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Causes of Glutathione Synthetase Deficiency and its Diagnosis

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Description

Glutathione Synthetase Deficiency (GSD) is a rare autosomal recessive metabolic disorder that prevents the production of glutathione [1]. Glutathione helps prevent damage to cells by neutralizing harmful molecules created during energy production. Glutathione also plays a role in the processing of drugs and compounds that cause cancer (carcinogens), as well as in the construction of DNA, proteins, and other important cellular components. This enzyme is part of the chemical process by which the body makes glutathione, a protein molecule involved in many cell processes. Glutathione synthetase deficiency is caused by changes (mutations) in the GSS gene and is inherited in an autosomal recessive manner. Glutathione also plays a role in the processing of drugs and cancer-causing compounds (carcinogens), and in the assembly of DNA, proteins, and other important cellular components [2].

Glutathione is a tripeptide composed of glutamate, cysteine and glycine. Glutathione is present in mill molar concentrations in most mammalian cells and is involved in several fundamental biological functions including free radical scavenging, xenobiotic and carcinogen detoxification, redox reactions, biosynthesis of DNA, proteins and leukotriene's, and neurotransmission/neuromodulation.

Classification of glutathione synthetase deficiency

Glutathione synthetase deficiency can be classified into three types: mild, moderate and severe. Mild glutathione synthetase deficiency usually leads to destruction of red blood cells (haemolytic anaemia). In addition, affected individuals may release large amounts of a compound called 5-oxoproline into their urine (5-oxoprolinuria). This compound builds up when glutathione is not processed properly in the cells [3].

Individuals with moderate glutathione synthetase deficiency [4] may experience symptoms beginning shortly after birth, including haemolytic anaemia, 5-oxoprolinuria, and increased levels of acid in the blood and tissues (metabolic acidosis). In addition to the features present in moderate glutathione synthetase deficiency, individuals affected by the severe form of this disorder may experience neurological symptoms [5]. These problems may include seizures; a general slowing down of physical reactions, movements and speech (psychomotor retardation); limited intellect; and a loss of coordination (ataxia). Some people with severe glutathione synthetase deficiency also develop recurrent bacterial infections [6].

Causes

Mutations in the GSS gene cause glutathione synthetase deficiency. The GSS gene provides instructions for making an enzyme called glutathione synthetase. This enzyme is involved in a process called the gamma-glut amyl cycle, which occurs in most cells in the body. This cycle is necessary to produce a molecule called glutathione. Glutathione protects cells from damage from unstable oxygen-containing molecules that are by-products of energy production.

Treatment

Treatment for individuals diagnosed with Glutathione Synthetase (GS) deficiency includes providing dietary supplements to correct metabolic acidosis and providing antioxidants such as vitamin E and vitamin C. A combination of sodium citrate and citric acid (Bicitra) can be taken orally medication and can maintain plasma bicarbonate levels within the reference range.

These are best ways to increase your glutathione levels naturally.

- Consume high sulphur foods.
- Increase vitamin C intake.
- Add selenium-rich foods to diet.

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- Eat foods naturally rich in glutathione.
- Supplementation with whey protein.
- Consider milk thistle.
- Try turmeric extract.
- Get enough sleep.

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