A Rare Genetic Disorder: Partial Trisomy on Chromosome 21

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A 7-year and 7-month-old boy presented with pubic hair. He was born at the 35th gestational week with a birth weight of 2400 g. He was diagnosed with Down syndrome (DS) in another center. Despite normal motor and mental development, he was under a special education in school since he was labeled with DS. Physical examination revealed dysmorphic findings, such as up-slanting palpebral fissures, epicanthus, low-set ears, and low nasal bridge, all suggesting DS. The height was 136 cm [>97th percentile, >2.7 standard deviation score (SDS)] and the weight was 31.6 kg (between 90 and 97th percentile, 2.7 SDS). Axillary hair was absent, while pubic hair appeared as stage 2 according to Tanner staging. Left testis was 2 mL and right testis was 1 mL. Complete blood count was normal, electrolytes were in normal ranges, 17-hydroxy progesterone (17-OHP) was 0.64 mg/mL, dehydroepiandrosterone sulfate was 191 µg/dL, and early morning cortisol level was 9.8 µg/dL. The bone age was 10 years. Echocardiography was normal. The patient was diagnosed with premature adrenarche. Despite the presence of typical appearance of Down syndrome, the patient had a significant tall stature. Repeated karyotype analyses revealed 47,XY,+2 (q22.11-qter), while Array CGH detected an increase of 10578411 base between 36103774 and 46682184 bases of the q22.11 and q22.3 bands of chromosome 21. Gene and gene regions which constitute the phenotypic features of DS are known to be located on the distal parts of the long arm of chromosome 21. The detected duplication in our patient was located on the distal part of the long arm of chromosome 21. This might be the reason of dysmorphic facial features. This case of partial trisomy of chromosome 21 was reported because of its scarcity.

Key words: Partial trisomy, chromosome 21, Down syndrome, phenotype

A Case of Type 1 Diabetes Mellitus with Klinefelter's Syndrome

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Type 1 diabetes mellitus (T1DM) is a model of chronic autoimmune disease beginning with genetic susceptibility in affected individuals and progressing to autoimmune destruction of β cells, precipitated by environmental insult. Many of the patients with T1DM have associated genetic disorders such as Klinefelter's syndrome (KS), Turner's syndrome, Down's syndrome, and Noonan's syndrome. This paper reports a case of 15-year-old boy diagnosed with T1DM accompanying KS, 47,XXY karyotype. A 14-year-old adolescent was admitted to The Hospital of Başkent University Faculty of Medicine for weight loss, polyuria, and polydipsia. He presented for evaluation of diabetic ketoacidosis. His fasting blood glucose (301 mg/dL) was markedly elevated, pH (7.32) and HCO_3 (10 mEq/L) levels were slightly low, and serum ketone test was positive due to ketoacidosis. The patient was born to a 27-year-old mother and was the first child of non-consanguineous parents. His sister was healthy, without remarkable medical history. There was no family history of diabetes or chromosomal abnormality, but his father and uncle suffered from rheumatoid arthritis. There are many studies that denote a dramatically increased risk of T2DM in KS. There are a few reports on KS accompanying T1DM. The present patient with T1DM was diagnosed as KS during his follow-up. Although he was at puberty on onset, on consecutive examination, we observed delay in puberty, such as lack of both secondary sexual characters and testicular volume increase. For differential diagnosis, chromosome analysis was performed. The result was 47,XXY karyotype, thus the diagnosis of KS was established. In conclusion, based on the findings presented in recent reports, KS should be considered in male patients with delayed puberty especially in the ones diagnosed with T1DM.

Key words: Type 1 diabetes mellitus, 47,XXY, diabetic ketoacidosis, Klinefelter's syndrome, puberty