Atypical presentation of cystic fibrosis – obese adolescent with hypertension and pseudo-Bartter’s syndrome

Atipična prezentacija cistične fibroze – gojazni adolescent sa hipertenzijom i pseudo-Barterovim sindromom

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Abstract

Introduction. Infants with cystic fibrosis may fail to thrive despite recommended caloric intake because of electrolyte disturbances caused by salt depletion resulting in hypochloremic metabolic alkalosis or pseudo-Bartter’s syndrome. In most patients reported symptoms began in infancy, but it may be an initial presentation of disease in a previously healthy adolescent. Case report. A 15-year-old boy was admitted for evaluation of recurrent episodes of malaise associated with dehydration and acute renal insufficiency. Laboratory analysis showed hypochloremic metabolic alkalosis with hyponatremia and hypokalemia. On admission the boy was obese, with body weight of 95.5 kg (> P97), height 174 cm (> P75), and body mass index of 31.2 kg/m² (> P95). Physical examination was inconclusive. Blood pressure holter monitoring proved significant systolic hypertension. Routine urinalysis, protein and electrolyte levels in urine were normal. Plasma renin and aldosteron were normal. Sweat chloride concentration was 63 mmol/L. Genetic testing confirmed the diagnosis of cystic fibrosis. Conclusion. To our knowledge, this is the first reported case of atypical presentation of cystic fibrosis in an adolescent presented with pseudo-Bartter’s syndrome and signs of obesity and hypertension. We suggest that every patient with hypochloremic metabolic alkalosis should be evaluated for cystic fibrosis.

Key words: cystic fibrosis; diagnosis; hypertension; obesity; adolescent.

Apstrakt


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resulting in hypochloremic metabolic alkalosis or pseudo-
Bartter’s syndrome 1. In most patients reported symptoms
began in infancy and mostly resolved with appropriate salt
intake at the age of four 1,2. Nevertheless, it may be an initial
presentation of disease in a previously healthy adolescent 3.

Case report

A 15-year-old boy was admitted for further evaluation
of recurrent episodes of malaise associated with dehydration
and acute renal insufficiency, requiring correction by intra-
venous infusion of fluids. These were first observed at the
age of 10, usually occurred during summer and were pro-
vided by physical efforts (sports training) which led to ex-
cessive sweating and malaise that sometimes resolved sponta-
neously. The boy was hospitalized twice in local hospital,
for more severe symptoms when laboratory analysis showed
hypochloremic metabolic alkalosis with hyponatremia and
hypokalemia.

He was born on term and was thriving normally. From the
age of 4 months, he was periodically treated with bron-
chodiators for acute respiratory infections associated with
wheezing. Recently he was sometimes complaining on dysp-
nea during exercise, but he had neither chronic sputum pro-
duction nor recurrent pneumonias. He had atopic dermatitis,
and positive family history of atopy. Skin prick tests for in-
halation allergens were negative. He did not have prolonged
neonatal jaundice, meconial ileus, greasy stools or rectal
prolapses. Histories of cyclic vomiting, dyspepsia, abdomi-
nal pain or dysuria were all negative.

On admission the boy was obese, with body weight of
95.5 kg (> P97), height 174 cm (> P95), body mass index of
31.2 kg/m² (> P95). Auscultatory findings over the chest were
normal. The rest physical examination was inconclusive.

Chest radiography showed no abnormalities. Spirome-
try, whole body plethysmography and impulse oscilometry
showed normal values. Ergospirometry results were above
normal values for age.

Pharyngeal aspirate was negative for bacterial patho-
gens. Blood gas analysis and oxygen saturation were within
normal range. Routine urinalysis, protein and electrolyte lev-
eels in urine were also normal. Plasma renin and aldosteron
were normal. Fecal elastase level was normal. Blood pres-
sure (BP) was elevated and subsequent holter monitoring
were normal. Plasma renin and aldosteron levels are elevated
and blood pressure is usually
within normal range, with normal sweat chloride level.

Hypertension found in our patient is probably caused by obesity.

Pseudo-Bartter’s syndrome is a metabolic disorder that
can be caused by CF, but also by uncontrolled diuretic and
laxative use, rigorous chloride-deficient diet, cyclic vomiting
and bulimia. Symptoms may include polyuria, polydipsia,
vomiting, frequent dehydration and salt craving 1,3. In one of
the largest cohort presented so far, median age at the pre-
sentation was 4 months 2.

First reported by Wagner et al. 7, G126D is a non-
common missense mutation located in exon 4 of the CFTR
gene. It was was reported in an infant who was compound het-
rozygote for deltaF508 and G126D mutations. This result
confirmed diagnosis of CF.

Further analyses were done. High resolution computed
tomography of the chest showed very mild cylindric bron-
chiectasis in middle and lingular lobes. Abdominal and heart
ultrasound were normal, without portal or pulmonary hyper-
tension. Ultrasound examination of testicles showed several
small cysts in the head of the both epididymis which was not
uncommon finding in male CF patients.

Discussion

In classical form of Bartter’s syndrome renal tubules
are unable to reabsorb electrolytes, which lead to its high
urine concentrations. In its several subtypes, plasma renin
and aldosteron levels are elevated and blood pressure is usu-
ally within normal range, with normal sweat chloride level.

Classical form of Bartter’s syndrome is also characterized by
the onset in early childhood 4,5. The findings in our patient,
such as late onset of symptoms, hypertension, normal renin
and aldosteron levels are not indicative for classical form of
Bartter’s syndrome. Hypertension found in our patient is
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It is shown that a long-term survival in CF is probably
not caused by residual CFTR function, and that it is possible
even with “severe” mutations, like the one found in our pa-
ient. It is proposed that mechanisms of a long-term survival
include genetic modifiers and environmental factors 8,9.

Conclusion

As to authors’ knowledge, this is the first reported case
of atypical presentation of CF in an adolescent presented
with pseudo-Bartter’s syndrome and clinical signs of obesity
and hypertension. We suggest that every patient with hypo-
chloremic metabolic alkalosis should be evaluated for CF.

REFERENCES


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