

Abstract Submission No.: A-0425**A case of Gitelman syndrome with SLC12A3 gene mutation****Yi-An Huang**, Huey-Liang Kuo

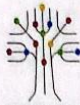
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Case Study : This was a 19-year-old female patient without past medical history, came to our hospital due to dysuria for 3 days. Progressive diffuse abdominal pain noted for a week. The pain was like squashed by the elephant, and had not related to meals or posture. Nausea sensation with poor appetite (<1/4 amount of regular diet) accompanied, but there was no vomiting or diarrhea. She took some analgesic agents from pharmacy and the pain had partial relieved. At emergent department stay, the vital signs were body temperature 36.9°C, pulse 87 beats per minute, respiratory rate 18 respirations per minute, and blood pressure 121/75mmHg. Lab analysis revealed no leukocytosis but mild elevated of hsCRP with value of 1.25*mg/dl (<1 mg/dl). Normal renal function with Creatinine but hypokalemia (K: 2.3* mmol/L) and hypomagnesemia(Mg 1.3*mg/dl) were found. Urine analysis showed pyuria. With diagnosis of right side Acute Pyelonephritis and hypokalemia, we gave her Cravit 750mg once a day and KCL 15meq once a day. For hypokalemia work-up, we checked serum data analysis as following: artery gas showed pH value 7.48 *, HCO₃ 36.5 mmol/L, osmolarity 294 mOsm/kg, ACTH(random) 9.32pg/ml(<46), Cortisol(random) 1.25 ug/dL, TSH 2.496 uIU/mL (0.34-5.60), free T4 0.98 ng/dL (0.54-1.40), Aldosterone: 0.83 ng/dL(6.8-17.3 ng/dL), PRA 0.14ng/mL/hr(supine: 0.32-1.84, stand for 2 hours: 0.60-4.18), and ARR 5.93 ng/dL per ng/mL/hour (<30). Urine data analysis showed osmolarity 258* mOsm/kg, Creatinine: 35.86 mg/dL, Urine Potassium(K) 7.2 mmol/L, and Urine Chloride(CL) 86 mmol/L. Suspected Gitelman syndrome, we sent her serum specimen or genetic analysis and found there were mutation point over c.536T>A, p.Val179Asp, c.2542G>A, and p.Adp848Asn of SLC12A3 gene.

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DIANTHUS MEDICAL GROUP



SOFIVA GENOMICS

SLC12A3基因檢測報告

慧智臨床基因醫學實驗室

採檢日期	2018-11-12	收件日期	2018-11-13	報告日期	2019-01-09
報告編號	病歷號碼	姓名	性別	身份證字號	出生日期
SLC12A3 3	14513689	柴慈茵	女	B223236717	1999-10-07
送檢單位	中國醫藥大學附設醫院內科部		送檢醫師	郭慧亮 醫師	
	404台中市北區育德路2號				
檢驗項目	SLC12A3 基因分析				

檢 驗 結 果

方 法

利用多組專一性引子進行PCR，並以核酸序列分析儀進行SLC12A3基因 (OMIM *600968) 之序列分析。

結 果

Exon-wide sequencing analysis of the SLC12A3 gene: [c.536T>A/c.2542G>A]

cDNA Level	c.536T>A	c.2542G>A
Protein Level	p.Val179Asp	p.Asp848Asn
Damage Prediction by PolyPhen-2 and SIFT database		
Database	p.V179D	p.D848N
PolyPhen-2	Damage score	0.992
PolyPhen-2	Damage score	1.000
SIFT	DAMAGING	DAMAGING
Reference	J Korean Med Sci. 2016;31:47-54.	Kidney Int. 2006;70:813-7.

經由檢驗結果顯示：具有SLC12A3基因c.536T>A, p.Val179Asp 及 c.2542G>A, p.Asp848Asn之突變點位 (Compound heterozygous)。

備 註

無。

技術員
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