Acatalasia-the catalase deficiency disease.

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Description

Catalase deficiency condition, commonly known as acatalasia, is a congenital disorder caused by abnormalities in the CAT gene. A severe deficit of an enzyme called catalase characterizes the disorder. Acatalasemia is a metabolic disorder (inborn metabolic error) characterised by reduced catalase activity in erythrocytes. Catalase is an antioxidant enzyme that breaks down hydrogen peroxide into oxygen and water. Infants with this condition are unable to breakdown endogenous or exogenous hydrogen peroxide, which builds up and causes oxidative stress. The soft tissues of the mouth and nasal mucosa are particularly vulnerable, resulting in ulceration, necrosis, and gangrene in severe cases.

Red blood cells, mucosa, skin, muscles, bone marrow, appendix, and liver tissue all contain catalase, which is a very effective enzyme. It converts hydrogen peroxide to water and oxygen gas. This procedure shields tissues from reactive oxygen species like peroxide produced by microorganisms (eg, *streptococci, pneumococci,* oral flora). Affected individuals are unable to breakdown exogenous or endogenous hydrogen peroxide due to the absence or substantial lack of catalase. Gingival hypoxemia, ulcerations, and necrosis of soft and hard tissues result from the buildup of these metabolites in periodontal tissues. The symptomatic form (Takahara illness) is the Japanese variation, in which the pathology is limited to the oral cavity. Because of advancements in oral hygiene, these consequences are rarely encountered in more recent cases of acatalasemia.

Type 2 diabetes is the most common kind of diabetes, and those with acatalasemia are more likely to get it. Type 2 diabetes affects a higher percentage of patients with acatalasemia than the general population, and the condition strikes at a younger age. Although catalase activity is reduced in numerous body tissues, including red blood cells, bone marrow, liver, and skin, only about half of those who are affected have symptoms, which include recurring infections of the gums and associated oral structures that can progress to gangrenous lesions. After puberty, such lesions are uncommon. Soft tissue damage caused by hydrogen peroxide causes mouth ulcers and gangrene in those with acatalasemia.

Furthermore, hydrogen peroxide damages pancreatic beta cells and inhibits insulin secretion. This could be the primary reason for Acatalasemia patients' increased vulnerability to diabetes mellitus. In children with oral ulcers, it should be evaluated in the differential diagnosis. When the patient's blood comes into touch with hydrogen peroxide, it turns brown and stops producing oxygen (seen as bubbling). Assays of the catalase enzyme reveal low levels. Clinical characteristics, pathologic laboratory test results, age, and oxidative stress-related illnesses are all linked to hereditary catalase impairment. It should be viewed as a complicating condition for ageing and oxidative stress, rather than a benign condition. Because Acatalasia is a hereditary illness, it may not be preventable.

It also focuses on new drugs and ways for preventing oxidative stress and diseases related with it. In terms of gene and enzyme therapy, animal studies show that implanting artificial cells with exogenous catalase can replace the non-functioning native enzyme and ameliorate acatalasemia symptoms.

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