Schaaf-Yang syndrome: Three cases report of MAGEL2 variation and literature review

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Abstract

Objective: To highlight the clinical characteristics and early genetic diagnosis of Schaaf-Yang syndrome (SYS).

Methods: Three cases with truncating mutations in MAGEL2 were identified and related literature were reviewed.

Results: Patient 1 and Patient 2, a newborn and a two-and-a-half-month-old infant, presented typical features of SYS with multiple facial dysmorphisms, tapering digits with camptodactyly of fingers, neonatal hypotonia, feeding difficulties and respiratory distress. Besides, patient 1 passed away at 19 days. By whole-exome sequencing, de novo heterozygous c.1996dupC pathogenic variants were detected in the two individuals. Patient 3 was a 30-month-old boy who showed dysmorphic facial features, delayed language development and cognitive deficits. A heterozygous c.1640-1641delTT mutation in MAGEL2 was detected in the patient and his father. To our knowledge, the variant detected in patient 3 has not been described for other patients with SYS previously.

Conclusions: Our results indicated that testing for mutations in MAGEL2 should be considered in children with developmental delay, intellectual disability, neonatal hypotonia, feeding difficulties, joint contracture, and autism spectrum disorder. And we highly suggest that MAGEL2 gene should be added to gene-panels or gene-filters in next-generation sequencing (NGS)-based diagnostics, which is of great significance for early diagnosis and early intervention of SYS patients.

References:

Topic: Growth and syndromes