e-ISSN: 0975-9506, p-ISSN: 2961-6093

Available online on www.ijpqa.com

International Journal of Pharmaceutical Quality Assurance 2026; 17(9); 376-379

Case Report

Fish Odour Syndrome (Trimethylaminuria) in a Rare Case of Obesity

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Received: 21-09-2026 / Revised: 23-10-2026 / Accepted: 28-10-2026
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Conflict of interest: Nil

Abstract:

Trimethylaminuria (TMAU), also known as Fish Odour Syndrome, is a rare condition in which the body cannot properly break down a chemical called trimethylamine (TMA). This happens because of a problem in the FMO3 enzyme. When TMA builds up, it causes a strong fish-like smell. We describe a 23-year-old man who had a persistent rotten, fishy body odour that did not improve even after using regular hygiene methods. This caused him a lot of stress and affected his confidence. Urine tests and genetic studies confirmed that he had an FMO3 gene mutation. The patient was treated with changes in diet and lifestyle, along with activated charcoal, certain vitamins, and probiotics. These steps helped reduce the smell and improved his symptoms. This case shows why early diagnosis is important. With proper testing and support from different healthcare professionals, people with TMAU can have better symptom control and improved quality of life.

Keywords: Trimethylaminuria, Fish Odour Syndrome, FMO3, Genetic Disorder.

DOI: 10.25258/ijpqa.17.9.56

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Introduction

Trimethylaminuria was firstly described in 1970 in a 6-year-old girl by Humbert and colleagues. [1] Primary trimethylaminuria (TMAU) or Fish Odour Syndrome, is a genetic disorder characterized by rotten fish like odour to body secretions like sweat, urine, saliva etc. Some patients even complain fishy odour to genital secretions also. The disease is characterised by inability to convert volatile trimethylamine (TMA) into its odourless form, trimethylamine N-oxide (TMAO). The disorder results from mutations in the FMO3 gene, which encodes flavin-containing hepatic monooxygenase 3 (FMO3), an enzyme responsible for the oxidation of TMA. The accumulation of TMA leads to a pervasive fish-like odour in bodily secretions, including sweat, breath, and urine. [2] There are other reasons for accumulation of TMA due to imbalance in TMA production and metabolism which are categorised in secondary trimethylaminuria like dietary overload, gut dysbiosis, hormonal etc. TMAU has significant psychosocial implications, often resulting in social withdrawal, anxiety, and depression. [3,4] Early diagnosis and management are crucial to mitigating symptoms and improving quality of life. We present a case of a young male diagnosed with Primary TMAU, highlighting clinical presentation, diagnostic approach, and management strategies.

Case Presentation

Patient Information: A 23-year-old male presented to the Obesity Clinic for weight reduction. On detailed history-taking, he reported a persistent rotten fish-like odour affecting his skin, breath and urine for the past five years. The odour was exacerbated by consumption of certain foods, including fish, eggs, dairy products, onions, garlic, and cruciferous vegetables. Despite using various antibacterial soaps and perfumes the odour persisted.

The patient had previously consulted dermatologists and general physicians, who attributed his symptoms to excessive sweating and recommended stronger deodorants and antibacterial treatments, which were ineffective.

He eventually developed social anxiety and depression, for which he consulted Psychiatrist and was prescribed antidepressants over the last three years. Due to effects of antidepressants, he started gaining weight. Over a period of 2 years patient became obese which was another concern for him

increasing psychological disturbances. The patient had no history of significant hepatic dysfunction or renal disease. He was not taking any medications apart from antidepressants and had no known food allergies or intolerances. There was no reported family history of similar symptoms or metabolic disorders.

On physical examination, BMI was 33.1kg/m2. Waist circumference was 112cm and Hip circumference was 104cm so calculated Waist-to-hip ratio (WHR) was 1.08.

Laboratory investigations shows Fasting blood glucose – 108mg/dl, Post prandial blood glucose-145mg/dl, HbA1c → 7.1%, Sr cholesterol →224 mg/dl, Sr Triglycerides- 276mg/dl. Liver function test (LFTs), Renal function tests (RFTs), Thyroid function test (TSH, T4, Complete blood count (CBC) was within normal limits Patients 24-hour urine sample was analysed for TMAO: Total TMA ratio, his free TMA was 45 µmol/mmol creatinine, TMA N-oxide (TMAO) 9 µmol/mmol creatinine so Total TMA (TMA + TMAO) 45 µmol/mmol creatinine. TMAO: Total TMA ratio = 16.7%. Significantly reduced TMAO: TMA ratio (16.7%), indicating impaired conversion of TMA to TMAO, consistent with severe primary Trimethylaminuria (FMO3 enzyme deficiency).

Patient was advised Ultrasound abdomen which showed Grade 1 Fatty Liver. Polysomnography (sleep study) was done to rule out Obstructive sleep apnoea.

Genetic Testing revealed a mutation in the FMO3 gene, confirming the diagnosis of TMAU.

Table 1:

Gene & Transcript	Ex-on/Intron number	Variant nomencla-ture (Variant depth/Total depth)	Zy-gosity	Classifi-cation	Disease	Inher-itance
FMO3 (NM_006894.6)	Exon 6	c.769G>A p.Val25Met (Depth-86X/182X)	Heter-ozy-gous	Pathogen-ic	TMA Trimethylaminu-ria	Autosomal recessive

Diagnosis: The patient was diagnosed with primary (genetic) Trimethylaminuria based on biochemical and genetic testing.

Differential Diagnosis: The differential diagnosis of TMAU includes certain inborn errors of metabolism like phenylketonuria (PKU), Tyrosinemia. Some systemic conditions that can cause abnormal body odour which include chronic liver diseases and chronic kidney diseases. Phenylketonuria results in uremic or ammonia-like odour in adults can be diagnosed by plasma amino acid analysis showing elevated phenylalanine levels, urine organic acid analysis, and genetic testing of the PAH gene.

Tyrosinemia, which produces a cabbage-like or rancid odour, is diagnosed by detecting elevated plasma tyrosine and methionine, measurement of succinyl acetone in urine, and genetic testing of FAH gene mutations. Other conditions which may mimic trimethylaminuria include poor oral hygiene, chronic infections, excessive sweating, dietary factors, and psychiatric conditions like olfactory reference syndrome. Careful history, systemic examination, metabolic workup, and confirmatory biochemical tests (urinary trimethylamine and trimethylamine-N-oxide ratio) were done to differentiate TMAU from these conditions.

Management and Outcome

Lifestyle Modifications: Patient was advised personalised diet plan focusing on reducing TMA rich food and weight loss. He was advised to avoid Choline rich foods, including seafood (especially cod, haddock, and shellfish), eggs, dairy products, soy products, and cruciferous vegetables (broccoli, cauliflower, cabbage, and Brussels sprouts).(5) Patient was encouraged to increase intake of foods rich in flavonoids like fresh fruits, berries, beans, turmeric, tomatoes, green tea etc. to enhance TMA metabolism. Dietary modifications like low calorie (1000kcal/day), low carbohydrate (120gm/day) and high fibre diet for weight reduction also advised. Avoid carnitine-rich and lecithin-rich supplements. Individualised exercise plan including cardio and resistance exercises. Bathing at least two times a day and use of slightly acidic soap to reduce skin odour. Changing clothes frequently also helps in reducing fishy odour. Soaps with a pH between 5.5–6.5 (slightly acidic) to help suppress bacterial growth and reduce TMA production on the skin. Patient was encouraged to use fragrance free soap as fragrances can sometimes mix with TMA odour and make it worse. Psychological support to address social anxiety, stress, and self-esteem issues.

Pharmacological and Supplementary Treatment: Patient was given Activated Charcoal (750 mg twice daily) to reduce gut absorption of TMA. Activated charcoal was given for 10 -15 day and stopped afterwards. Activated charcoal was not given continuously to avoid constipation and it may interfere with nutrient absorption. Activated charcoal was given as pulse therapy around any occasion or when patient used to notice increased

odour. Vitamin B2 (20 mg/day) was given to enhance FMO3 enzyme activity.(6) Vitamin C (1g/day) to increase urinary excretion of TMA. Probiotics (Lactobacillus acidophilus and Bifidobacterium bifidum) were given to alter gut microbiota and reduce TMA production. Patient was requested to continue antipsychotic medicines under supervision of psychiatrist. For Psychological Support patient underwent cognitive behavioural therapy (CBT) to address social anxiety and depressive symptoms. Peer support groups for individuals with metabolic disorders were recommended.

Genetic Counselling: Pretest counselling was done with detailed family history and pedigree charting.

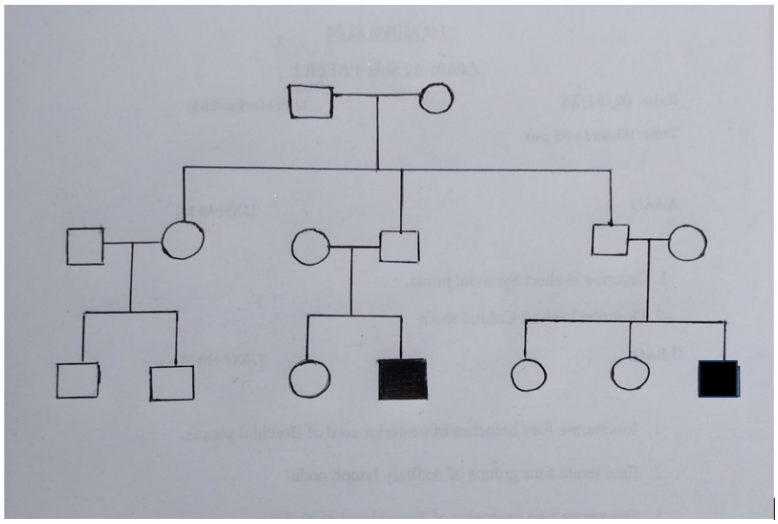


Figure 1: Pedigree chart. Diseased male patient

Post-test counselling will include explaining autosomal recessive inheritance pattern of FMO3 gene mutations. Both parents are likely carriers so recurrence risk in siblings is 25%. Patient was informed importance of pre conceptional counselling after his marriage. Patient was also informed that heterozygous carriers are typically asymptomatic but may have transient mild odour under stress/hormonal changes.

Follow Up and Outcome: After three months of lifestyle and pharmacological intervention, the patient reported a significant reduction in odour intensity. His weight reduced which improved confidence and social interactions, with reduced anxiety and depressive symptoms. His antidepressant dose was gradually tapered under psychiatric supervision.

Discussion

Trimethylaminuria (TMAU) results from a mutation in the FMO3 gene, leading to impaired oxidation of TMA into TMAO.(1) This defect results in excessive accumulation of TMA, which is excreted via sweat, urine, and breath, causing the characteristic fish-like odour. While TMAU is primarily genetic (autosomal recessive inheritance), secondary cases due to liver disease or specific dietary factors have been reported. [7,8] Patients with persistent malodour may be misdiagnosed with other conditions such as bromhidrosis (bacterial breakdown of sweat components), hyperhidrosis (excessive sweating leading to bacterial proliferation), liver or renal dysfunction, or metabolic disorders such as phenylketonuria. [9,10] Early testing, including basic lab tests and genetic tests, is important to clearly identify TMAU and separate it from other causes of body odour [2].TMAU can strongly affect a person's mental health. Many people with this condition feel embarrassed, avoid social situations, and struggle with low confidence, anxiety, or low mood. [3,11]. In this case, the patient's symptoms caused long-term emotional stress and also contributed to a delay in getting the right diagnosis. Because of this, mental health support should be a part of the overall treatment plan. There is no permanent cure for TMAU, so treatment mainly aims to reduce the smell and improve daily life. This includes diet changes, ways to improve enzyme function, changing the gut bacteria, and reducing how much TMA is absorbed in the body. [12]. Counselling or therapy can help people manage the emotional and social difficulties linked to the condition. Researchers are currently studying new approaches such as gene therapy for FMO3 changes, enzyme replacement, and personalised probiotics. These future treatments may offer better options for people with TMAU.

Conclusion

This case shows how important it is to identify trimethylaminuria (TMAU) as a genetic condition that can strongly affect a person's social and emotional well-being and may even contribute to

weight gain over time. Early diagnosis, lifestyle changes, and supportive counselling can greatly improve how patients feel and function. Doctors should consider TMAU in anyone who has a long-lasting body odour, so that the condition is not overlooked and the patient does not receive unnecessary treatments.

Consent: Informed consent was obtained from the patient for publication of this case report.

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