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## Ottava Giornata della Ricerca della Svizzera Italiana

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### Modulo per la sottomissione abstract di ricerca **STUDENTI MASTER**

**Titolo** (massimo **15 parole**)

Trimethylaminuria - the fish odor syndrome: two cases and review of the literature

**Autori** (cognome e iniziali, es: Grassi L.)

Jermini L., Ferrarini A., Ramelli G.P.

**Affiliazioni** (ospedale o istituto, servizio o reparto, indirizzo, es: Ospedale Regionale di Lugano, Servizio di angiologia, Lugano)

Department of Paediatrics EOC, Ospedale San Giovanni Bellinzona, Switzerland

**Testo** (massimo **250 parole**, preferibilmente in italiano (accettato anche in inglese), suddiviso in Introduzione, **Metodi, Risultati, Conclusioni e Finanziamento**)

#### Introduction

Trimethylaminuria (TMAU) is an autosomal recessive metabolic disorder, caused by a deficit in the enzymatic activity of FMO3 (Flavin-containing monooxygenase 3). Accumulation and consequentially increased excretion of trimethylamine are consequences of the impairment of enzymatic activity. A fish like smell, is the only clinical feature. TMAU is therefore also known as «fish odor syndrome». We report two pediatric patients (aged 5 and 10 years, respectively) with TMAU.

#### Cases report

The first case is a 10 years old boy, who from his birth emanated a strong smell of fish associated with febrile episodes, intake of certain foods or physical activities. The medical history and clinical picture as well as laboratory tests suggest a TMAU of genetic origin.

The second case is a 5 years old child, who presented within a few months of birth with a smell of fish. This occurred in particular after the ingestion of foods rich in choline and lecithin. The medical history and laboratory tests suggested a TMAU from substrates overload.

#### Discussion

Both patients show an unpleasant body odor without any other symptoms or signs. Faced with this clinical situation it is imperative to consider rare metabolic diseases as possible differential diagnosis. Additionally, acquiring the correct diagnosis may provide somatic relief for the patient and parents from a psychosocial perspective. Integrating the information related to our two cases with the literature's guidelines we were able to build a diagnostic algorithm for this disease; from medical history up until the molecularly diagnostic tests.

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**Visto superiore** (prego indicare Nome e Cognome del superiore)

Prof. Dr. med. Gian Paolo Ramelli

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**Criteria per sottomissione Abstract:**  
NO Case report  
NO Abstract senza nessun risultato  
VISTO da un superiore

**Invio Abstract**

