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Fish Odor Syndrome: A Case Report of Trimethylaminuria

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Trimethylaminuria is a rare, autosomal recessive, metabolic disorder that results in accumulation of trimethylamine (TMA), which smells like rotten fish. The chemical is excreted in sweat and urine owing to a deficiency in the enzyme flavin monooxygenase 3 (FMO3). We report a case of trimethylaminuria in a 12-year-old girl. The patient failed treatment with diet and hygiene modification, but achieved symptomatic improvement after a four-month course of metronidazole.

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Letter

Fish odor syndrome: a case report of trimethylaminuria

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Abstract

Trimethylaminuria is a rare, autosomal recessive, metabolic disorder that results in accumulation of trimethylamine (TMA), which smells like rotten fish. The chemical is excreted in sweat and urine owing to a deficiency in the enzyme flavin monooxygenase 3 (FMO3). We report a case of trimethylaminuria in a 12-year-old girl. The patient failed treatment with diet and hygiene modification, but achieved symptomatic improvement after a four-month course of metronidazole.

Keywords: Trimethylaminuria, Fish Odor Syndrome, Metabolic Disorders

Case synopsis

A 12-year-old girl was evaluated in the dermatology clinic for a chronic foul-smelling body odor. The patient's past medical history was significant for anxiety with generalized hyperhidrosis. Her symptoms flared during periods of anxiety and failed to improve with use of scented antiperspirant deodorants and frequent bathing. The patient was ostracized and severely ridiculed at school.

The patient was adopted, and her biologic family history was unknown. Physical examination was unremarkable except for noticeable foul-smelling odor.

The diagnosis of trimethylaminuria was confirmed by calculating the percent trimethylamine-N-oxide (TMAO) in the patient's urine. The patient's trimethylamine level was 80.7 μmole/mmole creatinine and the level of trimethylamine-N-oxide was 0.6 μmole/mmole creatinine from which the percent trimethylamine-N-oxide was calculated to be 0.7% (normal is considered >92%).

The patient experienced mild improvement on a choline, tyramine, and indole-restricted diet. Significant reduction in the odor was achieved after a four-month course of metronidazole 500 mg twice daily.

Discussion

Trimethylaminuria is a rare metabolic disorder resulting in accumulation of trimethylamine (TMA) in sweat and urine owing to enzymatic deficiency of flavin monooxygenase 3 (FMO3) caused by mutations in the FMO3 gene. Primary trimethylaminuria has an autosomal recessive inheritance pattern [1].

Normally, hepatic FMO3 oxidizes TMA, which smells like rotting fish, into trimethylamine-N-oxide (TMAO), which is odorless [1]. In trimethylaminuria, TMA is excreted through the sweat and urine, resulting in a characteristic fishy body odor [2]. Increased sweating related to exercise or anxiety exacerbates the odor [3].

Initial treatment includes restriction from foods containing choline, lecithin, and carnitine (saltwater fish, liver, egg yolk, soy beans, and legumes), which are precursors to TMA [1, 2]. The excessive consumption of milk has also been linked to increased TMA excretion [4]. Even with rigorous adherence to the restricted diet, the amount of TMA excreted fluctuates [3]. Frequent bathing and deodorants may help conceal the odor [5].

If clinical improvement is not accomplished through diet alone, metronidazole may be used to decrease microbial gut flora and reduce TMA production [2, 3, 5]. Neomycin, activated charcoal, and lactulose have been used in the past, but frequently cause diarrhea. To prevent treatment failure related to drug resistance, the administration of metronidazole for one- to two-week intervals interspersed with drug-free periods is advised [3].

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