CV for Departmental website

Name : Dr. Amita Moirangthem
Educational qualifications: MD (Pediatrics), DM (Medical Genetics)
Designation: Assistant Professor
Correspondence address: Department of Medical Genetics, Sanjay Gandhi Postgraduate Institute of Medical Sciences, Raebareli Road, Lucknow-226014, Uttar Pradesh, India
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Work Experience :

1. Six years in Medical Genetics including 3 years as faculty
2. More than 10 years in Pediatrics

Professional societies:

Life Member of the Society of Indian Academy of Medical Genetics (SIAMG)
Life Member of Indian Academy of Pediatrics (IAP)

Areas of clinical and research interests: Children with overgrowth disorders, arthrogryposis, neurodevelopmental disorders; analysis and interpretation of genomic sequencing data

Publications:

1. Masih S, **Moirangthem A**, Phadke SR. Renpenning syndrome in an Indian patient. Am J Med Genet A. 2020 Feb;182(2):293-295. doi: 10.1002/ajmg.a.61457. Epub 2019 Dec 16. PubMed PMID: 31840915.
2. Girisha KM, Bhavani GS, Shah H, **Moirangthem A**, Shukla A, Kim OH, Nishimura G, Mortier GR. Biallelic variants p.Arg1133Cys and p.Arg1379Cys in COL2A1: Further delineation of phenotypic spectrum of recessive Type 2 collagenopathies. Am J Med Genet A. 2020 Feb;182(2):338-347. doi: 10.1002/ajmg.a.61414. Epub 2019 Nov 22. PubMed PMID: 31755234.
3. **Moirangthem A**, Mandal K, Ghosh A, Phadke SR. Vici Syndrome with a Novel Mutation in EPG5. Indian Pediatr. 2019 Jul 15;56(7):603-605. PubMed PMID: 31333218.
4. **Moirangthem A**, Narayanan DL, Jacob P, Nishimura G, Mortier G, Girisha KM. Report of second case and clinical and molecular characterization of Eiken syndrome. *Clin Genet*. 2018 Nov;94(5):457-460.
5. Radhakrishnan P, **Moirangthem A**, Nayak SS, Shukla A, Mathew M, Girisha KM. Novel pathogenic variants in GBE1 causing fetal akinesia deformation sequence and severe neuromuscular form of glycogen storage disease type IV. Clin Dysmorphol. 2019 Jan;28(1):17-21. doi: 10.1097/MCD.0000000000000248. PubMed PMID: 30303820.

6. **Moirangthem A**, Phadke SR. Socio-demographic Profile and Economic Burden of Treatment of Transfusion Dependent Thalassemia. Indian J Pediatr. 2018 Feb;85(2):102-107. doi: 10.1007/s12098-017-2478-y. Epub 2017 Nov 9. PubMed PMID: 29119463.
7. **Moirangthem A**, Tuteja Bhatia M, Srivastava P, Mandal K, Rai A, Phadke SR. Expansion of the phenotypic spectrum in three families of methyl CpG-binding protein 2 duplication syndrome. *Clin Dysmorphol*. 2017 Apr;26(2):73-77.
8. Narayanan DL, Pandey H, **Moirangthem A**, Mandal K, Gupta R, Puri RD, Patil SJ, Phadke SR. Hotspots in PTPN11 Gene Among Indian Children With Noonan Syndrome. *Indian Pediatr*. 2017 Aug 15;54(8):638-643.
9. **Amita M**, Srivastava P, Agarwal D, Phadke SR Floating Harbor syndrome. *Indian J Pediatr.* 2016 Aug;83(8):896-7.
10. **Amita M**, Srivastava P, Mandal K, De S, Phadke SR. Fanconi-Bickel Syndrome: Another Novel Mutation in SLC2A2.. *Indian J Pediatr*. 2017 Mar;84(3):236-237.
11. Mandal K, Ray S, Saxena D, Srivastava P, **Moirangthem A**, Ranganath P, Gupta N, Mukhopadhyay S, Kabra M and Phadke S R. Pycnodysostosis: mutation spectrum in five unrelated Indian children. *Clinical Dysmorphology* 2016, 25:113–120

Research projects:

Sl no.	Title of project	Principal Investigator	Funding agency	Budget (Rs in lacs)	Year of starting	Duration (Ongoing / complete)
1.	Clinical and genotypic profile of arthrogryposis multiplex congenita	Dr Amita Moirangthem	SGPGI Research cell	4.9	2019	2 years (ongoing)