

Curriculum vitae

- **Name:** Dr Priyanka Upadhyai
- **Qualification:** PhD
- **Designation:** Associate Professor
- **Office address:** Department of Medical Genetics
Kasturba Medical College
Manipal Academy of Higher Education
Manipal 576104
Karnataka, India
- **Email:** priyanka.u@manipal.edu
- **Date of Birth:** 15th July, 1982
- **Summary of education and qualification**
 1. **Doctor of Philosophy:** Aug 2007 – May 2013. University of Pittsburgh, Pittsburgh, Pennsylvania, USA. Doctor of Philosophy in Biological sciences.
 2. **Master of Science:** Sep 2004 - Jul 2006. University of Calcutta, Kolkata, India. Master of Science in Biotechnology.
 3. **Bachelor of Science:** Aug 2001 – Jun 2004. University of Calcutta, Kolkata, India. Bachelor of Science (Hons.) in Zoology.
- **Postdoctoral Research experience**

University of Manchester, England, UK. Postdoctoral Research Associate
- **Employment History**

| Place | Post | Period |
|--|---------------------------------|------------------------------|
| Kasturba Medical College, Manipal Academy of Higher Education, Manipal | Associate Professor | October 2020 - Present |
| Kasturba Medical College, Manipal Academy of Higher Education, Manipal | Assistant Professor | March 2016 – October 2020 |
| University of Manchester, UK | Postdoctoral Research Associate | September 2013 – March 2016. |

- **Summary of Research interests**
 - *Orcid ID:* <http://orcid.org/0000-0002-4319-7283>
 - *Scopus ID:* 55871340200

- *Key areas*: cilia biology, microtubules, centrosomes, genetics of Mendelian disorders, transcriptional regulation of gene expression, *Drosophila* development and patterning, *Drosophila* models of human disease

- **Sponsored Projects as Principal Investigator**

1. Investigating the crosstalk between primary cilia and autophagy in chondrogenesis and its modulation by Fibroblast growth factor (FGF) signaling in FGFR3 related skeletal dysplasias *in vitro* (**ongoing**) by Indian Council for Medical Research (ICMR), Government of India.
Duration: 3 years (01/02/2021-31/01/2024)
2. Investigating the role of *IFT52* and *EXOC6B* in human biology and disease using cell-culture and *Drosophila* systems (ECR/2016/001475): Early Career Research award by Department of Science and Technology (DST) Science and Engineering Research Board (SERB), Govt. of India. (**completed**)
Duration: 3 years (30/3/2017 – 29/09/2020)

- **Sponsored Projects as Co-Principal Investigator/co-Investigator**

1. Transcriptional and behavioral mechanisms underlying social isolation stress in *Drosophila*. Funded by Department of Biotechnology (DBT), Govt. of India.
Duration: 3 years (2021-2024)
2. Virtual Centre for Functional Genomics of Novel Sequence variants in Genes for Rare Monogenic Disorders in India.
Technically approved for funding by Indian Council for Medical Research (ICMR), Govt. of India.
Duration: 3 years

- **Peer Reviewed Publications**

1. **Priyanka Upadhyai**, Gokul Suresh, Rahul Parit and Ranajit Das (2021). Genomic and Ancestral variation underlies the severity of COVID-19 clinical manifestation in individuals of European Descent. *MDPI Life* <https://www.mdpi.com/2075-1729/11/9/921>
2. **Priyanka Upadhyai**, Periyasami Radhakrishnan, Vishal Singh Guleria, Neethu Kausthubham, Shalini Nayak, Andrea Superti-Furga and Girisha KM (2021). Biallelic deep intronic variant c.5457+81T>A in *TRIP11* causes loss of function and results in achondrogenesis 1A. *Human Mutation* DOI: 10.1002/humu.24235
3. Puneeth H Somashekar, Parneet Kaur, Joshi Stephen, Vishal Singh Guleria, Rajagopal Kadavigere, Katta Mohan Girisha, Stephanie Bielas, **Priyanka Upadhyai**, Anju Shukla (2021). Bi-allelic missense variant, p.Ser35Leu in *EXOSC1* is associated with pontocerebellar hypoplasia. *Clinical Genetics* doi: 10.1111/cge.13928.
4. Puneeth H Somashekar, **Priyanka Upadhyai**, Dhanya Lakshmi Narayanan, Nutan Kamath, Shruti Bajaj, Katta Mohan Girisha, Anju Shukla (2020). Phenotypic diversity and genetic complexity of Pax3 related Waardenberg syndrome. *American Journal of Medical Genetics Part A* DOI: 10.1002/ajmg.a.61893
5. **Priyanka Upadhyai**^{*#}, Vishal Singh Guleria[#], Prajna Udupa (2020). Characterization of primary cilia features reveal cell-type specific variability in in vitro models of osteogenic and chondrogenic differentiation. *PeerJ* DOI 10.7717/peerj.9799 (*corresponding author, # equal contribution)

6. **Priyanka Upadhyai**, Eram Fatima Amiri, Vishal Singh Guleria, Stephanie L. Bielas, Katta Mohan Girisha and Anju Shukla (2020). Recurrent 1q21.1 deletion syndrome: report on variable expression, non-penetrance and review of literature. *Clin Dysmorphology* 29:127-131
7. Caroline Hoppe, Johnathan Bowles, Thomas G. Michington, Catherine Sutcliffe, **Priyanka Upadhyai**, Magnus Rattray, Hilary Ashe (2020) Modulation of promoter occupancy dictates the transcriptional response to graded BMP signaling levels in the *Drosophila* embryo. *Dev. Cell* S1534-5807(20)30581-5
8. Ranajit Das, Vladimir A Ivanisenko, Anastasia A Anashkina, **Priyanka Upadhyai** (2020). The story of the lost twins: decoding the genetic identities of the Kumhar and Kurcha populations from the Indian subcontinent. *BMC Genetics* <https://doi.org/10.1186/s12863-020-00919-2>
9. Puneeth H Somashekar, **Priyanka Upadhyai**, Anju Shukla, Katta Mohan Girisha (2020). Novel splice site and nonsense variants in *INVS* cause Infantile Nephronophthisis. *Gene*. 729, 144229
10. Ranajit Das, **Priyanka Upadhyai** (2019). Investigating the West Eurasian ancestry of Pakistani Hazaras. *Journal of Genetics*. 98:43
11. Ranajit Das, **Priyanka Upadhyai** (2019). Adaptation of the Geographic Population Structure (GPS) algorithm for biogeographical analyses of non-human individuals: a case study of wild and captive Gorillas. *BMC Bioinformatics*. 20(Suppl 1): 35
12. Anusha Uttarilli, Hitesh Shah, Bhavani GS, **Priyanka Upadhyai**, Anju Shukla, Katta Mohan Girisha (2019). Phenotyping and genotyping of skeletal dysplasias: Evolution of a center and a decade of experience in India. *Bone* 120: 204-211
13. Ranajit Das, **Priyanka Upadhyai** (2018). An ancestry informative marker set which recapitulates the known fine structure of populations in South Asia. *Genome Biology and Evolution* 10(9): 2408-2416 <https://doi.org/10.1093/gbe/evy182>
14. Ranajit Das, **Priyanka Upadhyai** (2017). Application of geographic population structure (GPS) algorithm for biogeographical analyses of populations with complex ancestries: a case study of South Asians from 1000 genomes project. *BMC Genetics*. 18 (Suppl 1): 109 doi: 10.1186/s12863-017-0579-2.
15. Ranajit Das, **Priyanka Upadhyai** (2017). Unraveling the population history of Indian Siddis. *Genome Biology and Evolution*. 9 (6): 1385-92. <https://doi.org/10.1093/gbe/evx095>
16. Anju Shukla, Malavika Hebbar, Anshika Srivastava, Rajagopal Kadavigere, **Priyanka Upadhyai**, Anil Kanthi, Oliver Brandau, Stephanie Bielas, Katta Mohan Girisha (2017). Homozygous p.(Glu87Lys) variant in *ISCA1* is associated with a multiple mitochondrial dysfunctions syndrome. *Journal of Human Genetics*. 62(7): 723-727 doi: 10.1038/jhg.2017.35.
17. Anju Shukla, **Priyanka Upadhyai**, Jhanvi Shah, Neethukrishna K, Stephanie Bielas, Katta Mohan Girisha (2017). Autosomal Recessive Spinocerebellar Ataxia 20: Report of a New Patient and Review of Literature. *European Journal of Medical Genetics*. 60 (2): 118-123

18. **Priyanka Upadhyai** and Gerard Campbell. (2013). Brinker possesses multiple mechanisms for repression because its primary corepressor, Groucho, may be unavailable in some cell-types. *Development*. 140, 4256-4265.

- **Conference Oral and Poster Presentations**

1. **Priyanka Upadhyai**, Michelle C do Rosario, Puneeth H Somashekar, Katta M Girisha, Anju Shukla. Bi-allelic missense variant p.Thr368Ala in *DLST* is associated with 2-oxoglutarate dehydrogenase complex deficiency related neurometabolic disorder. Presented as an e-poster at American Society of Human Genetics virtual conference 18-22 October 2021.
2. **Priyanka Upadhyai**, Radhakrishnan Periyasamy, Vishal Singh Guleria, Shalini Nayak, Katta M Girisha. Biallelic deep intronic variants c.1528-126A>G and c.5457+81T>A in *TRIP11* are associated with achondrogenesis 1A. Presented as an e-poster at European Society of Human Genetics virtual conference 28-31 August 2021.
3. Prajna Udupa, Vishal Singh Guleria, Girisha KM and **Priyanka Upadhyai**. 'Investigating the role of *IFT52/Osm6* associated with a novel human ciliopathy in *Drosophila*'. Presented as a **poster** at 5th Asia-Pacific *Drosophila* Research Conference from 6th-10th January 2020 at Pune, India
4. Prajna Udupa, Vishal Singh Guleria and **Priyanka Upadhyai**. 'Investigating the role of *IFT52/Osm6* associated with a novel human ciliopathy in *Drosophila*'. Presented as a **poster** at 26th European *Drosophila* Research conference from 5-8 September, 2019 at Lausanne, Switzerland
5. Vishal Singh Guleria, Prajna Udupa, Girisha, KM. and **Priyanka Upadhyai**. 'Developing *in vitro* and *Drosophila* models for novel ciliopathies to investigate the role of *IFT52* and *EXOC6B* in modulating primary cilia and allied regulation of skeletal differentiation'. Presented as a **platform talk** at Indian Society of Developmental Biology (InSDB) conference from 11-15 December, 2018 at IIT Kanpur, India.
6. Vishal Singh Guleria, Girisha, KM and **Priyanka Upadhyai**. 'Investigating the ciliopathy associated genes *IFT52* and *EXOC6B*: insights from *Drosophila*.' Presented as a **poster** at Indian *Drosophila* Research Conference (InDRC) 2017 from 6th-9th December, 2017 at IISER Bhopal, India.
7. **Priyanka Upadhyai** and Gerard Campbell. 'Why do transcriptional repressors recruit more than one co-repressor?' Presented at the 53rd Annual *Drosophila* Genetics Conference from 7th to 11th March, 2012 at Chicago, IL, USA as a **platform talk**.
8. **Priyanka Upadhyai** and Gerard Campbell. 'How do transcriptional repressors function: Insights from Brinker.' Presented at Science2011, held on 6th and 7th October 2011 at the University of Pittsburgh, Pittsburgh, USA as a **poster**.
9. **Priyanka Upadhyai** and Gerard Campbell. 'Investigating mechanisms of transcriptional repression: Insights from Brinker.' Presented at the 52nd Annual *Drosophila* Genetics Conference, held from 30th March to 3rd April, 2011 at San Diego, USA as a **poster**.

• Invited Talks

1. ‘Models of rare monogenic disorder’ at the International Conference on Human Genetics and Genomics, Yazd University, Iran on 2nd December, 2021.
2. ‘In vitro and *Drosophila* models of rare monogenic disorders’ at the ‘Science Unlimited’ webinar series organized by Yenepoya Research Centre (YRC), Yenepoya (Deemed to be University), Mangalore, India on 23rd November 2021.
3. ‘Why Fly: *Drosophila* as a model for Mendelian disorders.’ Presented on 15th July 2020 at the meeting of the Society for Indian Academy of Medical Genetics (SIAMG), India.
4. ‘Investigating molecular mechanisms underlying ciliopathies using in vitro and *Drosophila* models.’ Presented on 12th June 2020 at Research Talk Series by Manipal Academy of Higher Education, Manipal, India.
5. ‘Developing in vitro and *Drosophila* models for novel ciliopathies to investigate the role of IFT52 and EXOC6B in modulating primary cilia and allied regulation of skeletal differentiation.’ Presented at 45th National Conference of Association of Clinical Biochemists of India (ACBICON 2018) at Kasturba Medical College, Manipal, India.
6. ‘Gene editing strategies to decipher *Drosophila* development and functional models of human ciliopathies’. Presented at Dept. of Bioscience and Bioengineering, IIT Guwahati, Assam, India on 12th June, 2018.
7. “Hemoglobinopathies-A genetics perspective”. Presented at 43rd National Conference of Association of Clinical Biochemists of India (ACBICON 2016) on 12th December 2016: “Hemoglobinopathies-Fundamentals and Challenges”, held at Kasturba Medical College, Mangalore, Karnataka, India
8. “Overview of chromatin biology and histone code and what it means for regulation of gene expression. Linking histone modifications and chromatin remodeling to transcription”. Presented at Continual Medical Education (CME): Epigenetics – from genome to epigenome on 19th October 2016 at Kasturba Medical College, Manipal, Karnataka, India.
9. “Shining light on chromatin modifications as drug targets and epigenome editing by CRISPR/Cas9”. Presented at Continual Medical Education (CME): Epigenetics – from genome to epigenome on 19th October 2016 at Kasturba Medical College, Manipal, Karnataka, India.
10. ‘Utilization of gene editing to decipher early *Drosophila* development’ 23rd April, 2016. Presented at the Department of Biochemistry, Kasturba Medical College, Manipal, Karnataka, India.
11. ‘*Drosophila* enters the era of Genomic Engineering: A high-resolution dissection of BMP signaling’ on 6th January 2015. Presented at Bose Institute, Kolkata, India
12. “Decoding transcriptional repression using genomic engineering in *Drosophila*” on 24th June 2013. Presented at the Department of Zoology, West Bengal State University, Kolkata, India.

• Students Supervised

(A) Doctoral students:

1. Vishal Singh Guleria (*thesis guide*) under the TMA Pai PhD Scholarship program, Manipal Academy of Higher Education, Manipal
2. Neha Quadri (*thesis guide*) under the TMA Pai PhD Scholarship program, Manipal Academy of Higher Education, Manipal
3. Michelle do Rosario (*thesis co-guide*) under the project 'Genetic diagnosis of neurodevelopmental disorders in India' funded by National Institutes of Health (NIH), USA

(B) Junior research fellows:

1. Rahul Parit under the ICMR project entitled 'Investigating the crosstalk between primary cilia and autophagy in chondrogenesis and its modulation by Fibroblast growth factor (FGF) signaling in FGFR3 related skeletal dysplasias *in vitro*'
Duration: April 2021-present
2. Prajna Udupa under the SERB project entitled 'Investigating the role of *IFT52* and *EXOC6B* in human biology and disease using cell-culture and *Drosophila* systems'.
Duration: July 2018-March 2020

● **Academic Achievements**

1. Selected to participate in Young Investigators' Meeting (YIM) 2019 sponsored by EMBO, Department of Biotechnology, Government of India and CACTUS Communication from 6-10 March, 2019 at Guwahati, India as Young Investigator.
2. Awarded Early Career Research award by Department of Science and Technology (DST) Science and Engineering Research Board (SERB), Govt. of India in 2016.
3. Awarded the Andrew Mellon Pre-doctoral fellowship for the year 2012-13 by Dietrich School of Arts and Sciences, University of Pittsburgh, USA.
The Dietrich School of Arts and Sciences, University of Pittsburgh, offers the Andrew Mellon fellowships competitively to pre-doctoral students from its 38 constituent departments. The student must be first nominated by the department, the awarding decision is then made by a selection committee that is a mix of faculty from various departments of the graduate school of Arts and Sciences. The award decision is based on a project proposal that is a concise statement of the dissertation project and work intended to be accomplished during the fellowship year, three letters of recommendation, and the applicant's CV.
4. Ranked 2nd with distinction in BSc. Zoology (Hons.), University of Calcutta, India
5. Ranked 2nd with distinction in MSc. Biotechnology, University of Calcutta, India

● **Administrative and Professional Responsibilities**

- Departmental PhD program coordinator
- Coordinator for short-term training courses in Medical Genetics
- Departmental quality coordinator for ISO, NABH quality and environmental policy documentation.
- Co-faculty in-charge for overall Departmental research lab management, inventory and book-keeping.
- Serving on the Doctoral Advisory Committee (DAC) of three predoctoral students registered at the Dept. Medical Genetics, Kasturba Medical College, Manipal Academy of Higher Education.

- **Workshops and short-term courses attended/completed**

1. A basic online, hands-on image analysis course organized by Centre for Cellular and Molecular Platforms (C-CAMP) held on 14-16 September, 2020
2. An advanced, hands-on course on image analysis organized by Centre for Cellular and Molecular Platforms (C-CAMP) held on 24-30 September, 2020

- **Professional Development**

1. Participated in 9th Revised Basic Course Workshop in Medical Education Technologies, conducted by St. John's Medical College, Bengaluru, Nodal Center in Medical Education Technologies, at Kasturba Medical College, Manipal (21st-23rd March, 2018).
2. Participated in a half-day author workshop on 'How to Plan & Write Manuscripts and Get Published in Scientific Journals?' Organized by SEARCH – The Health Sciences Library and Dept. of Library and Information Sciences, Manipal Academy of Higher Education, Manipal, India in association with Springer Nature (7th July, 2016).
3. Participated in a half-day author workshop on 'How to Plan & Write Manuscripts and Get Published in Scientific Journals?' by Health Sciences library MAHE with Springer Nature (7th July, 2016).

- **Scientific service**

Journal Reviewer: Frontiers in Genetics, All Life (Taylor and Francis)

- **Continued Medical Education Programs and Conferences Organized**

1. Faculty and Joint Organizing Secretary at the 'Manipal Genetics Update V on Genomics of Neurodevelopmental Disorders' held in February 2018 at Manipal Academy of Higher Education, Manipal, India.
2. Faculty and Joint Organizing Secretary at the 'Manipal Genetics Update IV: Genetic Counseling' held in February 2017 at Manipal Academy of Higher Education, Manipal, India.
3. Faculty and part of Organizing Committee for the 'Manipal Genetics Update III: Genetic Disorders of the Bone' held in Dec 2016 at Manipal Academy of Higher Education, Manipal, India.