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Two Novel Mutations in Aquaporin 2 gene in a Korean Girl with Nephrogenic Diabetes Insipidus

Department of Pediatrics, Ewha Womans University Tongdae Mun Hospital,
Bioinformatics Team Idtech Inc.[1],
Department of Pediatrics, Seoul National University Children's Hospital[2]

Su Jin Cho, Yun Kyung Lee[1], Hee Kyung Kang[2], Il Soo Ha[2], Hae Il Cheong[2], Yong Choi[2]

Congenital nephrogenic diabetes insipidus (CNDI) is a rare inherited disorder characterized by the insensitivity of the distal nephron to the antidiuretic effect of vasopression. There are three inheritance patterns of CNDI; the X-linked recessive form associated with the arginine vasopression V2 receptor gene mutations, and the autosomal recessive and dominant forms associated with the aquaporin 2 gene mutations. The evaluation for polyuria and polydipsia in a Korean girl at the age of one month revealed no response to exogenous vasopression stimulation and confirmed the diagnosis of CNDI. Because being female with no family history, her disease was thought to be an autosomal recessive form. So, we anlayzed the aquaporin 2 gene and detected two heterozygous missense point mutations in the patient; 70Ala(GCC) to Asp(GAC) in exon 1 inherited from her father and 187Arg(CGC) to His(CAC) in exon 3 from her mother. The first mutation is located within the first NPA (asparagine-proline-alanine) motif of the aquaporin 2 molecule and the second one right after the second NPA motif. Both of the mutations are novel and first ones detected in Korea.