Opinion

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Experience of Skeletal Dysplasia in a Tertiary Referral Hospital in India

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India, being the second most populous country in the world, provides a huge opportunity for studying rare genetic Mendelian disorders. Autosomal recessive genetic bone disorders precipitate due to high rate of consanguineous marriages in several regions and communities. The recent advances in molecular techniques, specifically next generation sequencing is now available in the country which has led to the beginning of novel gene discovery notably for autosomal recessive disorders. However, further efforts are needed for functional characterization of novel mutations and genes. Though clinical genetic services are now available throughout the country, there is huge demand-supply mismatch.

Historic contributions from India in the field of skeletal dysplasias include recognition of Handigodu joint disease, Verma-Naumoff short-rib thoracic dysplasia syndrome and a few single case reports (Gorham like syndrome, handless footless imperforate anus cleft palate syndrome and a complex camptopolysyndactyly). A vast number of clinical collaborators have come together to publish mutations in large series of patients with progressive pseudorheumatoid dysplasia, Morquio syndrome, GM1 gangliosidosis, Hurler syndrome, Hunter syndrome, Torg-Winchester syndrome and osteogenesis imperfecta. Application of exome sequencing has resulted in discovery of new candidate genes for skeletal dysplasia including those for skeletal ciliopathies and a skeletal dysplasia with multiple joint laxity and dislocations. Several international collaborations have been initiated for the purpose of novel gene discovery and their functional characterization, thus working towards a better understanding of etiopathogenesis of these rare disorders.

Kasturba Medical College, Manipal has a specialized pediatric orthopedics team working with the medical geneticists in the field of skeletal dysplasia. They have excellent backup of national collaborators spread across the country. Our current research focusses on all skeletal dysplasias in general. Specific areas of interest are inherited arthropathies, autozygosity mapping of recessive skeletal disorders and vertebral segmentation defects. I would present our team's experience in the field and deliberate upon the opportunities for collaboration with Indian colleagues in the field of skeletal dysplasia.

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