Name :**Dr Ashwin B Dalal**
Designation : **Staff Scientist Grade VI**
Department/Institute: **Centre for DNA Fingerprinting and Diagnostics, Hyderabad**
Date of Birth :**4th December, 1975** Sex (M/F) **Male** SC/ST : **No**

Education (Post-Graduation onwards & Professional Career)

Sl No.	Institution Place	Degree Awarded	Year	Field of Study
1	Goa Medical College, Goa	MD	2002	Pediatrics
2	Sanjay Gandhi Postgraduate Institute of Medical Sciences, Lucknow	DM	2006	Medical Genetics

Position and Employment **(Starting with the most recent employment)**

From	To	Institution	Position
1/4/2016	Date	Centre for DNA Fingerprinting and Diagnostics, Hyderabad	Staff Scientist Grade VI
1/4/2011	31/3/2016	Centre for DNA Fingerprinting and Diagnostics, Hyderabad	Staff Scientist Grade V
2/4/2007	31/3/2011	Centre for DNA Fingerprinting and Diagnostics, Hyderabad	Staff Scientist Grade IV
22/7/2003	31/12/2006	Sanjay Gandhi Postgraduate Institute of Medical Sciences	Senior Resident (Medical Genetics)
8/5/2002	19/7/2003	Goa Medical College, Goa	Senior Resident

Professional Experience and Training relevant to the Project

1. Clinical experience in evaluation, counseling and management of patients with chromosomal disorders, congenital malformations and dysmorphic syndromes

Publications (Numbers only) - 97

Selected peer-reviewed publications (Ten best publications in chronological order)

1. Alber M, Kalscheuer VM, Marco E, Sherr E, Lesca G, Till M, Gradek G, Wiesener A, Korenke C, Mercier S, Becker F, Yamamoto T, Scherer SW, Marshall CR, Walker S, Dutta UR, Dalal AB, Suckow V, Jamali P, Kahrizi K, Najmabadi H, Minassian BA. ARHGEF9 disease: Phenotype clarification and genotype-phenotype correlation. **Neurol Genet.** 2017 May 26;3(3):e148.
2. Kar A, Phadke SR, Das Bhowmik A, Dalal A. Whole exome sequencing reveals a mutation in ARMC9 as a cause of mental retardation, ptosis, and polydactyly. **Am J Med Genet A**. 2018 Jan;176(1):34-40.
3. Shukla A, Das Bhowmik A, Hebbar M, Rajagopal KV, Girisha KM, Gupta N, Dalal A. Homozygosity for a nonsense variant in AIMP2 is associated with a progressive neurodevelopmental disorder with microcephaly, seizures, and spastic quadriparesis**. JHum Genet.** 2018 Jan;63(1):19-25.
4. Harms FL, Girisha KM, Hardigan AA, Kortüm F, Shukla A, Alawi M, Dalal A, Brady L, Tarnopolsky M, Bird LM, Ceulemans S, Bebin M, Bowling KM, Hiatt SM, Lose EJ, Primiano M, Chung WK, Juusola J, Akdemir ZC, Bainbridge M, Charng WL, Drummond-Borg M, Eldomery MK, El-Hattab AW, Saleh MA, Bézieau S, Cogné B, Isidor B, Küry S, Lupski JR, Myers RM, Cooper GM, Kutsche K. Mutations in EBF3 Disturb Transcriptional Profiles and Cause Intellectual Disability, Ataxia, and Facial Dysmorphism. **Am J Hum Genet.** 2017 Jan 5;100(1):117-127.
5. Ranganath P, Matta D, Bhavani GS, Wangnekar S, Jain JM, Verma IC, Kabra M, Puri RD, Danda S, Gupta N, Girisha KM, Sankar VH, Patil SJ, Ramadevi AR, Bhat M, Gowrishankar K, Mandal K, Aggarwal S, Tamhankar PM, Tilak P, Phadke SR, Dalal A. Spectrum of SMPD1 mutations in Asian-Indian patients with acid sphingomyelinase (ASM)-deficient Niemann-Pick disease. **Am J Med Genet A.** 2016 Oct;170(10):2719-30.

6. Hebbar M, Prasada L H, Bhowmik AD, Trujillano D, Shukla A, Chakraborti S, Kandaswamy KK, Rolfs A, Kamath N, Dalal A, Bielas S, Girisha KM. Homozygous deletion of exons 2 and 3 of NPC2 associated with Niemann-Pick disease type C. **Am J Med Genet A.** 2016 Sep;170(9):2486-9.
7. Phadke SR, Kar A, Bhowmik AD, Dalal A. Complex Camptosynpolydactyly and Mesoaxial synostotic syndactyly with phalangeal reduction are allelic disorders. **Am J Med Genet A.** 2016 ;170(6):1622-1625
8. Stephen J, Girisha KM, Dalal A, Shukla A, Shah H, Srivastava P, Kornak U, Phadke SR. Mutations in patients with osteogenesis imperfecta from consanguineous Indian families. **Eur J Med Genet**. 2015 Jan;58(1):21-7.
9. Aggarwal S, Bhowmik AD, Ramprasad VL, Murugan S, Dalal A. A splice site mutation in HERC1 leads to syndromic intellectual disability with macrocephaly and facial dysmorphism: Further delineation of the phenotypic spectrum. **Am J Med Genet A**. 2016 Jul;170(7):1868-73
10. Girisha KM, Kortüm F, Shah H, Alawi M, Dalal A, Bhavani GS, Kutsche K. A novel multiple joint dislocation syndrome associated with a homozygous nonsense variant in the EXOC6B gene. **Eur J Hum Genet.** 2016 Aug;24(8):1206-10

Research Support

Ongoing Research Projects

Sl No.	Title of Project	Funding Agency	Amount (lakhs)	Date of sanction and Duration
1	Whole Genome Sequencing for characterization of novel genes and de novo balanced chromosomal rearrangements in human genetic disorders Sanctioned	Science and Engineering Research Board, DST	72.39	3 years (2016-2019)

Completed Research Projects (State only major projects of last 3 years): None

Sl No.	Title of Project	Funding Agency	Amount (lakhs)	Date of sanction and Duration
1	Effect of Parental Psycho Education, Ethics of Research Participation, and Array Comparative Genomic Hybridization in Subjects with Mental Retardation (MR) and/or Autism	Department of Science and Technology	24.05	3 years (2011-2014)
2	Human exome sequencing for identification of novel genes in Mendelian disorders	Department of Biotechnology	38.29	3 years (2013-2016)
3	Multi Centric Collaborative Study Of The Clinical Biochemical and Molecular Characterization Of Lysosomal Storage Disorders In India: The Initiative For Research in Lysosomal Storage Disorders Task – Force Study (Joint Project from North, West and South	Department of Health Research and Indian Council of Medical Research	33.63	3 years (2015-2018)
4	Development and application of a next generation sequencing approach for molecular genetic analysis of lysosomal storage disorders	Department of Health Research	42.14	3 years (2014-2017)