Glutaric Acidemia type 1

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**Introduction**

Glutaric Acidemia type 1 is a hereditary disease in which the body is not able to properly break down some proteins and it leads to the accumulation of certain toxins and organic acid in the body. If the condition is not treated properly then it can cause death or brain defect. Glutaric acidemia type 1 is the synonym of glutaric aciduria type 1 and also known as GA-1, dicarboxylic aminoaciduria, Glutaryl-CoA dehydrogenase deficiency and Glutaraye-aspartate transport defect. The condition type is Organic Acid condition (Kölker, et.al, 2011).

The frequency of these diseases is that it affects one out of 40,000 babies in the United States and is common among the Amish population of the States. It is also common in the Indian population of Canada called Ojibwa and among Swedish people. It occurs due to the deficiency of glutaryl-CoA dehydrogenase and GCDH is used to code a mitochondrial matrix protein called a flavin adenine dinucleotide, which is helpful in the metabolism of L-tryptophan, L-hydroxylysine, and L-lysine (Hedlund, Longo, & Pasqual, 2006).

It is described by the accretion of glutaconic acid, 3-hydroxyglutaric acid and, and glutarylcarnitine. These can find out in the body fluid like urine, CSF and plasma and in the tissue by the help of mass spectrometry. A patient will release either a high amount of GA in the urine or low amount. Both types of patients will have an equal chance to get a striatal injury (Kölker, et.al, 2011).

It causes very unique neuropathy logical sequelae that are location and age specifics. It leads to front temporal atrophy that is the occurred when the temporal and frontal cortex is hindered. At the start of the diseases within six to eight months minor neurological symptoms appear and also added up by fever or by catabolic state and sometimes the permanent movement disorders occur due to the destruction of caudate and Putamen. Acute encephalopathy is the common cause of GA-1 and range from acute juvenile encephalopathy to brutally sudden death (Hoffmann, & Zschocke, 1999). In short, GA1 damages the brain in early childhood and it leads to very serious dystonic-dyskinetic disorder.

**Cause of Glutaric Acidemia type 1**

When the GCDH gene gets mutated then it causes Glutaric Acidemia type I. This gene provides the guideline to make glutaryl-CoA dehydrogenase enzyme and this enzyme processes the amino acid hydroxylysine, tryptophan and lysine. Mutation in this gene inhibits enzyme production and a defective enzyme is produced that is unable to function fully which leads to the intermediate breakdown of those amino acids and the products remains inside the body which causes severe complication like the brain damage or death (Kölker, et.al, 2011).

**Follow up testing:**

The diagnosis of GA1 starts with the screening of the newborn with the help of mass spectrometry in case of some family history of the diseases and the test can also be obtained if the child is having some neurological symptoms. The risk factors behind GA-I are the consanguineous marriage, deceased sibling and family history. GA-I can also be diagnosed with lab testing like ketosis, liver function test and hyperammonemia (Kölker, et.al, 2011).

All the new born in Ohio have to pass through different screening tests. They are process with hearing screening test, heart screening test and blood test to check the genetic diseases. All the babies have to participate in the new born screening according to the state policies and hospital needs to provide a formal report of the screening to the parents (“Ohio | Baby’s First Test | Newborn Screening | Baby Health,” n.d.).

**Early warning signs:**

A baby with GA would look the same at birth like a normal baby but some have larger heads and babies could be identified with newborn screening. A GA-1 baby who has not yet been treated will show the assigns of GA-1 within four months and two years after their birth. The early warning signs include poor appetite, Tiredness, longer sleeping’s, irritability, fever, vomiting, weak muscle tone, twitches, delay in growth, muscles would be rigid and the baby would have excessive sweetings. But the sign may vary from individual to individual (Hartley, Khwaja, & Verity, 2001).

**Treatment, and expected outcomes:**

The patient needs to get immediate treatment after diagnosing GA-1. Riboflavin (200–300 mg/d) and Oral L-Carnitine (100–200 mg/kg/d) are mostly recommended to such patient. The natural intake of protein should be limited to 0.5–1.5 gram per day. Special amino acid should be taken that must be free of lysine and should have a low level of tryptophan and the amount of the amino acid taken per day should be 3-3 gram. Further treatment includes anti-convulsion therapy, restricting the intake of protein for a day, high supplementation of carbohydrate and the correction therapy of acid and base( Hedlund, Longo, & Pasqual, 2006).

The treatment of GA-I could be possible in the first stage and the newborn screening has shown some good outcome and is a disease minimizing intervention. The favorable outcomes of the neurology also depend on the chosen therapy but the treatment of kidneys have not been impacted by any current therapy (Hartley, Khawaja, & Verity, 2001).

**Conclusion**

Glutaric acidemia is a very rare form of organic aciduria and the untreated patient will develop dystonia while they are going through infancy and resulting mortality or injury. The deficiency of glutaryl-CoA dehydrogenase is a main cause of GA-I and it is a hereditary disease. This defect results in the accommodation of a high level of 3-hydroxyglutaric, glutaric acid, and glutarylcarnitine

And glutaconic acid. These could be diagnosed with the help of mass spectrometry, or by gas chromatography. In some countries, the condition is identified with newborn screening and that has proved as the best mechanism for the treatment of GA-I. But most of the countries the patients could be diagnosed with the help of striatal injury and those are handled through various metabolic treatments. The treatment includes a diet with a minimum level of lysine and a carnitine supplementation is also given to the patient. Some countries have proper law that make their citizen bound to certain treatment. Like the Ohio State law ensures that all children must participate in the newborn screening test. Such initiatives from the government might decrease the risk of GA-I

**References**

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