Mild autistic spectrum disorder in a 33 year-old male Japanese patient with Temple syndrome

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Nothing to disclose

Introduction

Temple syndrome (TS14) caused by maternal uniparental disomy chromosome 14 (UPD(14)mat), paternal deletions and the imprinting defect affecting the 14q32.2 imprinted region is associated with non-specific symptoms such as growth failure, precocious puberty, obesity, and diabetes mellitus (DM).1,2 Some TS14 cases are misdiagnosed as having Prader–Willi syndrome (PWS).1 In TS14, patient’s intelligence quotient (IQ) is usually normal, and autism spectrum disorder (ASD) is a rare comorbidity.3

Case

A male patient was born at 39 weeks and was not small for his gestational age. He was clinically diagnosed with PWS owing to hypotonia during infancy. After infancy, he received no regular follow up. He exhibited precocious puberty, transient obesity, and DM. His final height was within normal limits at 159 cm (-2.0 SD). At 33 years of age, he visited our hospital to receive a genetic diagnosis and social welfare. He showed the normal methylation levels of the SNRPN-DMR on chromosome 15 and hypomethylation of the IG-DMR and MEG3-DMR at the 14q32.2 imprinted region without UPD(14)mat and maternal microdeletion involving the 14q32.2 imprinted region, and was diagnosed with TS14. At age 33, his total IQ was 97; verbal IQ was 104, and performance IQ was 88 (Wechsler Adult Intelligence Scale-III). Although his scores of ASD assessment scales (Pervasive Developmental Disorders ASD Rating Scale-Text Revision and Autism Spectrum Quotient) were low, we clinically diagnosed with ASD, with both verbal and nonverbal communication impairments.

B. Microsatellite analysis for chromosome 14

Left: summary of microsatellite results.
Right: representative microsatellite results.
N.I.; not informative

C. CGH+single nucleotide polymorphism (SNP) microarray profiles for chromosome 14.

We performed a single nucleotide polymorphism (SNP) array analysis the SurePrint G3 ISCA CGH+SNP Microarray Kit (catalog number G4890A, Agilent Technologies).

Conclusion

We report a patient who was diagnosed as TS14 at the age of 33, with post-natal growth failure, obesity and DM. Comorbidity of ASD was also diagnosed, which is the second case reported. This case also highlights the importance of genetic analysis for differentiating TS14 from PWS. Hypotonic infants with unknown etiology should be considered for genetic analysis of TS14. Additionally, long-term follow up is needed, not only to observe precocious puberty and DM, but also to identify problems associated with developmental disorders, such as ASD.

References


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