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Long-term outcome of Bartter syndrome

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Objectives: Bartter syndrome (BS) is a rare salt-wasting tubulopathy caused by mutations in genes encoding sodium, potassium, or chloride transporters of the thick ascending limb of Henle of the kidney. BS is characterized by polyuria, failure to thrive, hypokalemia, metabolic alkalosis, hyperreninemia and hyperaldosteronemia. Chronic kidney disease (CKD) might develop in BS as well. Potassium and/or sodium supplements, potassium sparing diuretics and NSAIDS are treatment options for BS. While presenting symptoms and initial management of BS are relatively well known, long-term outcome and treatment are still unclear.

Methods: In this study, we retrospectively reviewed genetically proven patients with BS to investigate their long-term and genotpye-phenotype correlation of BS.

Results: A total of 18 patients (M:F=10:8, median age 11.5) with BS were managed at our center. Genetic diagnoses were have homozygous or compound heterozygous mutations of the CLCNKB gene in 14 patients, SLC12A1 mutations in 3, and BSND mutation in 1. They were diagnosed as BS at their median age of 0.4 years [range 0.08-3] with chief complaints of growth retardation, poor oral intake, and electrolyte imbalance. All the patients had severe hypokalemia, metabolic alkalosis, and high renin/aldosterone levels at presentation. Median follow-up was 7 years. At the last visit, all the patients were taking potassium supplement (0.49-15.24 mEq/kg/day of K, median 3.68 mEq/kg/day). 14 patients were taking potassium sparing agents and 6 patients were taking NSAID. Their serum potassium was 3.4 mmol/L [range 2.3-4.5]. Estimated GFR (eGFR) was less than 60 mL/min/1.73 m2 in two patients. Nephrocalcinosis was observed in 10 patients and sensorineural hearing loss (SNHL) was observed in two patients. One patient was treated with growth hormone with favorable outcome.

Conclusions: In conclusion, Bartter syndrome patients need huge amount of potassium supplementation along with potassium sparing agent to maintain electrolyte imbalance, and some of the patients do progress to CKD.

Table 1. Clinical characteristics of 18 patients

	CLCNKB (n=14)	SLC12A1 (n=3)	BSND (n=1)	Total (n=18)
Sex, M:F	1:1	2:1	1:0	10:8
Initial sx, FTT	7/14	0/3	0/1	7/18
Age of onset(yr)	0.42	0.79	N/A	0.41
GH treatment	1/14	0/3	0/1	1/18
Nephrocalcinosis	6/14	3/3	1/1	10/18
SNHL	1/14	0/3	1/1	2/18

Table 2. Biochemical characteristics of 18 patients.

	CLCNKB (n=14)		SLC12A1 (n=3)		BSND (n=1)		Total (n=18)	
	before treatment	after treatment	before treatment	after treatment	before treatment	after treatment	before treatment	after treatment
Na	135.5 (121-141)	139 (134-142)	139 (134-142)	139(137-140)	135	135	135.5	139
K	2.9 (2-4.6)	3.15 (2.3-4.3)	3.5 (3.1-5.1)	3.7 (3.4-3.7)	3.3	4.5	3.1	3.4
Cl	92 (53-101)	96 (91-100)	97 (93-107)	101 (94-102)	96	93	94.5	96
Tco2	31 (22-48)	32 (27-38)	28 (23-29)	27 (21-31)	24	26	29	30
BUN	11 (3-21)	10.5 (5-39)	13 (10-17)	10 (8-17)	8	10	11	10
Cr	0.3 (0.17-0.6)	0.475 (0.44-1.59)	0.3 (0.24-0.5)	0.46 (0.4-0.89)	0.3	0.62	0.3	0.475
Urine Ca/Cr	0.265 (0.02-1.83)	0.24 (0.01-0.48)	1.43 (0.61-1.72)	0.65 (0.18-2.26)	0.64	0.48	0.47	0.29