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INTRODUCTION

Trimethylaminuria (TMAU) is a metabolic disorder that produces pronounced body odors resulting from the systemic accumulation of an excessive amount of unoxidized trimethylamine that is then excreted in the breath, sweat, and urine. The biochemical deficit causing this condition is the inability to completely metabolize trimethyamines (TMA) to TMA-oxide (TMAO) in the liver. Many researchers believe that there are several types of TMAU caused by a "spectrum" of changes in the gene which controls the formation of the flavin-containing monooxygenase 3 (FMO3) enzyme. In humans, this is an important liver enzyme that controls the metabolism of substances such as TMA. The most severe form of TMAU appears to be caused by mutations in the FMO3 gene; these mutations appear inherited in an autosomal recessive fashion. Studies are leading many researchers to conclude that the less severe forms of TMAU are caused by several non-benign genetic polymorphisms in the FMO3 gene. Genetic polymorphisms are changes in the gene structure that may be fairly common in the population; however, for reasons, which are not well understood, these changes lead to TMAU-symptoms in certain individuals.

It is estimated that as much as one percent of the U.S. population may suffer from TMAU, but its true incidence is not yet known. But whether it is one or one-tenth of one percent, we know that the condition affects people of both sexes and of all ages and races from around the world. Currently there are more than 300 people with a malodor disorder on the Trimethylaminuria Support Group's mailing list, with many more preferring to remain anonymous because of the often-associated stigma, negative and harassing behaviors targeted at some, and the general lack of medical and other support.

This bibliography was prepared in support of the Second Workshop on Trimethylaminuria* held at the National Institutes of Health (NIH) on March 15-16, 2002, in Bethesda, Maryland. The Workshop was sponsored by the NIH Office of Rare Diseases. Co-sponsors of the meeting included: the National Human Genome Research Institute; the National Center for Research Resources; the National Institute of Diabetes and Digestive and Kidney Diseases; the National Library of Medicine; and the Trimethylaminuria Support Group, Inc. (TSG). It was organized by TSG, Inc., and co-chaired by John Cashman, Ph.D., and Eileen Treacy, M.D. This meeting marked the first time large numbers of people with TMAU came together to get the critical facts needed to better understand its mechanisms and treatment options, and to learn how to manage life with TMAU. Goals of the meeting included educating people with TMAU, increasing general awareness, and stimulating new research into this life-disrupting condition.

Candidate citations for this bibliography were obtained from searching a wide of variety of online databases for the period January 1999 through August 2001. Although the search strategy was not limited by language or types of material, the resulting final bibliography consists primarily of citations to English-language journal articles and conference papers. Both human and animal studies have been included. Citations have been arranged into six broad categories; a citation appears in only one category. This publication also updates the bibliography prepared for the First International Workshop on Trimethylaminuria (Fish-Malodor Syndrome) held at the NIH in March 1999:

> Patrias, Karen; Pikus, Anita; McConnell, Harry; Mitchell, Steve, compilers. Trimethylaminuria (Fish- Malodor Syndrome) and the flavin monooxygenases [bibliography on the Internet]. Bethesda (MD): National Library of Medicine; 1999 Mar. (Current bibliographies in medicine; no. 99-2). 430 citations from January 1966 through December 1998. Available from: http://www.nlm.nih.gov/pubs/cbm/trimethylaminuria.html

The compilers which to thank Patti DeWild, Marcia Tucker, and Ed Hamilton for their valuable assistance in reviewing this updated bibliography.

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Patrias, Karen; Ahmed, Terry T.; Lambert, Debby, compilers. Trimethylaminuria and the flavin monooxygenases [bibliography on the Internet]. Bethesda (MD): National Library of Medicine (US); 2002 Mar [insert cited year month day]. (Current bibliographies in medicine; no. 2002-2). 163 citations from January 1999 through August 2001. Available from: http://www.nlm.nih.gov/pubs/cbm/trimethylaminuria_update.html

* Conference information:
http://rarediseases.info.nih.gov/news-reports/workshops/Trimethylaminuria010921_01.html

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Citations are formatted according to the rules established for *Index Medicus*®*. Sample journal and conference paper citations appear below. For journal articles written in a foreign language, the English translation of the title is displayed in brackets and the language of publication is shown by a three-letter abbreviation appearing at the end of the citation.

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Journal Article:

Authors *Article Title* *Abbreviated Journal Title* *Date*

Kashyap AS, Kashyap S. Fish odour syndrome. Postgrad Med J. 2000 May; 76(895):318-9. Available from: PubMed; PMID 10858113.

Volume Issue Pages *Availability*

Conference Papers Presented:

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Cha YN, Park CS, Lee KH, Chung WG. N-demethylation of caffeine by recombinant human flavin-containing monooxygenases. Paper presented at: 38th Annual Meeting of the Society of Toxicology; 1999 Mar 14-18; New Orleans, LA.

Title of Meeting *Date of Meeting* *Place of Meeting*

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*For details of the formats used for references, *see* the following publication:
Patrias, Karen. National Library of Medicine recommended formats for bibliographic citation. Bethesda (MD): The Library; 1991 Apr. Available from: NTIS, Springfield, VA; PB91-182030.

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APPENDIX:

SOURCES OF INFORMATION ON TRIMETHYLAMINURIA

Trimethylaminuria Support Group, Inc.
P.O. Box 3361
Grand Central Station
New York, NY 10163
212-678-2506
Trimeth411@aol.com

The Genetic and Rare Disease Information Center
P.O. Box 8126
Gaithersburg, MD 20898-8126
1-888-205-2311
TTY: 1-888-205-3223
Fax: 202-966-5689
E-mail: Gardinfo@nih.gov

The Office of Rare Diseases (ORD)
National Institutes of Health
31 Center Drive, MSC 2084
Building 31, Room 1B-19
Bethesda, MD 20892-2084
Telephone: 301-402-4336
Fax: 301-480-9655
E-mail: hh70f@nih.gov
http://rarediseases.info.nih.gov/

National Organization for Rare Disorders, Inc.
P.O. Box 8923
New Fairfield, CT 06812-8923
Phone: 203-746-6518; 1-800-999-6673
Fax: 203-746-6481
http://www.rarediseases.org