CURRICULUM VITAE

Name : DR. MOHAMMED FARUQ Designation : Senior Principal Scientist

Department/Institute/University: **CSIR-Institute of Genomics and Integrative Biology, Delhi**

Date of Birth: **11/05/1980** Sex (M/F) Male

EDUCATION (Post-Graduation onwards & Professional Career)

S. No.	Institution/ Place	Degree	Year	Field of Study
1	TD Medical College, Kerala University, Kerala	MBBS	Oct-2005	Medicine
2	AIIMS, New Delhi	Ph.D.	2014	Neurology

POSITION AND EMPLOYMENT (Starting with the most recent employment)

Sl No.	Institution / Place	Position	From (Date)	To (date)
1	CSIR-IGIB, New Delhi	Scientist" B	29.09.2006	29.9.2009
2	CSIR-IGIB, New Delhi	Scientist C	29.09.2009	29.9.2013
3	CSIR-IGIB, New Delhi	Senior Scientist (Scientist E1)	29.09.2013	29.09.2016
4	CSIR-IGIB, New Delhi	Principal Scientist (Scientist E2)	29.09.2016	29.09.2020
5	CSIR-IGIB, New Delhi	Senior Principal Scientist (Scientist F)	29.09.2020	Present

AWARDS AND RECOGNITION

- **Recipient of CSIR Young Scientist Award 2015 in Biological Sciences.**

PROFESSIONAL EXPERIENCE AND TRAINING RELEVANT TO THE PROJECT

Publications (Numbers only) 50+;

Book chapters: 3 Research Papers, Reports : 30

Patents : nil Others (Please specify) Best poster award in Intl. Conf .1

Selected presentations at National or International Conferences

1. **Invited Speaker at 26th Annual Conference of Indian Academy of Neurolgy, 2018, Raipur,** Frontiers in Neurogenetics, Explorative neurogenetics
2. **Invited speaker at 24th Annual Conference of Indian Academy of Neurology, 2016, Kolkata.** Advances in genetics of ataxias: Role of conventional versus next generation sequencing methods
3. **Invited speaker at NGBT, 2016, Cochin.** Genomics approach for Neurodegenerative Disorders: Cerebellar ataxia genetics as a prototype model
4. **Invited Speaker Genetics and Genomics of Neurological Disorders, ACBICON 2021.**
5. **Invited Speaker Genomics India Conference 2023, Bengaluru. Neurogenomics: Unraveling genic links to neurodegeneration in Session "Genomics in Research"**

Other Awards

Third prize for poster presentation on „Phenotype to Genotype correlations in spinocerebellar ataxias in Indian population: SCA2 as a case study." 2nd Asian and Oceanian Parkinson"s Disease and Movement disorders Congress (AOPMC), Delhi 2009.

Book Chapters

1. Avni Anand. **Faruq M**. Genetic Investigations CH-126 IAN Textbook of Neurology, 2e (Dr. Khadilkar and Dr. Gagandeep) 2023
2. Srivastava AK, **Faruq M.** Hereditary Ataxias: An overview and Indian Perspectives. Text Bookof Movement disorders, Edited by, Ashok Kumar, Jaypee Brothers Medical publishers Delhi 2014.pp.362-393.
3. Srivastava AK, **Faruq M.** Common Ataxias: An Indian perspective. Editors: Sunil K Narayan and Pratap Sancheti. Selected Topics in Tropical Neurology Publisher-Byword Books Pvt Ltd. Delhi 2015 pp 168-200.
4. Srivastava AK, **Faruq M.** Approach to Ataxia of Limbs and Gait. Editors: Sudesh Prabhakar and Gagandeep Singh. Differential Diagnosis in Neurology Publisher-Jaypee Brothers Medical Publishers. Delhi 2015 pp 251-268.

Membership

Member, Ataxia Study Group.Germany (http://www.ataxia-study-group.net/html/asg/imprint)

Ataxia Global Initiative.

10 Best Publications

1. Uppili, B., Sharma, P., Ahmad, I., Sahni, S., Asokachandran, V., Nagaraja, A. B., Srivastava, A. K., & **Faruq, M***. (2023). Sequencing through hyperexpanded Friedreich's ataxia-GAA repeats by nanopore technology: implications in genotype-phenotype correlation. *Brain communications*, *5*(2), fcad020. https://doi.org/10.1093/braincomms/fcad020
2. Sharma, P., Sonakar, A. K., Tyagi, N., Suroliya, V., Kumar, M., Kutum, R., Asokchandran, V., Ambawat, S., Shamim, U., Anand, A., Ahmad, I., Shakya, S., Uppili, B., Mathur, A., Parveen, S., Jain, S., Singh, J., Seth, M., Zahra, S., Joshi, A., ⋯ **Mohammed Faruq***, GOMED-Ataxia study group (2022). Genetics of Ataxias in Indian Population: A Collative Insight from a Common Genetic Screening Tool. *Advanced genetics (Hoboken, N.J.)*, *3*(2), 2100078. https://doi.org/10.1002/ggn2.202100078
3. Shamim U, Ambawat S, Singh J, Thomas A, Pradeep-Chandra-Reddy C, Suroliya V, Uppilli B, Parveen S, Sharma P, Chanchal S, Nashi S, Preethish-Kumar V, Vengalil S, Polavarapu K, Keerthipriya M, Mahajan NP, Reddy N, Thomas PT, Sadasivan A, Warrier M, Seth M, Zahra S,Mathur A, Vibha D, Srivastava AK, Nalini A, **Faruq M***. C9orf72 hexanucleotide repeat expansion in Indian patients with ALS: a common founder and its geographical predilection. *Neurobiol Aging*. 2020 Jan 3. pii: S0197-4580(19)30453-1.
4. Narang A, Uppilli B, Vivekanand A, Naushin S, Yadav A, Singhal K, Shamim U, Sharma P, Zahra S, Mathur A, Seth M, Parveen S, Vats A, Hillman S, Dolma P, Varma B, Jain V; TRISUTRA Ayurgenomics Consortium, Prasher B, Sengupta S, Mukerji M, **Faruq M*.** Frequency spectrum of rare and clinically relevant markers in multiethnic Indian populations (ClinIndb): A resource for genomic medicine in India.Hum Mutat. 2020 Nov;41(11):1833-1847. doi: 10.1002/humu.24102. Epub 2020 Sep 9.PMID: 32906206
5. Díaz-González F, Wadhwa S, Rodriguez-Zabala M, Kumar S, Aza-Carmona M, Sentchordi-Montané L, Alonso M, Ahmad I, Zahra S, Kumar D, Kushwah N, Shamim U, Sait H, Kapoor S, Roldán B, Nishimura G, Offiah AC, **Faruq M***, Heath KE.J. Biallelic cGMP-dependent type II protein kinase gene (*PRKG2*) variants cause a novel acromesomelic dysplasia. J. Med Genet. 2020 Oct 26:jmedgenet-2020-107177. doi: 10.1136/jmedgenet-2020-107177. Online ahead
6. Shakya S, Kumari R, Suroliya V, Tyagi N, Joshi A, Garg A, Singh I, Kalikavil Puthanveedu D, Cherian A, Mukerji M, Srivastava AK, **Faruq M***. Whole exome and targeted gene sequencing todetect pathogenic recessive variants in early onset cerebellar ataxia. *Clin Genet. 2019* Dec;96(6):566-574.
7. Erwin GS, Grieshop MP, Ali A, Qi J, Lawlor M, Kumar D, Ahmad I, McNally A, Teider N, Worringer K, Sivasankaran R, Syed DN, Eguchi A, Ashraf M, Jeffery J, Xu M, Park PMC, Mukhtar H, Srivastava AK, Faruq M, Bradner JE, Ansari AZ. Synthetic transcription elongationfactors license transcription across repressive chromatin *Science*. **2017** Dec 22;358(6370):1617-1622.

8. Srivastava AK, Takkar A, Garg A, **Faruq M***. Clinical behaviour of spinocerebellar ataxia type 12 and intermediate length abnormal CAG repeats in PPP2R2B. **Brain**. **2017** Jan;140(1):27-36.

9. Kumar D, Hussain A, Srivastava AK, Mukerji M, Mukherjee O, **Faruq M*.** Generation of three spinocerebellar ataxia type-12 patients derived induced pluripotent stem cell lines (IGIBi002-A,IGIBi003-A and IGIBi004-A). ***Stem Cell Res. 2018*** Aug;31:216-221.

10. Dhar MS, Marwal R, Vs R, Ponnusamy K, Jolly B, Bhoyar RC, Sardana V, Naushin S, Rophina M, Mellan TA, Mishra S, Whittaker C, Fatihi S, Datta M, Singh P, Sharma U, Ujjainiya R, Bhatheja N, Divakar MK, Singh MK, Imran M, Senthivel V, Maurya R, Jha N, Mehta P, A V, Sharma P, Vr A, Chaudhary U, Soni N, Thukral L, Flaxman S, Bhatt S, Pandey R, Dash D, **Faruq M**, Lall H, Gogia H, Madan P, Kulkarni S, Chauhan H, Sengupta S, Kabra S; Indian SARS-CoV-2 Genomics Consortium (INSACOG)‡, Gupta RK, Singh SK, Agrawal A, Rakshit P, Nandicoori V, Tallapaka KB, Sowpati DT, Thangaraj K, Bashyam MD, Dalal A, Sivasubbu S, Scaria V, Parida A, Raghav SK, Prasad P, Sarin A, Mayor S, Ramakrishnan U, Palakodeti D, Seshasayee ASN, Bhat M, Shouche Y, Pillai A, Dikid T, Das S, Maitra A, Chinnaswamy S, Biswas NK, Desai AS, Pattabiraman C, Manjunatha MV, Mani RS, Arunachal Udupi G, Abraham P, Atul PV, Cherian SS. Genomic characterization and epidemiology of an emerging SARS-CoV-2 variant in Delhi, India. Science. 2021 Nov 19;374(6570):995-999. doi: 10.1126/science.abj9932. Epub 2021 Oct 14.

11. Kumar P, Pandey R, Sharma P, Dhar MS, A V, Uppili B, Vashisht H, Wadhwa S, Tyagi N, Fatihi S, Sharma U, Singh P, Lall H, Datta M, Gupta P, Saini N, Tewari A, Nandi B, Kumar D, Bag S, Gahlot D, Rathore S, Jatana N, Jaiswal V, Gogia H, Madan P, Singh S, Singh P, Dash D, Bala M, Kabra S, Singh S, Mukerji M, Thukral L, **Faruq M***, Agrawal A, Rakshit P. Integrated genomic view of SARS-CoV-2 in India. Wellcome Open Res. 2020 Aug 3;5:184. doi: 10.12688/wellcomeopenres.16119.1. eCollection 2020.PMID: 32995557

Publications (80+ publications)

Ataxia and Neurology research

1. Uppili, B., Sharma, P., Ahmad, I., Sahni, S., Asokachandran, V., Nagaraja, A. B., Srivastava, A. K., & **Faruq, M***. (2023). Sequencing through hyperexpanded Friedreich's ataxia-GAA repeats by nanopore technology: implications in genotype-phenotype correlation. *Brain communications*, *5*(2), fcad020. https://doi.org/10.1093/braincomms/fcad020
2. Sharma P, Sonakar AK, Tyagi N, Suroliya V, Kumar M, Kutum R, Asokchandran V, Ambawat S, Shamim U, Anand A, Ahmad I, Shakya S, Uppili B, Mathur A, Parveen S, Jain S, Singh J, Seth M, Zahra S, Joshi A, Goel D, Sahni S, Kamai A, Wadhwa S, Murali A, Saifi S, Chowdhury D, Pandey S, Anand KS, Narasimhan RL, Laskar S, Kushwaha S, Kumar M, Shaji CV, Srivastava MVP, Srivastava AK, **Faruq M***; GOMED-Ataxia study group. Genetics of Ataxias in Indian Population: A Collative Insight from a Common Genetic Screening Tool. **Adv Genet** (Hoboken). 2022 Mar 10;3(2):2100078. doi: 10.1002/ggn2.202100078.
3. Garg D, Yoganathan S, Shamim U, Mankad K, Gulati P, Bonifati V, Botre A, Kalane U, Saini AG, Sankhyan N, Srivastava K, Gowda VK, Juneja M, Kamate M, Padmanabha H, Panigrahi D, Pachapure S, Udani V, Kumar A, Pandey S, Thomas M, Danda S, Iqbalahmed SA, Subramanian A, Pemde H, Singh V, **Faruq M*,** Sharma S. Clinical Profile and Treatment Outcomes of Hypermanganesemia with Dystonia 1 and 2 among 27 Indian Children. **Mov Disord Clin Pract**. 2022 Aug 12;9(7):886-899. doi: 10.1002/mdc3.13516.
4. Ahmad I, Goel D, Ghosh A, Kapoor H, Kumar D, Ramesh K, Ashley B, Deepika K, Shastry A, **Faruq M***. Generation of two induced pluripotent stem cell (iPSC) lines from patients with Duchenne muscular dystrophy (IGIBi006-A and IGIBi008-A) carrying exonic deletions in the dystrophin gene**. Stem Cell Res**. 2022 Oct;64:102927. doi: 10.1016/j.scr.2022.102927. Epub 2022 Sep 26.
5. Goyal M, **Faruq M**, Gupta A, Shrivastava D, Shamim U. The Clinical Diagnostic Utility of Array CGH in Children with Syndromic Microcephaly. **Ann Indian Acad Neurol**. **2022** Nov-Dec;25(6):1067-1074. doi: 10.4103/aian.aian_202_22. Epub 2022 Nov 17.
6. Narta K, Teltumbade MR, Vishal M, Sadaf S, Faruq M, Jama H, Waseem N, Rao A, Sen A, Ray K, Mukhopadhyay A. Whole Exome Sequencing Reveals Novel Candidate Genes in Familial Forms of Glaucomatous Neurodegeneration. **Genes (Basel). 2023** Feb 15;14(2):495. doi:

10.3390/genes14020495.

7. Agarwal A, Oinam R, Goel V, Sharma P, **Faruq M,** Garg A, Srivastava AK. "Ear of the Lynx" Sign in Hereditary Spastic Paraparesis (HSP) 76. **Mov Disord Clin Pract**. 2022 Nov 17;10(1):120-123. doi: 10.1002/mdc3.13606.
8. Rajeshwari M, Dhiman N, Chakrabarty B, Gulati S, Shamim U, **Faruq M**, Suri V, Sharma MC. X-linked Myopathy with Excessive Autophagy - A Rare Cause of Vacuolar Myopathy in Children. **Neurol India. 2022** Jul-Aug;70(4):1643-1648. doi: 10.4103/0028-3886.355110.
9. Nashi S, Polavarapu K, Bardhan M, Anjanappa RM, Preethish-Kumar V, Vengalil S, Padmanabha H, Geetha TS, Prathyusha PV, Ramprasad V, Joshi A, Chawla T, Unnikrishnan G, Sharma P, Huddar A, Uppilli B, Thomas A, Baskar D, Mathew S, Menon D, Arunachal G, **Faruq M**, Thangaraj K, Nalini A. Genotype-phenotype correlation and natural history study of dysferlinopathy: a single-centre experience from India. **Neurogenetics**. 2023 Jan;24(1):43-53. doi: 10.1007/s10048-022-00707-3. Epub 2022 Dec 29.
10. Mahajan S, Dhall A, Jassal B, Saluja A, **Faruq M**, Suri V, Rajan R, Vishnu VY, Sharma MC. Anoctamin-5 Muscular Dystrophy: Report of Two Cases with Different Phenotypes and Genotypes from the Indian Subcontinent. **Neurol India. 2022** Sep-Oct;70(5):2169-2173. doi: 10.4103/0028-3886.359155.
11. Panda S, Jain S, Dholakia D, Uppilli BR, **Faruq M**. Prolonged Episodic Dystonia in Tyrosine Hydroxylase Deficiency Due to Homozygous c.698G>A (p.Arg233His) Mutation-A Diagnostic Challenge. Mov Disord Clin Pract. 2022 Aug 10;9(8):1136-1139. doi: 10.1002/mdc3.13522.
12. Agarwal A, Das A, Pandit AK, Radhakrishnan DM, Rajan R, Faruq M, Srivastava AK. Allgrove Syndrome: A Frequently Under-Diagnosed ALS Mimic. Ann Indian Acad Neurol. 2022 May-Jun;25(3):500-501. doi: 10.4103/aian.aian_594_21. Epub 2021 Oct 22. PMID: 35936640; PMCID: PMC9350801.
13. Arshad F, Vengalil S, Nalini A, Polavarapu K, Shamim U, Jabeen S, Nagaraj C, Ramakrishnan S, **Faruq M***, Alladi S. Novel TBK1 variant associated with Frontotemporal Dementia overlap syndrome. Acta Neurol Scand. 2022 Apr;145(4):399-406. doi: 10.1111/ane.13562. Epub 2021 Nov 28. PMID: 34841512.
14. Vengalil S, Polavarapu K, Preethish-Kumar V, Nashi S, Arunachal G, Chawla T, Bardhan M, Mohan D, Christopher R, Bevinahalli N, Kulanthaivelu K, Nishino I, Faruq M, Nalini A. Mutation Spectrum of Primary Lipid Storage Myopathies. Ann Indian Acad Neurol. 2022 Jan-Feb;25(1):106-113. doi: 10.4103/aian.aian_333_21. Epub 2022 Feb 1. PMID: 35342266; PMCID: PMC8954319.
15. Sharma P, Sonakar AK, Goel V, Garg A, Srivastava AK, **Faruq M***. A Novel Co-existence of Spinocerebellar Ataxia 1 and Spinocerebellar Ataxia 2 Mutations in Indian Patients. Mov Disord Clin Pract. 2022 May 10;9(5):688-692. doi: 10.1002/mdc3.13464.
16. Tamuli D, Kaur M, Sethi T, Singh A, **Faruq M,** Jaryal AK, Srivastava AK, Kumaran SS, Deepak KK.Cortical and Subcortical Brain Area Atrophy in SCA1 and SCA2 Patients in India: The Structural MRI Underpinnings and Correlative Insight Among the Atrophy and Disease Attributes. **Neurol India.** 2021 Sep-Oct;69(5):1318-1325. doi: 10.4103/0028-3886.329596.
17. Mishra B, Rajan R, Gupta A, **Faruq M,** Shamim U, Parveen S, Garg A, Tripathi M, Vishnu VY, Singh MB, Bhatia R, Srivastava P. Cerebellar Ataxia in Adults with SQSTM1-Associated Frontotemporal Dementia-Amyotrophic Lateral Sclerosis Spectrum of Disorders. **Mov Disord Clin Pract.** 2021 Apr 28;8(5):800-802. doi: 10.1002/mdc3.13218. eCollection 2021 Jul.
18. Polavarapu K, Mathur A, Joshi A, Nashi S, Preethish-Kumar V, Bardhan M, Sharma P, Parveen S, Seth M, Vengalil S, Chawla T, Shingavi L, Shamim U, Nayak S, Vivekanand A, Töpf A, Roos A, Horvath R, Lochmüller H, Nandeesh B, Arunachal G, Nalini A, **Faruq M*.** A founder mutation in the GMPPB gene [c.1000G > A (p.Asp334Asn)] causes a mild form of limb-girdle muscular dystrophy/congenital myasthenic syndrome (LGMD/CMS) in South Indian patients. **Neurogenetics. 2021** Oct;22(4):271-285. doi: 10.1007/s10048-021-00658-1. Epub 2021 Aug 1.
19. Siddiqui S, Polavarapu K, Bardhan M, Preethish-Kumar V, Joshi A, Nashi S, Vengalil S, Raju S, Chawla T, Leena S, Mathur A, Nayak S, Mohan D, Shamim U, Prasad C, Lochmüller H**, Faruq M**, Nalini A.J Distinct and Recognisable Muscle MRI Pattern in a Series of Adults Harbouring an Identical GMPPB Gene Mutation. Neuromuscul Dis. 2022;9(1):95-109. doi: 10.3233/JND-200628.
20. Singh I, Swarup V, Shakya S, Kumar V, Gupta D, Rajan R, Radhakrishnan DM, **Faruq M,** Srivastava AK. Impact of SARS- CoV-2 Infection in Spinocerebellar Ataxia 12 Patients. **Mov Disord. 2021** Nov;36(11):2459-2460. doi: 10.1002/mds.28811. Epub 2021 Oct 7.

21. Agarwal A, Kaur H, Agarwal A, Nehra A, Pandey S, Garg A, **Faruq M**, Rajan R, Shukla G, Goyal V, Srivastava AK. Cognitive impairment in spinocerebellar ataxia type 12. Parkinsonism Relat Disord. 2021 Mar 13;85:52-56. doi: 10.1016/j.parkreldis.2021.03.010. Online ahead of print.PMID: 33740701

22. Shamim U, Ambawat S, Singh J, Thomas A, Pradeep-Chandra-Reddy C, Suroliya V, Uppilli B, Parveen S, Sharma P, Chanchal S, Nashi S, Preethish-Kumar V, Vengalil S, Polavarapu K, Keerthipriya M, Mahajan NP, Reddy N, Thomas PT, Sadasivan A, Warrier M, Seth M, Zahra S,Mathur A, Vibha D, Srivastava AK, Nalini A, **Faruq M***. C9orf72 hexanucleotide repeat expansion in Indian patients with ALS: a common founder and its geographical predilection. ***Neurobiol Aging***. 2020 Jan 3. pii: S0197-4580(19)30453-1.

23. Narang A, Uppilli B, Vivekanand A, Naushin S, Yadav A, Singhal K, Shamim U, Sharma P, Zahra S, Mathur A, Seth M, Parveen S, Vats A, Hillman S, Dolma P, Varma B, Jain V; TRISUTRA Ayurgenomics Consortium, Prasher B, Sengupta S, Mukerji M, **Faruq M*.** Frequency spectrum of rare and clinically relevant markers in multiethnic Indian populations (ClinIndb): A resource for genomic medicine in India.Hum Mutat. 2020 Nov;41(11):1833-1847. doi: 10.1002/humu.24102. Epub 2020 Sep 9.PMID: 32906206

24. Pathak P, Sharma MC, Jha P, Sarkar C, **Faruq M**, Jha P, Suri V, Bhatia R, Singh S, Gulati S, Husain M.J Mutational Spectrum of CAPN3 with Genotype-Phenotype Correlations in Limb Girdle Muscular Dystrophy Type 2A/R1 (LGMD2A/LGMDR1) Patients in India. Neuromuscul Dis. 2021;8(1):125-136. doi: 10.3233/JND-200547.PMID: 33337384

25. Mahadevan R, Bhoyar RC, Viswanathan N, Rajagopal RE, Essaki B, Suroliya V, Chelladurai R, Sankaralingam S, Shanmugam G, Vayanakkan S, Shamim U, Mathur A, Jain A, Imran M, **Faruq M**, Scaria V, Sivasubbu S, Kalyanaraman S. Genomic analysis of patients in a South Indian Community with autosomal dominant cortical tremor, myoclonus and epilepsy suggests a founder repeat expansion mutation in the SAMD12 gene. Brain Commun. 2020 Dec 19;3(1):fcaa214. doi: 10.1093/braincomms/fcaa214. eCollection 2021.PMID: 33501421

26. Kumar M, Wadhwa S, Tyagi N, Ahmad I, Kumar S, Sagar S, Zahra S, Kamai A, Shamim U, Kapoor S, **Faruq M*.**Stem Cell Res. Generation of induced pluripotent stem cell line (IGIBi007-A) from a patient with a novel acromesomelic dysplasia, PRKG2 type (AMDP). 2021 Apr 19;53:102340. doi: 10.1016/j.scr.2021.102340.

27. Díaz-González F, Wadhwa S, Rodriguez-Zabala M, Kumar S, Aza-Carmona M, Sentchordi-Montané L, Alonso M, Ahmad I, Zahra S, Kumar D, Kushwah N, Shamim U, Sait H, Kapoor S, Roldán B, Nishimura G, Offiah AC, **Faruq M***, Heath KE.J. Biallelic cGMP-dependent type II protein kinase gene (*PRKG2*) variants cause a novel acromesomelic dysplasia. J. Med Genet. 2020 Oct 26:jmedgenet-2020-107177. doi: 10.1136/jmedgenet-2020-107177. Online ahead of print.PMID: 33106379.

28. Goel D, Suroliya V, Shamim U, Mathur A, **Faruq M***. Spinocerebellar ataxia type 10 (SCA10):Mutation analysis and common haplotype based inference suggest its rarity in Indian population. ***NeurologicalSci***. 2019 Oct 24;17:100211.

29. Panda I, Ahmad I, Sagar S, Zahra S, Shamim U, Sharma S, **Faruq M*.** Encephalopathy due to defective mitochondrial and peroxisomal fission 2 caused by a novel MFF gene mutation in a young child. Clin Genet. 2020 Jun;97(6):933-937. doi: 10.1111/cge.13740. Epub 2020 Mar 24.PMID: 32181496

30. GUaRDIAN Consortium, Sivasubbu S, Scaria V.Genomics of rare genetic diseases-experiencesfrom India. Hum Genomics. 2019 Sep 25;14(1):52. doi: 10.1186/s40246-019-0215-5. Review.

31. Shakya S, Kumari R, Suroliya V, Tyagi N, Joshi A, Garg A, Singh I, Kalikavil Puthanveedu D, Cherian A, Mukerji M, Srivastava AK, **Faruq M***. Whole exome and targeted gene sequencing todetect pathogenic recessive variants in early onset cerebellar ataxia. *Clin Genet. 2019* Dec;96(6):566-574.

32. **Faruq M***, Kumar D, Wadhwa S, Shamim U, Mathur A, Parveen S, Garg A, Srivastava AK. Intrafamilial variable spastic paraplegia/ataxia/ALS phenotype linked to a novel KIF5A mutation.*Clin Genet. 2019* Sep;96(3):271-273.

33. Prasad S, Shamim U, Minj A, **Faruq M**, Pal PK. Manganism without Parkinsonism: IsolatedUnilateral Upper Limb Tremor in a Welder. *J Mov Disord.* 2019 May;12(2):135-137. doi: 10.14802/jmd.18068. Epub 2019 Apr 5.

34. Kumar D, Hussain A, Srivastava AK, Mukerji M, Mukherjee O, **Faruq M*.** Generation of three spinocerebellar ataxia type-12 patients derived induced pluripotent stem cell lines (IGIBi002-A,IGIBi003-A and IGIBi004-A). *Stem Cell Res. 2018* Aug;31:216-221.

35. Ramos A, Planchat M, Vieira Melo AR, Raposo M, Shamim U, Suroliya V, Srivastava AK, FaruqM, Morino H, Ohsawa R, Kawakami H, Bannach Jardim L, Saraiva-Pereira ML, Vasconcelos J, Santos C, Lima M. Mitochondrial DNA haplogroups and age at onset of Machado-Joseph disease/spinocerebellar ataxia type 3: a study in patients from multiple populations. *Eur J Neurol. 2019* Mar;26(3):506-512.

36. Erwin GS, Grieshop MP, Ali A, Qi J, Lawlor M, Kumar D, Ahmad I, McNally A, Teider N, Worringer K, Sivasankaran R, Syed DN, Eguchi A, Ashraf M, Jeffery J, Xu M, Park PMC, Mukhtar H, Srivastava AK, Faruq M, Bradner JE, Ansari AZ. Synthetic transcription elongationfactors license transcription across repressive chromatin *Science*. **2017** Dec 22;358(6370):1617-1622.

37. Kumari R, Kumar D, Brahmachari SK, Srivastava AK, Faruq M, Mukerji M. Paradigm for disease deconvolution in rare neurodegenerative disorders in Indian population: insights fromstudies in cerebellar ataxias. **J Genet. 2018** Jul;97(3):589-609.

38. Srivastava AK, Takkar A, Garg A, **Faruq M***. Clinical behaviour of spinocerebellar ataxia type 12 and intermediate length abnormal CAG repeats in PPP2R2B. **Brain**. **2017** Jan;140(1):27-36.

39. **Faruq M*,** Magaña JJ, Suroliya V, Narang A, Murillo-Melo NM, Hernández-Hernández O, Srivastava AK, Mukerji M A Complete Association of an intronic SNP rs6798742 with Origin of Spinocerebellar Ataxia Type 7-CAG Expansion Loci in the Indian and Mexican Population. **AnnHum Genet**. **2017** Sep;81(5):197-204.

40. Singh I, Swarup V, Shakya S, Goyal V, Faruq M, Srivastava AK. Single-step blood direct PCR:A robust and rapid method to diagnose triplet repeat disorders. **J Neurol Sci. 2017** Aug 15;379:49-54.

41. Singh I, Shakya S, Singh RK, Ahmad I, Goyal V, Shukla G, Srivastava MV, **Faruq M***, Srivastava AK. Iron related hemochromatosis (HFE) gene mutations in Friedreich Ataxiapatients. **Parkinsonism Relat Disord. 2017** Jan;34:71-72.

42. Singh I, **Faruq M***, Padma MV, Goyal V, Behari M, Grover A, Mukerji M, Srivastava AK. Investigation of mitochondrial DNA variations among Indian Friedreich's ataxia patients.**Mitochondrion**. **2015** Nov;25:1-5.

43. **Faruq M,** Srivastava AK, Singh S, Gupta R, Dada T, Garg A, Behari M, Mukerji M. Spinocerebellar ataxia 7 in Indian population: predilection of ATXN7-CAG expansion mutation inan ethnic population. **Indian J Med Res**. **2015** Feb;141(2):187-98.

44. **Faruq M*,** Srivastava AK, Suroliya V, Kumar D, Garg A, Shukla G, Behari M. Identification ofFXTAS presenting with SCA 12 like phenotype in India. **Parkinsonism Relat Disord**. **2014** Oct;20(10):1089-93.

45. **Faruq M,** Narang A, Kumari R, Pandey R, Garg A, Behari M, Dash D, Srivastava AK, Mukerji M. Novel mutations in typical and atypical genetic loci through exome sequencing in autosomalrecessive cerebellar ataxia families. **Clin Genet. 2014** Oct;86(4):335-41.

46. Vats A, Gourie-Devi M, Suroliya V, Verma S, Faruq M, Sharma A, Ganguly NK, Kukreti R, WajidS, Taneja V. J Analysis of C9orf72 repeat expansion in amyotrophic lateral sclerosis patients from North India. **Neurol Sci. 2017** Feb 15;373:55- 57.

Clinical Genomics and Genomics

1. Khandelwal P, Joshi A, Mathur A, Puraswani M, Gurjar BS, Sinha A, Hari P, Faruq M, Bagga A. Variants in complement genes are uncommon in patients with anti-factor H autoantibody-associated atypical hemolytic uremic syndrome. Pediatr Nephrol. 2023 Jan 9. doi: 10.1007/s00467-022-05862-1. Epub ahead of print. PMID: 36622444.
2. Sood A, Shamim U, Kharbanda OP, Kabra M, Gupta N, Mathur A, Joshi A, Parveen S, Zahra S, Sharma P, Seth M, Khan A, Faruq M, Mishra D. Next Generation Sequencing and Cytogenetic Based Evaluation of Indian Pierre Robin Sequence Families Reveals CNV Regions of Modest Effect and a Novel LOXL3 Mutation. **Cleft Palate Craniofac J. 2021 Nov 17**:10556656211052781. doi: 0.1177/10556656211052781.
3. Kumar A, Sharma R, Faruq M, Kumar M, Sharma S, Werner R, Hiort O, Vandana J. Clinical, Biochemical, and Molecular Characterization of Indian Children with Clinically Suspected Androgen Insensitivity Syndrome. **Sex Dev. 2022**;16(1):34-45. doi: 10.1159/000519047. Epub 2021 Oct 22.
4. Joshi A, Sinha A, Sharma A, Shamim U, Uppilli B, Sharma P, Zahra S, Parveen S, Mathur A, Chandan M, Tewari P, Khandelwal P, Hari P, Mukerji M, **Faruq M**, Bagga A; NephQuest Consortium. Next-Generation Sequencing for Congenital Nephrotic Syndrome: A Multi-Center Cross-Sectional Study from India. **Indian Pediatr. 2021** May 15;58(5):445-451.PMID: 33980730.
5. Naskar T, Faruq M, Banerjee P, Khan M, Midha R, Kumari R, Devasenapathy S, Prajapati B, Sengupta S, Jain D,
Mukerji M, Singh NC, Sinha S. Ancestral Variations of the PCDHG GeneCluster Predispose to Dyslexia in a Multiplex Family. **EBioMedicine**. **2018** Feb;28:168-179.
6. Juneja M, Shamim U, Joshi A, Mathur A, Uppili B, Sairam S, Ambawat S, Dixit R, **Faruq M*** Anovel mutation in SLC39A14 causing hypermanganesemia associated with infantile onset dystonia.. **J Gene**

Med. **2018** Mar 2:e3012.

7. Singh J, Muhammad PK, Jain S, Mathur A, Parveen S, Joshi A, Uppili B, Shaji CV, Kabeer KA,Menon S, **Faruq M***. A novel mutation in SGSH causing Sanfillipo type 3A Mucopolysaccharidoses in an Indian family. Mol Genet Metab Rep. 2018 Apr 18;15:124-126.

8. Jain S, Chaitanya V, **Faruq M***. A novel frameshift deletion in NAGLU causing sanfilipo type III-B in an Indian family.
Clin Case Rep. **2018** Oct 26;6(12):2399-2402.

9. Khera D, John J, Singh K, **Faruq M**. Tay-Sachs disease: a novel mutation from India. **BMJCase Rep. 2018** Dec 13;11(1). pii: e225916.

10. Prasad S, Shamim U, Minj A, Faruq M, Pal PK. Manganism without Parkinsonism: IsolatedUnilateral Upper Limb Tremor in a Welder. **J Mov Disord. 2019** May;12(2):135-137.

11. Netravathi M, Kumari R, Kapoor S, Dakle P, Dwivedi MK, Roy SD, Pandey P, Saini J, Ramakrishna A , Navalli D, Satishchandra P, Pal PK , Kumar A and **Faruq M** (*Correspondingauthors). Whole exome sequencing in an Indian family links Coats plus syndrome and dextrocardia with a homozygous novel CTC1 and a rare HES7 variation. **BMC Med Genet. 2015** Feb 10;16:5.

12. Hariprakash JM, Vellarikkal SK, Keechilat P, Verma A, Jayarajan R, Dixit V, Ravi R, SenthivelV, Kumar A, Sehgal P, Sonakar AK, Ambawat S, Giri AK, Philip A, Sivadas A, Faruq M, Bharadwaj D, Sivasubbu S, Scaria V. Pharmacogenetic landscape of DPYD variants in south Asian populations by integration of genome-scale data. **Pharmacogenomics. 2018** Feb;19(3):227-241.

13. Gowda VK, Bhat MD, Srinivasan VM, Prasad C, Benakappa A, Faruq M. A case of Canavandisease with microcephaly.
Brain Dev. 2016 Mar 15. pii: S0387-7604(16)30005-5.

Other national collaborative efforts

1. Mehdiratta K, Singh S, Sharma S, Bhosale RS, Choudhury R, Masal DP, Manocha A, Dhamale BD, Khan N, Asokachandran V, Sharma P, Ikeh M, Brown AC, Parish T, Ojha AK, Michael JS, Faruq M, Medigeshi GR, Mohanty D, Reddy DS, Natarajan VT, Kamat SS, Gokhale RS. Kupyaphores are zinc homeostatic metallophores required for colonization of *Mycobacterium tuberculosis*. **Proc Natl Acad Sci U S A**. 2022 Feb 22;119(8):e2110293119. doi: 10.1073/pnas.2110293119. PMID: 35193957; PMCID: PMC8872721.

2. Vasudevan A, Thergaonkar R, Mantan M, Sharma J, Khandelwal P, Hari P, Sinha A, Bagga A; Expert Group Of The Indian Society Of Pediatric Nephrology. Consensus Guidelines on Management of Steroid-Resistant Nephrotic Syndrome.Indian Pediatr. 2021 Jan 4:S097475591600278. Online ahead of print.PMID: 33408286

3. Kaur R, Attri SV, Saini AG, Sankhyan N, Singh S, **Faruq M**, Ramprasad VL, Sharda S, Murugan S. Seven novel genetic variants in a North Indian cohort with classical homocystinuria. Sci Rep. 2020 Oct 14;10(1):17299. doi: 10.1038/s41598-020-73475-5.PMID: 33057012 **.**

4. Kumar A, Sharma R, **Faruq M**, Suroliya V, Kumar M, Sharma S, Werner R, Hiort O, Jain V. Spectrum of Pathogenic Variants in SRD5A2 in Indian Children with 46,XY Disorders of Sex Development and Clinically Suspected Steroid 5α- Reductase 2 Deficiency Sex Dev. 2019;13(5-6):228-239. doi: 10.1159/000509812. Epub 2020 Sep 5.PMID: 32894851

5. Tyagi R, Kumar S, Dalal A, **Faruq M**, Mohanty M, Kaur P, Anand A. Repurposing Pathogenic Variants of *DMD* Gene and its Isoforms for DMD Exon Skipping Intervention.Curr Genomics. 2019 Nov;20(7):519-530. doi: 10.2174/1389202920666191107142754.PMID: 32655290

6. Passi GR, Shamim U, Rathore S, Joshi A, Mathur A, Parveen S, Sharma P, Crow YJ, **Faruq M.** An Indian child with Coats plus syndrome due to mutations in STN1. Am J Med Genet A. 2020 Sep;182(9):2139-2144. doi: 10.1002/ajmg.a.61737. Epub 2020 Jul 6.PMID: 32627942

7. Kumari K, Sharma MC, Kakkar A, Malgulwar PB, Pathak P, Suri V, Sarkar C, Chandra SP, **Faruq M.** mTOR pathway activation in focal cortical dysplasia. Ann Diagn Pathol. 2020 Jun;46:151523. doi: 10.1016/j.anndiagpath.2020.151523. Epub 2020 Apr 10.PMID: 32325422

8. Malgulwar PB, Nambirajan A, Pathak P, Faruq M, Rajeshwari M, Singh M, Suri V, Sarkar C, Sharma MC. C11orf95- RELA fusions and upregulated NF-KB signalling characterise a subsetof aggressive supratentorial ependymomas that express L1CAM and nestin. **J Neurooncol. 2018** Jan 22.

9. Jain D, Ramachandrappa VS, Singh V, Malik PS, Madan K, Faruq M, Guleria R Use of Exfoliative Specimens and Fine-Needle Aspiration Smears for Mutation Testing in LungAdenocarcinoma.. **Acta Cytol. 2017**;61(6):455- 461.

10. Malgulwar PB, Nambirajan A, Pathak P, Faruq M, Suri V, Sarkar C, Jagdevan A, Sharma BS,Sharma MC Study of β- catenin and BRAF alterations in adamantinomatous and papillary craniopharyngiomas: mutation analysis with immunohistochemical correlation in 54 cases.. **J Neurooncol. 2017** Jul;133(3):487-495.

11. Kumar P, Misra S, Kumar A, Faruq M, Shakya S, Vardhan G, Vivekanandhan S, Srivastava AK, Prasad K. Transforming growth factor-β1 (C509T, G800A, and T869C) gene polymorphisms and risk of ischemic stroke in North Indian population: A hospital-based case-control study. **AnnIndian Acad Neurol. 2017** Jan-Mar;20(1):5-12.

12. Kumari K, Sharma MC, Kakkar A, Malgulwar PB, Pathak P, Suri V, Sarkar C, Chandra SP,Faruq M, Gupta RK, Saran RK Role of mTOR signaling pathway in the pathogenesis of subependymal giant cell astrocytoma - A study of 28 cases.. **Neurol India. 2016** Sep- Oct;64(5):988-94.

13. Pathak P, Kumar A, Jha P, Purkait S, Faruq M, Suri A, Suri V, Sharma MC, Sarkar C. Genetic alterations related to BRAF- FGFR genes and dysregulated MAPK/ERK/mTOR signaling in adultpilocytic astrocytoma. **Brain Pathol. 2017** Sep;27(5):580-589. doi: 10.1111/bpa.12444. Epub 2017 Jan 6.

14. Kumar P, Kumar A, Sagar R, Misra S, Faruq M, Suroliya V, Vivekanandhan S, Srivastava AK,Prasad K. Association between interleukin-6 (G174C and C572G) promoter gene polymorphisms and risk of ischemic stroke in North Indian population: a case-control study. **Neurol Res. 2016** Jan;38(1):69-74.

15. Kumar P, Kumar A, Misra S, Sagar R, Faruq M, Suroliya V, Vivekanandhan S, Srivastava AK, Prasad K. Tumor necrosis factor-alpha gene polymorphisms and risk of ischemic stroke in northIndian population: A hospital based case-control study. **Meta Gene. 2015** Nov 17;7:34-9.

COVID research (2020-2022)

1. Dhar MS, Marwal R, Vs R, Ponnusamy K, Jolly B, Bhoyar RC, Sardana V, Naushin S, Rophina M, Mellan TA, Mishra S, Whittaker C, Fatihi S, Datta M, Singh P, Sharma U, Ujjainiya R, Bhatheja N, Divakar MK, Singh MK, Imran M, Senthivel V, Maurya R, Jha N, Mehta P, A V, Sharma P, Vr A, Chaudhary U, Soni N, Thukral L, Flaxman S, Bhatt S, Pandey R, Dash D, Faruq M, Lall H, Gogia H, Madan P, Kulkarni S, Chauhan H, Sengupta S, Kabra S; Indian SARS-CoV-2 Genomics Consortium (INSACOG)‡, Gupta RK, Singh SK, Agrawal A, Rakshit P, Nandicoori V, Tallapaka KB, Sowpati DT, Thangaraj K, Bashyam MD, Dalal A, Sivasubbu S, Scaria V, Parida A, Raghav SK, Prasad P, Sarin A, Mayor S, Ramakrishnan U, Palakodeti D, Seshasayee ASN, Bhat M, Shouche Y, Pillai A, Dikid T, Das S, Maitra A, Chinnaswamy S, Biswas NK, Desai AS, Pattabiraman C, Manjunatha MV, Mani RS, Arunachal Udupi G, Abraham P, Atul PV, Cherian SS. Genomic characterization and epidemiology of an emerging SARS-CoV-2 variant in Delhi, India. **Science. 2021** Nov 19;374(6570):995-999. doi: 10.1126/science.abj9932. Epub 2021 Oct 14.

2. Pathak AK, Mishra GP, Uppili B, Walia S, Fatihi S, Abbas T, Banu S, Ghosh A, Kanampalliwar A, Jha A, Fatma S, Aggarwal S, Dhar MS, Marwal R, Radhakrishnan VS, Ponnusamy K, Kabra S, Rakshit P, Bhoyar RC, Jain A, Divakar MK, Imran M, Faruq M, Sowpati DT, Thukral L, Raghav SK, Mukerji M. Spatio-temporal dynamics of intra-host variability in SARS-CoV-2 genomes. **Nucleic Acids Res. 2022 Feb** 22;50(3):1551-1561. doi: 10.1093/nar/gkab1297. PMID: 35048970; PMCID: PMC8860616.

3. Singh I, Swarup V, Shakya S, Kumar V, Gupta D, Rajan R, Radhakrishnan DM, Faruq M, Srivastava AK. Impact of SARS-CoV-2 Infection in Spinocerebellar Ataxia 12 Patients. Mov Disord. 2021 Nov;36(11):2459-2460. doi: 10.1002/mds.28811. Epub 2021 Oct 7. PMID: 34529277; PMCID: PMC8662285.

4. Mehta P, Sahni S, Siddiqui S, Mishra N, Sharma P, Sharma S, Tyagi A, Chattopadhyay P, Vivekanand A, Devi P, Khan A, Waghdhare S, Budhiraja S, Uppili B, Maurya R, Nangia V, Shamim U, Hazarika PP, Wadhwa S, Tyagi N, Dewan A, Tarai B, Das P, **Faruq M,** Agrawal A, Jha S, Pandey R. Respiratory Co-Infections: Modulators of SARS-CoV-2 Patients' Clinical Sub-Phenotype. **Front Microbiol. 2021** May 28;12:653399. doi: 10.3389/fmicb.2021.653399. eCollection 2021.

5. Kale P, Gupta E, Bihari C, Patel N, Rooge S, Pandey A, Bajpai M, Khillan V, Chattopadhyay P, Devi P, Maurya R, Jha N, Mehta P, Kumar M, Sharma P, Saifi S, Swaminathan A, Alam S, Uppili B, Faruq M, Agrawal A, Pandey R, Sarin SK. Vaccine Breakthrough Infections by SARS-CoV-2 Variants after ChAdOx1 nCoV-19 Vaccination in Healthcare Workers. Vaccines (Basel). 2021 Dec 31;10(1):54. doi: 10.3390/vaccines10010054. PMID: 35062715; PMCID: PMC8778656.

6. Ahmad M, Sharma P, Kamai A, Agrawal A, **Faruq M,** Kulshreshtha A. HRPZyme Assisted Recognition of SARS-CoV-2 infection by Optical Measurement (HARIOM). **Biosens Bioelectron. 2021** Sep 1;187:113280. doi: 10.1016/j.bios.2021.113280. Epub 2021 May 1.

7. Dhar MS, Asokachandran V, Uppili B, Tyagi N, Sharma P, Tiwari S, Srinivasan RV, Marwal R, Kanakan A, Khan AM, Pandey R, Jais M, Gogoi S, Shewale A, Nale T, Kabra S, **Faruq M**, Singh S, Agrawal A, Rakshit P. Reinfection or

   reactivation: Genome-based two distinct SNP profile of SARS-CoV2 repositivity in an Indian case. **J Med Virol**. 2021 Mar

22. doi: 10.1002/jmv.26948. Online ahead of print.PMID: 33749850

8. Bhoyar RC, Jain A, Sehgal P, Divakar MK, Sharma D, Imran M, Jolly B, Ranjan G, Rophina M, Sharma S, Siwach S, Pandhare K, Sahoo S, Sahoo M, Nayak A, Mohanty JN, Das J, Bhandari S, Mathur SK, Kumar A, Sahlot R, Rojarani P, Lakshmi JV, Surekha A, Sekhar PC, Mahajan S, Masih S, Singh P, Kumar V, Jose B, Mahajan V, Gupta V, Gupta R, Arumugam P, Singh A, Nandy A, P V R, Jha RM, Kumari A, Gandotra S, Rao V, **Faruq M**, Kumar S, Reshma G B, Varma G N, Roy SS, Sengupta A, Chattopadhyay S, Singhal K, Pradhan S, Jha D, Naushin S, Wadhwa S, Tyagi N, Poojary M, Scaria V, Sivasubbu S. High throughput detection and genetic epidemiology of SARS-CoV-2 using COVIDSeq next-generation sequencing. PLoS One. 2021 Feb 17;16(2):e0247115. doi: 10.1371/journal.pone.0247115. eCollection 2021.PMID: 33596239.

8. Kumar P, Pandey R, Sharma P, Dhar MS, A V, Uppili B, Vashisht H, Wadhwa S, Tyagi N, Fatihi S, Sharma U, Singh P, Lall H, Datta M, Gupta P, Saini N, Tewari A, Nandi B, Kumar D, Bag S, Gahlot D, Rathore S, Jatana N, Jaiswal V, Gogia H, Madan P, Singh S, Singh P, Dash D, Bala M, Kabra S, Singh S, Mukerji M, Thukral L, **Faruq M**, Agrawal A, Rakshit P. Integrated genomic view of SARS-CoV-2 in India. Wellcome Open Res. 2020 Aug 3;5:184. doi: 10.12688/wellcomeopenres.16119.1. eCollection 2020.PMID: 32995557

Research Project
As a Principle Investigator

1. Identification of blood based biomarkers in Friedeich's Ataxia using single cell transcriptomics. ICMR Funded project (2023-2026)
2. A large-scale comprehensive clinical and genomics approach for rare mutations and novel gene discovery in hereditary ataxias in India-CHANGER, ICMR Funded Project (2021-2024)
3. GenomeIndia, A DBT funded project (2020-2024)
4. Genomics and other Omics tools for Enabling Medical Decisions (GOMED), CSIR funded(2016-2018), 15 Crores
5. GOMED-Tech: Development, Translation and Commercialization of Genetic tests for prevalent genetic diseases in India, CSIR-Funded (2018-2020)
6. Elucidation of molecular pathogenesis in SCAs using IPS cell model and Next Generation Sequencing, CSIR funded GeNCODE, (2012-2017)

Collaborative project of other centres as Co-PI

1. **Co-Principal Investigator** in a DBT supported "Pediatric Renal Biology Program on Nephrotic Syndrome subproject entitled "Whole exome analysis for mutations in genes encoding key podocyte proteins" (2017 – 2022)

2. **Co-PI** in an ICMR supported "Advanced Centre for research in Pediatric kidney diseases toProf Arvind Bagga, AIIMS (2017 – 2022)

3. **Co-investigator** in ICMR supported project to AIIMS entitle Understanding Pathobiology of Multiple Sclerosis- Biomarkers, Genetic and Phenotypic Signatures (2018 – 2021)

4. **Co-Investigator** in ICMR supported project "A clinical and Molecular Characterization of early onset autosomal recessive cerebellar ataxias" (2012-2015)