

A Rare Chromosomal Disorder; Trisomy 4p

Merve Nur Hepokur¹, Zeynep Uzan Tatlı¹, Gül Direk¹, Leyla Akın¹, Nihal Hatipoğlu¹, Mustafa Kendirci¹,
Selim Kurtoğlu²

1. Erciyes University Faculty of Medicine, Department of Pediatric Endocrinology, Kayseri / Turkey
2. Memorial Hospital, Department of Pediatric Endocrinology and Neonatology, Kayseri / Turkey

INTRODUCTION

Duplication of 4p16.3 is a rarely described chromosomal disease, also known as trisomy 4p syndrome in the literature. Here we present a patient who admitted to our clinic with complaints of growth in hands and feet with delayed speech and mental retardation. We wanted to emphasize that overgrowth of hands and feet can rarely be a feature of 4p duplication syndrome.

CASE

A 15-year-old male patient admitted to our clinic with complaints of growth in hands and feet which have been noticed for the last 2 to 3 years. He had also delayed speech and psychiatric analyses was consistent with attention deficit hyperactivity disorder since he was 4 years old. Based on the history his parents were third degree relatives and he has a cousin who was followed up with the diagnosis of autism. On physical examination, his body weight was 76.9 kg (75-90p), height was 163.4 cm (10p) and head circumference was 57 cm (75p). He had frontal bossing, short neck, hypertelorism, broad nasal bridge, micrognathia, adenoid face and bilateral clinodactyly of fifth finger. His hands and feet were larger than normal. Difference between overarm and height measurements were above +2 SD, as like the ratio of upper and lower segments was + 2 SD. Other physical findings were normal.

In laboratory findings; basal growth hormone and insulin-like-growth factor-1 levels were within normal limits. Growth hormone suppression was provided with oral glucose tolerance test. Insulin resistance was not detected in clinical and laboratory findings. The IQ test was delayed moderately. The patient's pituitary MRI and visual field examination were also normal. He had no seizures but EEG findings were nonspecific. A genetic review revealed a duplication of 4p16.3p16.1.



CONCLUSION

Trisomy 4p is a very rare chromosomal disorder. As in this patient, facial dysmorphism, delayed speech and musculoskeletal abnormalities like clinodactyly may present in Trisomy 4p syndrome. In literature overgrowth of hand and feet have been reported in few cases, but to the best of our knowledge disproportionate growth has not been previously reported, as in the presented case

