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Hospital-based study of the spectrum of skeletal dysplasias in children in Northern India

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Children presenting with disproportionate short stature and obvious skeletal deformities were evaluated for skeletal dysplasia in a tertiary care hospital in Northern India. Fifty one children were included from March 2013 to March 2014 in this cross-sectional study. Data collection included detailed history, development, antenatal ultrasonogram, anthropometry and dysmorphic assessment. Skeletal survey/infantogram, thyroid profile, serum calcium, phosphate, alkaline phosphatase, urinary glycosaminoglycan and enzyme analysis were done in relevant cases. London Dysmorphology Database and European Skeletal dysplasia Registry were utilized. Molecular testing using PCR technique was conducted when feasible. Thirty five were males and 16 were females and maximum belonged to 6-10 year age group (27%). Twenty six (51%) were short trunk and 25 (49%) were short limb type, further grouped into Rhizomelic (72%), Mesomelic (19%) and Acromelic (9%). Forty five out of 51 skeletal dysplasias could be classified according to Spranger's atlas, most common being mucopolysaccharidosis (MPS) (23%) followed by Achondroplasia (15%) with a mean age of presentation 70 months and 29 months respectively. Parental consanguinity was present in 23.5%, etiological diagnosis obtained in 88% and molecular confirmation in 39% of cases. All Achondroplasia & Hypochondroplasia had the common hotspot in FGFR3, two novel mutations were found among the MPS and all cases of Desbuquois dysplasia belonged to the Kim variant. With team approach using clinical and radiological details, final diagnosis can be reached in a large proportion of skeletal dysplasias. Since majority do not have a definitive treatment, focus is on the prenatal diagnosis by ultrasonography & molecular methods.

Biography

Namita Ravikumar has completed her MBBS from Bangalore Medical College and Research Institute Bangalore, India and MD in Pediatrics from Maulana Azad Medical College, New Delhi, India. She has participated in many national and international conferences and presented scientific papers. She is a life member of Indian Academy of Pediatrics and currently working as Senior Resident in Indira Gandhi Institute of Child Health, Bangalore, India.

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