

Update On Skeletal Dysplasia

Tae-Joon Cho

Seoul National University, Seoul, Republic of Korea

Skeletal dysplasia is a heterogenous group of diseases caused by mutations of genes involving the metabolism of bone and cartilage tissue. Making a specific diagnosis is the starting point of care of the affected patients, however it is usually not an easy task, because of rarity of individual disease entity, subsequent limited exposure of clinicians, change or disappearance of the phenotype with age, and diversity of phenotype even with the same gene mutation. Thanks to the recent discovery of causative genes of many skeletal dysplasias and advancement of the genomics and sequencing technology, next generation sequencing is being established as a promising molecular diagnostic tool, although elaborate phenotyping remains as a critical step for diagnosis of skeletal dysplasia.

Orthopedic surgeons are the first line doctors confronting the patients with bone abnormality, are surgeons who correct limb and spine deformity, are the leader of total care for skeletal system, and can be the clinician-scientists conducting basic research on skeletal system.

Orthopaedic surgery is applied to skeletal dysplasia patients to correct and stabilize the thoracolumbar spine as well as cervical spine, to correct limb deformity, to preserve joint function as long as possible, to salvage the destroyed joints by arthroplasty, to stabilize weak limbs, and to lengthen the limbs for height. Judicious use of surgical intervention can improve general health condition of the patients, however natural course of the disease and long term effect of the surgical procedure, based upon correct and specific diagnosis of the disease, should be taken into consideration.