European Respiratory Society
Annual Congress 2013

Abstract Number: 1852

Publication Number: 3310

Abstract Group: 4.2. Sleep and Control of Breathing

Keyword 1: Apnoea / Hypopnea **Keyword 2:** Genetics **Keyword 3:** Children

Title: Trp64Arg polymorphism in children with obstructive sleep apnea (OSAS) and controls

Dr. Afroditi 14055 Sakellaropoulou afroditisak@gmail.com MD [1], Mrs. Styliani 14056 Pappa s_pappa@hotmail.com [1], Dr. Maria 14057 Papageorgiou m.papageorgiou@hotmail.gr MD [1], Prof. Dr Katerina 14058 Haidopoulou haidoka@med.auth.gr MD [2], Prof. Dr Maria 14059 Emporiadou eboriad@med.auth.gr MD [2], Dr. Evaggelia 14066 Dosiadi edosiadi@yahoo.gr MD [1] and Prof. Dr Maria 14075 Hatzistilianou nontas@topo.auth.gr MD [1]. [1] 2nd Department of Paediatrics, Medical School, Aristotle University of Thessaloniki, Thessaloniki, Greece, 54636 and [2] 4th Department of Paediatrics, Medical School, Aristotle University of Thessaloniki, Thessaloniki, Greece, 56429 .

Body: The b3-adrenergic receptor (ADRB3) plays a role in the regulation of metabolism. A point mutation at codon 64 of ADRB3 (Trp64Arg) is associated with obesity phenotypes. Its frequency varies considerably between the populations. Conflicting results have been reported about its role. Our study aimed to evaluate the clinical and polysomnographic characteristics and the incidence of Trp64Arg polymorphism in a group of children with OSAS and controls. The study group consisted of 42 children (2.5-14.5 years, 64.3% boys): 33.3% mild, 38.1% moderate and 28.6% severe OSAS. The control group comprised of 77 children with no history of OSA (59.74% girls). Heterozygous for the polymorphism Trp64Arg were 6 children with OSAS (14.3%) and 3/77 (3.89%) controls. The frequencies of genotypes and alleles of the polymorphism of the Trp64Arg in both groups were in equilibrium (p=1.00, χ^2=0.00). Most of the patients with OSAS, carriers of the polymorphism Trp64Arg, were males (p = 0,049). No differences exist concerning the control group (p>0.05). Patients with OSAS carrying the mutation compared with other patients with sleep apnea had higher: a) BMI z score (p= 0.215), b) Waist-Hip-Ratio (WHR), (p= 0.429), c) Waist to Hip Ratio (WHtR), (p= 0.228) and d) Conicity index (p= 0.172). No correlation between the presence of the Trp64Arg polymorphism and the lipid profile was recorded in both groups (p>0.05). The presence of the Trp64Arg polymorphism does not affect the characteristics of the sleep study in patients with sleep apnea (p> 0.05). The relationship between Trp64Arg polymorphism, sleep apnea and obesity is still unclear. Probably this polymorphism may be a marker for personalized weight loss in obese subjects.