Secondary hyperaldosteronism in the course of Cystic Fibrosis

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Authors declare nothing to disclose

Introduction

The electrolyte disorders in children are commonly considered as symptoms of the endocrine diseases concerning the **renin–angiotensin–aldosterone system (RAAS)** or antidiuretic hormone (ADH). Diseases such as diabetes insipidus (DI), congenital adrenal hyperplasia (CAH) are well known problems, while the **hyperaldosteronism** is much less observed.

The hyperaldosteronism can be primary (Conn's syndrome) as a result of too high production of aldosterone due to the adrenal cortex hyperplasia or tumors. The excessive secretion of aldosterone can be **secondary** due to the hyperactivation of the RAAS. The high concentration of aldosterone increases the sodium absorption in the renal tubules and causes **metabolic alkalosis with hypokalemia** and others electrolyte disorders.

Case description

July 2017 with **hot weather**, a **6-month-old girl** was admitted to the hospital because of the exacerbation of chronic cough. Failure to thrive and malnutrition was also significant despite the patient's good appetite reported by parents.

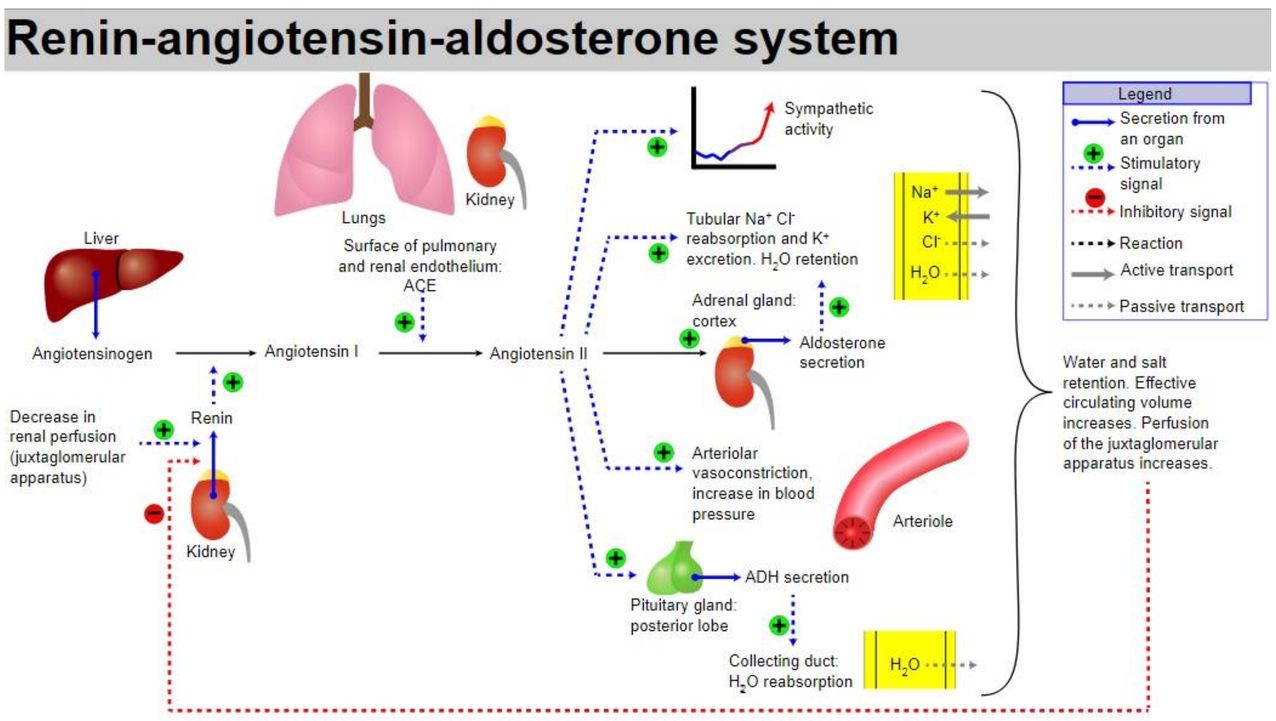
History	Physical examination
Chronic cough	Dyspnea
Recurrent respiratory system infections	Wheezing
Failure to thrive	Malnutrition
Breast-fed	

The bronchiolitis was suspected and treated with rapid good effect.

However the patient was referred to the department of the paediatric endocrinology due to **metabolic alkalosis with hypokalemia, hypochloremia and low concentration of plasma sodium with extremely high value of plasma aldosterone**.

pH	**7,563**	
HCO_3^-	**38,9**	mmol/L
Base Excess	**16,4**	mmol/L
K^+	**3,2**	mmol/L
Na^+	**135**	mmol/L
Aldosterone 1st day:	>>> **100**	ng/dl
Aldosterone 9th day:	>>> **100**	ng/dl

The initial intravenous correction was low-ffective and only the additional oral intake of potassium chloride within 7 days compensated the electrolyte abnormalities.



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Date	18.07	19.07	20.07	21.07	22.07	23.07	24.07	25.07	26.07	unit
pH	**7,563**	**7,536**	**7,509**	**7,523**	**5,513**	**7,502**	**7,476**	**7,442**	7,422	
HCO_3^-	**38,9**	**34,1**	**32,2**	**29,5**	27,6	27,4	24,3	**21,4**	20,8	mmol/L
BaseE	**16,4**	**11,5**	**9,2**	**6,7**	**4,6**	**4,2**	0,8	**-2,7**	-3,7	mmol/L
K+	**3,2**	**3,5**	4,3	**3,3**	4,2	4,1	5,3	**5,6**	5,3	mmol/L
Na+	**135**	**138**	139	**136**	139	137	138	**144**	142	mmol/L

Discussion

Clinical syptoms and the lab tests suggested the Bartter Syndrome (BS) – rare disease caused by the renal salt wasting due to a mutation of the ion channel in the loop of Henle. Nevertheless the associated features presented by the patient (chronic cough and malnutrition) indicated **Pseudo Bartter Syndrome (PBS)**. PBS develops due to salt wasting in varied mechanisms, one of them is the loss of electrolytes in **cystic fibrosis (CF)**.

The dysfunctional cystic fibrosis transmembrane regulator (CFTR) in the sweat ducts of CF patients is responsible for **excessive chloride and sodium loss, particularly during the warm seasons in infants exclusively breast-fed (low-salt level of human milk)**, as it was in case of our patient. The high secretion of aldosterone (activation of the RAAS) in the response to excessive skin salt loss causes increased renal loss of kalium and alkalosis.

Despite the negative screening test for CF the sweat test was perfomred in our patient. The twiced-repeated tests revealed elevated sweat chloride concentration. CF diagnosis was confirmed by the genotyping. It has exposed two rare CTFR mutations on different alleles of chromosome 7 (3849+10kbC->T; p.His199Arg). These mutations are usually connected with residual activity of the CFTR, later onset and longer life expectancy.

Finally the diagnosis of Pseudo Bartter Syndrome in the course of cystic fibrosis was made. The proper CF therapy was started and for now the girl is developing properly without electrolyte disorders and normal level of aldosterone.

Conclusions

Secondary hiperaldosteronism could be the the first symptom of CF very rarely (unusual electrolyte unbalance are 6,2% onset manifestations of CF according to United State Cystic Fibrosis Foundation Data Registry).

Present-day neonatal screening test for CF based on blood concentration of immunoreactive trypsinogen is the standard in the European Union. Nevertheless the 0,5% of patients could have false negative result.

CF patients have increased risk of electrolyte inbalance due to secondary hyperaldosteronism during the excessive loss of ions with sweat or diarrhoea.

References

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