**Correlation between the polymorphism thr92ala of the deiodinase type 2 enzyme and adaptive behavior in Down syndrome**

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**Introduction:** The present study aimed to identify a possible correlation between the presence of the Thr92Ala mutation of the enzyme deiodinase type 2 and adaptive behavior in children with Down syndrome (DS). Approximately 12 to 36% of the population presents homozygous for this polymorphism and positive correlations have already been found between mutations in D2 with intellectual deficiency, bipolarity and psychosis. It has recently been shown that this mutation leads to modifications in the cellular transcriptome in human brains that appear to relate to changes in pathways involved in the modulation of cognitive development and in Parkinson's disease, Alzheimer's and schizophrenia. DS is the most common autosomal aneuploidy, affecting on average 1/660 newborns and represents about 18% of the total number of mentally handicapped in specialized institutions. In addition to cognitive deficits, neuropathology, such as Alzheimer's, is common in individuals with DS [1]. Based on the data described, we asked whether the Thr92Ala-D2 mutation could contribute to changes in the adaptive behavior of individuals with DS.

**Methods:** To answer this question, we screened the presence of the Thr92Ala-D2 polymorphism in 29 children with DS, aged between 2 months and 18 years, and their adaptive behavior through the Vineland Adaptive Behavior Questionnaire. The genotype was determined by real-time PCR technique from the DNA obtained through collection of buccal epithelial cells by Swab.

**Results:** We found that the frequency in our population was 20.69% wild (TT), 55.17% heterozygous (AT) and 24.14% polymorphic (AA), a similar result to those found in other studies. The presence of the mutated allele in AA homozygosis when compared to the wild TT results in a significant worsening in low adaptive level frequency in the subdomains Domestic (83.33% AA and 14.28% TT), Community (100% AA and 42 , 85% TT), Interpersonal Relations (83.33% AA and 28.57% TT) and Social Rules (66.66% AA and 28.57% TT).

**Discussion:** The worsening in the adaptive behavior in the polymorphic DS children may be due to the altered expression of genes involved in the processing of beta-amyloid, in neuronal apoptosis, EGFR pathway, oxidative phosphorylation, and mitochondrial dysfunction. These pathways were altered in in the brain of typical patients with the polymorphism (AA) [2]. Together, these changes may interfere with cognitive functions explaining the differences observed between the DS with the polymorphism and DS without the polymorphism.

**Conclusion:** The present work confirmed that the frequency of the Thr92-Ala polymorphism in individuals with DS is similar to the population. More importantly, we found that the presence of the polymorphism impairs the functionality of these individuals.

**References:**[1] Van Duijn, G et al. Journal of Intellectual Disability Research 54 (11): 943–954, 2010.[2]Mcaninch, EA et al*.* The Journal of Clinical Endocrinology & Metabolism100: 920-933, 2015