

Dr Anju Shukla - Publications

1. 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 May 18. doi: 10.1002/ajmg.a.37164. [Epub ahead of print] PubMed PMID: 25988854.
2. 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. doi: 10.1016/j.gene.2015.04.078. Epub 2015 Apr 30. PubMed PMID: 25936995.
3. Shukla A, Mandal K, Patil SJ, Kishore Y, Phadke SR, Girisha KM. Co-occurrence of a de novo Williams and 22q11.2 microdeletion syndromes. *Am J Med Genet A*. 2015 Apr 21. doi: 10.1002/ajmg.a.37116. [Epub ahead of print] PubMed PMID: 25898978.
4. Nayak SS, Shukla A, Lewis L, Kadavigere R, Mathew M, Adiga PK, Vasudeva A, Kumar P, Shetty J, Shah H, Girisha KM. Clinical utility of fetal autopsy and its impact on genetic counseling. *Prenat Diagn*. 2015 Jul;35(7):685-91. doi: 10.1002/pd.4592. Epub 2015 Apr 5. PubMed PMID: 25763538.
5. Nayak SS, Shukla A, Girisha KM. Anomalies associated with single umbilical artery at perinatal autopsy. *Indian Pediatr*. 2015 Jan;52(1):73-4. PubMed PMID: 25638195.

6. Shukla A, Phadke SR. Chondrodysplasia punctata tibia metacarpal type: report of a 1.5 year old child with severe short stature and extensive calcific stippling. *Clin Dysmorphol*. 2015 Jul;24(3):118-21. doi: 10.1097/MCD.000000000000076. PubMed PMID: 25602717.
7. 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. doi: 10.1016/j.ejmg.2014.10.001. Epub 2014 Oct 24. PubMed PMID: 25450603.
8. Stephen J, Shukla A, Dalal A, Girisha KM, Shah H, Gupta N, Kabra M, Dabadghao P, Phadke SR. Mutation spectrum of COL1A1 and COL1A2 genes in Indian patients with osteogenesis imperfecta. *Am J Med Genet A*. 2014 Jun;164A(6):1482-9. doi: 10.1002/ajmg.a.36481. Epub 2014 Mar 25. PubMed PMID: 24668929.
9. Boggula VR, Shukla A, Danda S, Hariharan SV, Nampoothiri S, Kumar R, Phadke SR. Clinical utility of multiplex ligation-dependent probe amplification technique in identification of aetiology of unexplained mental retardation: a study in 203 Indian patients. *Indian J Med Res*. 2014 Jan;139(1):66-75. PubMed PMID: 24604040; PubMed Central PMCID: PMC3994742.
10. Shukla A, Taywade O, Stephen J, Gupta D, Phadke SR. Fibrodysplasia ossificans progressiva: three indian patients with mutation in the ACVR1 gene. *Indian J Pediatr*. 2014 Jun;81(6):617-9. doi: 10.1007/s12098-013-1117-5. Epub 2013 Aug 6. PubMed PMID: 23918320.