MYSTERIOUS DIAGNOSIS IN A CHILD WITH FAILURE TO THRIVE

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BACKGROUND

Mutations of several genes encoding the transporters involved in salt reabsorption in the thick ascending limb cause different types of Bartter syndrome (BS), with variable phenotypic expression and severity. Type I and II are the most severe presenting with polyhydramnios, prematurity and characteristically hypokalemia, metabolic alkalosis, polyuria and hypercalciuria

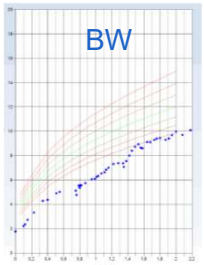
CASE REPORT

9 month old girl

Presentation
* Fever, vomiting, dehydration
* Imbalance of electrolytes despite fluid resuscitation

History
* Maternal polyhydramnios
* Prematurity (34 wks)
* Dysmaturity (birth weight 1,75kg)
* Feeding difficulties since 4 months

Na	154 mmol/l
K	2,8 mmol/l
Bicarbonate	28,7 mmol/l
Creatinin	0,55 mg/dl
Ca/creat urine	0,8
Renin	> 1645 pg/ml
Aldosteron	463 ng/dl



Failure to thrive

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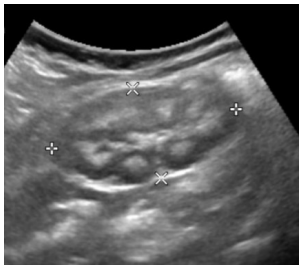

Nephrocalcinosis

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Polyuria
Frontal bossing
Wide nose bridge
Small hands
RR 120/85 mmHg

Hypothesis
*Bartter Syndrome (BS) type I –II
But
*Hypernatremia + hypertension ≠ BS

CLINICAL COURSE

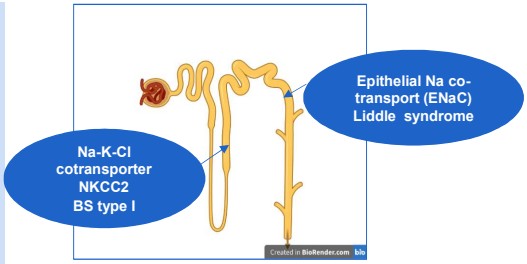
R/ Hyperhydration through nasogastric tube
K suppletion
Indomethacine
Hypercaloric nutrition

Growth improvement
Normalization of elektrolyte imbalance
Genetic analysis

Homozygous variant of SCL12A1gene:c.1216G>T(p.Asp406Tyr) (BS type I)
Heterozygous gain of function mutation of SCNN1Bgene: c.1904GA(p.Ser635Asn)(Liddle syndrome LS)

CONCLUSION

This is the first report of a girl with a phenotypic overlap between BS and LS. Genetic analysis revealed a homozygous variant of SLC12A1, a gene involved in BS type I. This rare variant is classified as pathogenic. However severe hypernatremia and hypertension are a very unusual finding in BS. The heterozygosity for SCNN1B, with subsequently enhanced renal sodium reabsorption, leads us to hypothesize that this variant may balance the renal salt wasting caused by BS.



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