Curriculum Vitae: Dr Girisha KM

Professor and Head
Department of Medical Genetics
Kasturba Medical College, Manipal
Manipal Academy of Higher Education
Manipal-576104, India
Telephone: +91 820 2923149
Email: girish.katta@manipal.edu

Education and Training

- MBBS, Government Medical College, Mysore, 1992-1998
- Diploma in Child Health, College of Physicians and Surgeons of Bombay, 2001
- MD (Pediatrics), Seth GS Medical College and KEM Hospitals, Mumbai, 1999-2002
- DM (Medical Genetics), Sanjay Gandhi Postgraduate Institute of Medical Sciences, Lucknow, 2003-2005

Current post and responsibilities

I am currently a Professor of Medical Genetics at Kasturba Medical College, Manipal since 16 July 2012. I head the department and look after the administration of the department. I am clinically qualified (paediatrician and clinical geneticist) and is clinically active. Our department provides consultations to patients and families with genetic disorders and birth defects, offers genomic testing (diagnostic) and is actively inloved in research. Our team runs a master of science in genetic counseling program in addition to doctoral training (PhD). We are in the process of getting accreditation for a residency program in clinical genetics.

Previous posts

- Associate Professor, Kasturba Medical College, Manipal, 1 July 2009 to 15 July 2012
- Assistant Professor, Kasturba Medical College, Manipal, 25 August to 30 June 2009
- Assistant Professor, Sanjay Gandhi Postgraduate Institute of Medical Sciences, Lucknow, 6 January 2007 to 19 August 2018

Memberships

- Member of International Skeletal Dysplasia Society (ISDS) Committee for the Nosology and Classification of Genetic Skeletal Disorders
- Founding member and current secretary of Indian Academy of Medical Genetics

- Life member of Indian Academy of Pediatrics and its Genetics Specialty Chapter
- Member of American College of Medical Genetics
- Member of Indian Society of Human Genetics
- Advisor for Organization for Rare Diseases India (ORDI)

Scientific activities

- Associate editor for American Journal of Medical Genetics Part A
- Reviewer for more than 10 journals
- Author of more than 161 scientific papers
- Organizer of five scientific meetings at Manipal (www.manipalgeneticsupdate.com)
- Supervisor for 7 PhD scholars (4 completed, 3 current)

Research projects

- 'Genotype and phenotype correlation of Indian patients with Morquio syndrome' sanctioned by Indian Council of Medical Research, 2009-2012
- 'Value of fetal autopsy and establishment of normal fetal radiologic anatomy' sanctioned by Indian Council of Medical Research, 2010-2013
- 'Evaluation of multiplex ligation dependent probe amplification (MLPA) for diagnosis and carrier detection in families with a dystrophinopathy' sanctioned by Department of Science and Technology, 2012-2015
- 'Multicentric Collaborative Study of the Clinical, Biochemical and Molecular Characterization of Lysosomal Storage Disorders in India: The Initiative for Research in Lysosomal Storage Disorders' sanctioned by Indian Council of Medical Research, 2015-2018
- 'Clinical and molecular evaluation of inherited arthropathies and multiple vertebral segmentation defects' sanctioned by Indian Council of Medical Research, 2015-2018
- 'Development and application of a next generation sequencing based gene panel for disorders with low bone mineral density' sanctioned by DBT-BMBF (Indo-German joint call), 2015-2018
- 'Application of autozygosity mapping and exome sequencing to identify genetic basis of disorders of skeletal development', sanctioned by Department of Science and Technology, 2015-2018
- 'Genetic Diagnosis of Heritable Neurodevelopmental Disorders with Exome Sequencing' sanctioned by National Institute of Health, USA, 2016-2018
- 'Improving the clinical care of children and young adults with Marfan syndrome and related disorders by molecular genetic testing through next-generation sequencing' sanctioned by ICMR-BMBF, 2017-2019

- ‘Does Postmortem Magnetic Resonance Imaging of Fetal Brain Help in Autopsy’ sanctioned by ICMR, 2017-2020

Selected publications (of about 162)

- Chang HR, Cho SY, Lee JH, Lee E, Seo J, Lee HR, Cavalcanti DP, Mäkitie O, Valta H, **Girisha KM** *et al*., Hypomorphic Mutations in TONSL Cause SPONASTRIME Dysplasia. Am J Hum Genet. PMID: 30773278 (Impact factor: 8.855)
- Van Bergen NJ, Guo Y, Rankin J, Paczia N, Becker-Kettern J, Kremer LS, Pyle A,Conrotte JF, Ellaway C, Procopis P, Prelog K, Homfray T, Baptista J, Baple E,Wakeling M, Massey S, Kay DP, Shukla A, **Girisha KM** *et al*., NAD(P)HX dehydratase (NAXD) deficiency: a novel neurodegenerative disorder exacerbated by febrile illnesses. Brain. 2019. PMID: 30576410 (Impact factor: 10.848)
- **Girisha KM**, von Elsner L, Neethukrishna K, Muranjan M, Shukla A, Bhavani GS, Nishimura G, Kutsche K, Mortier G. The homozygous variant c.797G>A/p.(Cys266Tyr) in PISD is associated with a Spondyloepimetaphyseal dysplasia with large epiphyses and disturbed mitochondrial function. Hum Mutat. 2019. PMID: 30488656 (Impact factor: 5.359)
- Uttarilli A, Shah H, Bhavani GS, Upadhyai P, Shukla A, **Girisha KM**. Phenotyping and genotyping of skeletal dysplasias: Evolution of a center and a decade of experience in India. Bone. 2019. PMID: 30408610 (Impact factor: 4.455)
- Szenker-Ravi E, Altunoglu U, Leushacke M, Bosso-Lefèvre C, Khatoo M, Thi Tran H, Naert T, Noelanders R, Hajamohideen A, Beneteau C, de Sousa SB, Karaman B, Latypova X, Başaran S, Yücel EB, Tan TT, Vlaminck L, Nayak SS, Shukla A, **Girisha KM**, Le Caignec C, Soshnikova N, Uyguner ZO, Vleminckx K, Barker N, Kayserili H, Reversade B. RSPO2 inhibition of RNF43 and ZNRF3 governs limb development independently of LGR4/5/6. Nature. 2018. PMID: 29769720 (Impact factor: 41.577)
- Shukla A, Hebbar M, Srivastava A, Kadavigere R, Upadhyai P, Kanthi A, Brandau O, Bielas S, **Girisha KM**. Homozygous p.(Glu87Lys) variant in ISCA1 is associated with a multiple mitochondrial dysfunctions syndrome. J Hum Genet. 2017. PMID: 28356563 (Impact factor: 2.942)
- **Girisha KM**, Shukla A, Trujillano D, Bhavani GS, Hebbar M, Kadavigere R, Rolfs A. A homozygous nonsense variant in IFT52 is associated with a human skeletal ciliopathy. Clin Genet. 2016. PMID: 26880018 (Impact factor: 3.512)
- **Girisha KM**, Harms FL , Hardigan AA, Kortüm F, Shukla A, Alawi M, Dalal A *et al*., Mutations in EBF3 Disturb Transcriptional Profiles and Cause Intellectual Disability, Ataxia, and Facial Dysmorphism. Am J Hum Genet. 2017. PMID: 28017373 (Impact factor: 8.855)

- Kiper POS, Saito H, Gori F, Unger S, Hesse E, Yamana K, Kiviranta R, Solban N, Liu J, Brommage R, Boduroglu K, Bonafé L, Campos-Xavier B, Dikoglu E, Eastell R, Gossiel F, Harshman K, Nishimura G, **Girisha KM**, Stevenson BJ, Takita H, Rivolta C, Superti-Furga A, Baron R. Cortical-Bone Fragility--Insights from sFRP4 Deficiency in Pyle's Disease. N Engl J Med. 2016. PMID: 27355534 (Impact factor: 79.258)
- **Girisha KM**, Kortüm F, Shah H, Alawi M, Dalal A, Bhavani GS, Kutsche K. A novel multiple joint dislocation syndrome associated with a homozygous nonsense variant in the EXOC6B gene. Eur J Hum Genet. 2016. PMID: 26669664. (Impact factor: 4.349)