A decade of experience of molecular testing for skeletal dysplasia in India

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Abstract:

Skeletal dysplasia team at Manipal comprises of a medical genetics unit with clinical and molecular testing facilities and a pediatric orthopedics service. The center provides comprehensive care for patients and families with a skeletal dysplasia, including testing, genetic counselling, management and prenatal diagnosis since 2008. The infrastructure has evolved from initial facility for targeted mutation analysis for common skeletal dysplasia to providing molecular testing for all known skeletal disorders as well as research based testing for several novel skeletal disorders through exome sequencing with in-house bioinformatics analysis.

Various national and international funding agencies supported our endeavor to investigate progressive pseudorheumatoid dysplasia, Morquio syndrome, GM1 gangliosidosis, osteogeneis imperfecta and multicentric osteolysis nodulosis arthropathy. These large series of clinical and mutational profiles of skeletal dysplasia have significantly enhanced the understanding of these disorders, capitalizing on the huge population of our country.

Current funded research projects focus on inherited arthropathies, vertebral segmentation defects, autozygosity mapping of skeletal dysplasia and conditions with low bone mineral density. Sanger sequencing of more than 20 genes is available. Also, exome sequencing is available for clinical and research testing in the centre which aids in providing a definitive diagnosis for a significant proportion of patients with skeletal dysplasia.

The center caters to the regional patients, and facilitates molecular testing for more than 50 clinical collaborators across the country. A well-established network has enabled us to confirm molecular diagnosis in 483 patients with skeletal dysplasia on research basis. We have been successful in delineation of new clinical phenotypes, new genotypes and contributing to the discovery of four new genes for skeletal dysplasia.