Analysis the relationship between clinical characteristics and genotype of six cases of Bartter syndrome and Gitelman syndrome in children

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OBJECTIVES

To summarize the children's clinical features, furosemide / hydrochlorothiazide loading test and genotype of Bartter syndrome and Gitelman syndrome; To guide the clinical diagnosis and selecting the target gene examination of Bartter and Gitelman syndrome through the clinical features and furosemide / hydrochlorothiazide loading test.

METHODS

Retrospective analysis of clinical and biochemical characteristics, furosemide / hydrochlorothiazide loading test and genetic testing results of 6 cases (all of them were confirmed by gene examinations) of Bartter syndrome and Gitelman syndrome in the period from 2012 to 2014.

Case	Age (year)	HtSDs	Hypomagnese-mia	The lowest serum potassium values (mmol/L)	Hypercalciuria	$\triangle FE_{cl}$ in the Hct loading test	$\triangle FE_{cl}$ in the furosem-ide loading test	Clinical diagnosis	Gene diagnosis
1	8.27	1.28	no	2.34	no	**< 2.3%**	—	G	no
2	10.76	-2.17	yes	2.07	no	—	**> 2.3%**	G	G
3	15.72	-1	yes	2.69	no	—	**> 2.3%**	G	G
4	2.46	-2.5	no	3.04	yes	**> 2.3%**	**< 2.3%**	B	B
5	0.91	-4.04	no	3.0	yes	—	—	B	B
6	1.04	-1.31	yes	2.1	no	**< 2.3%**	**> 2.3%**	G	G

RESULTS

6 patients came from 6 families, newly diagnosed at the age of 0.91-15.72 years old, and median age was 5.37 years. All patients had hypokalemic alkalosis with the normotensive, hyperreninemic hyperaldosteronism. Most of them had polydipsia, polyuria, and various degrees of growth retardation. The clinical diagnosis of Batter syndrome patients had hypercalciuria and the $\triangle FEcl < 2.3\%$ in Furosemide loading test. Meanwhile the Gitelman syndrome patients had hypomagnesemia and the $\triangle FEcl < 2.3\%$ in hydrochlorothiazide loading test. All the initial clinical diagnosis and genetic diagnosis is consistent at last.

CONCLUSIONS

According to the results of furosemide / hydrochlorothiazide loading test and clinical deta (hypomagnesemia, hypercalciuria), we can better carry out the preliminary diagnosis, and guide the selection of a target gene detection to save costs and medical resources.

References

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ESPE 2015 177--P2

Miscellaneous

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