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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

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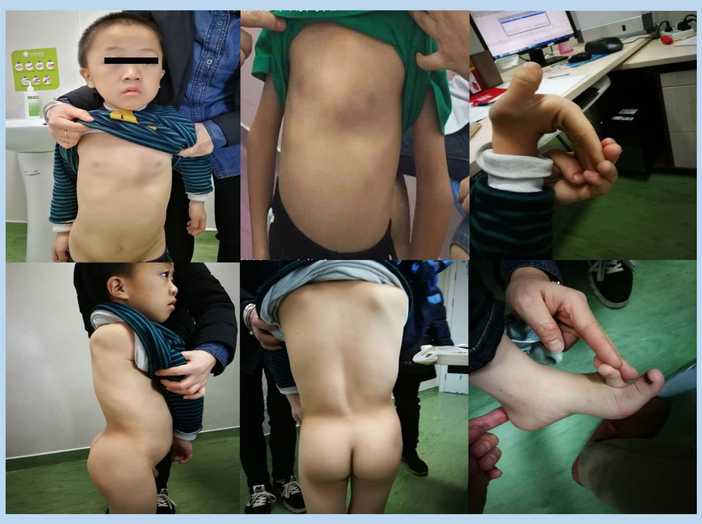

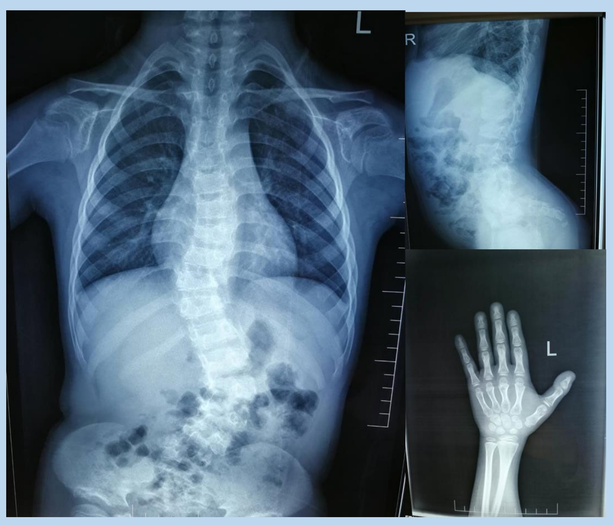

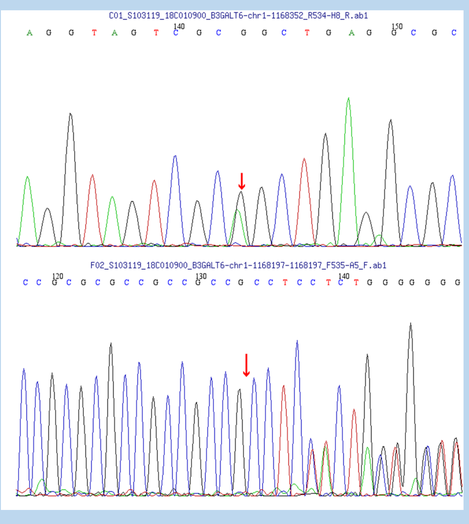

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