The First Case Report of SEMD-JL1 in China

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Background:
Spondyloepimetaphyseal dysplasia with joint laxity type 1 (SEMD-JL1) is a rare entity with a recessive inheritance. It is one of the genetic skeletal disorders (GSD) and B3GALT6 loss-of-function mutations were found in individuals with SEMD-JL1 from several families. However, there was no case described in China.

Case report:
An 8-year-old boy presented to our hospital with short stature, hyperlaxity with secondary spinal malalignment, ulnar subluxation and craniofacial alterations. The amplified DNA was captured with a disease related Gene Panel using biotinylated oligo-probes (MyGenostics GenCap Enrichment technologies). Molecular analyses did not show any other mutation but compound heterozygous variants in the B3GALT6 gene (c.694C>T and c.539_540insCCT), inherited from his parents. Then this boy was diagnosed SEMD-JL1. This is the first case report of SEMD-JL1 in China. While the c.539_540insCCT compound heterozygous mutation in B3GALT6 gene is not described before.

Conclusion:
SEMD-JL1 is caused by homozygous or compound heterozygous mutations in the B3GALT6 gene. We recommend that all the patients who have the clinical manifestations of GSD should undergo genetic analysis. This will be important for understanding the genetic laws of such diseases.

Reference