Beta-ureidopropionase deficiency

Beta-Ureidopropionase (βUP) deficiency is an autosomal recessive disease caused by abnormal changes in the pyrimidine-degradation pathway that causes excessive amounts of molecules called N-carbamyl-beta-aminoisobutyric acid and N-carbamyl-beta-alanine to be released in the urine. Neurological problems ranging from mild to severe also occur in some affected individuals.

People with a beta-ureidopropionase deficiency can have low muscle tone (hypotonia), seizures, speech difficulties, developmental delay, intellectual disability, and autistic behaviors that affect communication and social interaction. Some people with this condition have an abnormally small head size (microcephaly); they may also have brain abnormalities that can be seen with medical imaging. Deterioration of the optic nerve, which carries visual information from the eyes to the brain, can lead to vision loss in this condition.

In some people with beta-ureidopropionase deficiency, the disease causes no neurological problems and can only be diagnosed by laboratory testing.

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.