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Abstracts

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PCSK1 Heterozygous Gene Polymorphisms are Associated with Early Onset Morbid Childhood Class III Obesity Across Diverse Ethnic Groups

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Aim of the Study: Correlate genetic data of patients heterozygous of PCSK1 gene variations with the clinical phenotype.

Introduction: Heterozygous variants of the PCSK1 gene have been described in cases of early onset of morbid obesity in childhood. This gene encodes prohormone convertase 1/3 enzyme, a serine endoprotease expressed in neuroendocrine cells that converts inactive prohormones into functional hormones important in regulation of energy metabolism.

Methods: 293 cases of morbid obesity underwent genetic testing at Prevention Genetics after obtaining consent.

Results: All cases presented with early weight gain from 3-5 years of age, hyperphagia, acanthosis nigricans. 35 (12%) cases had heterozygous PCSK1 gene variations. 22 were females and 13 males: 15 Caucasian, 9 Hispanic, 6 Asian, and 5 African American. The age at diagnosis was 16.7 ± 9.4 (from 3 to 42 yrs.), BMI was extremely high 40.7 ± 4.6 kg/m².

24 cases of 35 (68.5%) carried the heterozygous polymorphism c.661A>G, which is predicted to result in an amino acid substitution p.Asn221Asp (rs6232). In other cases different PCSK1 variants were found: 2 cases c.818_820del, p.Asp273del; 2 cases c.1381G>A, p.Val461Met; 2 cases c.1918A>G, p.Thr640Ala; 2 cases c.28T>G, p.Cys10Gly; 1 case c.2099_2101del, p.Phe700del; 1 case c.1387G>A, p.Glu463Lys; 1 case c.760A>G, p.Ile254Val.

11 cases were only heterozygous for one PCSK1 gene variant, 14 cases in a combination of PCSK1 and one additional gene, 3 cases in combination with 2 additional genes, 5 cases in combination with 3 additional genes, 1 case each of 4 and 5 additional genes.

There were no cases of diabetes type 2. Intellectual development was normal in all patients, no evidence of autism, learning disability or mental retardation.

Only 2 patients had fatty liver, 2 had hypertension and another 2 had borderline elevated HbA1c of 5.9 and 6.2%. As a group the average Insulin was 24.91 ± 16.82 mIU/mL; TG of 133.3 ± 53.6 mg/dl, HDL of 43.5 ± 7.6 mg/dl. They had low-normal Leptin levels of 25.73 ± 14.5 ng/mL.

Conclusion: Genetic screening of early onset of obesity can identify patients with PCSK1 heterozygous gene polymorphisms. PCSK1 rs6232 is a frequent gene polymorphism associated with morbid obesity. Insulin resistance was not severe as expected for the degree of morbid obesity and leptin levels were relatively low. This information can help in the understanding of the natural history of PCSK1 gene carriers in diverse ethnic groups.

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Dietary and physical activity habits in children and adolescents in Greece

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Introduction: Obesity in childhood and adolescence is a major problem with many adverse consequences in public health due to its increasing prevalence, as well as the burden on the health system. Recently, the need to find effective intervention and prevention strategies for the management of obesity has led to the development of e-health technologies, which record behavioral data objectively and correlate them with factors that increase body mass index (BMI).

Objective: To determine the dietary and exercise habits of children and adolescents at the beginning of their participation in the study 'BigO: Big Data against Childhood Obesity' (<http://bigoprogram.eu>, Horizon2020, No. 727688).

Methods: Nine hundred (n=900) children and adolescents aged 8-18 years who attended the Out-patient Clinic for the Prevention and Management of Overweight and Obesity in Childhood and Adolescence participated in this cross-sectional study. Medical history and anthropometric measurements were obtained by a single trained observer and participants were clinically evaluated by a multidisciplinary management team. The data collection system included the BigO technology platform, which interfaces with a Smartphone and Smartwatch, and records data on diet, sleep and exercise objectively for each patient. Children's caregivers were asked to complete the self-administered food frequency questionnaire for children "ToyBox" and a physical activity questionnaire. The statistical analysis was carried out by the Statistical Package for Social Sciences (SPSS) software program.

Results: The study sample consisted of 900 children and adolescents of which 73.1% were obese, 24.7% overweight and 2.2% had normal BMI. A higher number of boys had obesity (76.3%/69.7%, p-value:0.031), while a higher number of girls were overweight (28.5%/21.1%, p-value:0.031). According to the descriptive data, the consumption of cereal without sugar and no added sugar was higher in all BMI categories than those with addition of sugar (p-value:0.025), although children with obesity tended to consume higher proportions of cereal than those with overweight and normal BMI (p-value:0.041). In all BMI categories, participants had large proportions of meat and poultry (p-value:0.031) while the consumption of potatoes was more frequent in obese than overweight children (p-value:0.029). Boys, consumed larger amount of water, light beverages, vegetables, meat, fried potatoes and chocolate spread than girls (p-value:<0.05). In both sexes, participants who never watched television during meals were more than those who watched (p-value:<0.05).