Biochemical and molecular diagnosis of glutaric aciduria type 1 in a black South African male child: case report.

Abstract:

Glutaric aciduria type 1 (GA-1) is an inborn error of metabolism caused by a deficiency of the mitochondrial enzyme glutaryl-Co enzyme A dehydrogenase. GA-1 is not uncommon amongst Caucasians but to the best of our knowledge, it has previously not been reported in black African children. We present a case of GA-1 in a black South African boy who was referred to hospital at the age of five years and ten 10 months with dyskinesia and dystonia accompanied by chorea and athetosis. Radiological examination revealed enlarged basal cisterns with bilateral fluid collection around the sylvian fissures suggestive of GA-1. Analysis of urine showed raised levels of glutaric acid at 520 micromol/mmol creatinine (normal <2.0), 3-hydroxyglutaric acid at 113 micromol/mmol creatinine (normal <3.0) and a low blood carnitine level of 31.5 micromol/l (normal 35-84). A definitive diagnosis was reached through DNA analysis which revealed homozygosity for an A293T mutation in the glutaryl-Co-enzyme A dehydrogenase (GCDH) gen