UPB1

The UPB1 gene provides instructions for making an enzyme called Beta-Ureidopropionase. This enzyme is involved in the breakdown of molecules called pyrimidines, which are building blocks of DNA and its chemical cousin RNA.

The beta-ureidopropionase enzyme is involved in the last step of the process that breaks down pyrimidines. This step converts N-carbamyl-beta-aminoisobutyric acid to beta-aminoisobutyric acid and also breaks down N-carbamyl-beta-alanine to beta-alanine, ammonia, and carbon dioxide. Both beta-aminoisobutyric acid and beta-alanine are thought to play roles in the nervous system. Beta-aminoisobutyric acid increases the production of a protein called leptin, which has been found to help protect brain cells from damage caused by toxins, inflammation, and other factors. Research suggests that beta-alanine is involved in sending signals between nerve cells (synaptic transmission) and in controlling the level of a chemical messenger (neurotransmitter) called dopamine.

Beta-Ureidopropionase (βUP) deficiency is an autosomal recessive disease caused by abnormal changes in the pyrimidine-degradation pathway. A study of Fang et al., from Tianjin Children's Hospital aimed to investigate the mutation of β-ureidopropionase gene (UPB1) gene and clinical features of 7 Chinese patients with βUP deficiency.

They reported 7 Chinese patients with βUP deficiency who were admitted at Tianjin Children's Hospital. Urine metabolomics was detected by gas chromatography-mass spectrometry (GC-MS). Then genetic testing of UPB1 was conducted by polymerase chain reaction (PCR) method.The patients presented with developmental delay, seizures, autism, abnormal magnetic resonance imaging, and significantly elevated levels of N-carbamyl-β-alanine and N-carbamyl-β-aminoisobutyric acid in urine. Subsequent analysis of UPB1 mutation revealed 2 novel missense mutations (c.851G>T and c.853G>A), 3 previously reported mutations including 2 missense mutations (c.977G>A and c.91G>A) and 1 splice site mutation (c.917-1 G>A).The results suggested that the UPB1 mutation may contribute to βUP deficiency. The c.977G>A is the most common mutation in Chinese population. [1)]

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Fang Y, Cai C, Wang C, Sun B, Zhang X, Fan W, Hu W, Meng Y, Lin S, Zhang C, Zhang Y, Shu J. Clinical and genetic analysis of 7 Chinese patients with β-ureidopropionase deficiency. Medicine (Baltimore). 2019 Jan;98(1):e14021. doi: 10.1097/MD.0000000000014021. PubMed PMID: 30608453.