THE PREVALENCE OF MELANOCORTIN-4 RECEPTOR GENE MUTATIONS IN TURKISH OBESE CHILDREN AND ADOLESCENTS

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BACKGROUND

•Melanocortin-4 receptor (MC4R) mutations are the most common known cause of monogenic obesity (1).

•Prevalence of *MC4R* mutations in children with severe obesity varies from 0.3% up to 6.3% (2).

•>150 different mutations have been reported (1).

OBJECTIVE

•To establish the prevalence of *MC4R* mutations in a group of Turkish obese children and adolescents with morbid obesity.

METHODS and SUBJECTS

MC4R gene was sequenced in **47** morbid non-syndromic obese children and adolescents (28 girls and 19 boys) aged **1-18 years**. Body weight, height and Body mass index (BMI), weight z-score, height z-scores and BMI z-scores were recorded using Turkish national anthropometric references (3).

Cases were included if BMI was **≥120 percent of the 95th percentile** values or **≥35 kg/m²** (whichever is lower). This corresponds to approximately the **99th percentile or BMI Z-score ≥2.33**

RESULTS

•Mean age was **13.2±4.1 years**, mean age at onset of obesity 5.1±2.1 years, mean height SD score1.21±0.93, mean BMI 40.0±8.8 and **BMI SD score 2.72±0.37.**

•In four cases **(8.5%)**, we detected three mutations one of which was **novel (c.870delG)** (Table 1).

•In addition, screening of family members revealed six more cases (one child, five adults) with a *MC4R* mutation.

Case	Genotype		Age (years)	Gender	BMI SDS	Height SDS	Age at onset of obesity (years)
1	c.496 G>A	Heterozygous	16	F	2.47	1,36	4
2	c.496 G>A	Heterozygous	8	M	3.05	3,4	3
3	c.870delG	Heterozygous	6	M	3.01	1.94	2
4	c.346_347delAG	Homozygous	10	F	3.07	1.06	1

Table 1. ***Genotypes and phenotype characteristics of mutation carriers***

- No differences were present regarding the anthropometric (BMI, height, and weight SD scores) and biochemical (fasting blood glucose, lipids and fasting blood insulin levels) between mutation carriers and noncarriers.

CONCLUSIONS

There is no published study regarding *MC4R* mutations in Turkish children and adolescents with morbid obesity. In the present study, prevalence of *MC4R* mutations was found to be 8.5%. We speculate that MC4R gene mutations are an important cause of morbid obesity with early onset in the Turkish children and adolescents as well.

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55ESPE 493--P2

Fat metabolism and Obesity

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DOI: 10.3252/pso.eu.55ESPE.2016

Poster presented at: ESPE 2016 PARIS