S, Cottrell T, Santos-Cortez RLP, Prijoles EJ, Bend R, Keren B, Mignot C, Nougues MC, Õunap K, Reimand T, Pajusalu S, Zahid M, Saqib MAN, Buratti J, Seaby EG, McWalter K, Telegrafi A, Baldridge D, Shinawi M, Leal SM, Schaefer GB, Stevenson RE, Banka S, Bonasio R, Fahrner JA. Delineation of a Human Mendelian Disorder of the DNA Demethylation Machinery: TET3 Deficiency. **Am J Hum Genet. 2020 Feb 6;106(2):234-245.**

18. Umair M, Bilal M, Ali RH, Alhaddad B, Ahmad F, Abdullah, Haack TB, Alfadhel M, **Ansar M**, Meitinger T, Ahmad W. Whole-exome sequencing revealed a nonsense mutation in STKLD1 causing non-syndromic pre-axial polydactyly type A affecting only upper limb. **Clin Genet. 2019 Aug;96(2):134-139**.

19. Khan A, Han S, Wang R, **Ansar M**, Ahmad W, Zhang X. Sequence variants in genes causing nonsyndromic hearing loss in a Pakistani cohort. **Mol Genet Genomic Med. 2019 Sep;7(9):e917.**

20. Parveen A, Mirza MU, Vanmeert M, Akhtar J, Bashir H, Khan S, Shehzad S, Froeyen M, Ahmed W, **Ansar M**, Wasif N. A novel pathogenic missense variant in CNNM4 underlying Jalili syndrome: Insights from molecular dynamics simulations. **Mol Genet Genomic Med. 2019 Sep;7(9):e902.**

21. Ullah I, Kakar N, Schrauwen I, Hussain S, Chakchouk I, Liaqat K, Acharya A, Wasif N, Santos-Cortez RLP, Khan S, Aziz A, Lee K, Couthouis J, Horn D, Kragesteen BK, Spielmann M, Thiele H, Nickerson DA, Bamshad MJ, Gitler AD, Ahmad J, **Ansar M**, Borck G, Ahmad W, Leal SM. Variants in KIAA0825 underlie autosomal recessive postaxial polydactyly. **Hum Genet. 2019 Jun;138(6):593-600.**

22. Liaqat K, Schrauwen I, Raza SI, Lee K, Hussain S, Chakchouk I, Nasir A, Acharya A, Abbe I, Umair M, **Ansar M**, Ullah I, Shah K; University of Washington Center for Mendelian Genomics, Bamshad MJ, Nickerson DA, Ahmad W, Leal SM. Identification of CACNA1D variants associated with sinoatrial node dysfunction and deafness in

additional Pakistani families reveals a clinical significance. **J Hum Genet. 2019 Feb;64(2):153-160.**

23. Schrauwen I, Giese AP, Aziz A, Lafont DT, Chakchouk I, Santos-Cortez RLP, Lee K, Acharya A, Khan FS, Ullah A, Nickerson DA, Bamshad MJ, Ali G, Riazuddin S, **Ansar M,** Ahmad W, Ahmed ZM, Leal SM. FAM92A Underlies Nonsyndromic Postaxial Polydactyly in Humans and an Abnormal Limb and Digit Skeletal Phenotype in Mice. **J Bone Miner Res. 2019 Feb;34(2):375-386.**

24. 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. Global genetic insight contributed by consanguineous Pakistani families segregating hearing loss. **Hum Mutat. 2019 Jan;40(1):53-72.**

25. Latif Z, Chakchouk I, Schrauwen I, Lee K, Santos-Cortez RLP, Abbe I, Acharya A, Jarral A, Ali I, Ullah E, Khan MN, Ali G, Tahir TH, Bamshad MJ, Nickerson DA, Ahmad W, **Ansar M**, Leal SM. Confirmation of the Role of DHX38 in the Etiology of Early-Onset Retinitis Pigmentosa. **Invest Ophthalmol Vis Sci. 2018 Sep 4;59(11):4552-4557.**

26. Liaqat K, Chiu I, Lee K, Chakchouk I, Andrade-Elizondo PB, Santos-Cortez RLP, Hussain S, Nawaz S, **Ansar M**, Khan MN, Basit S, Schrauwen I, Ahmad W, Leal SM. Novel missense and 3'-UTR splice site variants in LHFPL5 cause autosomal recessive nonsyndromic hearing impairment. **J Hum Genet. 2018 Nov;63(11):1099-1107.**

27. Santos-Cortez RLP, Khan V, Khan FS, Mughal ZU, Chakchouk I, Lee K, Rasheed M, Hamza R, Acharya A, Ullah E, Saqib MAN, Abbe I, Ali G, Hassan MJ, Khan S, Azeem Z, Ullah I, Bamshad MJ, Nickerson DA, Schrauwen I, Ahmad W, **Ansar M**, Leal SM. Novel candidate genes and variants underlying autosomal recessive neurodevelopmental disorders with intellectual disability. **Hum Genet. 2018 Sep;137(9):735-752.**

28. Fattahi Z, Sheikh TI, Musante L, Rasheed M, Taskiran II, Harripaul R, Hu H, Kazeminasab S, Alam MR, Hosseini M, Larti F, Ghaderi Z, Celik A, Ayub M, **Ansar M**, Haddadi M, Wienker TF, Ropers HH, Kahrizi K, Vincent JB, Najmabadi H. Biallelic missense variants in ZBTB11 can cause intellectual disability in humans. **Hum Mol Genet. 2018 Sep 15;27(18):3177-3188.**

29. Harripaul R, Vasli N, Mikhailov A, Rafiq MA, Mittal K, Windpassinger C, Sheikh TI, Noor A, Mahmood H, Downey S, Johnson M, Vleuten K, Bell L, Ilyas M, Khan FS, Khan V, Moradi M, Ayaz M, Naeem F, Heidari A, Ahmed I, Ghadami S, Agha Z, Zeinali S, Qamar R, Mozhdehipanah H, John P, Mir A, **Ansar M**, French L, Ayub M, Vincent JB. Mapping autosomal recessive intellectual disability: combined microarray and exome sequencing identifies 26 novel candidate genes in 192 consanguineous families. **Mol Psychiatry. 2018 Apr;23(4):973-984.**

30. Khan S, **Ansar M**, Khan AK, Shah K, Muhammad N, Shahzad S, Nickerson DA, Bamshad MJ, Santos-Cortez RLP, Leal SM, Ahmad W. A homozygous missense mutation in SLC25A16 associated with autosomal recessive isolated fingernail dysplasia in a Pakistani family. **Br J Dermatol. 2018 Feb;178(2):556-558.**

31. Shah K, Mehmood S, Jan A, Abbe I, Hussain Ali R, Khan A, Chishti MS, Lee K, Ahmad F, **Ansar M**; University of Washington Center for Mendelian Genomics, Shahzad S, Nickerson DA, Bamshad MJ, Coucke PJ, Santos-Cortez RLP, Spritz RA, Leal SM, Ahmad W. Sequence variants in nine different genes underlying rare skin disorders in 10 consanguineous families. **Int J Dermatol. 2017 Dec;56(12):1406-1413**.

32. Ullah E, Wu D, Madireddy L, Lao R, Ling-Fung Tang P, Wan E, Bardakjian T, Kopinsky S, Kwok PY, Schneider A, Baranzini S, **Ansar M**, Slavotinek A. Two missense mutations in SALL4 in a patient with microphthalmia, coloboma, and optic nerve hypoplasia. **Ophthalmic Genet. 2017 Jul-Aug;38(4):371-375.**

33. Shah K, **Ansar M**, Mughal ZU, Khan FS, Ahmad W, Ferrara TM, Spritz RA. Recessive progressive symmetric erythrokeratoderma results from a homozygous loss-of-function mutation of *KRT83* and is allelic with dominant monilethrix**. J Med Genet. 2017 Mar;54(3):186-189.**

34. Umair M, Rafique A, Ullah A, Ahmad F, Ali RH, Nasir A, **Ansar M**, Ahmad W. Novel homozygous sequence variants in the GDF5 gene underlie acromesomelic dysplasia

type-grebe in consanguineous families. **Congenit Anom (Kyoto). 2017 Mar;57(2):45-51**.

35. Ullah A, Kalsoom UE, Umair M, John P, **Ansar M**, Basit S, Ahmad W. Exome sequencing revealed a novel splice site variant in the ALX1 gene underlying frontonasal dysplasia.Clin Genet. 2017 Mar;91(3):494-498.

36. Ullah E, Nadeem Saqib MA, Sajid S, Shah N, Zubair M, Khan MA, Ahmed I, Ali G, Dutta AK, Danda S, Lao R, Ling-Fung Tang P, Kwok PY, **Ansar M**, Slavotinek A. Genetic analysis of consanguineous families presenting with congenital ocular defects. **Exp Eye Res. 2016 May;146:163-71.**

37. Chishti HM, Manica A, **Ansar M**, Eriksson A, Ajmal M, Hameed A. Inability of the most commonly used forensic genetic markers to distinguish between samples belonging to different ethnicities of Pakistan with diverse genetic background. Forensic Sci Int Genet. 2016 May;22:e7-8.

38. Shah K, Ali RH, **Ansar M**, Lee K, Chishti MS, Abbe I, Li B; University of Washington Center for Mendelian Genomics, Smith JD, Nickerson DA, Shendure J, Coucke PJ, Steyaert W, Bamshad MJ, Santos-Cortez RL, Leal SM, Ahmad W. Mitral regurgitation as a phenotypic manifestation of nonphotosensitive trichothiodystrophy due to a splice variant in MPLKIP. **BMC Med Genet. 2016 Feb 16;17(1):13.**

39. Santos-Cortez RL, Faridi R, Rehman AU, Lee K, **Ansar M**, Wang X, Morell RJ, Isaacson R, Belyantseva IA, Dai H, Acharya A, Qaiser TA, Muhammad D, Ali RA, Shams S, Hassan MJ, Shahzad S, Raza SI, Bashir ZE, Smith JD, Nickerson DA, Bamshad MJ; University of Washington Center for Mendelian Genomics, Riazuddin S, Ahmad W, Friedman TB, Leal SM. Autosomal-Recessive Hearing Impairment Due to Rare Missense Variants within S1PR2. **Am J Hum Genet. 2016 Feb 4;98(2):331-8.**

40. **Ansar M**, Jan A, Santos-Cortez RL, Wang X, Suliman M, Acharya A, Habib R, Abbe I, Ali G, Lee K, Smith JD; University of Washington Center for Mendelian Genomics, Nickerson DA, Shendure J, Bamshad MJ, Ahmad W, Leal SM. Expansion of the spectrum of ITGB6-related disorders to adolescent alopecia, dentogingival abnormalities and intellectual disability. Eur J Hum Genet. 2016 Aug;24(8):1223-7

41. Ahmad F, **Ansar M**, Mehmood S, Izoduwa A, Lee K, Nasir A, Abrar M, Mehmood S, Ullah A, Aziz A; University of Washington Center for Mendelian Genomics, Smith JD, Shendure J, Bamshad MJ, Nicekrson DA, Santos-Cortez RL, Leal SM, Ahmad W. A novel missense variant in the PNPLA1 gene underlies congenital ichthyosis in three consanguineous families. J Eur Acad Dermatol Venereol. 2016 Dec;30(12):e210-e213.

42. Ullah R, **Ansar M**, Durrani ZU, Lee K, Santos-Cortez RL, Muhammad D, Ali M, Zia M, Ayub M, Khan S, Smith JD, Nickerson DA, Shendure J, Bamshad M, Leal SM, Ahmad W. Novel mutations in the genes TGM1 and ALOXE3 underlying autosomal recessive congenital ichthyosis. Int J Dermatol. 2016 May;55(5):524-30.

43. Ali RH, Mahmood S, Raza SI, Aziz A, Irfanullah, Naqvi SK, Wasif N, **Ansar M**, Ahmad W, Shah SH, Khan BT, Zaman Q, Gul A, Wali A, Ali G, Khan S, Khisroon M, Basit S. Genetic analysis of Xp22.3 micro-deletions in seventeen families segregating isolated form of X-linked ichthyosis. J Dermatol Sci. 2015 Sep 28. pii: S0923-1811(15)30047-5.

44. **Ansar M**, Raza SI, Lee K, Irfanullah, Shahi S, Acharya A, Dai H, Smith JD, Shendure J, Bamshad MJ, Nickerson DA, Santos-Cortez RL, Ahmad W, Leal SM. A homozygous missense variant in type I keratin KRT25 causes autosomal recessive woolly hair. J Med Genet. 2015 Oct;52(10):676-80.

45. **Ansar M**, Santos-Cortez RL, Saqib MA, Zulfiqar F, Lee K, Ashraf NM, Ullah E, Wang X, Sajid S, Khan FS, Amin-Ud-Din M; University of Washington Center for Mendelian Genomics, Smith JD, Shendure J, Bamshad MJ, Nickerson DA, Hameed A, Riazuddin S, Ahmed ZM, Ahmad W, Leal SM. Mutation of ATF6 causes autosomal recessive achromatopsia. **Hum Genet. 2015 Sep;134(9):941-50.**

46. Habib R, **Ansar M**, Mattheisen M, Shahid M, Ali G, Ahmad W, Betz RC. A Novel Locus for Ectodermal Dysplasia of Hair, Nail and Skin Pigmentation Anomalies Maps to Chromosome 18p11.32-p11.31. **PLoS One. 2015 Jun 26;10(6):e0129811.**

47. Rehman AU, Santos-Cortez RL, Drummond MC, Shahzad M, Lee K, Morell RJ, **Ansar M**, Jan A, Wang X, Aziz A, Riazuddin S, Smith JD, Wang GT, Ahmed ZM, Gul K, Shearer AE, Smith RJ, Shendure J, Bamshad MJ, Nickerson DA; University of Washington Center for Mendelian Genomics, Hinnant J, Khan SN, Fisher RA, Ahmad W, Friderici KH, Riazuddin S, Friedman TB, Wilch ES, Leal SM. Challenges and solutions for gene identification in the presence of familial locus heterogeneity. **Eur J Hum Genet. 2015 Sep;23(9):1207-15.**

48. Rafiq MA, Leblond CS, Saqib MA, Vincent AK, Ambalavanan A, Khan FS, Ayaz M, Shaheen N, Spiegelman D, Ali G, Amin-Ud-Din M, Laurent S, Mahmood H, Christian M, Ali N, Fennell A, Nanjiani Z, Egger G, Caron C, Waqas A, Ayub M, Rasheed S, Forgeot d'Arc B, Johnson A, So J, Brohi MQ, Mottron L, **Ansar M**, Vincent JB, Xiong L. Novel VPS13B Mutations in Three Large Pakistani Cohen Syndrome Families

Suggests a Baloch Variant with Autistic-Like Features. **BMC Med Genet. 2015 Jun 25;16:41.**

49. Saqib MA, Nikopoulos K, Ullah E, Sher Khan F, Iqbal J, Bibi R, Jarral A, Sajid S, Nishiguchi KM, Venturini G, **Ansar M**, Rivolta C. Homozygosity mapping reveals novel and known mutations in Pakistani families with inherited retinal dystrophies. **Sci Rep. 2015 May 6;5:9965.**

50. Ravesh Z, El Asrag ME, Weisschuh N, McKibbin M, Reuter P, Watson CM, Baumann B, Poulter JA, Sajid S, Panagiotou ES, O'Sullivan J, Abdelhamed Z, Bonin M, Soltanifar M, Black GC, Amin-ud Din M, Toomes C, **Ansar M**, Inglehearn CF, Wissinger B, Ali M. Novel C8orf37 mutations cause retinitis pigmentosa in consanguineous families of Pakistani origin. **Mol Vis. 2015 Mar 7;21:236-43.**

51. Mehmood S, Jan A, Muhammad D, Ahmad F, Mir H, Younus M, Ali G, Ayub M, **Ansar M**, Ahmad W. Mutations in the lipase-H gene causing autosomal recessive hypotrichosis and woolly hair. **Australas J Dermatol**. **2015 Aug;56(3):e66-70.**

52. Law R, Dixon-Salazar T, Jerber J, Cai N, Abbasi AA, Zaki MS, Mittal K, Gabriel SB, Rafiq MA, Khan V, Nguyen M, Ali G, Copeland B, Scott E, Vasli N, Mikhailov A, Khan MN, Andrade DM, Ayaz M, **Ansar M**, Ayub M, Vincent JB, Gleeson JG. Biallelic truncating mutations in FMN2, encoding the actin-regulatory protein Formin 2, cause nonsyndromic autosomal-recessive intellectual disability. **Am J Hum Genet. 2014 Dec 4;95(6):721-8.**

53. Khan MA, Rupp VM, Orpinell M, Hussain MS, Altmüller J, Steinmetz MO, Enzinger C, Thiele H, Höhne W, Nürnberg G, Baig SM, **Ansar M**, Nürnberg P, Vincent JB, Speicher MR, Gönczy P, Windpassinger C. A missense mutation in the PISA domain of HsSAS-6 causes autosomal recessive primary microcephaly in a large consanguineous Pakistani family. **Hum Mol Genet. 2014 Nov 15;23(22):5940-5949.**

54. Chishti HM, **Ansar M**, Ajmal M, Hameed A. Application of Short Tandem Repeat markers in diagnosis of chromosomal aneuploidies and forensic DNA investigation in Pakistan. **Gene. 2014 Sep 15;548(2):217-22.**

55. Khan MA, Rupp V, Khan MA, Khan MP, **Ansar M**, Windpassinger C. Genetic analysis of a consanguineous Pakistani family with Leber congenital amaurosis identifies a novel mutation in GUCY2D gene. **J Genet. 2014 Aug;93(2):527-30.**

56. Santos-Cortez RL, Lee K, Giese AP, **Ansar M**, Amin-Ud-Din M, Rehn K, Wang X, Aziz A, Chiu I, Hussain Ali R, Smith JD; University of Washington Center for Mendelian Genomics, Shendure J, Bamshad M, Nickerson DA, Ahmed ZM, Ahmad W,

Riazuddin S, Leal SM. Adenylate cyclase 1 (ADCY1) mutations cause recessive hearing impairment in humans and defects in hair cell function and hearing in zebrafish. **Hum Mol Genet. 2014 Jun 15;23(12):3289-98.**

57. Ali RH, Habib R, Ud-Din N, Khan MN, **Ansar M**, Ahmad W. Novel mutations in the gene HOXC13 underlying pure hair and nail ectodermal dysplasia in consanguineous families. **Br J Dermatol. 2013 Aug;169(2):478-80.**

58. Memon MM, Raza SI, Basit S, Kousar R, Ahmad W, **Ansar M**. A novel WDR62 mutation causes primary microcephaly in a Pakistani family. **Molecular Biology Reports (2013), Jan;40(1):591-595.**

59. Mir H, Khan S, Arif MS, Ali G, Wali A, **Ansar M**, Ahmad W. Mutations in the gene phospholipase C, delta-1 (PLCD1) underlying hereditary leukonychia. **European Journal of Dermatology (2012) 22(6):736-739.**

60. Riazuddin S, Belyantseva IA, Giese AP, Lee K, Indzhykulian AA, Nandamuri SP, Yousaf R, Sinha GP, Lee S, Terrell D, Hegde RS, Ali RA, Anwar S, Andrade-Elizondo PB, Sirmaci A, Parise LV, Basit S, Wali A, Ayub M, **Ansar M**, Ahmad W, Khan SN, Akram J, Tekin M, Riazuddin S, Cook T, Buschbeck EK, Frolenkov GI, Leal SM, Friedman TB, Ahmed ZM. Alterations of the CIB2 calcium- and integrin-binding protein cause Usher syndrome type 1J and nonsyndromic deafness DFNB48. **Nat Genet. 2012 Sep 30;44(11):1265-71.**

61. Khan MA, Rafiq MA, Noor A, Hussain S, Flores JV, Rupp V, Vincent AK, Malli R, Ali G, Khan FS, Ishak GE, Doherty D, Weksberg R, Ayub M, Windpassinger C, Ibrahim S, Frye M, Ansar M, Vincent JB. Mutation in NSUN2, which encodes an RNA methyltransferase, causes autosomal-recessive intellectual disability. **Am J Hum Genet. 2012 May 4; 90(5):856-63.**

62. Lee K, Khan S, Islam A, **Ansar M**, Andrade PB, Kim S, Santos-Cortez RL, Ahmad W, Leal SM. Novel TMPRSS3 variants in Pakistani families with autosomal recessive nonsyndromic hearing impairment. **Clin Genet. 2012 Jul; 82(1):56-63.**

63. Khan S, Basit S, Zimri FK, Ali N, Ali G, **Ansar M**, Ahmad W. A novel homozygous missense mutation in it WNT10B in familial split-hand/foot malformation. **Clin Genet. 2012 Jul;82(1):48-55.**

64. Lee K, **Ansar M**, Andrade PB, Khan B, Santos-Cortez RL, Ahmad W, Leal SM. Novel CLDN14 mutations in Pakistani families with autosomal recessive non-syndromic hearing loss. **Am J Med Genet A. 2012 Feb; 158A(2):315-21.**

65. Umm-E-Kalsoom, Basit S, Kamran-Ul-Hassan Naqvi S, **Ansar M**, Ahmad W. Genetic mapping of an autosomal recessive postaxial polydactyly type A to chromosome 13q13.3-q21.2 and screening of the candidate genes. **Hum Genet. 2012 Mar;131(3):415-22.**

66. Saqib MA, Awan BM, Sarfraz M, Khan MN, Rashid S, **Ansar M**. Genetic analysis of four Pakistani families with achromatopsia and a novel S4 motif mutation of CNGA3. **Jpn J Ophthalmol. 2011 Nov;55(6):676-80.**

67. Ansar M, Lee K, Naqvi SK, Andrade PB, Basit S, Santos-Cortez RL, Ahmad W, Leal SM. A new autosomal recessive nonsyndromic hearing impairment locus DFNB96 on chromosome 1p36.31-p36.13. **J Hum Genet. 2011 Dec; 56(12):866-8.**

68. Kousar R, Hassan MJ, Khan B, Basit S, Mahmood S, Mir A, Ahmad W, **Ansar M**. Mutations in WDR62 gene in Pakistani families with autosomal recessive primary microcephaly. **BMC Neurol. 2011 Oct 1; 11(1):119.**

69. Rafiq MA, Kuss AW, Puettmann L, Noor A, Ramiah A, Ali G, Hu H, Kerio NA, Xiang Y, Garshasbi M, Khan MA, Ishak GE, Weksberg R, Ullmann R, Tzschach A, Kahrizi K, Mahmood K, Naeem F, Ayub M, Moremen KW, Vincent JB, Ropers HH, **Ansar M**, Najmabadi H. Mutations in the Alpha 1,2-Mannosidase Gene, MAN1B1, Cause autosomal-Recessive Intellectual Disability. **Am J Hum Genet 2011; 89(1):176-82.**

70. Ali G, Lee K, Andrade PB, Basit S, Santos-Cortez RL, Chen L, Jelani M, **Ansar M**, Ahmad W, Leal SM. Novel Autosomal Recessive Nonsyndromic Hearing Impairment Locus DFNB90 Maps to 7p22.1-p15.3. **Hum Hered. 2011;71(2):106-112.**

71. Khan S, Habib R, Mir H, Umm-E-Kalsoom, Naz G, Ayub M, Shafique S, Yamin T, Ali N, Basit S, Wasif N, Kamran-Ul-Hassan Naqvi S, Ali G, Wali A, **Ansar M**, Ahmad W. Mutations in the LPAR6 and LIPH genes underlie autosomal recessive hypotrichosis/woolly hair in 17 consanguineous families from Pakistan. **Clin Exp Dermatol. 2011 Aug;36(6):652-4.**

72. Sang L, Miller JJ, Corbit KC, Giles RH, Brauer MJ, Otto EA, Baye LM, Wen X, Scales SJ, Kwong M, Huntzicker EG, Sfakianos MK, Sandoval W, Bazan JF, Kulkarni P, Garcia-Gonzalo FR, Seol AD, O'Toole JF, Held S, Reutter HM, Lane WS, Rafiq MA, Noor A, **Ansar M**, Devi AR, Sheffield VC, Slusarski DC, Vincent JB, Doherty DA, Hildebrandt F, Reiter JF, Jackson PK. Mapping the NPHP-JBTS-MKS Protein Network Reveals Ciliopathy Disease Genes and Pathways. **Cell. 2011 May 13;145(4):513-28.**

73. Basit S, Iqbal Z, Umicevic-Mirkov M, Kamran Ul-Hassan Naqvi S, Coenen M, **Ansar M**, van Bokhoven H, Ahmad W. A novel deletion mutation in proteoglycan-4 underlies camptodactyly-arthropathy-coxa-vara-pericarditis syndrome in a consanguineous pakistani family. **Arch Med Res. 2011 Feb;42(2):110-4.**

74. Khan MA, Rafiq MA, Noor A, Ali N, Ali G, Vincent JB, **Ansar M**. A novel deletion mutation in the TUSC3 gene in a consanguineous Pakistani family with autosomal recessive nonsyndromic intellectual disability. **BMC Med Genet. 2011 Apr 22;12(1):56.**

75. Wasif N, Naqvi SK, Basit S, Ali N, **Ansar M**, Ahmad W. Novel mutations in the keratin-74 (KRT74) gene underlie autosomal dominant woolly hair/hypotrichosis in Pakistani families. **Hum Genet. 2011 129(4):419-424.**

76. Basit S, Lee K, Habib R, Chen L, Umm-E-Kalsoom, Santos-Cortez RL, Azeem Z, Andrade P, **Ansar M**, Ahmad W, Leal SM. DFNB89, a novel autosomal recessive nonsyndromic hearing impairment locus on chromosome 16q21-q23.2. **Hum Genet. 2011 129(4):379-385.**

77. 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. Loss-of-Function Mutations of ILDR1 Cause Autosomal-Recessive Hearing Impairment DFNB42. **Am J Hum Genet. 2011 Feb 11;88(2):127-37.**

78. 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, Van Camp G, Riazuddin S, Friedman TB, Riazuddin S. Functional Null Mutations of MSRB3 Encoding Methionine Sulfoxide Reductase Are Associated with Human Deafness DFNB74. **Am J Hum Genet. 2011 7;88(1):19-29. Impact Factor 10.6**

79. Nicholas AK, Khurshid M, Désir J, Carvalho OP, Cox JJ, Thornton G, Kausar R, **Ansar M**, Ahmad W, Verloes A, Passemard S, Misson JP, Lindsay S, Gergely F, Dobyns WB, Roberts E, Abramowicz M, Woods CG. WDR62 is associated with the spindle pole and is mutated in human microcephaly. **Nature Genetics 2010 Nov; 42 (11):1010-4.**

80. Rafiq MA, **Ansar M**, Marshall CR, Noor A, Shaheen N, Mowjoodi A, Khan MA, Ali G, Amin-Ud-Din M, Feuk L, Vincent JB, Scherer SW. Mapping of three novel loci for non-syndromic autosomal recessive mental retardation (NS-ARMR) in consanguineous families from Pakistan. **Clinical Genetics 2010; 78(5): 478-483.**

81. Kousar R, Nawaz H, Khurshid M, Ali G, Khan SU, Mir H, Ayub M, Wali A, Ali N, Jelani M, Basit S, Ahmad W, **Ansar M**. Mutation Analysis of the ASPM Gene in 18 Pakistani Families With Autosomal Recessive Primary Microcephaly. **Journal of Child Neurology 2010; 25(6) 715-720.**

82. Basit S, Ali G, Wasif N, **Ansar M**, Ahmad W. Genetic mapping of a novel hypotrichosis locus to chromosome 7p21.3-p22.3 in a Pakistani family and screening of the candidate genes. **Hum Genet 2010 Aug;128(2):213-20**.

83. Schraders M, Lee K, Oostrik J, Huygen PL, Ali G, Hoefsloot LH, Veltman JA, Cremers FP, Basit S, **Ansar M**, Cremers CW, Kunst HP, Ahmad W, Admiraal RJ, Leal SM, Kremer H. Homozygosity Mapping Reveals Mutations of GRXCR1 as a Cause of Autosomal-Recessive Nonsyndromic Hearing Impairment. **Am J Hum Genet. 2010 86(2):138-147.**

84. Kalsoom UE, Habib R, Khan B, Ali G, Ali N, **Ansar M**, Ahmad W. Mutations in lipase H gene underlie autosomal recessive hypotrichosis in five Pakistani families. **Acta Derm Venereol. 2010;90(1):93-94.**

85. Azeem Z, Naqvi SK, **Ansar M**, Wali A, Naveed AK, Ali G, Hassan MJ, Tariq M, Basit S, Ahmad W. Recurrent mutations in functionally-related EDA and EDAR genes underlie X-linked isolated hypodontia and autosomal recessive hypohidrotic ectodermal dysplasia. **Archives of Dermatology Research. 2009 Sep; 301(8):625-629.**

86. Schultz JM, Khan SN, Ahmed ZM, Riazuddin S, Waryah AM, Chhatre D, Starost MF, Ploplis B, Buckley S, Velásquez 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, Morell RJ. Noncoding mutations of HGF are associated with nonsyndromic hearing loss, DFNB39. **American Journal of Human Genetics. 2009 Jul; 85(1):25-39. Impact Factor 10.6**

87. Chishti MS, Lee K, McDonald ML, Hassan MJ, **Ansar M**, Ahmad W, Leal SM. Novel autosomal recessive non-syndromic hearing impairment locus (DFNB71) maps to chromosome 8p22-21.3. **Journal of Human Genetics (2009) Mar 54 (3): 141-144.**

88. Naz G, Khan B, Ali G, Azeem Z, Wali A, **Ansar M**, Ahmad W. Novel missense mutations in lipase H (LIPH) gene causing autosomal recessive hypotrichosis (LAH2). **Journal of Dermatological Science 54 (2009) 12–16.**

89. Basit S, Naqvi SK, Wasif N, Ali G, **Ansar M**, Ahmad W. A novel insertion mutation in the cartilage-derived morphogenetic protein-1 (CDMP1) gene underlies Grebe-type chondrodysplasia in a consanguineous Pakistani family. **BMC Med Genet. 2008 Nov 27; 9:102.**

90. Attya Bhatti, Lee K, McDonald ML, Hassan MJ, Gutala R, **Muhammad Ansar**, Wasim Ahmad, Leal SM. Mapping of a new autosomal recessive non-syndromic hearing impairment locus (DFNB45) to chromosome 1q43-q44. **Clin Genet. 2008 Apr 73 (4): 395-398.**

91. Hassan MJ, Mahmood S, Ali G, Bibi N, Waheed I, Rafiq MA, **Muhammad Ansar**, Wasim Ahmad. Intragenic deletions in the dystrophin gene in 211 Pakistani Duchenne muscular dystrophy patients. **Pediatr Int. 2008 Apr 50(2):162-166**.

92. Collin RW, Kalay E, Tariq M, Peters T, van der Zwaag B, Venselaar H, Oostrik J, Lee K, Ahmed ZM, Caylan R, Li Y, Spierenburg HA, Eyupoglu E, Heister A, Riazuddin S, Bahat E, **Ansar M**, Arslan S, Wollnik B, Brunner HG, Cremers CW, Karaguzel A, Ahmad W, Cremers FP, Vriend G, Friedman TB, Riazuddin S, Leal SM, Kremer H. Mutations of ESRRB encoding estrogen-related receptor beta cause autosomal-recessive nonsyndromic hearing impairment DFNB35**. American Journal of Human Genetics. 2008 Jan 82(1):125-138.**

93. Peter John, Muhammad Tariq, Muhammad Arshad Rafiq, Muhammad Amin-Ud-Din, Dost Muhammad, Ishrat Waheed, **Muhammad Ansar**, Wasim Ahmad. Recurrent intragenic deletion mutation in desmoglein 4 gene underlies autosomal recessive hypotrichosis in two Pakistani families of Balochi and Sindhi origins. **Archives of Dermatology Research 2006; 298(3):135-137.**

94. Abdul Wali, **Muhammad Ansar**, Muhammad Nasim Khan, W Ahmad. Atrichia with papular lesions resulting from a novel insertion mutation in the human hairless gene. **Clinical Experimental Dermatology 2006; 31(5):695-698.**

95. Sajid Malik, Amir Ali Abbasi, **Muhammad Ansar**, Wasim Ahmad, MC Koch, KH Grzeschik. Genetic Heterogeneity of synpolydactyly: a novel locus SPD3 maps to chromosome 14q11.2-q12. **Clinical Genetics 2006; 69(6):518-524.**

96. Asma Gul, Muhammad Jawad Hassan, Saqib Mahmood, Wenje C, Safa R, Muhammad Imran Naseer, Lisa D, Noor Muhammad, Muhammad Arshad Rafiq, **Muhammad Ansar**, Muhammad Salman Chishti, Ghazanfar Ali, Teepu Siddique, Wasim Ahmad. Genetic studies of autosomal recessive primary microcephaly in 33 Pakistani families: novel sequence variants in ASPM gene. **Neurogenetics 2006; 7 (2): 105-110.**

97. Muhammad Jawad Hassan, Santos RL, Muhammad Arshad Rafiq, Chahrour MH, Pham TL, Muhammad Wajid, Hijab N, Wambangco M, Lee K, **Muhammad Ansar**, Yan K, Wasim Ahmad, Leal SM. A novel autosomal recessive non-syndromic hearing impairment locus (DFNB47) maps to chromosome 2p25.1-p24.3. **Human Genetics 2006; 118:605–610.**

98. Santos RL, Muhammad Wajid, Muhammad Nasim Khan, McArthur N, Pham TL, Bhatti A, Lee K, Saba Irshad, Asif Mir, Yan K, Chahrour MH, **Muhammad Ansar**, Ahmad W, Leal SM. Novel sequence variants in the TMC1 gene in Pakistani families with autosomal recessive hearing impairment. **Human Mutation 2005; 26:396.**

99. Muhammad Aslam, Muhammad Wajid, Maria H. Chahrour, **Muhammad Ansar**, Sayedul Haque, Thanh L. Pham, Regie P. Santos, Kai Yan, Wasim Ahmad, Suzanne M. Leal. A novel autosomal recessive nonsyndromic hearing impairment locus (DFNB42) maps to chromosome 3q13.31-q22.3. **American Journal of Medical Genetics 2005; 133A, 18 – 22.**

100. Asif Mir, **Muhammad Ansar**, Maria H. Chahrour, Thanh L. Pham, Muhammad Wajid, Sayedul Haque, Kai Yan, Wasim Ahmad, Suzanne M. Leal. Mapping of a novel autosomal recessive nonsyndromic deafness locus (DFNB46) to

chromosome 18p11.32-p11.31. **American Journal of Medical Genetics 2005; 133A, 23 – 26.**

101. **Muhammad Ansar**, Maria Chahrour, Muhammad Amin Ud Din, Muhammad Arshad, Sayed-ul-Haque, Thanh L Pham, Kai Yan, Wasim Ahmad, Suzanne M Leal. DFNB44, a Novel Autosomal Recessive Non-Syndromic Hearing Impairment Locus, Maps to Chromosome 7p14.1-q11.22. **Human Heredity. 2004; 57:195-199.**

102. Muhammad Arshad Rafiq, **Muhammad Ansar**, Saqib Mahmood, Sayedul Haque, Muhammad Faiyaz ul Haque, Suzanne M Leal, Wasim Ahmad. A recurrent intragenic deletion mutation in DSG4 gene in three Pakistani families with autosomal recessive hypotrichosis. **Journal of Investigative Dermatology (2004) 123: 247 - 248**.

103. Muhammad Arshad Rafiq**, Muhammad Ansar**, Thanh L Pham, Mohammad Amin-ud-Din, Maqsood Anwar, Sayedul Haque, Maria Chahrour, Kai Yan, Suzanne M. Leal, Wasim Ahmad. Localization of a novel locus for hereditary nail dysplasia to chromosome 17q25.1-17q25.3. **Clinical Genetics (2004) 66: 73 - 78.**

104. **Muhammad Ansar**, Mohammad Amin-ud-Din, Muhammad Arshad, Muhammad Sohail, Mohammad Faiyaz ul Haque, Sayedul Haque, Wasim Ahmad, Suzanne M. Leal. A novel autosomal recessive non-syndromic deafness locus (DFNB35) maps to14q24.1-14q24.3 in large consanguineous kindred from Pakistan. **European Journal of Human Genetics (2003) 11: 77 - 80.**

105. **Muhammad Ansar**, Mohammad Ramzan, Thanh L. Pham, Kai Yan, Syed Muhammad Jamal, Sayedul Haque, Wasim Ahmad, Suzanne M. Leal. Localization of A Novel Autosomal Recessive Non-Syndromic Hearing Impairment Locus (DFNB38) to 6q26-q27 in a Consanguineous Kindred from Pakistan. **Human Heredity (2003) 55: 71 –74.**

106. Muhammad Arshad Rafique, **Muhammad Ansar**, Syed Muhammad Jamal, Muhammad Sohail, Mohammad Faiyaz-Ul-Haque, Sayedul Haque, Suzanne M. Leal, Wasim Ahmad. A Locus for Hereditary hypotrichosis localized to Human Chromosome 18q21.1. **European Journal of Human Genetics (2003) 11: 623 - 628.**

107. Muhammad Wajid, Amir Ali Abbasi, **Muhammad Ansar**, Thanh L. Pham, Kai Yan, Sayedul Haque, Wasim Ahmad, Suzanne M. Leal. DFNB39, a recessive form of sensorineural hearing impairment, maps to chromosome 7q11.22-q21.12. **European Journal of Human Genetics (2003) 11: 812–815.**