A Novel Hexokinase Mutation in a Child Presenting with Transfusion Dependent Haemolytic Anaemia

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Abstract

Hexokinase (HK) is one of the key regulatory enzymes for glycolysis. HK catalyses the phosphorylation of glucose to glucose – 6 – phosphate resulting in irreversible transfer of high-energy phosphate from ATP to glucose. HK deficiency is a rare red cell enzyme defect. There are approximately 30 cases and 10 mutations of HK deficiency reported till date. Many patients remain undiagnosed due to mild manifestations of the disease. Here we report a case of a 3-year-old boy who presented with haemolytic anaemia and was found to have HK deficiency with a novel mutation at exon 1 of HK1 gene.

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