

Key Publications

1. Sait H, Srivastava P, Gupta N, Kabra M, Kapoor S, Ranganath P, Rungsung I, Mandal K, Saxena D, Dalal A, Roy A, Pabbati J, Phadke SR. Phenotypic and genotypic spectrum of CTSK variants in a cohort of twenty-five Indian patients with pycnodysostosis. *Eur J Med Genet.* 2021 Jul;64(7):104235.
2. Puri RD, Setia N, N V, Jagadeesh S, Nampoothiri S, Gupta N, Muranjan M, Bhat M, Girisha KM, Kabra M, Verma J, Thomas DC, Biji I, Raja J, Makkar R, Verma IC, Kishnani PS. Late onset Pompe Disease in India - Beyond the Caucasian phenotype. *NeuromusculDisord.* 2021 May;31(5):431-441.
3. Gabra P, Jana M, Naranje P, Gupta N, Kabra M, Gupta AK, Yadav R. Spine radiograph in dysplasias: A pictorial essay. *Indian J Radiol Imaging.* 2020 Oct-Dec;30(4):436-447.
4. Correa ARE, Endrakanti M, Naini K, Kabra M, Gupta N. Hydrops fetalis in PKD1L1-related heterotaxy: Report of two fetuses and expanding the phenotypic and molecular spectrum. *Ann Hum Genet.* 2021 May;85(3-4):138-145.
5. Kausthubham N, Shukla A, Gupta N, Bhavani GS, Kulshrestha S, Das Bhowmik A, Moirangthem A, Bijarnia-Mahay S, Kabra M, Puri RD, Mandal K, Verma IC, Bielas SL, Phadke SR, Dalal A, Girisha KM. A data set of variants derived from 1455 clinical and research exomes is efficient in variant prioritization for early-onset monogenic disorders in Indians. *Hum Mutat.* 2021 Apr;42(4):e15-e61.
6. Anand V, Shukla G, Gupta N, Gupta A, Sapra S, Gulati S, Pandey RM, Pandey S, Kabra M. Association of Sleep Apnea With Development and Behavior in Down Syndrome: A Prospective Clinical and Polysomnographic Study. *Pediatr Neurol.* 2021 Mar;116:7-13.
7. Nampoothiri S, Yesodharan D, Bhattacharjee A, Ahamed H, Puri RD, Gupta N, Kabra M, Ranganath P, Bhat M, Phadke S, Radha Rama Devi A, Jagadeesh S, Danda S, Sylaja PN, Mandal K, Bijarnia-Mahay S, Makkar R, Verma IC, Dalal A, Ramaswami U. Fabry disease in India: A multicenter study of the clinical and mutation spectrum in 54 patients. *JIMD Rep.* 2020 Aug 15;56(1):82-94.
8. Elmonem MA, Belanger-Quintana A, Bordugo A, Boruah R, Cortès-Saladelafont E, Endrakanti M, Giraldo P, Grünert SC, Gupta N, Kabra M, Knerr I, Krämer J, Kuster A, Levtchenko E, Ngu LH, Rovira-Remisa MM, Sass JO, Sykut-Cegielska J, Tummolo A, van den Heuvel LP. The impact of COVID-19 pandemic on the diagnosis and management of inborn errors of metabolism: A global perspective. *Mol Genet Metab.* 2020 Nov;131(3):285-288.
9. Dudani P, Mahajan S, Gupta N, Kabra M, Bhari N. Stippled keratoderma and nail dystrophy associated with hyperkeratotic pustular lesions in a 2-year-old boy. *Pediatr Dermatol.* 2020 Sep;37(5):e64-e66.

10. Aggarwal B, Kabra M, Gupta N. Report of an Indian Family with Sengers Syndrome. *Indian J Pediatr.* 2021 Jan;88(1):92.
11. Pasumarthi D, Gupta N, Sheth J, Jain SJMN, Rungsung I, Kabra M, Ranganath P, Dalal A. Identification and characterization of 30 novel pathogenic variations in 69 unrelated Indian patients with Mucopolysaccharidosis Type II and Type III. *J Hum Genet.* 2020 Nov;65(11):971-984.
12. Kaur R, Correa ARE, Thakur S, Kabra M, Gupta N. Methylene Tetrahydrofolate Reductase Deficiency. *Indian J Pediatr.* 2020 Nov;87(11):951-953.
13. Kaur R, Siddiqui I, Mathur V, Jana M, Kabra M, Gupta N. Bi-allelic loss-of- function novel variants in LTBP3-related skeletal dysplasia: Report of first patient from India. *Am J Med Genet A.* 2020 Aug;182(8):1944-1946.
14. Gupta A, Kabra M, Gupta N. Duchenne Muscular Dystrophy- Where Genetic Testing is Inevitable and Vital! *Indian J Pediatr.* 2020 Jul;87(7):487-488.
15. Gupta N, Langeh N, Sridharan A, Kabra M. Identification of a Novel 19-bp Deletion Mutation in LTBP4 Using Exome Sequencing in Two Siblings with Autosomal Recessive Cutis Laxa Type 1C. *J Pediatr Genet.* 2020 Jun;9(2):125-131.
16. Perveen S, Gupta N, Kumar M, Kaur P, Chowdhury MR, Kabra M. Spectrum of amyloglucosidase mutations in Asian Indian patients with Glycogen storage disease type III. *Am J Med Genet A.* 2020 May;182(5):1190-1200.
17. Malhotra R, Shukla R, Kabra M, Gupta Y, Jyotsna VP, Khadgawat R. Impact of parental origin of X-chromosome on clinical and biochemical profile in Turner syndrome. *J PediatrEndocrinolMetab.* 2020 Sep 25;33(9):1155-1163.
18. Sudarshan S, Atin Kumar, Gupta A, Bhari N, SethuramanG, Kabra M, Roy Chowdhury M, et al. Mutation spectrum of Tuberous Sclerosis Complex patients in Indian population. *J Pediatr Genet.* 2020;
19. Puri RD, Setia N, N V, Jagadeesh S, Nampoothiri S, Gupta N, Muranjan M, Bhat M, Girisha KM, Kabra M, Verma J, Thomas DC, Biji I, Raja J, Makkar R, Verma IC, Kishnani PS. Late onset Pompe Disease in India - Beyond the Caucasian phenotype. *NeuromusculDisord.* 2021 Feb 16:S0960-8966(21)00041-9.
20. Gabra P, Jana M, Naranje P, Gupta N, Kabra M, Gupta AK, Yadav R. Spine radiograph in dysplasias: A pictorial essay. *Indian J Radiol Imaging.* 2020 Oct-Dec;30(4):436-447.
21. Correa ARE, Endrakanti M, Naini K, Kabra M, Gupta N. Hydrops fetalis in PKD1L1-related heterotaxy: Report of two fetuses and expanding the phenotypic and molecular spectrum. *Ann Hum Genet.* 2021 Mar 2.
22. Kausthubham N, Shukla A, Gupta N, Bhavani GS, Kulshrestha S, Das Bhowmik A, Moirangthem A, Bijarnia-Mahay S, Kabra M, Puri RD, Mandal K, Verma IC, Bielas SL, Phadke SR, Dalal A, Girisha KM. A data set of variants derived from 1455 clinical and research exomes is efficient in variant prioritization for early-onset monogenic disorders in Indians. *Hum Mutat.* 2021 Apr;42(4):e15-e61.
23. Patil K, Gupta N. Lipoprotein Lipase Deficiency: Diet is the Key! *Indian J Pediatr.* 2021 Feb;88(2):111-112.

24. Anand V, Shukla G, Gupta N, Gupta A, Sapra S, Gulati S, Pandey RM, Pandey S, Kabra M. Association of Sleep Apnea With Development and Behavior in Down Syndrome: A Prospective Clinical and Polysomnographic Study. *Pediatr Neurol*. 2021 Mar;116:7-13.
25. Nampoothiri S, Yesodharan D, Bhattacharjee A, Ahamed H, Puri RD, Gupta N, Kabra M, Ranganath P, Bhat M, Phadke S, Radha Rama Devi A, Jagadeesh S, Danda S, Sylaja PN, Mandal K, Bijarnia-Mahay S, Makkar R, Verma IC, Dalal A, Ramaswami U. Fabry disease in India: A multicenter study of the clinical and mutation spectrum in 54 patients. *JIMD Rep*. 2020 Aug 15;56(1):82-94.
26. Elmonem MA, Belanger-Quintana A, Bordugo A, Boruah R, Cortès-Saladelafont E, Endrakanti M, Giraldo P, Grünert SC, Gupta N, Kabra M, Knerr I, Krämer J, Kuster A, Levtchenko E, Ngu LH, Rovira-Remisa MM, Sass JO, Sykut-Cegielska J, Tummolo A, van den Heuvel LP. The impact of COVID-19 pandemic on the diagnosis and management of inborn errors of metabolism: A global perspective. *Mol Genet Metab*. 2020 Nov;131(3):285-288.
27. Tekendo-Ngongang C, Owosela B, Fleischer N, Addissie YA, Malonga B, Badoe E, Gupta N, Moresco A, Huckstadt V, Ashaat EA, Hussen DF, Luk HM, Lo IFM, Hon-Yin Chung B, Fung JLF, Moretti-Ferreira D, Batista LC, Lotz-Esquivel S, Saborio-Rocafort M, Badilla-Porras R, PenonPortmann M, Jones KL, Abdul-Rahman OA, Uwineza A, Prijoles EJ, Ifeorah IK, LlamasPaneque A, Sirisena ND, Dowsett L, Lee S, Cappuccio G, Kitchin CS, Diaz-Kuan A, Thong MK, Obregon MG, Mutesa L, Dissanayake VHW, El Ruby MO, Brunetti-Pierri N, Ekure EN, Stevenson RE, Muenke M, Kruszka P. Rubinstein-Taybi syndrome in diverse populations. *Am J Med Genet A*. 2020 Dec;182(12):2939-2950.
28. Dudani P, Mahajan S, Gupta N, Kabra M, Bhari N. Stippled keratoderma and nail dystrophy associated with hyperkeratotic pustular lesions in a 2-year-old boy. *Pediatr Dermatol*. 2020 Sep;37(5):e64-e66.
29. Gangodkar P, Khadilkar V, Raghupathy P, Kumar R, Dayal AA, Dayal D, Ayyavoo A, Godbole T, Jahagirdar R, Bhat K, Gupta N, Kamalanathan S, Jagadeesh S, Ranade S, Lohiya N, Oke RL, Ganesan K, Khatod K, Agarwal M, Phadke N, Khadilkar A. Clinical application of a novel next generation sequencing assay for CYP21A2 gene in 310 cases of 21- hydroxylase congenital adrenal hyperplasia from India. *Endocrine*. 2021 Jan;71(1):189-198.
30. Aggarwal B, Kabra M, Gupta N. Report of an Indian Family with Sengers Syndrome. *Indian J Pediatr*. 2021 Jan;88(1):92.
31. Pasumarthi D, Gupta N, Sheth J, Jain SJMN, Rungsung I, Kabra M, Ranganath P, Aggarwal S, Phadke SR, Girisha KM, Shukla A, Datar C, Verma IC, Puri RD, Bhavsar R, Mistry M, Sankar VH, Gowrishankar K, Agrawal D, Nair M, Danda S, Soni JP, Dalal A. Identification and characterization of 30 novel pathogenic variations in 69 unrelated Indian patients with Mucopolidosis Type II and Type III. *J Hum Genet*. 2020 Nov;65(11):971-984.
32. Kaur R, Correa ARE, Thakur S, Kabra M, Gupta N. Methylene Tetrahydrofolate Reductase Deficiency. *Indian J Pediatr*. 2020 Nov;87(11):951-953.
33. Kaur R, Siddiqui I, Mathur V, Jana M, Kabra M, Gupta N. Bi-allelic loss-of-function novel variants in LTBP3-related skeletal dysplasia: Report of first patient from India. *Am J Med Genet A*. 2020 Aug;182(8):1944-1946.

34. Gupta A, Kabra M, Gupta N. Duchenne Muscular Dystrophy- Where Genetic Testing is Inevitable and Vital! *Indian J Pediatr.* 2020 Jul;87(7):487-488.
35. Gupta N, Langeh N, Sridharan A, Kabra M. Identification of a Novel 19-bp Deletion Mutation in LTBP4 Using Exome Sequencing in Two Siblings with Autosomal Recessive Cutis Laxa Type 1C. *J Pediatr Genet.* 2020 Jun;9(2):125-131.
36. Perveen S, Gupta N, Kumar M, Kaur P, Chowdhury MR, Kabra M. Spectrum of amyloglucosidase mutations in Asian Indian patients with Glycogen storage disease type III. *Am J Med Genet A.* 2020 May;182(5):1190-1200.
37. Mameli C, Zichichi G, Mahmood N, Elalaoui SC, Mirza A, Dharmaraj P, Burrone M, Cattaneo E, Sheth J, Gandhi A, Kochar GS, Alkuraya FS, Kabra M, Mercurio G, Zuccotti G. Natural history of non-lethal Raine syndrome during childhood. *Orphanet J Rare Dis.* 2020 Apr 16;15(1):93.
38. Correa ARE, Mishra P, Kabra M, Gupta N. Epigenetic Abnormalities of 11p15.5 Region in Beckwith-Wiedemann Syndrome - A Report of Eight Indian Cases. *Indian J Pediatr.* 2020 Mar;87(3):175-178.
39. Vats P, Dabas A, Jain V, Seth A, Yadav S, Kabra M, Gupta N, Singh P, Sharma R, Kumar R, Polipalli SK, Batra P, Thelma BK, Kapoor S. Newborn Screening and Diagnosis of Infants with Congenital Adrenal Hyperplasia. *Indian Pediatr.* 2020 Jan 15;57(1):49-55.
40. Kruszka P, Addissie YA, Tekendo-Ngongang C, Jones KL, Savage SK, Gupta N, Sirisena ND, Dissanayake VHW, Paththinige CS, Aravena T, Nampoothiri S, Yesodharan D, Girisha KM, Patil SJ, Jamuar SS, Goh JC, Utari A, Sihombing N, Mishra R, Chitrakar NS, Iriele BC, Lulseged E, Megarbane A, Uwineza A, Oyenusi EE, Olopade OB, Fasanmade OA, Duenas-Roque MM, Thong MK, Tung JYL, Mok GTK, Fleischer N, Rwegera GM, de Herreros MB, Watts J, Fieggen K, Huckstadt V, Moresco A, Obregon MG, Hussen DF, Ashaat NA, Ashaat EA, Chung BHY, Badoe E, Faradz SMH, El Ruby MO, Shotelersuk V, Wonkam A, Ekure EN, Phadke SR, Richieri-Costa A, Muenke M. Turner syndrome in diverse populations. *Am J Med Genet A.* 2020 Feb;182(2):303-313.
41. Sudarshan S, Kumar M, Kaur P, Kumar A, G S, Sapra S, Gulati S, Gupta N, Kabra M, Roy Chowdhury M. Decoding of novel missense TSC2 gene variants using in-silico methods. *BMC Med Genet.* 2019 Oct 26;20(1):164.
42. Gupta SK, Aggarwal A, Shaw M, Gulati GS, Kothari SS, Ramakrishnan S, Saxena A, Devagourou V, Talwar S, Sharma S, Gupta N, Airan B, Anderson RH. Clarifying the anatomy of common arterial trunk: a clinical study of 70 patients. *Eur Heart J Cardiovasc Imaging.* 2019 Oct 19. pii: jez255.
43. Gupta N, Kazi ZB, Nampoothiri S, Jagdeesh S, Kabra M, Puri RD, Muranjan M, Kalaivani M, Rehder C, Bali D, Verma IC, Kishnani PS. Clinical and Molecular Disease Spectrum and Outcomes in Patients with Infantile-Onset Pompe Disease. *J Pediatr.* 2020 Jan;216:44-50.e5.
44. Niceta M, Barbuti D, Gupta N, Ruggiero C, Tizzano EF, Graul-Neumann L, Barresi S, Nishimura G, Valenzuela I, López-Grondona F, Fernandez-Alvarez P, Leoni C, Zweier C, Tzschach A, Stellacci E, Del Fattore A, Dallapiccola B, Zampino G, Tartaglia M. Skeletal abnormalities are common features in Aymé-Gripp syndrome. *Clin Genet.* 2020 Feb;97(2):362-369.

45. Gupta N, Yadav S, Gurramkonda VB, VI R, Sg T, Kabra M. First report of THOC6 related intellectual disability (Beaulieu Boycott Innes syndrome) in two siblings from India. *Eur J Med Genet.* 2019 Aug 14;103742.
46. Yadav S, Thakur S, Kohlhase J, Bhari N, Kabra M, Gupta N. Report of Two Novel Mutations in Indian Patients with Rothmund-Thomson Syndrome. *J Pediatr Genet.* 2019 Sep;8(3):163-167.
47. Gupta N, Correa ARE, Jana M, Kabra M. Report of a Novel Homozygous Nonsense DDR2 Mutation in an Indian Adult Male with Spondylo-meta-epiphyseal Dysplasia, Short Limb-Abnormal Calcification Type. *J Pediatr Genet.* 2019 Sep;8(3):153-156.
48. Mistri M, Mehta S, Solanki D, Kamate M, Gupta N, Kabra M, Puri R, Girisha K, Hariharan S, Nampoothiri S, Sheth F, Sheth J. Identification of novel variants in a large cohort of children with Tay-Sachs disease: An initiative of a multicentric task force on lysosomal storage disorders by Government of India. *J Hum Genet.* 2019 Oct;64(10):985-994.
49. Kaur R, Gupta N. Hemolytic Anemia and Neurological Manifestations – An Uncommon Combination. *Indian J Pediatr.* 2019 Aug;86(8):673-674.
50. Verma P, Kapoor S, Kalaivani M, Vats P, Yadav S, Jain V, Thelma BK; Science and Engineering Research Board – Newborn Screening Initiative Group (SERB-NBS) members. An Optimal Capillary Screen Cut-off of Thyroid Stimulating Hormone for Diagnosing Congenital Hypothyroidism: Data from a Pilot Newborn Screening Program in Delhi. *Indian Pediatr.* 2019 Apr 15;56(4):281-286.
51. Satapathy AK, Pandey S, Chaudhary MR, Bagga A, Kabra M, Uwe K, Gupta N. Report of Another Mutation Proven Case of Carbonic Anhydrase II Deficiency. *J Pediatr Genet.* 2019 Jun;8(2):91-94.
52. Narayanan DL, Matta D, Gupta N, Kabra M, Ranganath P, Aggarwal S, Phadke SR, Datar C, Gowrishankar K, Kamate M, Jain JMN, Dalal A. Spectrum of ARSA variations in Asian Indian patients with Arylsulfatase A deficient metachromatic leukodystrophy. *J Hum Genet.* 2019 Apr;64(4):323-331.
53. Kaur R, Mishra P, Kumar S, Sankar MJ, Kabra M, Gupta N. Apert syndrome with congenital diaphragmatic hernia: another case report and review of the literature. *Clin Dysmorphol.* 2019 Apr;28(2):78-80.
54. Langawi Mona AI, Byrnes C, Davies JC, Hamouda S, Kabra M, Rached SZ, Sands D, Shteinberg M, Taylor-Cousar J, Tullis E, Wainwright C. "Go for It, Dream Big, Work Hard and Persist": A Message to the Next Generation of CF Leaders in Recognition of International Women's Day 2020 <https://doi.org/10.1016/j.jcf.2020.02.021>
55. Perveen S, Gupta N, Kumar M, Kaur P, Chowdhury MR, Kabra M. Spectrum of amyloglucosidase mutations in Asian Indian patients with Glycogen storage disease type III. *Am J Med Genet A* 2020 Mar 28.
56. Kumar A, Jain V, Chowdhury MR, Kumar M, Kaur P, Kabra M. Pathogenic / likely pathogenic variants in SHOX, GHR and IGFALS genes among Indian children with idiopathic short stature. *Journal of Pediatric Endocrinology and Metabolism.* 2019;33: Issue 1

57. Vanathi M, Shukla R, Balakrishnan P, Dwivedi R, Gupta N, Tandon R. Evaluation of thrombospondin-1 gene polymorphisms in corneal allograft rejection in Asian Indian patients. *Indian J Ophthalmol*. 2020;68(4):565–572.
58. Sachdeva A, Jain P, Gunasekaran V, Mahay SB, Mukherjee S, Hagerman R, Shankar S, Kapoor S, Kedia SN; Indian Academy of Pediatrics Consensus in Diagnosis and Management of Fragile X Syndrome Committee. ; Indian Academy of Pediatrics Consensus in Diagnosis and Management of Fragile X Syndrome Committee. Consensus Statement of the Indian Academy of Pediatrics on Diagnosis and Management of Fragile X Syndrome in India. *Indian Pediatr*. 2019 Mar 15;56(3):221-228.
59. Singh A, Lodha R, Shastri S, Sethuraman G, Sreedevi KN, Kabra M, Kabra SK. Aquagenic Wrinkling of Skin: A Screening Test for Cystic Fibrosis. *Indian Pediatr*. 2019 Feb 15;56(2):109-113.
60. Jauhari P, Saini AG, Suthar R, Sankhyan N, Rehder C, Kishnani P, Gupta N, Kabra M, Singhi P. Thenar Hypertrophy and Electrical Myotonia in Pompe Disease. *J Clin Neuromuscul Dis*. 2019 Mar;20(3):135-137.
61. Meena H, Jana M, Singh V, Kabra M, Jain V. Growth Pattern and Clinical Profile of Indian Children with Classical 21-Hydroxylase Deficiency Congenital Adrenal Hyperplasia on Treatment. *Indian J Pediatr*. 2019 Jan 30.
62. Narayanan DL, Matta D, Gupta N, Kabra M, Ranganath P, Aggarwal S, Phadke SR, Datar C, Gowrishankar K, Kamate M, Jain JMN, Dalal A. Spectrum of ARSA variations in Asian Indian patients with Arylsulfatase A deficient metachromatic leukodystrophy. *J Hum Genet*. 2019 Apr;64(4):323-331.
63. Kaur R, Mishra P, Kumar S, Sankar MJ, Kabra M, Gupta N. Apert syndrome with congenital diaphragmatic hernia: another case report and review of the literature. *Clin Dysmorphol*. 2019 Apr;28(2):78-80.
64. Sheth J, Mistri M, Bhavsar R, Pancholi D, Kamate M, Gupta N, Kabra M, Mehta S, Nampoorthi S, Thakker A, Jain V, Shah R, Sheth F. Batten disease: biochemical and molecular characterization revealing novel PPT1 and TPP1 gene mutations in Indian patients. *BMC Neurol*. 2018 Dec 12;18(1):203.
65. Verma IC, Puri R, Venkataswamy E, Tayal T, Nampoorthi S, Andrew C, Kabra M, Bagga R, Gowda M, Batra M, Hegde S, Kaul A, Gupta N, Mishra P, Subramanian JG, Lingaiah S, Akhtar R, Kidangan F, Chandran R, Kiran C, Ravi Kumar GR, Ramprasad VL, Kadam P. Single Nucleotide Polymorphism-Based Noninvasive Prenatal Testing: Experience in India. *J Obstet Gynaecol India*. 2018 Dec;68(6):462-470..
66. Singh PK, Sharma S, Ghosh M, Shastri SS, Gupta N, Kabra M. Spectrum of GJB2 gene variants in Indian children with non-syndromic hearing loss. *Indian J Med Res*. 2018 Jun;147(6):615-618.
67. Natarajan H, Kumar L, Bakhshi S, Sharma A, Velpandian T, Kabra M, Gogia A, Ranjan Biswas N, Gupta YK. Imatinib trough levels: a potential biomarker to predict cytogenetic and molecular response in newly diagnosed patients with chronic myeloid leukemia. *Leuk Lymphoma*. 2019 Feb;60(2):418-425.

68. Gupta N, Verma G, Kabra M, Bijarnia-Mahay S, Ganapathy A. Identification of a case of SRD5A3-congenital disorder of glycosylation (CDG1Q) by exome sequencing. *Indian J Med Res.* 2018 Apr;147(4):422-426.
69. Yenamandra VK, Vellarikkal SK, Chowdhury MR, Jayarajan R, Verma A, Scaria V, Sivasubbu S, Ray SB, Dinda AK, Kabra M, Sharma VK, Sethuraman G. Genotype-Phenotype Correlations of Dystrophic Epidermolysis Bullosa in India: Experience from a Tertiary Care Centre. *Acta Derm Venereol.* 2018 Oct 10;98(9):873-879.
70. Tewari VV, Mehta R, Sreedhar CM, Tewari K, Mohammad A, Gupta N, Gulati S, Kabra M. A novel homozygous mutation in POLR3A gene causing 4H syndrome: a case report. *BMC Pediatr.* 2018 Apr 4;18(1):126..
71. ICMR Task Force on Inherited Metabolic Disorders. Newborn Screening for Congenital Hypothyroidism and Congenital Adrenal Hyperplasia. *Indian J Pediatr.* 2018 Nov;85(11):935-940.
72. Puri RD, Kapoor S, Kishnani PS, Dalal A, Gupta N, Muranjan M, Phadke SR, Sachdeva A, Verma IC, Mistry PK; Gaucher Disease Task Force. Diagnosis and Management of Gaucher Disease in India - Consensus Guidelines of the Gaucher Disease Task Force of the Society for Indian Academy of Medical Genetics and the Indian Academy of Pediatrics. *Indian Pediatr.* 2018 Feb 15;55(2):143-153.
73. Agarwal M, Bakhshi S, Dwivedi SN, Kabra M, Shukla R, Seth R. Cyclin dependent kinase inhibitor 2A/B gene deletions are markers of poor prognosis in Indian children with acute lymphoblastic leukemia. *Pediatr Blood Cancer.* 2018 Jun;65(6):e27001.
74. Lallar M, Rai A, Srivastava P, Mandal K, Gupta N, Kabra M, Phadke SR. Molecular Testing of MECP2 Gene in Rett Syndrome Phenotypes in Indian Girls. *Indian Pediatr.* 2018 Jun 15;55(6):474-477.
75. Somashekar PH, Girisha KM, Nampoothiri S, Gowrishankar K, Devi RR, Gupta N, Narayanan DL, Kaur A, Bajaj S, Jagadeesh S, Lewis LES, Shailaja S, Shukla A. Locus and allelic heterogeneity and phenotypic variability in Waardenburg syndrome. *Clin Genet.* 2019 Mar;95(3):398-402.
76. Saini I, Bagri N, Gupta N. My Phenotype speaks: please do not harm me with biopsy needle. *Acta Reumatol Port.* 2018 Apr-Jun;43(2):156-158.
77. Kruszka P, Porras AR, de Souza DH, Moresco A, Huckstadt V, Gill AD, Boyle AP, Hu T, Addissie YA, Mok GTK, Tekendo-Ngongang C, Fieggen K, Prijoles EJ, Tanpaiboon P, Honey E, Luk HM, Lo IFM, Thong MK, Muthukumarasamy P, Jones KL, Belhassan K, Ouldin K, El Bouchikhi I, Bouguenouch L, Shukla A, Girisha KM, Sirisena ND, Dissanayake VHW, Paththinige CS, Mishra R, Kisling MS, Ferreira CR, de Herreros MB, Lee NC, Jamuar SS, Lai A, Tan ES, Ying Lim J, Wen-Min CB, Gupta N, Lotz-Esquivel S, Badilla-Porras R, Hussen DF, El Ruby MO, Ashaat EA, Patil SJ, Dowsett L, Eaton A, Innes AM, Shotelersuk V, Badoe Ě, Wonkam A, Obregon MG, Chung BHY, Trubnykova M, La Serna J, Gallardo Jugo BE, Chávez Pastor M, Abarca Barriga HH, Megarbane A, Kozel BA, van Haelst MM, Stevenson RE, Summar M, Adeyemo AA, Morris CA, Moretti-Ferreira D, Linguraru MG, Muenke M. Williams-Beuren syndrome in diverse populations. *Am J Med Genet A.* 2018 May;176(5):1128-1136.
78. Aggarwal B, Gupta N. Familial Hypercholesterolemia: Nip the Evil in the Bud. *Indian J Pediatr.* 2018 May;85(5):331-332.

79. Agarwal M, Bakhshi S, Dwivedi SN, Kabra M, Shukla R, Seth R. Cyclin dependent kinase inhibitor 2A/B gene deletions are markers of poor prognosis in Indian children with acute lymphoblastic leukemia. *Pediatr Blood Cancer*. 2018 Feb 15.
80. Lallar M, Rai A, Srivastava P, Mandal K, Gupta N, Kabra M, Phadke SR. Molecular Testing of MECP2 Gene in Rett Syndrome Phenotypes in Indian Girls. *Indian Pediatr*. 2018 Feb 9. pii: S097475591600115.
81. Gaucher Disease Task Force, Puri RD, Kapoor S, Kishnani PS, Dalal A, Gupta N, Muranjan M, Phadke SR, Sachdeva A, Verma IC, Mistry PK. Diagnosis and Management of Gaucher Disease in India - Consensus Guidelines of the Gaucher Disease Task Force of the Society for Indian Academy of Medical Genetics and the Indian Academy of Pediatrics. *Indian Pediatr*. 2018 Feb 15;55(2):143-153.
82. Sinha R, Singh P, Saini NK, Kumar A, Pathak R, Chandolia A, Garima K, Tyagi G, Chopra M, Prasad AK, Raj HG, Bose M. Methyl-accepting chemotaxis like Rv3499c (Mce4A) protein in Mycobacterium tuberculosis H37Rv mediates cholesterol-dependent survival. *Tuberculosis (Edinb)*. 2018 Mar;109:52-60.
83. 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 quadriplegia. *J Hum Genet*. 2018 Jan;63(1):19-25.
84. Gupta N, Jain V. Prader Willi Syndrome - A Common Epigenetic Cause of Syndromic Obesity. *Indian J Pediatr*. 2017 Nov;84(11):809-810.
85. Mishra A, Devi S, Saxena R, Gupta N, Kabra M, Chowdhury MR. Frequency of primary mutations of Leber's hereditary optic neuropathy patients in North Indian population. *Indian J Ophthalmol*. 2017 Nov;65(11):1156-1160.
86. Jain V, Kabra M. The Unusual Story of an Infant with Congenital Adrenal Hyperplasia. *Indian Pediatr*. 2017 Sep 15;54(9):781-782.
87. Singh PK, Ghosh M, Sharma S, Shastri S, Gupta N, Chowdhury MR, Anand A, Kabra M. Identification of a novel homozygous mutation in transmembrane channel like 1 (TMC1) gene, one of the second-tier hearing loss genes after GJB2 in India. *Indian J Med Res*. 2017 Apr;145(4):492-497.
88. Gupta N, Tewari VV, Kumar M, Langeh N, Gupta A, Mishra P, Kaur P, Ramprasad V, Murugan S, Kumar R, Jana M, Kabra M. Asparagine Synthetase deficiency-report of a novel mutation and review of literature. *Metab Brain Dis*. 2017 Dec;32(6):1889-1900.
89. Jana M, Nair N, Gupta AK, Kabra M, Gupta N. Pelvic radiograph in skeletal dysplasias: An approach. *Indian J Radiol Imaging*. 2017 Apr-Jun;27(2):187-199.
90. Aathira R, Gulati S, Tripathi M, Shukla G, Chakrabarty B, Sapra S, Dang N, Gupta A, Kabra M, Pandey RM. Prevalence of Sleep Abnormalities in Indian Children With Autism Spectrum Disorder: A Cross-Sectional Study. *Pediatr Neurol*. 2017 Sep;74:62-67.
91. Gupta S, Mukherjee A, Khadgawat R, Kabra M, Lodha R, Kabra SK. Bone Mineral Density of Indian Children and Adolescents with Cystic Fibrosis. *Indian Pediatr*. 2017 Jul 15;54(7):545-549.
92. Harivenkatesh N, Kumar L, Bakhshi S, Sharma A, Kabra M, Velpandian T, Gogia A, Shastri SS, Gupta YK. Do polymorphisms in MDR1 and CYP3A5 genes influence the risk of

- cytogenetic relapse in patients with chronic myeloid leukemia on imatinibtherapy? *Leuk Lymphoma*. 2017 Sep;58(9):1-9.
93. Kumar A, Jaryal A, Gulati S, Chakrabarty B, Singh A, Deepak KK, Pandey RM, Gupta N, Sapra S, Kabra M, Khajuria R. Cardiovascular Autonomic Dysfunction in Children and Adolescents With Rett Syndrome. *Pediatr Neurol*. 2017 May;70:61-66.
 94. Harivenkatesh N, Kumar L, Bakhshi S, Sharma A, Kabra M, Velpandian T, Gogia A, Shastri SS, Biswas NR, Gupta YK. Influence of MDR1 and CYP3A5 genetic polymorphisms on trough levels and therapeutic response of imatinib in newly diagnosed patients with chronic myeloid leukemia. *Pharmacol Res*. 2017 Jun;120:138-145.
 95. Chowdhury MR, Chauhan S, Dabral A, Thelma BK, Gupta N, Kabra M. Validation of Polymerase Chain Reaction-Based Assay to Detect Actual Number of CGG Repeats in FMR1 Gene in Indian Fragile X Syndrome Patients. *J Child Neurol*. 2017 Mar;32(4):371-378.
 96. Yenamandra VK, Vellarikkal SK, Kumar M, Chowdhury MR, Jayarajan R, Verma A, Scaria V, Sivasubbu S, Ray SB, Dinda AK, Kabra M, Kaur P, Sharma VK, Sethuraman G. Application of whole exome sequencing in elucidating the phenotype and genotype spectrum of junctional epidermolysis bullosa: A preliminary experience of a tertiary care centre in India. *J Dermatol Sci*. 2017 Apr;86(1):30-36.
 97. Dar L, Namdeo D, Kumar P, Thakar A, Kant S, Rai S, Singh PK, Kabra M, Fowler KB, Boppana SB. Congenital Cytomegalovirus Infection and Permanent Hearing Loss in Rural North Indian Children. *Pediatr Infect Dis J*. 2017 Jul;36(7):670-673.
 98. Das Bhowmik A, Gupta N, Dalal A, Kabra M. Whole exome sequencing identifies a homozygous nonsense variation in ALMS1 gene in a patient with syndromic obesity. *Obes Res Clin Pract*. 2017 Mar - Apr;11(2):241-246.
 99. Amit Kumar Satapathy, Alec Correa, Madhulika Kabra, Sabrina Eichler, Arndt Rolfs, Manisha Jana, Neerja Gupta. Ethylmalonic encephalopathy masquerading as malabsorption syndrome - A case report. *Meta Gene* 13 (2017) 115–118
 100. Mishra A, Devi S, Saxena R, Gupta N, Kabra M, Chowdhury MR. Frequency of primary mutations of Leber's hereditary optic neuropathy patients in North Indian population. *Indian J Ophthalmol* 2017; 65: 11; 1156-1160.