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CASE REPORT OF GENETICALLY CAUSED SEVERE HYPOKALEMIA

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Case Study : Introduction A decrease in serum potassium below 3.5 mmol/L is called a hypokalemic change, and this change is a common mineral disorder. Usually due to secondary causes, in some cases there is a decrease in serum potassium of unknown cause. There are many types of tests to diagnose them, as well as genetic testing in many cases. If a large amount of potassium is excreted in the urine and hypokalemia occurs, differential diagnosis should be made from a group of diseases related to changes in the function of the tubules. Common causes of hypokalemia in renal tubule disease include osmotic diuresis, renal tubular acidosis, aldosterone hyperactivity, Gitalman syndrome, and Bartter's syndrome, and the differential diagnosis should be based on clinical signs and symptoms and laboratory tests. Purpose To determine the cause of hypokalemia caused by tubular disorders. Materials and methods Identification of gene mutations by 3B-Exome, Proband technology The result Based on our case questionnaire and actual examination and analysis, it was necessary to distinguish between Bartter and Gitalman syndromes based on the fact that there was a lot of potassium excretion in the urine, as well as the normal level of calcium excretion in the urine. Genetic testing revealed a heterozygous recessive mutation in the autosomal SLC12A3 gene that may be the cause of Gitalman syndrome, and Gitalman syndrome was confirmed as the disease was found to be autosomal recessive. Conclusion It is difficult to distinguish Gitalman syndrome, which is manifested by changes with hypokalemia, from other diseases, and some of the symptoms of this disease are likely to be different in all cases, which requires genetic analysis. In our case, the mutation of the SLC12A3 gene of sodium and hydrogen channels was detected, and the diagnosis was confirmed is there.

GS.png

Genetic testing: 2023/07/18

ORDER INFO

Physician	Institution	Sample
Name: Hee Gyung Kang	Name: Seoul National University Hospital Address: 101 Daehak-ro, Jongno-Gu, Seoul, Seoul, Korea, Republic of Korea (03080)	Type: EDTA blood Ordered on: 2023-07-10 Collected on: 2023-07-08 Accessioned on: 2023-07-18

CLINICAL INFO

Hypokalemia

RESULTS

POSITIVE

Gitelman syndrome [OMIM: 263800]

Gene	Variant	Classification
SLC12A3	**Genomic Position:** 16-56920314-G-A (GRCh37) **DNA:** NM_001126108.2:c.1964G>A **Protein:** NP_001119580.2:p.Arg655His **Zygosity:** Heterozygous **Inheritance:** Unknown	Pathogenic
SLC12A3	**Genomic Position:** 16-56924229-CG-C (GRCh37) **DNA:** NM_001126108.2:c.2332del **Protein:** NP_001119580.2:p.Glu778ArgfsTer8 **Zygosity:** Heterozygous **Inheritance:** Unknown	Likely pathogenic

Potential compound heterozygous pathogenic and likely pathogenic variants were identified in *SLC12A3*. *SLC12A3* is associated with autosomal recessive 'Gitelman syndrome (OMIM: 263800)'. As one of the variants has never been reported in other patients, functional analysis is recommended. Parental testing is also recommended to check if the two variants are in *trans*.