
Molecular Diagnosis for Skeletal Dysplasia in Korea

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Molecular diagnosis of skeletal dysplasia is challenging because skeletal dysplasia is a complex group of more than 400 conditions with extreme clinical and molecular heterogeneity. Previously, gene-by-gene approach has been applied for molecular diagnosis of skeletal dysplasia and target genes for analysis were selected based on clinical and radiological findings. Recent advances in next-generation sequencing (NGS) technologies have made disease-targeted gene panels, whole exome (WES), or genome sequencing (WGS) a realistic option for molecular diagnosis of skeletal dysplasia. In addition, the novel sequencing technologies have boosted the discovery pace of novel disease genes. Before the WES/WGS is available, a priori information on the causative genes that might underlie a genetic condition is a prerequisite for

molecular diagnostics but, theoretically, WES/WGS does not require any information on candidate genes. Although disease-targeted gene panel testing or WES/WGS is still expensive, it may be a cost-effective approach for molecular diagnosis of some genetic disorders with extensive genetic heterogeneity such as hearing impairments, muscular disorders, Charcot-Marie-Tooth disease, and skeletal dysplasia. In addition, WES/WGS may find unexpected mutations in genes known to cause different conditions from the initial diagnosis. However, there are still many hurdles that should be overcome before implementing gene panel testing or WES/WGS in clinical practice for molecular diagnostics. In this talk, I'll present recent advances in molecular diagnosis for skeletal dysplasia.

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