Clinical Genetics

AB082. Glycogen storage disease IXa in a Filipino patient

Sylvia Estrada[1,2]

[1]Department of Pediatrics (Metabolic Genetics), University of the Philippines Manila, Manila, Philippines; [2]National Institutes of Health Manila (Institute of Human Genetics), Manila, Philippines

Background: Glycogen storage disease (GSD) type IX, due to the deficiency of hepatic phosphorylase b kinase, results in liver enlargement, growth retardation and fasting ketosis. Though, many patients with this disorder are asymptomatic and do not require treatment, a number of patient can develop liver cirrhosis and growth retardation. Early diagnosis and treatment with cornstarch may prevent liver cirrhosis and promote better growth and development. This is a case report of the first documented GSD IXa in a Filipino boy who was evaluated for suboptimal growth.
Methods: MT is the 2nd child of healthy and non-consanguineous parents. He was noted to have a prominent abdomen since birth, but without organomegaly. He had been reported slightly shorter than his peers, hence was brought to medical attention at 9 years of age.
Results: Diagnostic evaluation revealed no Kayser-Fleischer ring, but hepatomegaly with mildly elevated serum alanine aminotransferase (ALT), aspartate aminotransferase (AST), and ammonia levels. Liver biopsy revealed microvesicular steatosis. Whole exome sequencing with specific analysis of the phosphorylase kinase alpha-2 subunit (*PHKA2*) gene documented hemizygous state of a previously reported variant, c.3614 C>T (p.P1205L). Diet therapy with cornstarch was started and the patient is on regular follow up.
Conclusions: DNA analysis prompted by clinical and pathologic findings led to the definitive diagnosis of GSD type IX.

Keywords: Glycogen storage disease IXa (GSD); hepatomegaly; poor growth; cornstarch

doi: 10.21037/atm.2017.s082

Cite this abstract as: Estrada S. Glycogen storage disease IXa in a Filipino patient. Ann Transl Med 2017;5(Suppl 2):AB082. doi: 10.21037/atm.2017.s082