Name: Dr. Kausik Mandal

Degree: MBBS, MD (Pediatrics), DM (Medical Genetics)

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Education		
Course	**College/University**	**Period of Study**
MBBS	NRS Medical college, Calcutta	1992 to 1999
MD (PEDIATRICS)	Maulana Azad Medical College, Delhi University	May 2000 to April 2003
D.M. (Medical Genetics)	SGPGIMS, Lucknow	July 2006 to June 2009
Current and previous relevant positions including academic appointments (Most current position first):		
Month and Year	**Title**	**Institution/Company, Country**
July 2019 to present	Additional Professor, Medical Genetics	SGPGIMS, Lucknow, India
July 2016 to June 2019	Associate Professor, Medical Genetics	SGPGIMS, Lucknow, India
June 2013 to June 2016	Assistant Professor, Medical Genetics	SGPGIMS, Lucknow, India

June 2011 to Sept 2011	Assistant Professor (Pediatrics) and Consultant, Medical Genetics	VIMS, R K Mission, Kolkata, India
October 2010 to June 2011	Assistant Professor (Pediatrics)	College of Medicine and JNM hospital, Kalyani, India
August 2009 to July 2010	Assistant Professor (Medical Genetics)	Christian Medical College, Vellore, India
July 2006 to June 2009	Senior Resident (Medical Genetics)	SGPGIMS, Lucknow, India
May/ June2003 to June 2006	Senior Resident (Pediatrics)	MAMC and KSCH, New Delhi, India

Experience:
Around 14 years experience in Medical Genetics including Pre-natal testing

Clinical trial

Title	Funding Agency	Institution	Duration and Date of start
ENGAGE trial (Co-investigator): trial on "Small molecule" for Gaucher disease	Genzyme	CMC, Vellore, India	2009-2010

Research projects in which Principal Investigator

Title	Funding Agency	Duration and Date of start
Spectrum of PTPN11 mutation in Indian subjects with Noonan syndrome	Intramural	2 years (15.2. 2014) Completed March 2016
Study of genetic causes of childhood overgrowth and search for new genes	ICMR	Under process (Concept note accepted)
Monogenic causes of Paediatric Kidney diseases: A multicentre collaborative study from North, East and North-East India	DBT	Under process

To evaluate the role of CAG repeat polymorphism of the androgen receptor in young females of UP origin [age 18-25 years] suffering from Polycystic Ovary Syndrome.	Extramural (UPCST)	Under review

Research projects in which co-investigator

Title	Funding Agency	Duration and Date of start
Plasma exosomal micro RNA and proteome profiling in autism spectrum disorder children's: probing for biomarkers	SERB	August 2018 (3 years)
The Indian Movement disorder and Biobank	DBT	
Genomic studies into defects of human limb development	DBT	3 years (3.3.2015)
Immunocytochemical Novel Rabbit Monoclonal antibody HER2/neu expression and gene amplication with FISH on cell blocks of breast carcinoma	Intramural	2 years (15.2 2014) Completed March 2016
Functional Validation of Novel Sequence Variants in Monogenic Disorders to prove their pathogenicity	DST	3 years (June 2016)
Utility of single nucleotide polymorphism genotyping array in identification of genetic etiology in patients with autosomal recessive disorder with consanguinity	Intramural	2 years (15.2 2014)
Role of WNT pathway signaling module RNF43, ZNFR3 in colorectal cancer	Intramural	2 years (2017)
Genetic and immunological characteristics and skeletal muscle dysfunction in patients with hypoparathyroidism	Intramural	October, 2018
Understanding pathogenesis of Takayasu arteritis - transcriptomics applied to peripheral blood mononuclear cells (PBMC) and involved vessels, and the role of various T-cell subsets	Extramural (ICMR)	3 years (April 2019)
Genetic characterization of idiopathic	ICMR	Under consideration

hypoparathyroidism using a whole exome sequencing approach		
Pathophysiology of clinical outcomes and FGF23 production in patients with phosphaturic mesenchymal tumours	SERB	Under consideration
Genotype phenotype correlation of neuroendocrine tumours	Intramural	Jan 2019

Memberships:

1. Society for Indian Academy of Medical Genetics: Executive and Founder member
2. Indian Academy of Pediatrics (Genetics Chapter): Executive member
3. Molecular pathology association of India: Executive member

Important indexed publications:

1. Congenital Nephrotic Syndrome in India in the Current Era: A Multicenter Case Series. Sinha R, Vasudevan A, Agarwal I, Sethi SK, Saha A, Pradhan S, Ekambaram S, Thaker N, Matnani M, Banerjee S, Sharma J, Singhal J, Ashraf S, Mandal K. Nephron. 2019 Oct 25:1-9.
2. Computer-aided Facial Analysis in Diagnosing Dysmorphic Syndromes in Indian Children. Narayanan DL, Ranganath P, Aggarwal S, Dalal A, Phadke SR, Mandal K. Indian Pediatr. 2019 Dec 15;56(12):1017-1019.
3. Vici Syndrome with a Novel Mutation in EPG5. Moirangthem A, Mandal K, Ghosh A, Phadke SR. Indian Pediatr. 2019 Jul 15;56(7):603-605.
4. Novel mutations and spectrum of the disease of NR0B1 (DAX1)-related adrenal insufficiency in Indian children. Gupta S, Joshi K, Zaidi G, Sarangi AN, Mandal K, Bhavani N, Pavithran PV, Pillai MG, Singh SK, Godbole T, Bhatia V, Bhatia E. J Pediatr Endocrinol Metab. 2019 Jun 20.
5. Congenital Chylothorax in a Neonate with Cornelia de Lange Syndrome: A Rare Complication Managed with a Novel Indigenously Prepared Milk Formulation. Gupta A, Naranje KM, Singh A, Pandita A, Gupta G, Mandal K, Pradhan M. Indian J Pediatr. 2019 Jul;86(7):645-647.

6. Cytogenetic microarray in structurally normal and abnormal foetuses: a five year experience elucidating increasing acceptance and clinical utility. Lallar M, Srivastava P, Rai A, Saxena D, Mandal K, Phadke SR. J Genet. 2019 Mar;98(1).
7. Endocrine Manifestations of Von Hippel-Landau Disease.(Review article). Pradhan R, George N, Mandal K, Agarwal A, Gupta SK. Indian J Endocrinol Metab. 2019 Jan-Feb;23(1):159-164.
8. Expanding the phenotype in autosomal dominant mental retardation-24: a novel variation in DEAF1 gene. Sharma P, Gambhir PS, Phadke SR, Mandal K. Clin Dysmorphol. 2019 Apr;28(2):94-97.
9. Study of the association of forkhead box P3 (*FOXP3*) gene polymorphisms with unexplained recurrent spontaneous abortions in Indian population. Mishra S, Srivastava A, Mandal K, Phadke SR. J Genet. 2018 Jun;97(2):405-410.
10. Phenotypic characterization of derivative 22 syndrome: case series and review. Saxena D, Srivastava P, Tuteja M, Mandal K, Phadke SR. J Genet. 2018 Mar;97(1):205-211. Review.
11. Connexin 26 (GJB2) Mutations Associated with Non-Syndromic Hearing Loss (NSHL). Mishra S, Pandey H, Srivastava P, Mandal K, Phadke SR. Indian J Pediatr. 2018 Dec;85(12):1061-1066
12. Molecular Testing of MECP2 Gene in Rett Syndrome Phenotypes in Indian Girls. Lallar M, Rai A, Srivastava P, Mandal K, Gupta N, Kabra M, Phadke SR. Indian Pediatr. 2018 Jun 15;55(6):474-477. Epub 2018 Feb 9.
13. Next Generation Sequencing in Diagnosis of MLPA Negative Cases Presenting as Duchenne/ Becker Muscular Dystrophies (2017)Indian Journal of Pediatrics
14. Noonan syndrome in diverse populations (2017) American Journal of Medical Genetics, Part A
15. Expansion of the phenotypic spectrum in three families of methyl CpG-binding protein 2 duplication syndrome. Moirangthem A, Tuteja Bhatia M, Srivastava P, Mandal K, Rai A, Phadke SR. Clin Dysmorphol. 2017 Apr;26(2):73-77.
16. Hotspots in PTPN11 Gene Among Indian Children With Noonan Syndrome. Indian pediatrics Narayanan DL, Pandey H, Moirangthem A, Mandal K, Gupta

R, Puri RD, Patil SJ, Phadke SR.Indian Pediatr. 2017 Aug 15;54(8):638-643. Epub 2017 Jun 4.

17. Cover Image, Volume 173A, Number 9, September 2017. American journal of medical genetics. Part A
18. Spondyloepiphyseal dysplasia Omani type: CHST3 mutation spectrum and phenotypes in three Indian families. Srivastava P, Pandey H, Agarwal D, Mandal K, Phadke SR. Am J Med Genet A. 2017 Jan;173(1):163-168.
19. Fanconi-Bickel Syndrome: Another Novel Mutation in SLC2A2. Amita M, Srivastava P, Mandal K, De S, Phadke SR. ndian J Pediatr. 2017 Mar;84(3):236-237.
20. Novel mutations in the transmembrane natriuretic peptide receptor NPR-B gene in four Indian families with acromesomelic dysplasia, type Maroteaux.Srivastava P, Tuteja M, Dalal A, Mandal K, R Phadke S. J Genet. 2016 Dec;95(4):905-909.
21. Identification and characterization of 20 novel pathogenic variants in 60 unrelated Indian patients with mucopolysaccharidoses type I and type II. Uttarilli A, Ranganath P, Matta D, Md Nurul Jain J, Prasad K, Babu AS, Girisha KM, Verma IC, Phadke SR, Mandal K, Puri RD, Aggarwal S, Danda S, Sankar VH, Kapoor S, Bhat M, Gowrishankar K, Hasan AQ, Nair M, Nampoothiri S, Dalal A. Clin Genet. 2016 Dec;90(6):496-508
22. Novel sequence variations in the thymidine phosphorylase gene causing mitochondrial neurogastrointestinal encephalopathy. Karyampudi A, Srivastava P, Mandal K, Yadav P, Ghoshal UC, Verma A, Phadke SR. Clin Dysmorphol. 2016 Oct;25(4):156-62.
23. Pycnodysostosis: mutation spectrum in five unrelated Indian children. Mandal K, Ray S, Saxena D, Srivastava P, Moirangthem A, Ranganath P, Gupta N, Mukhopadhyay S, Kabra M, Phadke SR. Clin Dysmorphol. 2016 Apr 18.
24. Bidirectional ventricular tachycardia of unusual etiology. Praloy Chakraborty, Bhavna Kaul, Kausik Mandal, H.S. Isser, Sandeep Bansal, Anandaraja Subramanian. Indian Pacing Electrophysiol J. 2016 Feb 23;15(6):296-9.

25. Incessant left ventricular tachycardia of unusual etiology. Chakraborty P, Isser HS, Arava S, Mandal K. Indian Pacing Electrophysiol J. 2016 May - Jun;16(3):104-106.
26. Spectrum of SMPD1 mutations in Asian-Indian patients with acid sphingomyelinase (ASM)-deficient Niemann-Pick disease. 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. Am J Med Genet A. 2016 Oct;170(10):2719-30.
27. Complex chromosomal rearrangement involving five chromosomes: deciphering genomic imbalances in an apparently balanced chromosomal translocation. Mandal K, Agarwal M, Boggula VR, Patil SJ, Phadke SR. Clin Dysmorphol. 2016 Apr;25(2):63-7
28. Hunter Syndrome in Northern India: Clinical features and Mutation Spectrum. Narayanan DL , Srivastava P, Mandal K, Gambhir PS, Phadke SR. Indian Pediatr. 2016;53:134-6.
29. Cartilage Hair Hypoplasia: Two Unrelated Cases with g.70 A > G Mutation in RMRP Gene. Narayanan DL, Shukla A, Siddesh AR, Stephen J, Srivastava P, Mandal K, Phadke SR. Indian J Pediatr. 2016 Feb 1. [Epub ahead of print]
30. Smith-Magenis Syndrome: Face Speaks. Gupta R, Gupta N, Nampoothiri S, Mandal K, Kishore Y, Sharma P, Kabra M, Phadke SR. Indian J Pediatr. 2015 Dec 17. [Epub ahead of print]
31. Potter's sequence: A story of the rare, rarer and the rarest. Sarkar S, DasGupta S, Barua M, Ghosh R, Mondal K, Chatterjee U, Datta C. Indian J Pathol Microbiol. 2015 Jan-Mar;58(1):102-4.
32. Co-occurrence of a de novo Williams and 22q11.2 microdeletion syndromes. Shukla A, Mandal K, Patil SJ, Kishore Y, Phadke SR, Girisha KM. Am J Med Genet A. 2015 Apr 21. doi: 10.1002/ajmg.a.37116. [Epub ahead of print]
33. Prenatal diagnosis in India is not limited to sex selection. Dalal AB, Ranganath P, Phadke SR, Kabra M, Danda S, Puri RD, Sankar VH, Gupta N, Patil SJ, Mandal K, Tamhankar P, Aggarwal S, Agarwal M.

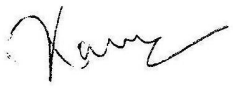
34. Partial trisomy of chromosome 15q and partial monopsony of 6q due to maternal balanced translocation. Singla S, Mandal K, Sharma S, Chhapola V. J Pediatr Neurosci. 2014 May;9(2):178-81.
35. Patient with mutation in the matrix metalloproteinase 2 (MMP2) gene - a case report and review of the literature. Ekbote AV, Danda S, Zankl A, Mandal K, Maguire T, Ungerer K. J Clin Res Pediatr Endocrinol. 2014;6(1):40-6.
36. A novel X-chromosomal microdeletion encompassing congenital hemidysplasia with ichthyosiform erythroderma and limb defects. Raychaudhury T, George R, Mandal K, Srivastava VM, Thomas M, Bornholdt D, Grzeschik KH, Koehler A. Pediatr Dermatol. 2013 Mar-Apr;30(2):250-2.
37. Fanconi- Bickel Syndrome: mutation in an Indian patient. Ekbote AV, Mandal K, Agarwal I, Sinha R, Danda S. Indian J Pediatr. 2012 Jun;79(6):810-2.
38. Prenatal diagnosis of Apert Syndrome in Second Trimester – A Case Report : Phadke S R, Mandal K, Ranganath P; Perinatology; 2012; 12:4; 159-62
39. Aetiologic spectrum of mental retardation & developmental delay in India. Aggarwal S, Bogula VR, Mandal K, Kumar R, Phadke SR. Indian J Med Res. 2012 Sep;136(3):436-44.
40. Familial 18p deletion syndrome and 18p partial trisomy inherited from a mother with balanced translocation. Koshy B, Mandal K, Srivastava VM, Loius PT, Danda S. Clin Dysmorphol. 2011 Jul;20(3):148-51.
41. Homocysteine, fibrinogen and lipid profile in children of young adults with coronary artery disease. Khalil A, Mandal K, Khalil S, Mallika V. Indian Pediatr. 2011 Feb;48(2):156-7.
42. Vitamin D Receptor Gene Polymorphisms in Indian Children with Idiopathic Nephrotic Syndrome Tabrez Jafar, Gaurav Tripathi, Abass A. Mehndi, Kaushik Mandal, Sanjeev Gulati, Raj K Sharma, Vinod. P. Baburaj, Shaily Awasthi, Suraksha Agrawal. Int J Hum Genet, 2009; 9(1): 49-55. Published online: 04 Sep 2017
43. Does cytokine gene polymorphism affect steroid responses in idiopathic nephrotic syndrome? Tripathy G, Jafar T, Mandal Kaushik et al. Indian J Med Sci, 62(10), October 2008: 383-91

44. Use of Multiplex Ligation-Dependent Probe Amplification (MLPA) in screening of subtelomeric regions in children with idiopathic mental retardation. Mandal K, Boggula VR, Borkar M, Agarwal S, Phadke SR. Indian J Pediatr. 2009 Oct;76(10):1027-31.
45. Fabry disease: a treatable lysosomal storage disorder. Phadke SR, Mandal K, Girisha KM. Natl Med J India. 2009 Jan-Feb;22(1):20-2.
46. Milder form of pachydermoperiostosis: a report of four cases. Girisha KM, Mandal K, Phadke SR. Clin Dysmorphol. 2009 Apr;18(2):85-9.
47. Congenital swan neck deformity of fingers with syndactyly. Mandal K, Phadke SR, Kalita J. Clin Dysmorphol. 2008 Apr;17(2):109-11.
48. Berardinelli-Seip congenital lipodystrophy. Mandal K, Aneja S, Seth A, Khan A. Indian Pediatr. 2006 May;43(5):440-5.
49. Acute transverse myelitis following hepatitis E virus infection. Mandal K, Chopra N. Indian Pediatr. 2006 Apr;43(4):365-6.

Book Chapters:

- PG Textbook of Pediatrics; SECTION 3: Metabolic Disorders; Chapter3.13 Carbohydrate Metabolism Defects
- Pediatric Office Practice: Chapter on Genetic and metabolic disorders
- Genetics in Tropics: Tropical Pediatrics
- Congenital anomalies of brain and spinal cord: Pediatric neurology

The above entries are true to the best of my knowledge



Dr Kausik Mandal (Dated:20/03/2020)