

Department of Bioscience and Bioengineering Indian Institute of Technology Jodhpur

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Dr. Mitali Mukerji, FNASc, FAScProfessor and Head

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Letter of Recommendation

It gives me immense pleasure to nominate **Dr Mohammed Faruq**, a Senior Principal Scientist at the CSIR-Institute of Genomics and Integrative Biology (IGIB), Delhi for the **Sun Pharma Science Foundation Research Awards 2023**. Presently, I am a Professor and Head of Department of Bioscience and Bioengineering at IIT Jodhpur. Earlier, for over two decades, I worked in CSIR-IGIB and was heading the department of Genomics and Molecular Medicine.

I have known Dr Faruq for over a decade and half now since he joined CSIR-IGIB as a young scientist in our functional genomics group. Also as a fresh MBBS graduate, he joined for a PhD program in hereditary ataxia under the joint mentorship of mine and Prof Achal Srivastava at the Department of Neurology at AIIMS, Delhi. Faruq is a rare clinician scientist who works at the cutting edge of clinical genomics and has made prominent contributions in the area of rare hereditary disorders especially in hereditary ataxias. Through his efforts spanning over 15 years, he has been able to demonstrate Bed side to Bench research for hereditary ataxias at par with any international lad. AIIMS runs a unique ataxia clinic in the country which has a referral from all over the country, a major credit of which goes to him. His major research contribution in rare disorder research in the country includes:

- Setting up a paradigm to develop an algorithm for reducing the time to diagnosis and rapid cost-effective assays (long GAA repeats sequencing on Nanopore tech, Brain Communication 2023).
- Discovery of **novel molecular etiologies** using state-of-art genomics like the **NGS technologies** since its early days in **2012**.
- Establishment of one of the largest repositories of Indian hereditary ataxia in the world comprising of >7000 patients for research, discovery and translation.
- Addition of novel SCA subtypes in Indian population (Clinical genetics 2013, 2018), novel genes and mutations and development of targeted panels that resolves more than 75% of ataxias in India.
- Continued and sustained support to AIIMS to run a unique ataxia clinic in the country

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- that has led to publication of the largest data series (n~5500) of hereditary ataxias from India (Advanced Genetics, 2022) in multi-institutional collaboration.
- Detailed and systematic characterization of a uniquely prevalent form of SCA subtype
 "SCA12" in Indian population (Brain 2015). Nearly 500 families of SCA12 have been
 traced to a unique founder and a world's first SCA12 resource of pluripotent stem cell
 derived from diseased cell lines such as of SCA12 (Stem cell Research 2021)

Dr Faruq has also been able to make some prominent contributions to research through international collaborative efforts. His group demonstrated and validated a novel synthetic molecule that can overcome molecular deficiency for FRDA using patient derived cell lines in collaboration with Wisconsin University which was published in Science in 2017. Using his genomics expertise, he discovered a novel acromesomelic skeletal dysplasia by a novel gene cGMP-dependent type II protein kinase gene (PRKG2) in equal collaboration with a Spain group (Journal of Medical Genetics, 2022). He leads the transcriptomic and genomics analysis work in a collaborative work has led to a prominent publication "Kupyaphores are zinc homeostatic metallophores required for colonization of Mycobacterium tuberculosis" in PNAS 2022.

Dr Faruq has shown enormous team spirit and prodigious leadership qualities and has been instrumental in setting up the next generation sequencing platforms and its applications for various research activities in CSIR-IGIB and with other collaborating partners. He also runs a unique GOMED programme (http://gomed.igib.in) that provides tests to over 300 genetic disorders to the clinicians Pan-India. This has provided over 45000 tests to more than 11000 patients and many of these tests have been licensed to Dr Lal Path Lab pvt Ltd through Technology Transfer and brought royalty worth 35 lakhs to the institute. Through his clinical network he has enabled capacity building in this space for more than 100 PhDs as well as medical graduates.

Since March 2020, his lab was the site for RT-PCR for **Delhi Govt Covid-19 clusters surveillance** (Science 2021) and first 100 genomes of covid sequences were submitted by this lab from India. He also contributed in **methodology** development of a colorimetric based SARS CoV2 detection system, HRPzyme based for SARS-CoV2 genomes sequencing (HARIOM Biosens Bioelectron. 2021, IF~13.0).

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It is commendable that the entire work done by him has been in India. I am optimistic of his commitment and perseverance to excel in his endeavors. He is a role model of sorts for aspiring medical graduates who would like to take up a career with an interdisciplinary outlook in precision medicine. I very strongly recommend Dr Mohammed Faruq for the Sun Pharma Science Foundation Research award.

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