Glutaric aciduria type I (GA1) is a rare hereditary metabolic disorder caused by a deficiency of the mitochondrial enzyme glutaryl-CoA dehydrogenase (GCDH). It is in the group of disorders known as cerebral organic acidemias. Individuals with this condition have deficiency or absence of GCDH enzyme that is involved in the lysine metabolism. GCDH deficiency results in increased concentrations of potentially neurotoxic metabolites, glutaric acid (GA), 3-hydroxy glutaric acid (3-OH-GA) and glutaconic acid within body tissues, especially within the brain. Two arbitrary biochemical subtypes have been defined, high (HE) and low excretors (LE), depending on the amount of GA in the urine. Newborns may show unspecific clinical signs like enlarged head circumference (macrocephaly) or decreased muscle tone (hypotonia). Without treatment, most affected children develop an acute encephalopathic crisis following febrile illness episodes or other catabolic conditions resulting in bilateral striatal injury and consequently, dystonic movement disorder. Cognitive outcome has not been systematically studied, but severe cognitive dysfunction is rarely seen. Sometimes babies with GA1 have been mistaken to have been abused because they present with subdural and/or retinal hemorrhages. GA1 is included in the newborn screening panel in a growing number of countries which is essential for early intervention. Importantly, patients with the low excreting phenotype may be missed by newborn screening. GA1 is a rare inborn error of metabolism that affects males as often as females. It has been estimated that there are about 140 patients with this type of organic aciduria in the United States. GA1 occurs in approximately 1 of every 100,000 births. Five genetic isolates are known with a high carrier frequency (up to 1:10) and incidence (up to 1:250 newborns): the Old Order Amish Community in Lancaster County, Pennsylvania, United States, the Oji-Cree First Nations in Manitoba and Western Ontario, Canada, the Irish Travelers in the Republic of Ireland and United Kingdom, the Lumbee in North Carolina, United States and the Xhosa in South Africa.