

Undiagnosed trimethylaminuria may explain many cases of personal malodor

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Scientists from the Monell Center report that approximately one third of patients with unexplained body malodor production test positive for the metabolic disorder trimethylaminuria (TMAU). A definitive diagnosis offers relief to these individuals, as symptoms of TMAU can hinder social and workplace interactions and cause psychological distress. But once the disease is identified, these debilitating symptoms can be ameliorated using changes in diet and other approaches.

"[Health care professionals](#) must arrive at a [correct diagnosis](#) to suggest appropriate treatment," said study lead author Paul M. Wise, Ph.D., a sensory psychologist at Monell. "This research raises awareness of both the disease and also the proper methods of diagnosis and treatment."

TMAU is a genetically-transmitted disease that inhibits the ability of an enzyme to metabolize or transform trimethylamine (TMA), a [chemical compound](#) produced naturally from many foods. TMA has a foul, fishy odor. At lower concentrations, it may be perceived as unpleasant or "garbage-like."

Production of TMA is associated with foods rich in the dietary constituent, choline. Such foods include eggs, certain legumes, wheat germ, [saltwater fish](#) and organ meats.

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

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. This makes the disease difficult to diagnose, as patients can appear to be odor-free when they consult a health professional.

In the study, published online in *The American Journal of Medicine*, the authors tested 353 patients who had contacted the Monell Center because of unexplained personal malodor production. The offensive odors persisted despite good personal hygiene and the underlying causes could not be identified by medical and dental professionals.

Testing at Monell included a choline challenge, in which each patient ingests a set amount of choline and urinary TMA levels are measured over the next 24 hours using sophisticated chemical instrumentation. A high level of urinary TMA confirmed a diagnosis of TMAU in 118 individuals.

Monell's malodor evaluation protocol has the unique advantage of combining sensory evaluation with analytical chemistry techniques. In the current study, sensory findings described a high degree of variation in the quality and severity of malodors associated with TMAU. The authors point out that such individual differences in odor production indicate that the choline challenge test is necessary to confirm the diagnosis of TMAU.

"Although the scientific and popular literature typically describes TMAU sufferers as smelling fishy, our sensory exams demonstrated this not to be so," said study author George Preti, Ph.D., an analytical organic chemist at Monell. "The odors are diverse and only after a choline challenge do the most severe cases have a fish-like odor."

Understanding the genetic and dietary bases of these individual

differences will be one of several subjects for future research.

TMAU is classified as a "rare disease," meaning that it affects less than 200,000 people in the United States. However, its actual incidence remains questionable, due in part to the inconclusive diagnostic techniques used previously.

The current study was funded primarily by private and patient donations. "Only a few laboratories worldwide study TMAU and very little government or foundation funding is available," said Wise. "Yet our research is advancing clinical knowledge while also producing relief and hope for those who suffer from malodor production."

Finally, the scientists note that 65 percent of the afflicted individuals examined did not test positive for TMAU and that future studies will seek to identify other causes of malodor production.

Provided by Monell Chemical Senses Center

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