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Wiedemann-Steiner Syndrome Caused by Novel Mutation of KMT2A Gene: A Report of Two Cases

Pingping Zhang, Jie Fang, Dongmei Gan, Yueying Feng, Yudan Zhao and Shuxia Ding

Department of Pediatric Endocrinology, Women and Children's Hospital Affiliated to Ningbo University, 339 Liuting Street, Haishu District Ningbo, Zhejiang 315012, China

KEYWORDS Growth Deficiency and Mental Retardation with Facial Dysmorphism. KMT2A. Hirsutism. Precocious Puberty. Molecular Epidemiology. Mutagenesis. Exome Sequencing

ABSTRACT Wiedemann-Steiner Syndrome (WDSTS, OMIM 605130) is a rare autosomal dominant disorder. This case report describes two related cases and explores new mutation sites, aiming to provide new reference cases for clinical use. This report presents two female pediatric cases characterised by hirsutism, facial deformities, and intellectual disabilities, investigated via molecular genetic analysis. Patient 1 showed precocious puberty, hirsutism, facial deformities, congenital heart disease, astigmatism, pilonidal sinus, growth retardation, and intellectual disability. Patient 2 exhibited severe growth and developmental delays, astigmatism, facial deformities, and hirsutism. Whole-exome sequencing identified a KMT2A:c.666delA (p.Gly223Glufs*9) variant in Patient 1 and KMT2A:c.5664+1G>C in Patient 2. Patient 1 received gonadotropin-releasing hormone analogs for puberty control and growth hormone therapy, resulting in improved growth and metabolic rates. These findings expand the KMT2A mutation spectrum and highlight the value of genetic testing for children with growth retardation, hirsutism, and intellectual disabilities.