

## **Dr Girisha KM - 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. 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.
4. 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.
5. Daly SB, Shah H, O'Sullivan J, Anderson B, Bhaskar S, Williams S, Al-Sheqaih N, Mueed Bidchol A, Banka S, Newman WG, Girisha KM. Exome Sequencing Identifies a Dominant TNNT3

Mutation in a Large Family with Distal Arthrogyriposis. *Mol Syndromol*. 2014 Aug;5(5):218-28. doi: 10.1159/000365057. Epub 2014 Jul 8. PubMed PMID: 25337069; PubMed Central PMCID: PMC4188168.

6. 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. doi: 10.1002/ajmg.a.36735. Epub 2014 Sep 22. PubMed PMID: 25252036.
7. 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.
8. Girisha KM, Bidchol AM, Kamath PS, Shah KH, Mortier GR, Mundlos S, Shah H. A novel mutation (g.106737G>T) in zone of polarizing activity regulatory sequence (ZRS) causes variable limb phenotypes in Werner mesomelia. *Am J Med Genet A*. 2014 Apr;164A(4):898-906. doi: 10.1002/ajmg.a.36367. Epub 2014 Jan 29. PubMed PMID: 24478176.
9. Dalal A, Bhavani G SL, Togarrati PP, Bierhals T, Nandineni MR, Danda S, Danda D, Shah H, Vijayan S, Gowrishankar K, Phadke SR, Bidchol AM, Rao AP, Nampoothiri S, Kutsche K, Girisha KM. Analysis of the WISP3 gene in Indian families with progressive pseudorheumatoid dysplasia. *Am J Med Genet A*. 2012 Nov;158A(11):2820-8. doi: 10.1002/ajmg.a.35620. Epub 2012 Sep 17. PubMed PMID: 22987568.

10. Girisha KM, Abdollahpour H, Shah H, Bhavani GS, Graham JM Jr, Boggula VR, Phadke SR, Kutsche K. A syndrome of facial dysmorphism, cubital pterygium, short distal phalanges, swan neck deformity of fingers, and scoliosis. *Am J Med Genet A*. 2014 Apr;164A(4):1035-40. doi: 10.1002/ajmg.a.36381. Epub 2014 Jan 23. PubMed PMID: 24458843.