

AB126. Genetic skeletal dysplasias in Thailand: twenty-five years experiences at Siriraj Hospital

Pornswan Wasant, Nithiwat Vattanawicharn, Achara Sathienkitkanchai, Somporn Liammongkolkul

Division of Medical Genetics, Department of Pediatrics, Faculty of Medicine Siriraj Hospital, Mahidol University, Bangkok, Thailand

Background: Genetic skeletal dysplasias are a heterogeneous group of disorders associated with abnormalities in the size and shape of the limbs, trunk, and/or skull that frequently result in disproportionate short stature. There are well over 100 distinct skeletal dysplasias that have been classified primarily on the basis of their clinical or radiographic characteristics.

Methods: The Genetic Skeletal Dysplasia Clinic was established at the Division of Medical Genetics, Department of Pediatrics, Faculty of Medicine Siriraj Hospital, Mahidol University in Bangkok since 1992, the only one of its kind in Thailand. Main objective of the clinic is to educate Thai pediatricians and orthopedic surgeons in training (from multicenter) about various genetic skeletal disorders; and how to care and treat pediatric patients with

skeletal dysplasias appropriately.

Results: There are numerous cases: achondroplasia, hypochondroplasia, pseudoachondroplasia, atelosteogenesis, pyknodysostosis, spondylo-epiphyseal dysplasia congenita, spondylo-meta-epiphyseal dysplasia, osteogenesis imperfecta, chondroectodermal dysplasia (Ellis-van-Creveld), cleidocranial dysostosis, thanatophoric dysplasia, rhizomelic chondrodysplasia punctata, trichorhinophalangeal syndrome, mucopolysaccharidoses, (Hurler, Hunter, Sanfilippo, Morquio, Maroteaux Lamy, Sly), osteopetrosis, campomelic dysplasia, Stickler syndrome, Kniest dysplasia, Nail-patella syndrome, diastrophic dysplasia, short-rib polydactyly syndrome, Larsen syndrome, multiple epiphyseal dysplasia, metaphyseal dysplasia and hypophosphatemic rickets.

Conclusions: The diagnoses are established at the clinical, radiological and biochemical level. However, there are limitations in confirmation at the molecular level in developing countries. Multidisciplinary approach and genetic counseling are provided.

Keywords: Skeletal dysplasias; genetic clinic; Thailand

doi: 10.21037/atm.2017.s126

Cite this abstract as: Wasant P, Vattanawicharn N, Sathienkitkanchai A, Liammongkolkul S. Genetic skeletal dysplasias in Thailand: twenty-five years experiences at Siriraj Hospital. *Ann Transl Med* 2017;5(Suppl 2):AB126. doi: 10.21037/atm.2017.s126