

## The Possible Role of Mitochondrial Uncoupling Protein 2 (*UCP2*) Gene on the Development of Metabolic Syndrome and Platelet Count in Obese Children/Adolescents - A Preliminary Study

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**Introduction:** The aim of this study was to determine the potential relationship between polymorphisms of the uncoupling protein 2 (*UCP2*) gene and metabolic syndrome (MS) and platelet count in obese children/adolescents.

**Method:** One-hundred unrelated obese children and adolescents (57 of them with MS) were selected. MS was defined according to the "National Cholesterol Education Program" criteria. The -866G >A and 45 bp insertion/deletion (I/D) polymorphisms of the human *UCP2* gene were genotyped using the polymerase chain reaction-restriction fragment length polymorphism method. A p-value <0.05 was considered statistically significant.

**Results:** The mean age of 100 obese patients (55 male/45 female) was 11.32±3.82 years (range 3-17 years). For the -866 G>A polymorphism, the distributions of G/G, G/A, and A/A genotypes were respectively 22.8%, 49.1%, and 28.1% in patients with MS compared to 9.3%, 60.5%, and 30.2% in patients without MS (p>0.05). The allele frequencies of G and A were 47.4% and 52.6% in patients with MS and 39.5% and 60.5% in patients without MS (p>0.05). The distributions of D/D, D/I, and I/I genotypes for the I/D polymorphism were 52.6%, 24.6%, and 22.8% in patients with MS compared to 37.2%, 39.5%, and 23.3% in patients without MS (p>0.05). The allele frequencies of D and I were 64.9% and 35.1% in patients with MS and 57.0% and 43.0% in patients without MS (p>0.05). Platelet count was found significantly higher in patients with MS carrying I allele (p=0.004).

**Conclusion:** This study could not verify the potential role of *UCP2* gene polymorphisms on the development of MS in childhood obesity. However, I allele of *UCP2* gene may have a possible role, as a risk factor, in the development of atherothrombosis in obese children with MS. Since this is a preliminary study, further investigations with larger populations are needed to confirm the exact role(s) of this gene in obese children.

**Key words:** Childhood, obesity, metabolic syndrome, atherothrombosis, polymorphism

## Wolfram (DIDMOAD) Syndrome - Report of Two Siblings

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**Introduction:** Wolfram syndrome (WS), also known as DIDMOAD is a rare autosomal recessive syndrome characterised by juvenile onset of diabetes mellitus, optic nerve atrophy, diabetes insipidus, sensorineural deafness, renal tract and neurological abnormalities. WS is caused mainly by biallelic mutations in the *WFS1* gene, which encodes wolframin. Wide tissue distribution of wolframin and many mutations in the wolframin gene resulting in Wolfram syndrome may contribute to different phenotypes and the unusual combinations of clinical features.

Mostly the first presentation might be type 1 diabetes. A detailed analysis of the patient's medical history and a review of the literature suggest that many cases of WS may remain undiagnosed due to misdiagnosis as type 1 DM. Herein, we reported two siblings with WS who diagnosed as type 1 diabetes firstly.

**Case:** The first case is 16 year-old boy who diagnosed type 1 DM when 3.5 years old, During follow-up, growth hormone deficiency and cataract developed when he was 13 years old. After that, in two years, optic atrophy and sensorineural (SN) hearing loss and diabetes insipidus were diagnosed.

The patient's 7-year-old sister; diagnosed type 1 diabetes mellitus in the age of 4.5 years and optic atrophy was recognized 6 months later. Both patients underwent genetic analysis and IVS4 + 1G> A homozygous mutation was detected.

In conclusion, in type 1 diabetic children, accompanying symptoms such as cataract, short stature, hearing problems and polyuria in normoglycemic state must alert the clinician for WS.

**Key words:** Wolfram syndrome, type 1 diabetes, cataracts, short stature, deafness