

gene mutations (4 missense: p.T93M, p.R352W, p.Y432C, p.E448K; 2 nonsense: p.W151X, p.Q259X; and one splice site: c.831 + 1G > C) were detected. p.T93M was the most frequent (57% of all alleles) mutation. Two of the seven mutations (p.Q259X, c.831 + 1G > C) were defined for the first time in this study.

This study defines the mutation spectrum and genotype phenotype correlation of *DHCR7* gene within the Turkish SLOS patients. As seen in other Mediterranean populations, p.T93M mutation was the most frequent mutation observed in our patients.

(P-55)

Anthropometric Measurements and Complications of Achondroplasia Patients

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Achondroplasia is the most common reason of inherited disproportionate short stature. It is caused by mutations in the *FGFR3* (fibroblast growth factor receptor-3) gene. In this study, we aimed to evaluate the anthropometric measurements and complications in achondroplasia patients.

In this study, using Sanger sequencing or next-generation sequence analysis, *FGFR3* gene mutations were detected in 29 patients (achondroplasia/hypochondroplasia: 15/14) between 2012 and 2016. Nine of the 15 achondroplasia patients and one of the 14 hypochondroplasia patients had been followed by both Pediatric Genetics and Pediatric Endocrinology Subdivisions of Ege University Medical Faculty. Fifty percent of patients were female and 50% were male. Median age was 27 months (min 9-max 96 months). Anthropometric measurements of the patients were found to be in normal ranges using growth curves specific for achondroplasia patients reported by Horton et al. When evaluated according to normal growth curves, mean standard deviations of height, weight, and head circumference were $-4.45 (\pm 1.59)$, $-1.38 (\pm 1.15)$, and $1.99 (\pm 1.20)$, respectively. In one patient, foramen magnum stenosis and cervicospinal junction compression were observed. Another patient had mental retardation and epilepsy.

It has been considered that growth of achondroplasia patients can be maintained in the limits of achondroplasia patients when appropriate follow-up is performed. They should be carefully evaluated for neurological and orthopedic complications.

(P-56)

Mutation Spectrum of *GCK*, *HNF1A*, and *HNF1B* in MODY Patients and 40 Novel Mutations

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MODY (maturity onset diabetes of the young) is a monogenic diabetes mellitus caused by pancreatic beta cell dysfunction. It has been classified into 9 groups according to the underlying molecular etiology. Mutations in the genes encoding the nuclear transcription factor 1 homeobox A (*HNF1A*) and the enzyme glucokinase (*GCK*) are the most common causes of MODY. Additionally, *HNF1B* gene is responsible for 5% of the disease. The aim of this study was to investigate the mutation spectrum of *GCK*, *HNF1A*, and *HNF1B* genes in MODY patients.

Molecular test results of 152 patients carrying mutations in *GCK*, *HNF1A*, or *HNF1B* genes were evaluated. Rate of mutations detected in *GCK*, *HNF1A*, and *HNF1B* genes were 84%, 13%, and 3%, respectively. Fifty-seven different mutations (40 missense, 8 nonsense, 7 frameshift, 1 in-frame deletion, and one splice site) in *GCK*, 15 different mutations (11 missense, 3 frameshift, and one 3' UTR) in *HNF1A* and 4 different mutations (2 missense, one frameshift, and one indel) in *HNF1B* were found. Thirty-three, 5, and 2 mutations were detected as novel mutations in *GCK*, *HNF1A*, and *HNF1B* genes, respectively.

Definition of molecular etiology in MODY patients is important for giving appropriate genetic counseling and disease management. The most commonly affected gene has been found to be *GCK* gene among the MODY patients studied. In the genes *GCK*, *HNF1A*, and *HNF1B*, 40 mutations have been defined for the first time in this study.

(P-57)

MEN 2A Family

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MEN 2 is a rare genetic disorder with autosomal dominant inheritance. Here, we present a family in which MEN2A was detected in the index case and two brothers had detected pheochromocytoma and medullary thyroid cancer.

MEN 2A index case;

Index case: A thirty-six-year-old male patient presented with headache, sweating, and palpitations. Urine catecholamines