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The case Bartter Syndrome with novel mutation in CLCNKB gene

Bilal Haider Shamsi and Liu Yong Lin
Xi'an Medical University, China

The female child of Chinese origin was born of a non-consanguineous marriage, SVD, premature with the birth weight of 1450 g, as the elder amongst the twins of test-tube pregnancy to a G1P2 mother at 29+4 weeks of gestation. The Apgar score was 9 at 1st min and 10 at 5th min. At the age of 8-months the child who had already been treated indoor, three times before, for repeated infections and malnutrition, presented to the hospital for 10 days of anorexia. The child appeared severely malnourished weighing 5 kg only. She had low urine specific gravity <1.005), metabolic alkalosis (serum bicarbonate 29 mmol/l, pH 7.67, BE=9 mmol/L), hyponatremia (Na 136.9 mmol/L), hypocalcemia (Ca 1.44 mmol/L), hypokalemia (2.96 mmol/ L), and hypochloremia (CL 86.50 mmol/L). With the above-mentioned findings, she was clinically diagnosed as BS type III. The polymerase chain reaction (PCR) for amplifying DNA sequences and direct sequencing of all the exons of CLCKB gene was performed using peripheral blood genomic DNA. All the primers were designed according to the sequence of NG_013079.1. The sample was analyzed for CLCNKB gene showed homozygous mutation: c.655+2 T>A (coding region 655+2 nucleotide thymine to adenine), resulting in amino acid splicing mutation. The present study has found a novel mutation, including one already reported SNP, which would enrich the human gene mutation database (HGMD) and provide valuable references to the genetic counseling and diagnosis.

drhydi@outlook.com