Employee No. 18544

1. Name: (first name) AKHTAR; (middle name) --; (surname) ALI

2. Designation: Assistant professor

3. Academic Qualifications:

Sr.	Degree	Institution	Year
1.	B.Sc.	Dr R.M.L. Awadh University, Faizabad, (U.P.), India	2001
2.	M.Sc.	Department of Zoology, Banaras Hindu University, Varanasi, India	2004
3.	Ph.D.	Department of Zoology, Banaras Hindu University, Varanasi, India	2010

4. Area of Specialization: (brief writeup, 200 words)

i. Clinical Molecular Genetics.
ii. Genetic Diagnostics.
iii. Identification of genetic and environmental risk factors for orofacial cleft disorders.
iv. Genomic analysis of congenital limb malformations.

5. Contact Information:

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6. Projects Undertaken as PI/ Co PI:

S. No	Name of the project	Duration	Source of Funding	Amount of Funding (Rs)
1.	Tran-differentiation of human umbilical cord tissue derived mesenchymal stem cells into neurogenic cell lineage: implication for hypoxic ischemic brain damage **Role: Co-I**	2016-2019	UGC	1000000.00
2.	Molecular Genetic Exploration of Congenital Limb Malformations and Limb Development **Role: PI**	2013-2016	SERB-DST	2250000.00
3.	Identification of the role of Bmpr1b gene in digit chondrogenesis **Role: PI**	2014-2015	UGC-UPE	200000.00
4.	Chromosomal and molecular genetic analysis of facial disorders **Role: PI**	2010-2011	UGC grant for new faculty under 11^{th} plan	270000.00

7. Awards/ Recognitions if any:

INVITED LECTURES

- 'Molecular Tools In Genetic Diagnosis: Recent Advancements'. Workshop on 'Maternal and Newborn Care : Issues and Challenges', March 01-07, 2017, Interdisciplinary School of Life Sciences (ISLS), B.H.U. Varanasi.
- Genomic analysis of congenital malformations. 10th Biennial conference of the Indo-Nepal association for peripheral nerve surgery (Nervecon-2014 and Lymphocon-2014). Institute of Medical Sciences, Banaras Hindu University and Apex Hospital, Varanasi. November 05-07, 2014.
- International Conference on Environmental Biology and Ecological Modeling (ICEBEM-2014), February 24-26, 2014 Visva-Bharati, Santiniketan – 731 235, India.
- Understanding Cleft Genetics. INDOCLEFTCON 2013, Nagpur Jan 17-19, 2013.
- Mapping genes and identification of risk factors for genetic disorders. Department of Drvyaguna, Faculty of Ayurveda, Institute of Medical Sciences, Banaras Hindu University, Varanasi, Jan 02, 2013.
- Genomics of Orofacial Cleft Disorders. Symposium on Population Genetics and Chromatin Dynamics. January 22-23, 2012, Banaras Hindu University, Varanasi.

WORKSHOP FACULTY

- Workshop on 'Maternal and Newborn Care : Issues and Challenges', March 01-07, 2017, Interdisciplinary School of Life Sciences (ISLS), B.H.U. Varanasi.
- Real-Time PCR. IBPRO-APRC Associate School of Neuroscience, Nov 2013, Banaras Hindu University, Varanasi.

8. List of 10 major Publications: (in order of importance)

1. Patel R, Tripathi FM, Singh SK, Rani A, Bhattacharya V, Ali A. (2014). A novel *GLI3*c.750delC truncation mutation in a multiplex Greig Cephalopolysyndactyly

Syndrome family with an unusual phenotypic combination in a patient. *Meta Gene* 2:880-887.

2. Patel R, Singh CB, Bhattacharya V, Singh SK, Ali A. (2016). GLI3 mutations in syndromic and non-syndromic polydactyly in two Indian families. Congenit Anom 56:94-97.
3. Patel R, Zenith RK, Chandra A, Ali A. (2017). Novel Mutations in the Crystallin Gene in Age-Related Cataract Patients from a North Indian Population. Mol Syndromol. 8:179-186.
4. Ali A, Singh SK, Raman R (2009). *MTHFR* 677TT alone and *IRF6* 820GG together with *MTHFR* 677CT, but not *MTHFR* A1298C, are risks for nonsyndromic cleft lip with or without cleft palate in an Indian population. *Genet Test Mol Biomarkers* 13:355-360.
5. Ali A, Singh SK, Raman R (2009). Coding Region of *IRF6* Gene may not be Causal for Van der Woude syndrome in Cases from India. *The Cleft Palate-Craniofac J* 46:541-544.
6. Kumari P, Ali A, Sukla KK, Singh SK, Raman R (2013). Lower incidence of nonsyndromic cleft lip with or without cleft palate in females: Is homocysteine a factor? *J Biosci* 38:21-6. Gupta S, Bhaskar PK, Bhardwaj R, Chandra A, Chaudhry VN, Chaudhry P, Ali A, Mukherjee A, Mutsuddi M (2014). *MTHFR* C677T predisposes to POAG but not to PACG in a North Indian population: a case control study. *PLoS ONE* 23;9:e103063.
7. Jaiswal SK, Kumar A, Ali A, Rai AK. (2015) A Male child with mosaic supernumerary marker characterised as isochromosome 18p by cytogenetic microarray having severe manifestations. *Gene* 559:94-98.
8. Jaiswal SK, Upadhyay A, Ali A, Upadhyay SK, Kumar A, Rai AK. (2016). Two Familial Cases of Robertsonian Transloacations 13; 14 and Its Clinical Consequences. J Genet Syndr Gene Ther 7: 283.
9. Keshri V, Prakash D, Kumar S, Maurya O P S, Chandra A, Ali A. (2015). Association of α-crystallin gene mutation with Age related Cataract J. Adv. Res. HealthCare & Med. Inform. 2: 5-11.
10. Singh CB, Patel R, Rani A, Singh SK, Ali A. (2017). Split-hand and foot malformation-3 (SHFM3). *J Sc Res BHU.* 61:103-108.

9. Additional Information/ Achievements:

Involved in the molecular diagnosis and genetic counselling of the referred cases from the University hospital, at the Centre for Genetic Disorders, Institute of Science, BHU.

10. Full List of Publications:

JOURNALS

1. Patel R, Singh CB, Rani A, Singh SK, Ali A. (2017). GLI3 mediated polydactyly: a review. *J Sc Res BHU.* 61:109-117.
2. Singh CB, Patel R, Rani A, Singh SK, Ali A. (2017). Split-hand and foot malformation-3 (SHFM3). *J Sc Res BHU.* 61:103-108.
3. Patel R, Zenith RK, Chandra A, Ali A. (2017). Novel Mutations in the Crystallin Gene in Age-Related Cataract Patients from a North Indian Population. Mol Syndromol. 8:179-186.
4. Jaiswal SK, Upadhyay A, Ali A, Upadhyay SK, Kumar A, Rai AK. (2016). Two Familial Cases of Robertsonian Transloacations 13; 14 and Its Clinical Consequences. J Genet Syndr Gene Ther 7: 283.
5. Patel R, Singh CB, Bhattacharya V, Singh SK, Ali A. (2016). GLI3 mutations in syndromic and non-syndromic polydactyly in two Indian families. Congenit Anom 56:94-97.

6. Keshri V, Prakash D, Kumar S, Maurya O P S, Chandra A, Ali A. (2015). Association of α-crystallin gene mutation with Age related Cataract J. Adv. Res. HealthCare & Med. Inform. 2: 5-11.
7. Jaiswal SK, Kumar A, Ali A, Rai AK. (2015) A Male child with mosaic supernumerary marker characterised as isochromosome 18p by cytogenetic microarray having severe manifestations. *Gene* 559:94-98.
8. Patel R, Tripathi FM, Singh SK, Rani A, Bhattacharya V, Ali A. (2014). A novel *GLI3*c.750delC truncation mutation in a multiplex Greig Cephalopolysyndactyly Syndrome family with an unusual phenotypic combination in a patient. *Meta Gene* 2:880-887.
9. Gupta A, Tewari P, Agrawal NK, Patel R, Ali A, Byadgi PS. (2014) Kapha and Kapha-Pittaja type of body constitution and *MTHFR* C677T are strong risks for Type2 Diabetes Mellitus. *Ind J Hum Genet* (*in press*).
10. Rani A , Ali A, Rai AK. (2014). Globozoospermia –A Rare Cause of Male Infertility. *Ind J Res* 8:9-11.
11. Gupta S, Bhaskar PK, Bhardwaj R, Chandra A, Chaudhry VN, Chaudhry P, Ali A, Mukherjee A, Mutsuddi M (2014). *MTHFR* C677T predisposes to POAG but not to PACG in a North Indian population: a case control study. *PLoS ONE* 23;9:e103063.
12. Kumari P, Ali A, Sukla KK, Singh SK, Raman R (2013). Lower incidence of nonsyndromic cleft lip with or without cleft palate in females: Is homocysteine a factor? *J Biosci* 38:21-6.
13. Ali A, Singh SK, Raman R (2009). *MTHFR* 677TT alone and *IRF6* 820GG together with *MTHFR* 677CT, but not *MTHFR* A1298C, are risks for nonsyndromic cleft lip with or without cleft palate in an Indian population. *Genet Test Mol Biomarkers* 13:355-360.
14. Ali A, Singh SK, Raman R (2009). Coding Region of *IRF6* Gene may not be Causal for Van der Woude syndrome in Cases from India. *The Cleft Palate-Craniofac J* 46:541-544.
15. Mishra V, Siddhartha, Rai M, Tilak V, Rai AK, Ali A. Status of BCR-ABL fusion transcripts in an Indian population. (In press).
16. Kumari P, Ali A. Singh SK, Chaurasia A, *Raman R*: Genetic heterogeneity in Van der Woude Syndrome: Identification of *NOL4* and *IRF6* haplotype from the noncoding regions as candidates in two families. *J. Genet.* (under revision).

BOOK CHAPTERS:

1. Dubey PK, Tripathi A, Ali A. Assisted Reproductive Techniques in Infertility Treatment: Opportunities and Challenges. In: R. Singh, K. Singh, (eds.). *Male Infertility: Understanding, Causes and Treatment*. 1st ed. Springer Nature; (*In press*). (DOI 10.1007/978-981-10-4017-7_27).

CONFERENCE PROCEEDINGS/ABSTRACTSPUBLISHED IN JOURNALS:

1. Ali A. Molecular Tools In Genetic Diagnosis: Recent Advancements'. Workshop on 'Maternal and Newborn Care : Issues and Challenges', March 01-07, 2017, Interdisciplinary School of Life Sciences (ISLS), B.H.U. Varanasi.
2. Singh CB, Mishra V, Samrat S, Patel R, Rai M, Tilak V, Ali A. Importance of rare BCR-ABL fusion transcript analysis in Chronic myeloid leukaemia. International conference on Updates in Cancer Prevention and Research (ICUCPR-2017), Babasaheb Bhimrao Ambedkar University, Lucknow, February 14-16, 2017.
3. Singh CB, Mishra V, Samrat S, Patel R, Ali A. A comprehensive review on BCR-ABL fusion transcripts in chronic myeloid leukemia. International conference on Updates in Cancer Prevention and Research (ICUCPR-2017), Babasaheb Bhimrao Ambedkar University, Lucknow, February 14-16, 2017.

4. Singh CB, Mishra V, Samrat S, Patel R, Ali A. International Symposium on Integrative Physiology and Comparative Endocrinology (ISIPCE-2016) at Department of Zoology, Institute of Science, Banaras Hindu University, Varanasi, February 12-14, 2016
5. 5th Annual Conference of Molecular Pathology Association of India (MPAI 2016): international symposium on 'Integration of Genetics & Genomics in Laboratory Medicine' at department of Hematology, PGIMER, Chandigarh, March 12-13, 2016.
6. Gupta P, Chaturvedi TP, Pratap CB, Tapadia M, Ali A. Evaluation of MSX1 gene for malocclusion. 8th International Orthodontic Congress 2015. 27-30 September 2015, London.
7. Singh CB, Ali A. Regulatory role of SALL1 and BMPR1B in limb development. UGC-UPE mini symposium 28 feb 2015, BHU, Varanasi.
8. Singh CB, Ali A. BMPR1B Signalling in HEK293 Cells. ISHG-2015: International Symposium on 'Genomics in Health and Disease' & 40th Annual Conference of Indian Society in Human Genetics at National Institute of Immunohaematology (ICMR), Mumbai. January 28-30, 2015.
9. Patel R, Bhattacharya V, Singh SK, Ali A. Syndromic and non-syndromic polydactyly presented at 40th Annual Conference of the Indian Society of Human Genetics: Genomics in Health & disease organized by National Institute of Immunohaematology (ICMR), Mumbai, on 28th – 30th January 2015. Indian Journal of human genetics (2014) 1:20 S69.
10. Pandey H, Rai AK, Das P, Ali A. An update on Duchenne Muscular Dystrophy. Indian society of human genetics meeting-2014.
11. Gupta V, Yadav AK, Ali A, Rai AK, Das P. Dermatoglyphic Patterns Study in Morton's Toe/Royal Toe. Indian society of human genetics meeting-2014.
12. Singh CB, Mishra V, Samrat S, Rai M, Tilak V, Rai AK and Ali A. Prevalence of BCR-ABL fusion transcripts in Chronic Myeloid Leukemia patients in Eastern Uttar Pradesh. XXXVIII All India Cell Biology Conference and International Symposium on 'Cellular Response to Drug' at Central Drug Research Institute, Lucknow (Decemb er 10-12, 2014) .
13. Ali A. Genomic analysis of congenital malformations. 10th Biennial conference of the Indo-Nepal association for peripheral nerve surgery (Nervecon-2014 and Lymphocon-2014). Institute of Medical Sciences, Banaras Hindu University and Apex Hospital, Varanasi. November 05-07, 2014.
14. Ali A Patel R, Jaiswal SK, Singh SK, Raman R. Discontinuous microdeletion at 1p13.3 involving *NBPF4* but not *ALX3* in a patient with severe frontonasal dysplasia. 64th Annual meeting of American Society of Human Genetics, (ASHG 2014), October 17-22, 2014, San Diego, California, USA.
15. Kumari P, Ali A, Singh SK, Raman R. Genetic heterogeneity in Van der Woude syndrome. 64th Annual meeting of American Society of Human Genetics, (ASHG 2014), October 17-22, 2014, San Diego, California, USA.
16. Byadgi PS, Gupta A, Tewari P, Ali A "Kapha and Kapha-Pittaja type of body constitution and MTHFR 677CC are strong risks for Type2 Diabetes Mellitus. 16th International Ayurveda symposium and Research Seminar (IRSA 2014), September 12-14, 2014, European Academy for Ayurveda, Birstein, Germany.
17. Ali A. Gene-Environmental Interactions as the Causes for Orofacial Cleft Disorders. Invited lecture abstract in: International Conference on Environmental Biology and Ecological Modelling (ICEBEM-2014), February 24-26, 2014 Visva-Bharati, Santiniketan – 731 235, India.
18. Patel R, Ali A. 'Mapping genes for Split-Hand/Foot malformation (SHFM3)'. Abstract in: International Symposium on Karyotype to Haplotype and Beyond. Banaras Hindu University, Varanasi, December 08-10, 2013.

19. Ali A, Singh SK, Murthy J. INDOCLEFTCON-2013, Understanding Cleft Genetics. Nagpur, Jan 17-19, 2013.

20. Patel R, Ali A. Mutational analysis of SALL1 gene and associated disorders. International symposium on developmental and complex disorders, and 38th annual conference of the Indian society of human genetics, Banaras Hindu University, Dec 09-11, 2012; pp 98.

21. Rani A, Ali A, Rai A. Absent acrosome in all sperms- a rare cause of infertility. International symposium on developmental and complex disorders, and 38th annual conference of the Indian society of human genetics, Banaras Hindu University, Dec 09-11, 2012; pp 102.

22. Kumari P, Ali A, Singh SK, Raman R. Mapping loci involved in a van der Woude syndrome affected family. International symposium on developmental and complex disorders, and 38th annual conference of the Indian society of human genetics, Banaras Hindu University, Dec 09-11, 2012; pp 94.

23. Singh S, Lata S, Tiwari KN, Ali A, Upadhyay R. Protective effects of aqueous extracts of phyllanthus fraternus on cyclophosphamide induced DNA damage in spermatozoa of mice. International symposium on developmental and complex disorders, and 38th annual conference of the Indian society of human genetics, Banaras Hindu University, Dec 09-11, 2012; pp106.

24. Gupta A, Byadgi PS, Tewari P, Ali A. Association of MTHFR C677T, A1298C and CF7L2-IVS3 CT with prakriti and risk for type2 diabetes mellitus. Banaras Hindu University, International symposium on developmental and complex disorders, and 38th annual conference of the Indian society of human genetics, Dec 09-11, 2012; pp110.

25. Keshari VK, Chaurasia N, Chandra A, Maurya OPS, Ali A. Genetic analysis of patients with cataract. International symposium on developmental and complex disorders, and 38th annual conference of the Indian society of human genetics, Banaras Hindu University, Dec 09-11, 2012; pp 117.

26. Ali A. Genome-wide linkage scan for split-hand/foot malformation in an Indian family reveals a locus within 433kb at 10q25.1-q25.3. International conference on genes, genetics and genomics: today and tomorrow-human concerns, and XXXVII annual conference of the Indian society of human genetics, Panjab University, Chandigarh, March 03-05, 2012; pp 103.

27. Ali A, Raman R. Genome-wide Linkage Scan in an Indian Family with Split-hand/foot Malformation narrowed down the SHFM3 region to 10q25.1-q25.3. American Society of Human Genetics, Montreal, Canada, October 11-15, 2011.

28. Bothra A, Paul D, Singh N, Tiwari SK, Poudel SB. Rai AK, Ali A, Singh K. Cytogenetic Analysis of Human Genetic Disorders. International conference on Genomics, Genetic Diseases and Diagnostics, and XXXVI Annual Conference of the Indian Society of Human Genetics, February 14-16, 2011.

29. Chandra A, Dixit A, Bhardwaj R, Maurya OPS, Mutsuddi M, Ali A. Association of *MTHFR* C677T polymorphism with primary open angle glaucoma in north Indian population. 69th Annual Conference of All India Ophthalmological Society, February 3-6, 2011, Gujrat University Convention Centre, Ahmedabad.

30. Kumari P, Ali A, Singh SK, Raman R. Association of Homocysteine and Homocysteine pathway genes (RFC1, MTHFR) with NSCL/P in a population from Eastern India. International symposium on genetic and epigenetic basis of complex diseases. CCMB, Hyderabad, Dec 5-7, 2009.

31. Ali A, Singh SK, Raman R. Plasma Homocysteine Concentration and MTHFRC677T and A1298C mutations in the etiology of Nonsyndromic Cleft Lip with or Without Cleft Palate. 34th Annual Conference of the Indian Society of Human Genetics, March 2009, New Delhi, pp 124.

32. Ali A, Singh SK, Raman R. Analysis of IRF6 gene in Van der Woude syndrome from an Indian population. 58th Annual Meeting of American Society of Human Genetics, 2008, pp 431.
Also In: Ali A, Singh SK, Raman R. Analysis of IRF6 gene in Van der Woude syndrome from an Indian population. (Abstract) Am. J. Hum. Genet. (suppl.): A, 2008.

33. Ali A, Singh SK, Raman R. Novel variants IVS1+3900 A>G, IVS6+27 C>G and 263 T>C in non-coding regions of *Interferon Regulatory Factor 6 gene in* Van der Woude syndrome patients from Indian population. XXXI All India Cell Biology Conference & Symposium on "Stem Cells: Application and Prospects", Dec 14-16, 2007, BHU, Varanasi.

34. Ali A, Singh SK, Raman R. *MTHFR* 677TT and *MTHFR* 677 CT combined with *IRF6* 820GG are risk factors for nonsyndromic cleft lip with or without cleft palate. XXXII Annual Conference of Indian Society of Human Genetics and International Symposium on Deconstructing Human Diseases: The Genomic Advantage, 2007, IICB, Kolkata.

35. Ali A, Singh SK, Raman R. Association of G820A of *IRF6* gene with Nonsyndromic cleft lip with or without cleft palate in Indian population: A cases-control study. "International symposium on Human Genomics and Public Health" and "XXXI Annual Conference of Indian Society of Human Genetics, 2006, JNU, New Delhi.

36. Ali A. "Donate Eyes: Give Life" Poster Competition, 16th Oct. 2004, Department of Ophthalmology, IMS, BHU.