

КРАТКОЕ СООБЩЕНИЕ

CASE HR 010144 INCREASED HAIR K/NA RATIO OF CYSTIC FIBROSIS

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ABSTRACT. A case of cystic fibrosis in a six years old girl was considered. Hair sodium, potassium concentrations in the patient were found long term tending to group at their low adequate reference values or beyond them. However, the K/Na ratio was dramatically increased being eight and six times above normal in 2017 and 2021, respectively. This condition is probably associated with the loss of body electrolytes with sweat, which is characteristic of cystic fibrosis. Considering the fact that diagnosing cystic fibrosis is a complex problem, the results suggest that measuring hair K, Na and the K/Na ratio may provide a noninvasive and widely accessible initial screening test for cystic fibrosis.

KEYWORDS: cystic fibrosis, diagnostics, sodium, potassium, hair analysis.

Recently I was approached by a lady asking for help if her susceptibly autistic daughter is overexposed to the environmental metals. Our patient was a six years old girl 113 cm tall and weighing 19 kg; her height was just two mm below the Geigy 50 percentiles (1). Hair multi bioelement profile analysis was done at the Center for Biotic Medicine (CBM), Moscow, Russia, an internationally acknowledged commercial and research analytical laboratory specialized in medical bioelements (electrolytes, trace elements, and ultra trace elements). Her mother provided two hair samples, one from this, 2021 year, and the other from the year 2017 when her daughter was two years old. The 2017 hair was collected and saved when the girl had serious respiratory troubles diagnosed as pulmonary asthma and for what she was treated with corticosteroids.

The results of both the multi bioelement hair profile showed no traces of metal poisoning. Both profiles are available from the author upon an e-mail request. Indeed, hair bioelement concentrations tend to group at their low adequate reference values, or some of them falling even beyond it. Indeed, our girl had deficient and borderline low sodium concentration in 2017 and 2021 hair samples, respectively.

At the same time, hair potassium concentrations were at the low adequate nutritional status. Our girl had

overt lung corticosteroid responsive asthma in 2017, and now has difficulties of bowel movement. This medical history of pulmonary and gastrointestinal troubles drew our attention to cystic fibrosis (CF).

Cystic fibrosis is an inherited disease of the exocrine glands, primarily affecting gastrointestinal tract and respiratory systems, and usually characterized with chronic obstructive pulmonary disease (COPD), exocrine pancreatic insufficiency and abnormally high sweat electrolytes, notably increased chloride sweat excretion (2). The gene responsible for CF is localized to 250 000 base pairs of genomic DNA on chromosome 7q (the long arm). It encoded a membrane associated protein named the cystic fibrosis transmembrane regulator (CFTR), which is a cAMP regulator of chlorine channel that regulates Cl and Na transport across epithelial membranes.

The results of our Case HR 010144 study over the 2017 and 2021 time periods showed marginal sodium deficiency combined with low adequate potassium nutritional status. However, the K/Na ratio was dramatically increased. Indeed, the observed K/Na ratios were eight and six times above normal in 2017 and 2021, respectively. Evidently, the physiological coupling of chloride and sodium metabolism should be considered if we are confronted with excessive chloride sweat loss in CF.

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Table 1. Hair potassium, sodium, and K/Na ratio at the age of two and six years of life ($\mu\text{g}\cdot\text{g}^{-1}$)

Element	Age (Y)		
	Two (2017)	Six (2021)	Low limit (3)
K	154	158	20.6
Na	47.57	68.2	60.9
K/Na	3.24	2.32	0.400

Diagnosing CF is a complex medical diagnosis problem. This disease has a variety of degrees of severity since infancy, and what may remain unrecognized until in adult life, or even remain undiagnosed (4). The available CF diagnostic tests are protean, but not specific. The pancreatic deficiency of cystic fibrosis is especially dangerous because it goes with lipase deficiency what impairs the absorption of lipid solu-

ble vitamins A, D, E, and K, and what may impair children growth and development. The most severe pancreatic insufficiency of CF may lead to diabetes mellitus (5). Here presented results indicate that measuring hair potassium, sodium and their K/Na ratio may provide a noninvasive and widely accessible initial diagnostic test before more sophisticated genetic testing for CFTR and its mutations are indicated.

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CASE HR 010144. ПОВЫШЕННОЕ ОТНОШЕНИЕ К/НА В ВОЛОСАХ ПРИ МУКОВИСЦИДОЗЕ

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РЕЗЮМЕ. Рассматривается случай муковисцидоза у шестилетней девочки. Было обнаружено, что концентрация натрия и калия в волосах пациентки в течение длительного времени имела тенденцию к снижению до нижней границы нормы или за ее пределы. При этом соотношение К/Na было резко увеличено, превышая норму в 8 и 6 раз в 2017 и 2021 гг. соответственно. Это состояние, вероятно, связано с потерей электролитов с потом, характерной для муковисцидоза. Учитывая тот факт, что диагностика муковисцидоза является сложной проблемой, результаты показывают, что определение К, Na и соотношения К/Na в волосах может обеспечить неинвазивный и широкодоступный начальный скрининговый тест на муковисцидоз.

КЛЮЧЕВЫЕ СЛОВА: муковисцидоз, диагностика, натрий, калий, анализ волос.

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