Employee No. 18514

1. Name: (first name) Amit (middle name) Kumar (surname) Rai

2. Designation: Assistant professor

3. Academic Qualifications:

Sr.	Degree	Institution	Year
	B.Sc	Banaras Hindu University, Varanasi, India	1994
	M.Sc.	Banaras Hindu University, Varanasi, India	1996
	Ph.D.	Banaras Hindu University, Varanasi, India	2004

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

i. Human Cytogenetics.
ii. Human genotype and phenotype correlations.
iii. Molecular characterizations of chromosomal disorders in humans.

5. Contact Information:

Centre for Genetic Disorders, Institute of Science,
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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.	Genome scan of congenital defects **Role: PI**	2013	DST-PURSE, BHU	50000.00
2.	Financial support **Role: PI**	2016	DST-PURSE, BHU	100000.00
3.	Financial support **Role: PI**	2015	BHU	200000.00
4.	Chromosomal and Molecular Genetic analysis of Facial Disorders **Role: PI**	2010-2013	XI Plan Research Grant for New Faculty, UGC-BHU	270000.00

7. Awards/ Recognitions if any: Nil.

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

i. **Rai AK**, Singh S, Mehta S, Kumar A, Pandey LK, Raman R. *MTHFR* C677T and A1298C polymorphisms are risk factors for Down's syndrome in Indian mothers. J Hum Genet, 2006, 51:278–283. **IF 2.462**

ii. Sukla KK, Jaiswal SK, **Rai AK**, Mishra OP, Gupta V, Kumar A, and Raman R. Role of folate-homocysteine pathway gene polymorphisms and nutritional cofactors in Down syndrome: A triad study. Hum. Reprod., 2015, 30:1982-1993. **IF 4.569**

iii. Jaiswal SK, Sukla KK, A Chauhan, Lakhotia AR, A Kumar, **Rai AK**. Choline metabolic pathway gene polymorphisms and risk for Down syndrome: An association study in a population with folate-homocysteine metabolic impairment. European Journal of Clinical Nutrition, 2016, 1–6. (accepted for publication on August 26, 2016). **IF 2.935**

iv. Jaiswal SK, Sukla KK, Mishra SK, Lakhotia AR, Kumar A, **Rai AK**. Association of Genetic Polymorphisms in Genes Involved at the Branch Point of Nucleotide Biosynthesis and Remethylation with Down Syndrome Birth Risk: A Case-Control Study. J Mol Genet Med, 2016, 10:1-9. **IF 1.2**

v. Pooja Chauhan, Jaiswal SK, Lakhotia AR, **Rai AK**. Molecular cytogenetic characterization of two Turner syndrome patients with mosaic ring X chromosome. J Assist Reprod Genet, 2016, 33:1161–1168. (accepted for publication on June 20, 2016). **IF 1.858**

vi. Jaiswal SK, Sukla KK, Kumari N, Lakhotia AR, Kumar A, **Rai AK**. Maternal risk for Down syndrome and polymorphisms in the Promoter Region of *DNMT3B* gene: A case – control study. Birth defect research part A: Clinical and Molecular Teratology, 2015, 103:298–304. **IF 2.211**

vii. Jaiswal SK, Kumar A, A. Ali, **Rai AK**. Co-occurrence of mosaic supernumerary isochromosome 18p and intermittent 2q13 deletions in a child with multiple congenital anomalies. GENE, 2015, 559:94–98. **IF 2.138**

viii. Jaiswal SK, Sukla KK, Gupta V, **Rai AK**. Overlap of Patau syndrome and Pierre Robin syndrome along with abnormal metabolism: An interesting case. Journal of Genetics, 2014, 93:865–868. **IF 1.093**

ix. Jaiswal SK, Upadhyay A, Upadhyay SK, Kumar A, **Rai AK**. Two familial cases of Robertsonian transloacations 13;14 and its clinical consequences. Journal of Genetic Syndromes & Gene Therapy, 2015, 6:1-4. **IF 1.808**

x. Gupta V, Sandeep H S, **Rai A**, Shukla J. Functional and radiological assessment of arthropathy in Indian children with haemophilia. Haemophilia, 2013, 19:e358--e396. **IF 2.603**

9. Additional Information/ Achievements:

1. Postdoctoral Fellow **(**July-August, 2007**)** Department of Pathology, Columbia Medical School, Columbia University, New York-10032 USA

2. Postdoctoral Fellow **(**January-June, 2007**)** Cytokine Research Laboratory (Unit 143), Department of Experimental Therapeutics, UT M.D. Anderson Cancer Center, 1515 Holcombe Boulevard, Houston, Texas 77031 USA

3. Research Associate (RA) as three years in India as RA (December, 2003-October, 2006)

4. Member of Organizing committee, International Symposium on developmental and complex Disorders and 38th Annual conference of Indian Society of Human Genetics: Genomics and Community Health, December 9-11, 2012, Dept of Zoology, BHU

5. Member of Organizing committee, International symposium on Karyotype to Haplotype and beyond, December 8-10, 2013, Dept of Zoology, BHU

10. Full List of Publications:

1. Jaiswal SK, Sukla KK, A Chauhan, Lakhotia AR, A Kumar, **Rai AK**. Choline metabolic pathway gene polymorphisms and risk for Down syndrome: An association study in a population with folate-homocysteine metabolic impairment. European Journal of Clinical Nutrition, 2016, 1–6. (accepted for publication on August 26, 2016). **IF 2.935**
2. Pooja Chauhan, Jaiswal SK, Lakhotia AR, **Rai AK**. Molecular cytogenetic characterization of two Turner syndrome patients with mosaic ring X chromosome. J Assist Reprod Genet, 2016, 33:1161–1168. (accepted for publication on June 20, 2016). **IF 1.858**
3. Jaiswal SK, Sukla KK, Mishra SK, Lakhotia AR, Kumar A, **Rai AK**. Association of Genetic Polymorphisms in Genes Involved at the Branch Point of Nucleotide Biosynthesis and Remethylation with Down Syndrome Birth Risk: A Case-Control Study. J Mol Genet Med, 2016, 10:1-9. **IF 1.2**
4. Jaiswal SK, Upadhyay A, Upadhyay SK, Kumar A, **Rai AK**. Two familial cases of Robertsonian transloacations 13;14 and its clinical consequences. Journal of Genetic Syndromes & Gene Therapy, 2015, 6:1-4. **IF 1.808**
5. Karthickeyan SMK, **Rai AK**, Tirumurugaan KG, Kanakaraj P. Comparative fluorescence in situ hybridisation (FISH) mapping of class I and II MHC genes on Bos indicus and Bubalus bubalis chromosomes. The Indian Journal of Animal Sciences, 2015, 85. (published on Aug 01, 2015). **IF 0.160**
6. Sukla KK, Jaiswal SK, **Rai AK**, Mishra OP, Gupta V, Kumar A, and Raman R. Role of folate-homocysteine pathway gene polymorphisms and nutritional cofactors in Down syndrome: A triad study. Hum. Reprod., 2015, 30:1982-1993. **IF 4.569**
7. Jaiswal SK, Kumar A, A. Ali, **Rai AK**. Co-occurrence of mosaic supernumerary isochromosome 18p and intermittent 2q13 deletions in a child with multiple congenital anomalies. GENE, 2015, 559:94**–**98. **IF 2.138**
8. Jaiswal SK, Sukla KK, Kumari N, Lakhotia AR, Kumar A, **Rai AK**. Maternal risk for Down syndrome and polymorphisms in the Promoter Region of *DNMT3B* gene: A case – control study. Birth defect research part A: Clinical and Molecular Teratology, 2015, 103:298–304. **IF 2.211**
9. S Das, V Tilak, V Gupta, A Singh, M Kumar, **A Rai**. Clinical, hematological, and cytogenetic profile of aplastic anemia. The Egyptian Journal of Haematology, 2015, 40:3-10.
10. Jaiswal SK, Sukla KK, Gupta V, **Rai AK**. Overlap of Patau syndrome and Pierre Robin syndrome along with abnormal metabolism: An interesting case. Journal of Genetics, 2014, 93:865–868. **IF 1.093**
11. Gupta V, Sandeep H S, **Rai A**, Shukla J. Functional and radiological assessment of arthropathy in Indian children with haemophilia. Haemophilia, 2013, 19:e358--e396. **IF 2.603**

12. Srivasatva L, Chandel IS, Rai A K, Rastogi A and Srivastava P. Optimization studies on the growth of human chondrocyte. Indian Journal of Biotechnology, 2013, 12:483-488. **IF 0.386**
13. Chandra A, Rasal A, Maurya OP Singh, Prakash D, **Rai A K**. Human Limbal Stem cell Culture: In-vitro study. UP Journal of Ophthalmology, 2011, 01:10-20.
14. Chandra A, Singh M K, Singh V P, **Rai A K**, Maurya OP Singh. A live cysticercosis in anterior chamber leading to glaucoma secondary to pupilary block: Case report. Journal of Glaucoma, 2007, 16(2):271-273. **IF 1.865**
15. Tilak V, Rai M, Singh V.P, **Rai AK**, Rajiva Raman. Hepatocellular carcinoma presenting as leukemoid reaction-A rare entity. Journal of Indian Medical Association, 2007, 105:462-5.
16. KARTHICKEYAN S M K, **RAI A K**, KANAKARAJ P: Fluorescent in situ hybridization of pBOLA, DRA and DRB3 genes to Buffalow (Bubalus bubalis) chromosome by tyaramide signal amplification. Buffalow journal, 2007, 23:127-132.
17. KARTHICKEYAN S M K, **RAI A K**, TIRUMURUGAAN K G, KANAKARAJ P (Tamil Nadu Veterinary and Animal Sciences Univ, Chennai, Tamil Nadu-600 007): **Localization of MHC class I and II genes in zebu and crossbred cattle**. Indian J Anim Sci, 2007, 77:625-6. **IF 0.160**
18. **Rai AK**, Singh S, Mehta S, Kumar A, Pandey LK, Raman R. *MTHFR* C677T and A1298C polymorphisms are risk factors for Down's syndrome in Indian mothers. J Hum Genet, 2006, 51:278–283. **IF 2.462**

CONFERENCE PROCEEDINGS/ABSTRACTSPUBLISHED IN JOURNALS:

1. Pooja Chauhan, Sushil Kumar jaiswal, **Amit Kumar Rai**. Polymorphism rs6166 in Follicular Stimulating Hormone Receptor associated as a risk factor for primary amenorrhoea. Abstract, International symposium on "Integration of genetics & genomics in laboratory medicine" (MPAI 2015) March, 12-13, 2016, PGIMER, Chandigarh, India
2. Pooja Chauhan, **Amit Kumar Rai.** A novel phenotype in Complete Androgen Insensitivity Syndrome due to ΔPhe583 in Androgen Receptor gene. Abstract, International symposium on " Genomics in Health and Disease" (ISHG 2015), January, 28-30, 2015, NIIH (ICMR), Mumbai, India
3. Sushil Kumar Jaiswal, Anjali Rani Lakhotia, Ashok Kumar, **Amit Kumar Rai**. *Thymidylate synthase* gene polymorphisms in 5'- and 3-untranslated region associated with predisposition of mothers for Down syndrome child birth in north India: a case–control analysis. Abstract, December 10-12, 2014, XXXVIII AICBC, CDRI, Lucknow, UP, India
4. 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. Indian Society of Cell Biology (ISCB 2014), Dec 10-12, 2014, Lucknow, India.
5. Sushil Kumar Jaiswal, Krishna Kishore Sukla, Neha Kumari, Anjali Rani Lakhotia, Ashok Kumar, **Amit Kumar Rai**. Promoter polymorphisms of *DNMT3B* gene and risk for Down syndrome: A case – control study. Abstract, International Symposium on Genetic Analysis Translational and Developmental and Annual Meeting of Society for Biotechnologist (India), November 20-22, 2014, WU, Ward wan, West Bengal, India
6. Jaiswal SK, Kumar A, Gupta V, Rani A, **Rai AK**. Maternal gene polymorphisms of folate metabolism as genetic risk factor for Down syndrome in North Indian population. Published in Molecular Cytogenetics 2014, 7(Suppl 1):P120. **IF. 2.140**)

7. Sushil Kumar Jaiswal, Ashok Kumar, Vineeta Gupta and **Amit Kumar Rai**. A rare case of trisomy chromosome 18p due to supernumerary marker chromosome. Abstract, Haplotype to karyotype and beyond: December, 08-10, 2013, BHU, Varanasi, UP, India
8. Pooja Chauhan, Sushil Kumar Jaiswal, **Amit Kumar Rai.** X-Chromosome deletion mapping in Turner syndrome patients with ring chromosome. Abstract, Haplotype to karyotype and beyond: December, 08-10, 2013, BHU, Varanasi, UP, India
9. Sushil Kumar Jaiswal, Ashok Kumar, Vineeta Gupta and **Amit Kumar Rai.** Unusual Cases of Chromosomal Disorder diagnosed by Cytogenetics Microarray. Abstract, 38th ISHG 2013, BHU, Varanasi, UP, India
10. Sushil Kumar Jaiswal, Ashok Kumar, **Amit Kumar Rai.** Identification of chromosomal subtypes in Down syndrome by using molecular Cytogenetics. Abstract, 37th ISHG March 03-05, 2012, Punjab University, Chandigargh, Punjab, India
11. Abhishek Chandra **Amit Kumar Rai,** Om Prakash Maurya, Rajiva Raman (2007) Limbal Cells Transplantation in Cornea: A BHU Experience. Abstract, XXXI All India Cell Biology Conference and Symposium on Stem Cells: Application and Prospects (December 14-16, 2007), Centre of Advanced Study, Department of Zoology, Banaras Hindu University, Varanasi-221 005, India
12. Attended Indo-US Symposium on Genetic Disorders, Focus on Hemoglobinopathies (October 29-31 2006), Banaras Hindu University, Varanasi, Uttar Pradesh, India
13. **Amit Kumar Rai**, Satya Singh, Rajiva Raman (2006) Occurrence of Down Syndrome and neural tube defects in the same family: identification of cases of folic acid resistance. Abstract-101, Pp-80, International Symposium and XXX India Society of Human Genetics Conference, NCAHG, School of Life Sciences, Jawaharlal Nehru University, New Delhi 110067, India
14. **Amit Kumar Rai** and Rajiva Raman (2006) Cytogenetic and molecular analyses of cases from eastern Uttar Pradesh and adjoining areas (2002-2005). Abstract-133, Pp-98, International Symposium and XXX India Society of Human Genetics Conference, NCAHG, School of Life Sciences, Jawaharlal Nehru University, New Delhi 110067, India
15. **Amit Kumar Rai** and Rajiva Raman (2002) Parental Origin and Meiotic Stage of Nondisjunction in Down Syndrome Patients in Adjoining Areas of Varanasi. Abstract-O2, Pp-50, XXVI All India Cell Biology Conference and Symposium, ACTREC, Cancer Research Institute, Navi Mumbai, India