

# ***Curriculum vitae (cv)***

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**Name:** Dr. Akshi Spolia

## **Educational qualifications**

1. BSc honors' Biotechnology from Sher-e-Kashmir University of Agricultural Sciences and Technology (SKUAST-J)
2. MSc Biotechnology from Shri Mata Vaishno Devi University (SMVDU)
3. PhD Biotechnology from Shri Mata Vaishno Devi University (SMVDU)

**Ph.D. thesis title:** Genetic Characterization of some Monogenic Disorders in the population of Jammu and Kashmir.

**Specialization:** Ph.D. Biotechnology (specialization in Human Genetics).

## **Ph.D. Research experience:**

- My doctoral research was focused on the "Genetic Characterization of some Monogenic Disorders in the population of Jammu and Kashmir". Research has genetically screen three Monogenic disorders from Jammu and Kashmir region, namely Neurofibromatosis Type 1 (NF1), Pantothenate kinase-associated neurodegeneration (PKAN), and Spinocerebellar ataxia (SCA); through aid of certain advance molecular biological tools. The advanced techniques used to characterize these disorders were Next Generation Sequencing (NGS), Sanger sequencing, Data interpretation of NGS and sanger sequencing, validation of the results using certain Bioinformatic tools.

## **Equipment/Experimental handling skills**

1. Handling of human samples, DNA isolation (from blood, saliva), PCR amplification, Gel Electrophoresis (Agarose, PAGE), Mass Array, RT PCR, Biosafety cabinet (Laminar Air Flow), Microscope, Autoclave, Centrifuge, Weighing balance, Micropipettes, Bioreactors etc.
2. Expertise in advanced molecular biological techniques such as Next Generation Sequencing (NGS), Sanger sequencing, Data analysis (NGS, Sanger), expertise in certain bioinformatics tools etc. Experience in conducting genetic counseling.

## **Teaching Experience:**

I have worked as Academic arrangement Lecturer at Govt. Degree College (Boys), Department of Biotechnology, Udhampur, J&K.

Guest Faculty in Biotechnology Department, Shri Krishan Chander Government Degree College, Poonch, J&K Conducted theory and Practical Classes of various semesters against allotted work, also conducted examinations and prepared results.

## Academic Qualifications

CLASS	SCHOOL	BOARD	SUBJECTS	MARKS (%)
10th	Seventh Day Adventist Senior Secondary School, Jalandhar (2007)	ICSE Board	Science, English, Hindi, Environmental Education, History, Civics & Geography, Mathematics, Commercial Applications	70.71
12th	St. Peters Higher Secondary School, Jammu (2009)	STATE Board	Physics, Chemistry, Biology, Environmental Sciences, General English	80.9

DEGREE	UNIVERSITY (SESSION)	SUBJECTS STUDIED	CGPA
B.Sc. (Honours) Biotechnology	Sher- E- Kashmir University of Agricultural Sciences and Technology, Jammu, J&K (2010-2014)	Genetics, Plant Biotechnology, Animal Biotechnology, Microbiology, Immunology, Statistics, Biochemistry, Bioinformatics, Environmental Sciences, Molecular biology, Biophysics, Biomathematics, Industrial Biotechnology etc	6.91
M.Sc. Biotechnology	Shri Mata Vaishno Devi University, Katra, J&K (2014-2016)	Plant Biotechnology, Animal Biotechnology, Microbiology, Immunology, Statistics, Biochemistry, Principles in Human Genetic disorders, Bioinformatics, Medical Genetics etc	8.17
Ph.D. Biotechnology	Shri Mata Vaishno Devi University Katra, J&K (2016-2022)	Title: Genetic Characterization of some Monogenic Disorders in the population of Jammu and Kashmir	

## **Publications**

1. Cost effective Whole Exome Sequencing economically discovers pathogenic variation causing Neurofibromatosis type 1 in a family from Jammu and Kashmir (2023). **Scientific Reports**
2. Genetic Characterization of Pantothenate Kinase Associated Neurodegeneration (PKAN) in a Consanguineous Family from Jammu and Kashmir, India (2022). **International journal of Human Genetics**
3. Understanding Rare Genetic Diseases in Low Resource Regions Like Jammu and Kashmir-India (2020). **Frontiers in Genetics**
4. A case report on a novel MT-ATP6 gene variation in atypical mitochondrial Leigh syndrome associated with bilateral basal ganglia calcifications (2018). **Mitochondrion**
5. Missense Variation in TPP1 Gene causes Neuronal Ceroid Lipofuscinosis Type 2 in a Family from Jammu and Kashmir-India (2021). Preprints doi: 10.20944/preprints202107.0661.v1

## **Database submission**

The variant and data of Neurofibromatosis Type 1 disorder is available in the ClinVar database repository with ClinVar accession number: SCV001762282.1.

## **Other experience**

- 6 months M.Sc. dissertation from Shri Mata Vaishno Devi University on topic entitled “Development of Procedures to obtain good Quality Human Genomic DNA from saliva and Buccal cells: A non-invasive Approach”.
- Experience in providing guidance to M.Sc. dissertation students during my Ph.D.

## **Awards and Fellowship**

- Qualified national level examination “Graduation Aptitude Test in Engineering” GATE, India, 2016 and awarded with distinction medal in MSc Biotechnology.

## ***Conference Presentations and Workshops attended***

### **Presentations**

1. Poster presentation titled “Family with NEUROFIBROMATOSIS TYPE 1 identified in Jammu and Kashmir, in India”, in 42<sup>nd</sup> Annual Meeting of Indian Society of Human Genetics & International Symposium on Trends in Human Genetic Research & Management, held from March 2-4, 2017, at the Indian Institute of Science, Bangalore-560012, India.
2. Poster presentation titled “Genetic characterization of Spinocerebellar ataxia reported as extended familial case in Jammu and Kashmir, India”, in 9<sup>th</sup> international Meeting Next Gen Genomics, Biology, Bioinformatics and Technologies (NGBT) conference, held from 30<sup>th</sup> September-2<sup>nd</sup> October, 2019, Mumbai, India.
3. Presented e-poster titled “Genetic characterization of Pantothenate kinase- associated neurodegeneration (PKAN) in the family from Jammu and Kashmir” in 6<sup>th</sup> National ISIEM 2021 Conference, held from 28<sup>th</sup>-29<sup>th</sup> August 2021.
4. Poster presentation titled “Genetic characterization of a case of Myopathy/Muscular Dystrophy reported in the state of Jammu and Kashmir”, in 43<sup>rd</sup> Annual Meeting of Indian Society of Human Genetics Population and Medical Genomics, held from March 12<sup>th</sup> -14<sup>th</sup>, 2018, at CSIR-Centre for Cellular and Molecular Biology, Hyderabad, India.
5. Oral presentation titled “Identification of a familial case of NEUROFIBROMATOSIS TYPE 1 in Jammu and Kashmir”, in India International Conference on Emerging Trends in Biosciences and Chemical Technology, held from 3<sup>rd</sup> -5<sup>th</sup> December, 2022, at Shri Mata Vaishno Devi University, Jammu & Kashmir, India.
6. Presented work titled “Genetic characterization of Myopathy/Muscular Dystrophy identified in the state of J&K, at 5<sup>th</sup> International Conference on Recent Trends & Advancement in Engineering and Technology, held from 25<sup>th</sup> -26<sup>th</sup> October, 2018, at Shri Mata Vaishno Devi University, Jammu & Kashmir, India.
7. Presented poster titled “Chilblains: An overview of a cold-Manifested Vasculopathy”, at 3<sup>rd</sup> International Conference on Recent Trends & Advancement in Engineering and Technology, held from 17<sup>th</sup>-18<sup>th</sup> November 2016, at Shri Mata Vaishno Devi University, Jammu & Kashmir, India.
8. Presented poster titled “Identification of a familial case of Neurofibromatosis type 1 in Jammu and Kashmir, India”, at 4<sup>th</sup> International Conference on Recent Trends & Advancement in Engineering and Technology, held from 03<sup>rd</sup>-04<sup>th</sup> November, 2017, at Shri Mata Vaishno Devi University, Jammu & Kashmir, India.

## **Conferences and workshops**

- Attended hands-on session on Unity and Cohesion in Academics Writing , during “Research Day” organized by Shri Mata Vaishno Devi University , on 25 September 2019.
- Attended One Day seminar on “RAMAN SPECTROSCOPY AND MICRO ARRAY PRINTING” organized by Shri Mata Vaishno Devi University (SMVDU) ,Katra.
- Attended J&K State Science, Technology and Innovation Council sponsored one day workshop on “Scientific Innovation & Intellectual Property Rights” organized by Department of Biotechnology (DoBT) , Shri Mata Vaishno Devi University (SMVDU), Katra , J&K on 27 March 2016.
- Attended CME on Endocrine Malignancies organized at Shri Mata Devi Narayana Superspeciality Hospital , Kakryal Katra ,J&K on 21 April 2018.
- Participated as volunteer in the National Symposium on Recent Trends in Biotechnology and Drug Discovery held at Shri Mata Vaishno Devi University , Katra during 30-31 March 2016.
- Participated on Training on “GCS3000Dx and CytoScan Optima Assay”, held at SoBT, Shri Mata Vaishno Devi University, Katra from 22nd to 24th November 2022.
- Participated in a 3 Day Workshop on “Research Methodologies and Data Analysis” organized by Human Resource Development Centre (H.R.D.C) ,S.M.V.D.U Katra.
- Attended two days National workshop on “Diagnostic cytogenetics”, held from 26-27<sup>th</sup> feb 2018, at Shri Mata Vaishno Devi University, Katra.
- Attended and presented in 42<sup>nd</sup> Annual Meeting of Indian Society of Human Genetics & International Symposium on Trends in Human Genetic Reasearch & Management, held from March 2-4, 2017, at the Indian Institute of Science, Bangalore-560012, India.
- Attended and presented in 9<sup>th</sup> international Meeting Next Gen Genomics, Biology, Bioinformatics and Technologies (NGBT) conference, held from 30 <sup>th</sup> September-2<sup>nd</sup> October, 2019, Mumbai, India.
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- Attended and presented in 5<sup>th</sup> International Conference on Recent Trends & Advancement in Engineering and Technology, held from 25<sup>th</sup> -26<sup>th</sup> october, 2018, at Shri Mata Vaishno Devi University, Jammu & Kashmir, India.
- Attended and presented in 3<sup>rd</sup> International Conference on Recent Trends & Advancement in Engineering and Technology, held from 17<sup>th</sup>-18<sup>th</sup> november, 2016, at Shri Mata Vaishno Devi University, Jammu & Kashmir, India.

## ***Reference***

- Dr. Ekta Rai, Associate Professor, School of life sciences, Jawaharlal Nehru University, Delhi [ektarai@mail.jnu.ac.in](mailto:ektarai@mail.jnu.ac.in) and [ekta.rai@smvdu.ac.in](mailto:ekta.rai@smvdu.ac.in)
- Dr. Swarkar Sharma, Professor, Department of Zoology, Central University of Jammu, J&K, [swarkar.sharma@smvdu.ac.in](mailto:swarkar.sharma@smvdu.ac.in), [swarkar.molb@cujammu.ac.in](mailto:swarkar.molb@cujammu.ac.in)
- Dr. Arshia Angural, Assistant Professor, Department of Medical Genetics, JSS Medical College and Hospital, JSS Academy of Higher Education and Research Mysuru, Karnataka, [arshia.angural@yahoo.co.in](mailto:arshia.angural@yahoo.co.in)

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