

George Preti, PhD  
Member, Monell Chemical Senses Center

## TRIMETHYLAMINURIA

Trimethylaminuria (TMAU), formerly known as “Fish-Odor Syndrome,” is a genetically-transmitted metabolic disorder that causes patients to give off an unpleasant body odor.

People with TMAU are missing or have a defect in the enzyme that allows them to completely metabolize (break down) trimethylamine (TMA), a chemical compound found naturally in many foods. TMA has a foul, fishy odor. At low concentrations, it may be perceived as unpleasant or “garbage-like.”

The principal symptoms of TMAU stem from accumulation of excess TMA – and its associated unpleasant odor – which is then excreted from the body in urine, sweat, saliva, and breath.

A spectrum of changes to the gene coding for the critical enzyme, known as flavin-containing monooxygenase enzyme 3 (FMO3), results in varied degrees of impairment of FMO3’s ability to metabolize TMA. Depending upon the specific type of gene mutation, the amount of unmetabolized TMA and **the related odor varies from individual to individual**. Only about 10-15% of patients with TMAU have an odor that can be characterized as “fishy.”

Excessive production of TMA is linked to the intake of choline-rich foods, including eggs; certain legumes such as soy, kidney beans, etc; wheat germ; saltwater fish; and organ meats, including liver. Importantly, **TMA production and associated odor symptoms depend on what foods recently have been eaten and therefore may occur in irregular and seemingly unpredictable intervals**. Because many foods contain choline, it usually is difficult for patients and their family members to make a connection between symptoms and food intake.

Symptoms may include foul body odors, halitosis (bad breath) and/or dysguesia (bad taste or mouth feel). These can produce social embarrassment. Often, symptoms can only be temporarily relieved by normal hygiene procedures. Further, the unpleasant symptoms are often sporadic in occurrence and seemingly subjective. These characteristics, combined with a lack of knowledge of the disease and its etiology among health professionals, often lead to misdiagnosis of poor hygiene, psychiatric problems and/or referrals to other specialists.

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Consequently, a resulting and persistent problem afflicting most patients are psychosocial ones caused by sporadic, unexplained malodor production and by the unpleasant and unforgiving reactions of people in schools and the workplace. Several reports in the literature have described other complications occurring in conjunction with TMAU; these include seizure, skin rashes, sarcoidosis and Prader-Willi and Noonan's syndromes. However, the majority of patients have normal mental and physical abilities, lead normal lives, and have good hygiene practices.

TMAU is diagnosed using a biochemical test that measures the amount of TMA in the urine following ingestion of 12-16 oz of juice containing 5 g of added choline. A definitive diagnosis often is reassuring to many patients. Currently there is no cure for TMAU. However, current treatment options, including avoidance of choline-containing foods, can help reduce odor production for those afflicted with the disorder.