Etiologies of Short Stature in Pediatric Endocrine Clinic in Northwest Region (Trakya) of Turkey

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Background: Short stature (SS) is one of the common disorders referred for investigation of an endocrine disorder. The etiologies of SS vary and are commonly grouped into pathological and nonpathological disorders. Despite standard clinical and laboratory evaluation, a pathological diagnosis is not reached in 50-90% cases.

Aim: The aim of this study was to determine the etiologies and describe the characteristic of short stature patients who were followed up by Pediatric Endocrinology Clinic and to compare factors between normal variant short stature (NVSS) and growth hormone deficiency (GHD).

Methods: The clinical and laboratory data of a total of 309 patients who were diagnosed as having SS during a six year period (2011-2017) were retrospectively analyzed based on the hospital records. Short stature was defined as height below – 2 standart deviation score (SDS) by gender and age based on population data of Turkish children.

Results:
• There were 169 (54.7%) boys and 140 girls (45.3%) with a mean age of 10.8±3.1 years.
• The proportion of boys to girls was 1.2:1.
• Of 309 patients, 65.1% (201 cases; 116M, 85F) were NVSS; that is familial short stature (16.2%), constitutional delay of growth and puberty (CDGP) (29.8%) or combination of both (19.1%).
• Pathological short stature was found in 34.9% of the cases, of which 4.5% disproportional short stature (28.5% of them achondroplasia).
• Endocrinological causes accounted for 18% of short stature. GHD was found in 17.4% of total cases and it comprises 96% among endocrinological causes.
• The common etiologies in severe SS (height SDS < -2.5) were CDGP (14.2%) and CDGP-familial SS (13.2%).
• In the moderate SS group (height SDS between -2 and -2.5), CDGP was the most common etiology (15.5%).
• Among the syndromic SS, Turner syndrome was the most common (3.6%) followed by Noonan syndrome (1%). Celiac disease was found in 1% of total cases.

Conclusions: In this study, NVSS was the most common etiology of short stature, followed by growth hormone deficiency and syndromes in accordance with literature. NVSS was also the common cause of the severe SS.