

List of Publications (Last 5 years)

1. Dröge C, Bonus M, Baumann U, Klindt C, Lainka E, Kathemann S, Brinkert F, Grabhorn E, Pfister ED, Wenning D, Fichtner A, Gotthardt DN, Weiss KH, McKiernan P, **Puri RD**, Verma IC, Kluge S, Gohlke H, Schmitt L, Kubitz R, Häussinger D, Keitel V. Sequencing of FIC1, BSEP and MDR3 in a large cohort of patients with cholestasis revealed a high number of different genetic variants. *J Hepatol*. 2017 Jul 18
2. 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 Jun 4
3. Verma J, Thomas DC, Jhingan G, **Puri RD**, Verma IC. MoM cutoffs for variables, an important tool for multivariate analysis and accurate interpretation of preeclampsia risk in high-risk pregnancy at 11-13(+6) weeks gestation. *Hypertens Pregnancy*. 2016 Nov;35(4):548-558
4. Soni JP, **Puri RD**, Jetha K, Bhavani GS, Chaudhary M, Kohli S, Verma IC. Infantile Systemic Hyalinosis: Novel Founder Mutation in the Initiation Codon among "Malis (Farmers)" in Jodhpur. *Indian J Pediatr*. 2016 Nov;83(11):1341-1345. Epub 2016 Oct 18. PubMed PMID: 27753005.
5. Verma J, Thomas DC, Jhingan G, **Puri RD**, Verma IC. MoM cutoffs for variables, an important tool for multivariate analysis and accurate interpretation of preeclampsia risk in high-risk pregnancy at 11-13+6 weeks gestation. *Hypertens Pregnancy*. 2016 Nov;35(4):548-558.
6. Soni JP, **Puri RD**, Jetha K, Bhavani GS, Chaudhary M, Kohli S, Verma IC. Infantile Systemic Hyalinosis: Novel Founder Mutation in the Initiation Codon among "Malis (Farmers)" in Jodhpur. *Indian J Pediatr*. 2016 Nov;83(11):1341-1345.
7. **Puri RD**, Tuteja M, Verma IC. Genetic Approach to Diagnosis of Intellectual Disability. *Indian J Pediatr*. 2016 Oct;83(10):1141-9.
8. **Puri RD**, Kabra M. Editorial: New Horizons in Genetic Diagnosis in Pediatric Practice: The Excitement and Challenges! *Indian J Pediatr*. 2016 Aug 11.
9. Vyas B, **Puri RD**, Namboodiri N, Saxena R, Nair M, Balakrishnan P, Jayakrishnan MP, Udyavar A, Kishore R, Verma IC. Phenotype guided characterization and molecular analysis of Indian patients with long QT syndromes. *Indian Pacing Electrophysiol J*. 2016 Jan-Feb;16(1):8-18
10. 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 Jun 24. doi: 10.1002/ajmg.a.37817. [Epub ahead of print]

11. Gupta D, Bijarnia-Mahay S, Kohli S, Saxena R, **Puri RD**, Shigematsu Y, Yamaguchi S, Sakamoto O, Gupta N, Kabra M, Thakur S, Deb R, Verma IC. Seventeen Novel Mutations in PCCA and PCCB Genes in Indian Propionic Acidemia Patients, and Their Outcomes. *Genet Test Mol Biomarkers*. 2016 Jul;20(7):373-82
12. **Puri RD**, Kotecha U, Lall M, Dash P, Bijarnia-Mahay S, Verma IC. Is the diagnostic yield influenced by the indication for fetal autopsy? *Am J Med Genet A*. 2016 Aug;170(8):2119-26.
13. **Puri RD**. Next Generation Sequencing in the Clinic. *Indian J Pediatr* (2016) 83: 281
14. Roosing S, Romani M, Isrie M, Rosti RO, Micalizzi A, Musaev D, Mazza T, Al-Gazali L, Altunoglu U, Boltshauser E, D'Arrigo S, De Keersmaecker B, Kayserili H, Brandenberger S, Kraoua I, Mark PR, McKanna T, Van Keirsbilck J, Moerman P, Poretti A, Puri R, Van Esch H, Gleeson JG, Valente EM. Mutations in CEP120 cause Joubert syndrome as well as complex ciliopathy phenotypes. *J Med Genet*. 2016 May 6. pii: jmedgenet-2016-103832..
15. Uttarilli A, Ranganath P, Matta D, Md Nurul Jain J, Prasad C K, Babu A S, Girisha KM, Verma IC, Phadke SR, Mandal K, **Puri RD**, Aggarwal S, Danda S, H SV, Kapoor S, Bhat M, Gowrishankar K, Hasan AQ, Nair M, Nampoothiri S, Dalal A. Identification and Characterization of 20 Novel Pathogenic Variants in 60 unrelated Indian patients with Mucopolysaccharidoses (MPS) type I and type II. *Clin Genet*. 2016 May 5. doi: 10.1111/cge.12795. [Epub ahead of print]
16. Vyas B, **Puri RD**, Namboodiri N, Nair M, Sharma D, Movva S, Saxena R, Bohora S, Aggarwal N, Vora A, Kumar J, Singh T, Verma IC. KCNQ1 mutations associated with Jervell and Lange-Nielsen syndrome and autosomal recessive Romano-Ward syndrome in India-expanding the spectrum of long QT syndrome type 1. *Am J Med Genet A*. 2016 Jun;170(6):1510-9.
17. Verma J, Thomas DC, Kasper DC, Sharma S, Puri RD, Bijarnia-Mahay S, Mistry PK, Verma IC. Inherited Metabolic Disorders: Efficacy of Enzyme Assays on Dried Blood Spots for the Diagnosis of Lysosomal Storage Disorders. *JIMD Rep*. 2016 Mar 24. [Epub ahead of print]
18. Uttarilli A, Ranganath P, Jain SJ, Prasad CK, Sinha A, Verma IC, Phadke SR, **Puri RD**, Danda S, Muranjan MN, Jevalikar G, Nagarajaram HA, Dalal AB. Novel mutations of the arylsulphatase B (ARSB) gene in Indian patients with mucopolysaccharidosis type VI. *Indian J Med Res*. 2015 Oct;142(4):414-25
19. Verma IC, **Puri RD**. Global burden of genetic disease and the role of genetic screening. *Semin Fetal Neonatal Med*. 2015 Oct;20(5):354-63.
20. Verma J, Thomas DC, Sharma S, Jhingan G, Saxena R, Kohli S, **Puri RD**, Bijarnia S, Verma IC. Inherited metabolic disorders: prenatal diagnosis of lysosomal storage disorders. *Prenat Diagn*. 2015 Nov;35(11):1137-47.

21. Bhavani GS, Shah H, Dalal AB, Shukla A, Danda S, Aggarwal S, Phadke SR, Gupta N, Kabra M, Gowrishankar K, Gupta A, Bhat M, **Puri RD**, Bijarnia-Mahay S, Nampoothiri S, Mohanasundaram KM, Rajeswari S, Kulkarni AM, Kulkarni ML, Ranganath P, Ramadevi AR, Hariharan SV, Girisha KM. [Novel and recurrent mutations in WISP3 and an atypical phenotype](#). Am J Med Genet A. 2015 Oct;167A(10):2481-4.
22. Verma J, Thomas DC, Sharma S, Jhingan G, Singh A, Hsiao KJ, Schoonderwoerd K, **Puri RD**, Verma IC. [Inherited metabolic disorders: Quality management for laboratory diagnosis](#). Clin Chim Acta. 2015 Jul 20;447:1-7
23. Bidchol AM, Dalal A, Trivedi R, Shukla A, Nampoothiri S, Sankar VH, Danda S, Gupta N, Kabra M, Hebbar SA, Bhat RY, Matta D, Ekbote AV, **Puri RD**, Phadke SR, Gowrishankar K, Aggarwal S, Ranganath P, Sharda S, Kamate M, Datar CA, Bhat K, Kamath N, Shah H, Krishna S, Gopinath PM, Verma IC, Nagarajaram HA, Satyamoorthy K, Girisha KM. [Recurrent and novel GLB1 mutations in India](#). Gene. 2015 Aug 10;567(2):173-81.
24. Bijarnia-Mahay S, Movva S, Gupta N, Sharma D, **Puri RD**, Kotecha U, Saxena R, Kabra M, Mohan N, Verma IC. [Molecular Diagnosis of Hereditary Fructose Intolerance: Founder Mutation in a Community from India](#). JIMD Rep. 2015;19:85-93.
25. 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. [Prenatal diagnosis in India is not limited to sex selection](#). Genet Med. 2015 Jan;17(1):88.
26. Bidchol AM, Dalal A, Shah H, S S, Nampoothiri S, Kabra M, Gupta N, Danda S, Gowrishankar K, Phadke SR, Kapoor S, Kamate M, Verma IC, **Puri RD**, Sankar VH, Devi AR, Patil SJ, Ranganath P, Jain SJ, Agarwal M, Singh A, Mishra P, Tamhankar PM, Gopinath PM, Nagarajaram HA, Satyamoorthy K, Girisha KM. [GALNS mutations in Indian patients with mucopolysaccharidosis IVA](#). Am J Med Genet A. 2014 Nov;164A(11):2793-801
27. Gupta D, Bijarnia-Mahay S, Saxena R, Kohli S, Dua-Puri R, Verma J, Thomas E, Shigematsu Y, Yamaguchi S, Deb R, Verma IC. [Identification of mutations, genotype-phenotype correlation and prenatal diagnosis of maple syrup urine disease in Indian patients](#). Eur J Med Genet. 2015 Sep;58(9):471-8
28. Puri RD. Fetal Dysmorphology. Puri, R.D. J. Fetal Med. (2015) 2: 151.
29. Dash, P., Puri, R.D., Goyal, M. et al. **Absent/Hypoplastic Fetal Nasal Bone and Its Association with Aneuploidies**. J. Fetal Med. (2015) 2: 75.
30. **Sunita Bijarnia-Mahay1 • Ratna D. Puri1 • Udhaya Kotecha1 • Pratima Dash1 • Swasti Pal1 • Meena Lall1 • Surbhi Mahajan1 • Pushpa Saviour1 • Preeti Paliwal1 • Ashok Baijal2 • Nandita Dimri2 • Nidhish Sharma2 • Ishwar C. Verma1 Outcome of Prenatally-Detected Fetal Ventriculomegaly.**
31. Bashyam MD, Chaudhary AK, Kiran M, Nagarajaram HA, Devi RR, Ranganath P, Dalal A, Bashyam L, Gupta N, Kabra M, Muranjan M, Puri RD, Verma IC, Nampoothiri S, Kadandale JS. 2014. Splice, insertion-deletion and nonsense mutations that perturb the phenylalanine hydroxylase transcript cause phenylketonuria in India. [J Cell Biochem](#). Mar;115 (3):566-74
32. Puri RD, Verma IC. Role of radiographs in Fetal Autopsy. 2014.J.Fetal Med.1: 7-9

33. Kotecha UH, Movva S, Sharma D, Verma J, Puri RD, Verma IC. Molecular evaluation of a novel missense mutation & an insertional truncating mutation in SUMF1 gene. *Indian J Med Res.* 2014 Jul;140(1):55-9.
34. Bashyam MD, Chaudhary AK, Kiran M, Reddy V, Nagarajaram HA, Dalal A, Bashyam L, Suri D, Gupta A, Gupta N, Kabra M, Puri R, Ramadevi R, Kapoor S, Danda S. 2013. Molecular analyses of novel ASAH1 mutations causing Farber lipogranulomatosis: analyses of exonic splicing enhancer inactivating mutation. *Clin Genet.* Nov 8. doi: 10.1111/cge.12316
35. Bashyam MD, Chaudhary AK, Kiran M, Reddy V, Nagarajaram HA, Dalal A, Bashyam L, Suri D, Gupta A, Gupta N, Kabra M, Puri RD, Ramadevi R, Kapoor S, Danda S. Molecular analyses of novel ASAH1 mutations causing Farber lipogranulomatosis: analyses of exonic splicing enhancer inactivating mutation. *Clin Genet.* 2014 Dec;86(6):530-8.
36. Bashyam MD, Chaudhary AK, Sinha M, Nagarajaram HA, Ramadevi R, Ranganath P, Dalal A, Bashyam L, Gupta N, Kabra M, Muranjan M, Puri RD, Verma IC, Nampoothiri S, Kadandale JS. Splice, insertion-deletion and nonsense mutations that potentially perturb the Phenylalanine hydroxylase transcript are a major cause for phenylketonuria in India. *J Cell Biochem.* 2014 Mar;115(3):566-74.
37. Singla D, Gujral K, Nandita Dimrib, Ratna Puric, Jyotsana Verma, I.C. Verma Contingent screening for Down syndrome: A pilot study. *Current Medicine Research and Practice* Volume 4, Issue 5, September–October 2014, Pages 201–205
38. Nahar R, Deb R, Saxena R, Puri RD, Verma IC. Variability in CYP2C9 allele frequency: A pilot study of its predicted impact on warfarin response among healthy South and North Indians. *Pharmacol Rep.* 2013;65(1):187-94
39. Dua V, Yadav SP, Kumar V, Khan AA, Puri R, Verma IC, Flanagan SE, Ellard S, Sachdeva A. Thiamine Responsive Megaloblastic Anemia With a Novel SLC19A2 Mutation Presenting With Myeloid Maturational Arrest. *Pediatr Blood Cancer.* 2013 Mar 19
40. Nahar R, Puri RD, Saxena R, Verma IC. Do parental Perceptions and Motivations towards Genetic testing and Prenatal Diagnosis for Deafness vary in different Cultures? *American journal of Medical Genetics.* In Press
41. Agarwal N, Balani S, Arya S, Puri RD. Non Invasive Management of Rhesus Alloimmunization. *International Journal of Infertility & Medicine.* 2013, Vol 4.No 2: 59-61
42. Chung SK, Bode A, Cushion TD, Thomas RH, Hunt C, Wood SE, Pickrell WO, Drew CJ, Yamashita S, Shiang R, Leiz S, Longhardt AC, Raile V, Weschke B, **Puri RD**, Verma IC, Harvey RJ, Ratnasinghe DD, Parker M, Rittey C, Masri A, Lingappa L, Howell OW, Vanbellighen JF, Mullins JG, Lynch JW, Rees MI. GLRB is the third major gene of effect in hyperekplexia. *Hum Mol Genet.* 2013 Mar 1;22(5):927-40.