

## DR. SWARKAR SHARMA

Professor (Associate)

निदेशक / Director

आण्विक जीवविज्ञान केंद्र / Centre for Molecular Biology

जम्मू केंद्रीय विश्वविद्यालय / Central University of Jammu

बागला-सुचानी, सांबा / Bagla-Suchani, Samba

जम्मू और कश्मीर, भारत-181143 / Jammu and Kashmir,

Phone: +91-9419955636, [swarkar.sharma@gmail.com](mailto:swarkar.sharma@gmail.com), [swarkar.molb@cuammu.ac.in](mailto:swarkar.molb@cuammu.ac.in)

ORCID: 0000-0001-8857-5906, ResearcherID: B-8096-2008, Scopus Author ID: 55675277600

**Research Areas:** Human Genetics, Genomics, Molecular Genetics and Diagnostics, Medical Genetics  
**Research and Teaching Experience (Post PhD):** 16 years

### Research Highlights (details at [www.jkdna.in](http://www.jkdna.in))

- My PhD thesis work [2003-2008] depicted the role of mtDNA and Human evolutionary history in developing Type 2 diabetes.
- In Postdoc [2008-2012], worked to understand the Genetics of Adolescent Idiopathic Scoliosis (AIS) in Caucasian populations exploiting Genome Wide Association studies (GWAS) and Next Generation Sequencing (NGS) methodologies and for the first time, implicated Neurological pathways are involved in etiology of AIS. In addition, found many candidate genes for AIS.
- The first discovery of a Gene and respective mutations responsible for causing the rare disorder called "Osteofibrous Dysplasia" exploiting next-generation sequencing. Further evaluated functional consequences of the variations and elucidated the role of genes in the disease.
- Using Next Generation Sequencing Technologies (Whole Exome Sequencing) characterised more than five decades old, sporadic severe skeletal disorder as Progressive Pseudorheumatoid Dysplasia and also identified the gene mutations causing it in a village in Jammu and Kashmir, India.
- I am working to understand better the structure of the populations with futuristic aims to develop tools for disease risk predictions and personalised medicine. Project JKDNA with the main goal of aggregating and harmonising Genomic data from the population of J&K, generated through candidate gene study approaches, high throughput methodologies for Genome-wide Association Studies (GWAS), genome sequencing projects (Next Generation Sequencing), published resources or public datasets, and making open data available for the use of wider scientific community, especially researchers in J&K and India. In addition, developing point-of-care molecular diagnostics assays.
- The future goals are: 1. to work on various populations of India; 2. Explore the metabolic health of Populations 3. Point of care devices and assays for human diseases.

### Employment Details: Position and Employment

| Sr.No. | Institution/Place   | Designation                | From Date | To Date       |
|--------|---|----------------------------|-----------|---------------|
| 1      | Texas Scottish Rite Hospital, Dallas, USA & UT Southwestern Medical Center, Dallas, Tx, USA | PostDoc Research Associate | May 2008  | November 2012 |

|    |                                   |                     |            |            |
|----|-----------------------------------|---------------------|------------|------------|
| 2  | Shri Mata Vaishno Devi University | Assistant Professor | Nov 2012   | April 2023 |
| 3. | Central University of Jammu       | Associate Professor | April 2023 | Present    |

### Education

|              |   |            |
|--------------|---|------------|
| <b>Ph.D.</b> | Guru Nanak Dev University, Amritsar; Human Genetics<br>Dissertation: "Genetic Diversity and Screening of Type 2 Diabetes Susceptibility in Different Population Groups of North West India" | March 2008 |
| <b>M.Sc.</b> | Guru Nanak Dev University, Amritsar; Human Genetics   | April 2002 |
| <b>B.Sc.</b> | University of Delhi; Zoology Hons.  | April 2000 |

### Awards/Honors Details

1. John S. Appelton Spine Research Award 2009 by Texas Scottish Rite Hospital, Dallas, USA
2. Russel A. Hibbs Basic Science Award 2012, by Scoliosis Research Society, USA
3. Enlisted amongst Leading Health Professionals of the World (2014) by the International Biographical Centre, Cambridge, UK for contribution to Medical Genetics.
4. National Leadership Awards 2020: Best Entrepreneurial Empowerment by Confederation of Indian Micro, Small and Medium Enterprises (CIMSME)

### Projects:

| S.no | Title of the Project  | Funding Agency   | Amount           | Duration |
|------|---|--|------------------|----------|
| 1.   | Analyses of Modern Human DNA to understand genomic formation of population groups from J&K-India. | National Geographic Society, USA                           | \$30,000 (USD)   | 2 years  |
| 2.   | Whole Genome genotyping of Himalayan populations in North Western India                           | New York University in Abu Dhabi, Dubai<br>(Collaborative) | \$20,000 (USD)   | 2 years  |
| 3.   | Transdisciplinary Research and Innovation Hub at SMVDU  | Higher Education Department J&K                            | 2.5 crores (INR) | 1 year   |
| 4.   | Genetic Diversity and Evolutionary Perspectives in Population Groups of Jammu and Kashmir         | National Geographic USA                                    | \$50,000 (USD)   | 3 Years  |
| 5.   | Genetic perspectives of Type 2 Diabetes susceptibility in   | SERB, DST, Gol   | 28.5 Lakh (INR)  | 3 years  |

| population group of Jammu and Kashmir, India |  |                                 |                  |         |
|--|--|---------------------------------|------------------|---------|
| 6.   | Grant in AID under NIDHI SSS to SMVDU-TBIC | NSTEDB DST                      | 2.1 crores (INR) | 2 years |
| 7.   | NIDHI PRAYAS to SMVDU TBIC                 | NSTEDB DST                      | 2.2 crores (INR) | 2 years |
| 8.   | Technology Innovation Hub by SMVDU         | Higher Education Department J&K | 20 Lakh (INR)    | 1 year  |

### Research Outcome Summary:

|                               |     |  |      |
|-------------------------------|-----|--|------|
| Research Papers Published:    | >50 | Cumulative Impact factor (JCR, WOS) :        | >350 |
| Ph.Ds Awarded in Supervision: | 5,  | PG Dissertations Awarded in Supervision:     | 39   |
| Patents: 5, Book Chapters:    | 3,  | Invited Talks and Conference Presentations : | 28   |

### Patents Applications:

1. 201811041582 An assay for Telomere Length Measurement (Published)
2. 201811020588 A Polymerase Chain Reaction (PCR) kit and process for making the same (Published)
3. 201711043149 A Novel Strategy to Increase the Biogas Production (Published)
4. 202011010352 Green synthesis of silver nanoparticles (Published)
5. 202011015826 A system and a method for the diagnosis of an infectious disease (Granted)

### Highlighted Publications of the last 10 years.

1. Spolia A, Angural A, Sharma V, Shipra, Razdan S, Dhar MK, Mahajan A, Verma V, Pandita KK, Sharma S, Rai E. Cost-effective Whole Exome Sequencing discovers pathogenic variant causing Neurofibromatosis type 1 in a family from Jammu and Kashmir, India. **Sci Rep. 2023;13(1):7852. \*Corresponding Author**
2. Genetic Characterisation of Pantothenate Kinase Associated Neurodegeneration (PKAN) in a Consanguineous Family from Jammu and Kashmir, India. **Int J Hum Genet (2022)**  
DOI: [10.31901/24566330.2022/22.03.828](https://doi.org/10.31901/24566330.2022/22.03.828) \*Corresponding Author
3. Rana S, Ali S, Wani HA, Mushtaq QD, **Sharma S**, Rehman MU. Metabolic syndrome and underlying genetic determinants systematic review. **J Diabetes Metab Disord. 2022 Mar 3;21(1):1095-1104.**
4. Shah R, Khaitan PG, Pandita TK, Rafiq A, Abrol D, Suri J, Kaul S, Kumar R, **Sharma S**. Gastric cancer in Jammu and Kashmir, India: A review of genetic perspectives. **J Cancer Res Ther. 2022 Jul-Sep;18(4):873-879. \*Corresponding Author**
5. Singh H, Shipra, Sharma V, Sharma I, Sharma A, Modeel S, Gupta N, Gupta G, Pandita AK, Butt MF, Sharma R, Pandita S, Singh V, Rai E, Ikegawa S, Sharma S. (2022) The first study of epidemiology of adolescent idiopathic scoliosis shows lower prevalence in females of Jammu and Kashmir, India. **Am J Transl Res. 2022 15;14(2):1100-1106. \*Corresponding Author**
6. Angural, A.; Ponnusamy, K.; Langeh, D.; Kumari, M.; Spolia, A.; Rai, E.; Sharma, A.; Pandita, K.K.; Sharma, S. (2021) Missense Variation in *TPP1* Gene causes Neuronal Ceroid Lipofuscinosis Type 2 in a Family from Jammu and Kashmir-India **Preprints 2021, 2021070661 (doi: 10.20944/preprints202107.0661.v1) \*Corresponding Author**

7. Sethi I, Bhat GR, Kumar R, Rai E, Sharma S.(2021) Dual labeled fluorescence probe based qPCR assay to measure the telomere length. **Gene. 2021 Jan 30;767:145178. doi: 10.1016/j.gene.2020.145178. \*Corresponding Author**
8. Sharma, S., Singh, I., Haider, S., Malik, M.Z., Ponnusamy, K., Rai, E. RE: ACE2 Homodimerization Affects Binding of SARS-CoV-2 Spike Protein (reply to Yan et. al. 2020. **Structural basis for the recognition of SARS-CoV-2 by full-length human ACE2. Science, Vol. 367, Issue 6485, pp. 1444-1448). \*Corresponding Author**
9. Sethi, I., Sharma, V., Sharma, I., Singh, G., Bhat, G. R., Bhanwer, A., Sharma, S., & Rai, E. (2020). Telomere Maintenance Genes are associated with Type 2 Diabetes Susceptibility in Northwest Indian Population Group. **Scientific Reports, 10(1), 6444. https://doi.org/10.1038/s41598-020-63510-w \*Corresponding Author**
10. Angural, A., Spolia, A., Mahajan, A., Verma, V., Sharma, A., Kumar, P., Dhar, M. K., Pandita, K. K., Rai, E., & Sharma, S. (2020). Review: Understanding Rare Genetic Diseases in Low Resource Regions Like Jammu and Kashmir - India. **Frontiers in genetics, 11, 415. \*Corresponding Author**
11. Freda Lalrohlu, Varun Sharma, Indu Sharma, Ekta Rai, John Zohmingthanga, Vanlal Hruaii, Swarkar Sharma, & Nachimuthu Senthil Kumar (2020). Genotyping of T2D susceptible genes in a high risk North-East Indian population **Obesity Medicine, 17, 100162.**
12. Shah, R., Sharma, V., Singh, H., Sharma, I., Bhat, G., Shah, I., Iqbal, B., Rafiq, R., Nissa, N., Muzaffar, M., Rasool, M., Lone, G., Kaul, S., Lone, M., Rai, E., Dar, N., & Sharma, S. (2020). LRFN2 gene variant rs2494938 provides susceptibility to esophageal cancer in the population of Jammu and Kashmir. **Journal of Cancer Research and Therapeutics. \*Corresponding Author**
13. Lalrohlu F, Sharma V, Sharma I, Singh H, Kour G, Sharma S, Yuman, Zohmingthanga J, Vanlalhruaii, Rai E, Singh V, Senthil Kumar N & Sharma S. MACF1 gene variant rs2296172 is associated with T2D susceptibility in Mizo population from Northeast India. **Int J Diabetes Dev Ctries (2020). https://doi.org/10.1007/s13410-019-00788-1 \*Corresponding Author**
14. Sharma S, Lalrohlu F, Sharma V, Sharma I, Sharma S, Javed Parihar T, Zohmingthanga J, Singh V, Sharma S, Senthil Kumar N& Rai E. Candidate gene association study of UCP3 variant rs1800849 with T2D in Mizo population of Northeast India. **Int J Diabetes Dev Ctries (2020). https://doi.org/10.1007/s13410-020-00812-9\*Corresponding Author**
15. Palit SP, Patel R, Jadeja SD, Rathwa N, Mahajan A, Ramachandran AV, Dhar MK, Sharma S, Begum R. A genetic analysis identifies a haplotype at adiponectin locus: Association with obesity and type 2 diabetes. **Sci Rep. 2020 Feb 19;10(1):2904. doi: 10.1038/s41598-020-59845-z. Erratum in: Sci Rep. 2020 Apr 27;10(1):7017. PMID: 32076038; PMCID: PMC7031532.**
16. Sudan J, Singh R, Sharma S, Salgotra RK, Sharma V, Singh G, Sharma I, Sharma S, Gupta SK, Zargar SM. ddRAD sequencing-based identification of inter-genepool SNPs and association analysis in Brassica juncea. **BMC Plant Biol. 2019 Dec 30;19(1):594. doi: 10.1186/s12870-019-2188-x. PMID: 31888485; PMCID: PMC6937933.**
17. Xu C, Yang X, Zhou H, Li Y, Xing C, Zhou T, Zhong D, Lian C, Yan M, Chen T, Liao Z, Gao B, Su D, Wang T, Sharma S, Mohan C, Ahituv N, Malik S, Li QZ, Su P. A novel ZRS variant causes preaxial polydactyly type I by increased sonic hedgehog expression in the developing limb bud. **Genet Med. 2020 Jan;22(1):189-198. doi: 10.1038/s41436-019-0626-7. Epub 2019 Aug 9. PMID:31395945; PMCID: PMC6944640.**
18. Angural A, Sharma I, Pandoh P, Sharma V, Spolia A, Rai E, Singh V, Razdan S, Pandita KK, Sharma S. A case report on a novel MT-ATP6 gene variation in atypical mitochondrial Leigh

- syndrome associated with bilateral basal ganglia calcifications. **Mitochondrion**. 2018 Jun 18. pii: S1567-7249(17)30343-4. doi:10.1016/j.mito.2018.06.005. \*Corresponding Author
19. Sharma I, Sharma V, Khan A, Kumar P, Rai E, Bamezai RNK, Vilar M, Sharma S. Ancient Human Migrations to and through Jammu Kashmir- India were not of Males Exclusively. **Nature Scientific Reports** 2018;8(1):851. doi: 10.1038/s41598-017-18893-8. \*Corresponding Author
  20. Varun Sharma, Itty Sethi, Indu Sharma, Gurvinder Singh, Ankit Mahajan, Arshia Angural, A. J. S. Bhanwer, Manoj K. Dhar, K. K. Pandita, Vinod Singh Ekta Rai, Swarkar Sharma. Replication of MACF1 gene variant rs2296172 with type 2 diabetes susceptibility in the Bania population group of Punjab, India. **Int J Diabetes Dev Countries** (2017). <https://doi.org/10.1007/s13410-017-0598-6> \*Corresponding Author
  21. Angural A, Singh I, Mahajan A, Pandoh P, Dhar MK, Kaul S, Verma V, Rai E, Razdan S, Kishore Pandita K, **Sharma S**. A variation in PANK2 gene is causing Pantothenate kinase-associated Neurodegeneration in a family from Jammu and Kashmir - India. **Nature Scientific Reports** 2017 Jul 5;7(1):4834. doi: 10.1038/s41598-017-05388-9. \*Corresponding Author
  22. Sahakyan, H., B. Hooshiar Kashani, R. Tamang, A. Kushniarevich, A. Francis, M. D. Costa, A. K. Pathak, Z. Khachatryan, I. Sharma, M. van Oven, J. Parik, H. Hovhannisyan, E. Metspalu, E. Pennarun, M. Karmin, E. Tamm, K. Tambets, A. Bahmanimehr, T. Reisberg, M. Reidla, A. Achilli, A. Olivieri, F. Gandini, U. A. Perego, N. Al-Zahery, M. Houshmand, M. H. Sanati, P. Soares, E. Rai, J. Sarac, T. Saric, V. Sharma, L. Pereira, V. Fernandes, V. Cerny, S. Farjadian, D. P. Singh, H. Azakli, D. Ustek, N. Ekomasova Trofimova, I. Kutuev, S. Litvinov, M. Bermisheva, E. K. Khusnutdinova, N. Rai, M. Singh, V. K. Singh, A. G. Reddy, H. V. Tolk, S. Cvjetan, L. B. Lauc, P. Rudan, E. N. Michalodimitrakis, N. P. Anagnou, K. I. Pappa, M. V. Golubenko, V. Orekhov, S. A. Borinskaya, K. Kaldma, M. A. Schauer, M. Simionescu, V. Gusar, E. Grechanina, P. Govindaraj, M. Voevoda, L. Damba, **S. Sharma**, L. Singh, O. Semino, D. M. Behar, L. Yepiskoposyan, M. B. Richards, M. Metspalu, T. Kivisild, K. Thangaraj, P. Endicott, G. Chaubey, A. Torroni and R. Villems (2017). "Origin and spread of human mitochondrial DNA haplogroup U7." **Nature Scientific Reports** 7: 46044.
  23. Sharma V, Sharma I, Sethi I, Mahajan A, Singh G, Angural A, Bhanwer AJ, Dhar MK, Singh V, Rai E, **Sharma S\*** (2017) "Replication of newly identified type 2 diabetes susceptible loci in Northwest Indian population" **Diabetes Res Clin Pract**. doi: 10.1016/j.diabres.2017.02.013. \*corresponding author
  24. Itty Sethi, G.R. Bhat, Vinod Singh, Rakesh Kumar, AJS Bhanwer, Rameshwar N. K. Bamezai, **Swarkar Sharma\*** Ekta Rai\*. (2016) Role of Telomeres and Associated Maintenance Genes in Type 2 Diabetes Mellitus: A Review. **Diabetes Research and Clinical Practice**. doi: 10.1016/j.diabres.2016.10.015 \*Corresponding Author
  25. Shafat Ali, Shazia Nafis, Ponnusamy Kalaiarasan, Ekta Rai, **Swarkar Sharma\***, Rameshwar NK Bamezai\*(2016) Understanding Genetic Heterogeneity in Type 2 Diabetes by Delineating Physiological Phenotypes – SIRT1 and its Gene Network in Impaired Insulin Secretion. **Rev Diabet Stud**, 13(1):17-34 \*Corresponding Author
  26. Ekta Rai, Ankit Mahajan, Parvinder Kumar, Arshia Angural, Manoj K Dhar, Sushil Razdan, Kumarasamy Thangaraj, Carol A. Wise, Shiro Ikegawa, Kamal Kishore Pandita, **Swarkar Sharma** (2016) Whole Exome Screening Identifies Novel and Recurrent WISP3 Mutations Causing Progressive Pseudorheumatoid Dysplasia in Jammu and Kashmir-India. **Nature Scientific Reports**, 13;6:27684. doi: 10.1038/srep27684. \*Corresponding Author
  27. Mary J. Gray\*, Peter Kannu\*, **Swarkar Sharma\***, Christine Neyt\*, Dongping Zhang, Nandina Paria, Philip B. Daniel, Heather Whetstone, Hans-Georg Sprenger, Philipp Hammerschmidt, Angela Weng, Lucie Dupuis, Rebekah Jobling, Roberto Mendoza-Londono, Michael Dray, Peiqiang Su, Megan J. Wilson, Raj P. Kapur, Edward F. McCarthy, Ben A. Alman, Andrew

- Howard, Gino R. Somers, Christian R. Marshall, Simon Manners, Adrienne M. Flanagan, Karl Rathjen, Lori A. Karol, Haemish Crawford, David M. Markie, Jonathan J. Rios, Carol A. Wise, Stephen P. Robertson (2015). Mutations Preventing Regulated Exon Skipping of MET Cause Osteofibrous Dysplasia. **American Journal of Human Genetics** **97(6)**, 3: 837–847 \*First Shared Author
28. Sharma S, Londono D, Eckalbar WL, Gao X, Zhang D, Mauldin K, Kou I, Takahashi A, Matsumoto M, Kamiya N, Murphy KK, Cornelia R; TSRHC Scoliosis Clinical Group; Japan Scoliosis Clinical Research Group, Herring JA, Burns D, Ahituv N, Ikegawa S, Gordon D, Wise CA. (2015) A PAX1 enhancer locus is associated with susceptibility to idiopathic scoliosis in females. **Nature Communication** **2015 Mar 18;6:6452**. doi: [10.1038/ncomms7452](https://doi.org/10.1038/ncomms7452).
  29. Londono D\*, Kou I\*, Johnson TA\*, Sharma S\*, Ogura Y, Tsunoda T, Takahashi A, Matsumoto M, Herring JA, Lam TP, Wang X, Tam EM, Song YQ, Fan YH, Chan D, Cheah KS, Qiu X, Jiang H, Huang D; Japanese Scoliosis Clinical Research Group, TSRHC IS Clinical Group, the International Consortium for Scoliosis Genetics, Su P, Sham P, Cheung KM, Luk KD, Gordon D, Qiu Y, Cheng J, Tang N, Ikegawa S, Wise CA (2014) A meta-analysis identifies adolescent idiopathic scoliosis association with LBX1 locus in multiple ethnic groups **J Med Genet.** **2014 Apr 10**. doi: [10.1136/jmedgenet-2013-102067](https://doi.org/10.1136/jmedgenet-2013-102067). \*First Shared Author
  30. Varun Sharma, Indu Sharma, Vishav Pratap Singh, Sonali Verma, Anil Pandita, Vinod Singh, Ekta Rai, Swarkar Sharma (2014) mtDNA G10398A Variation Provides Risk to Type 2 Diabetes in Population Group from Jammu Region of India. **MetaGene** **2014 Apr 13**. doi:[10.1016/j.mgene.2014.02.003](https://doi.org/10.1016/j.mgene.2014.02.003). \*Corresponding author.
  31. Mahajan A, Sharma S, Dhar MK, Bamezai RNK (2013) Risk factors of type 2 diabetes in population of Jammu and Kashmir, India. **Journal of Biomedical Research** **27 (5)**, 372
  32. Ikuyo Kou, Yohei Takahashi, Todd A Johnson, Atsushi Takahashi, Long Guo, Jin Dai, Xusheng Qiu, Swarkar Sharma, Aki Takimoto, Yoji Ogura, Hua Jiang, Huang Yan, Katsuki Kono, Noriaki Kawakami, Koki Uno, Manabu Ito, Shohei Minami, Haruhisa Yanagida, Hiroshi Taneichi, Naoya Hosono, Taichi Tsuji, Teppei Suzuki, Hideki Sudo, Toshiaki Kotani, Ikuho Yonezawa, Douglas Londono, Derek Gordon, John A Herring, Kota Watanabe, Kazuhiro Chiba, Naoyuki Kamatani, Qing Jiang, Yuji Hiraki, Michiaki Kubo, Yoshiaki Toyama, Tatsuhiko Tsunoda, Carol A Wise, Yong Qiu, Chisa Shukunami, Morio Matsumoto & Shiro Ikegawa (2013) Genetic variants in GPR126 are associated with adolescent idiopathic scoliosis. **Nature Genetics** **45 (6)**, 676-679
  33. Shafat Ali, Rupali Chopra, Siddharth Manvati, Yoginder Singh, Nabodita Koul, Anita Behura, Ankit Mahajan, Prabodh Sehajpal, Subash Gupta, Manoj K Dhar, Chainy Gagan BN, Amarjit S. Bhanwar, Swarkar Sharma\*, Rameshwar NK Bamezai\* (2013) "Replication of Type 2 diabetes candidate genes variations in three geographically unrelated Indian population groups" **PLoS one.** **8(3): e58881**. \*Corresponding author.
  34. Wenjie Gao, Yan Peng, Guoyan Liang, Anjing Liang, Wei Ye, Liangming Zhang, Swarkar Sharma, Peiqiang Su, Dongsheng Huang (2013) Association between Common Variants near LBX1 and Adolescent Idiopathic Scoliosis Replicated in the Chinese Han Population. **PLoS One** **8(1): e53234**
  35. Rai E, Sharma S\*, Kaul S, Jain K, Matharoo K, Bhanwer AS, Bamezai RN\* (2012) The interactive effect of SIRT1 promoter region polymorphism on type 2 diabetes susceptibility in the North Indian population. **PLoS One.****7(11):e48621**. \*Corresponding author.
  36. Liang G, Gao W, Liang A, Ye W, Peng Y, Zhang L, Sharma S, Su P, Huang D (2012). Normal leptin expression, lower adipogenic ability, decreased leptin receptor and hyposensitivity to Leptin in Adolescent Idiopathic Scoliosis. **PLoS One;****7(5):e36648**.

37. Londono D, Buyske S, Finch SJ, **Sharma S**, Wise CA, Gordon D. (2012) TDT-HET: a new transmission disequilibrium test that incorporates locus heterogeneity into the analysis of family-based association data. **BMC Bioinformatics. 20;13:13.**
38. Carol A Wise, **Swarkar Sharma**, Xiaochong Gao, Douglas Londono, Kristen N Mauldin, January M Brandon, Vanessa King, Dongping Zhang, Derek Gordon, John A Herring (2012) Genome-wide association studies of adolescent idiopathic scoliosis suggest genes encoding axon guidance molecules as candidates for disease susceptibility **Journal of Bone and Joint Surgery - British Volume 01/2012; 94-B(SUPP XXVII):46**

**Membership Academics bodies/Societies/ other Professional bodies:**

- Founder President J&K Society of Biologists, India (2 years)
- Life Member International Consortium for Scoliosis Genetics (ICSG USA)
- Member National Society of Genetic Counselors, USA (Annual)
- Member National Geographic Society, USA
- Member American Society of Human Genetics, USA (Annual)
- Member Human Genome Organisation (HUGO), Singapore (Annual)
- Life Member and Executive Council member (2019-2021) Indian Society of Human Genetics, India
- Life member Society for Indian Academy of Medical Genetics, India
- Vice-President and President Society of Human Genetics, GNDU, Amritsar (2001-2003)
- CEO SMVDU- Technology Business Incubation Centre (SMVDU TBIC Aug 2015-Jan 2022)
- Founder Member Board of Directors, CUJammu SITE Council (Central University of Jammu Startup Incubation and Technology Enablement Council) (2023-Present)

**Scientific Advisor Startups:**

- Biodroid innovations Pvt Ltd(<https://biodroidinnovations.com>)
- B-ezy diagnostics Pvt Ltd (<https://b-ezy.com/>)
- Key2Genes (<https://key2genes.com/>)

**Invited Talks/Conference Presentations**

1. Underlying Challenges in Unravelling Genetics of Rare Diseases in India: Our learnings from J&K, 2nd Rare Genetic Diseases Research Summit-2023 (REDRESS 2023), at TIGS, Bengaluru (23<sup>rd</sup>-24<sup>th</sup> November, 2023)
2. Understanding Genetics and Genomics of Human Population of Jammu and Kashmir-India: Project JkDNA, International scientific conference: 23rd-ADNAT-"Translating Human Evolutionary History to Precision Medicine" Banaras Hindu University, Varanasi, India (March 10th-12th ,2023)
3. Project JK-DNA to understand Unique Human Gene Pool of Jammu and Kashmir-India: towards Precision Medicine, International Conference on Advances in Molecular Diagnostics and Precision Medicine 2022, Anna University, Chennai. (October 15-17, 2022)
4. Understanding Unique Gene Pool and Risk of Genetic Disorders in Populations of Jammu and Kashmir-India: Project JK-DNA, Online International Conference Biomolecules to Biome at Department of Life Sciences Presidency University, Kolkata India (August 24-25, 2022)
5. Host genes can explain differential SARS-CoV-2 entry and high population specific differences in clinical outcomes of COVID-19., Annual Meeting of American Society of Human Genetics, USA (October 27-30, 2020) ,USA

6. Understanding Rare Genetic Disease: the untold stories from Jammu & Kashmir., Recent advances in Biosciences (online), Department of Zoology Sree Chaitanya College Habra, Kolkatta (June 8, 2020)
7. Y chromosomal and mitochondrial DNA diversity in Jammu and Kashmir-India questions "Aryan Invasion hypothesis"., NextGen Genomics, Biology, Bioinformatics and Technologies (NGBT) Conference , SciGenom Research Foundation, Mumbai (30 Sept-2 October, 2019)
8. Scoliosis in Indian Populations, International Consortium for Spinal Genetics, Development and Disease , Karolinska Institute, Stockholm, Sweden (September, 5-7, 2019)
9. IPR: How to protect Your Research, One Week Faculty Development Programme on Research Methodology using R and E views, FDC, Shri Mata Vaishno Devi University (May, 21-25, 2019)
10. Procedure for filing Patent, Four week Orientation Programme, FDC, Shri Mata Vaishno Devi University (April, 1-30, 2019)
11. Intellectual Property Rights, Four week Orientation Programme, FDC, Shri Mata Vaishno Devi University (April, 1-30, 2019)
12. Unique and Conserved gene pool...Jammu & Kashmir, India, Annual Meeting of American Society of Human Genetics at San Diego, USA (16 to 20 October 2018) , San Diego, USA
13. Role of Genetics in Diseases, CME on Endocrine Malignancies, SMVDNSH, JAMMU (April, 21, 2018)
14. Unique Gene Pool and Higher Risk of Rare Genetic Disorders in Populations of Jammu and Kashmir-India: Next Generation Sequencing Methodologies to Rescue., NextGen Genomics, Biology, Bioinformatics and Technologies (NGBT) Conference , SciGenom Research Foundation, Jaipur Rajasthan (30 Sept-2 October, 2018)
15. A novel MT-ATP6 gene variation resulting in an atypical case of Leigh syndrome., 43rd Annual Meeting of the Indian Society of Human Genetics (Population and Medical Genomics), CSIR-CCMB, Hyderabad (12 to 14 March, 2018)
16. Panel discussion presented on Scoliosis Scenario in Indian populations., Genomic Approached to Understanding and Treating Scoliosis Conference, Texas Scottish Rite Hospital for Children, Dallas, Texas, USA (16-18 March, 2017)
17. Rare Disorders in Population of Jammu and Kashmir India., Recent Advances in molecular genetics with new biomedical insights, INSA and Israel Academy of Sciences and Humanities (February, 12-13, 2018)
18. Faculty in workshop for Genetics, 2nd Masterclass in Gynaecology workshop, SMVDNSH, Jammu (October, 7, 2017)
19. Geographic Isolation & Endogamous Practices Preserved Unique Gene Pool and Provide Higher Risk of Rare Genetic Disorders in Populations of Jammu & Kashmir., 2nd International Conference on Founder Populations, Kochi, Kerala (November, 9-12, 2017 )
20. High Mitogenomes Diversity in Jammu and Kashmir Defi esthat Ancient Human Migrations in India were of Males Exclusively., NextGen Genomics, Biology, Bioinformatics and Technologies (NGBT) Conference , SciGenom Research Foundation , Bhubaneswar, Orissa (2-4 October, 2017)
21. Next Generation Sequencing in Identification and Characterization of Rare Genetic Disorders., 42nd Indian Society of Human Genetics Annual Meeting and International Conference, IISC-Bangalore (2-4 March, 2017)
22. WES helps characterize the mysterious skeletal disorder of avillage in Jammu and Kashmir, India , 13th International Congress of Human Genetics, Kyoto International Conference Centre, Japan (April, 3-7, 2016)



23. Panel discussion talk on role of genetics in cardiac disorders, National Seminar on Cardiac Sciences, SMVDN, Superspeciality Hospital (December, 3, 2016)
24. Whole Exome Sequencing in Characterizing Mysterious Crippling Disorder of Arai Village of Jammu and Kashmir., 10th J&K Science Congress, University of Jammu (March, 14-16, 2015)
25. Whole Exome Sequencing helps characterize a rare highly penetrant familial disorder, turning epidemic in a village of Jammu and Kashmir, India. , 40th Annual Conference of Indian Society of Human Genetics, National Institute of Immunohaematology (ICMR) Mumbai (January, 28-30, 2015)
26. Characterization of Mysterious Crippling Disorder of Arai Village of J&K, India. , NextGen Genomics, Biology, Bioinformatics and Technologies (NGBT) Conference, SciGenom Research Foundation , Bangalore (17-19 November, 2014)
27. Genetics of Human Disorders : Is it needed in Indian populations, Multi disciplinary national conference science Colloquium: Emerging trends in basic and applied sciences, DAV college, Jalandhar, Punjab (March, 6-7, 2014)
28. Exome sequencing in idiopathic scoliosis reveals rare variants in VANG1, a planar cell polarity gene involved in axial development. , 62nd Annual session of 'American Society of Human Genetics' (ASHG-2012), San Francisco, California , USA (Nov 6-10, 2012 )
29. Genome-wide studies of idiopathic scoliosis susceptibility implicate genes encoding neural cell adhesion proteins., 60th Annual session of 'American Society of Human Genetics' (ASHG-2010), Washington DC, USA (Nov 2-6, 2010)
30. Neural cell adhesion protein CHL1 is associated with susceptibility for idiopathic scoliosis., Genomics of common diseases, The Wellcome Trust Conference Centre, Cambridge, UK (September, 23-26, 2009)
31. Polymorphic Alu Insertions and the Genetic Structure of Population Groups from Punjab, Northwest India. , 58th Annual Meeting of ASHG, Pennsylvania Convention Centre in Philadelphia (Nov 11-15, 2008)
32. The Indian origin of paternal haplogroup R1a1\*, 5th International Symposium on Genetics, Health and disease , GNDU, Amritsar (Feb 17-19, 2008)

#### References:

##### 1. Prof. Carol Wise

Eugene McDermott Center for Human Growth and Development  
University of Texas Southwestern Medical Center  
5323 Harry Hines Blvd, Dallas, TX 75390, USA

[Carol.wise@tsrh.org](mailto:Carol.wise@tsrh.org)

##### 2. Prof. RNK Bamezai

Retired Professor,  
Former Dean, School of Life Sciences  
Jawaharlal Nehru University, New Delhi 110067, India

[rameshwarbamezai@gmail.com](mailto:rameshwarbamezai@gmail.com)

##### 3. Prof. AJS Bhanwer

Retired Professor, Guru Nanak Dev University  
Professor and Head, Department of Genetics  
Sri Guru Ram Das University of Health Sciences  
Mehta Road, Amritsar 143 501, India

[ajsbanwer@gmail.com](mailto:ajsbanwer@gmail.com)