

Diagnosis and treatment of tyrosinemia type I: a US and Canadian consensus group review and recommendations.

Chinsky et al., Genetics in Medicine, 2017. PMID: 28771246

Summary of conclusions using the Appraisal of Guidelines for REsearch & Evaluation (AGREE) Instrument (AGREE II) tool: the AGREE II tool is intended for guideline developers to conduct an internal assessment to ensure that their guidelines are sound or to evaluate guidelines from other groups for potential adaptation to their own context.

Number of Appraisers: 2

Endorsed by the Garrod Guideline committee on April 13, 2023

Summary: This document is constructed for metabolic physicians and dietitians by metabolic physicians and dietitians for the diagnosis, management, and monitoring of individuals with tyrosinemia type I.

Please find our evaluation based on the following domains:

1. **Scope and purpose.** The target population, care goals, and expected benefits of recommendations are clearly defined.
2. **Stakeholders' involvement.** Written by metabolic physicians and dietitians without clear and direct (preferences and views) of the target population. Guidelines are intended for use by metabolic clinics.
3. **Rigor of development-** The guideline paper is very strong in outlining the search methods, clearly indicating criteria for the selection of the evidence, linking this to existing evidence, and considering benefit and harm. The strength and limitations of existing evidence were not discussed; however, it was specifically noted by the reviewers that there was a limited body of evidence to evaluate. The recommendations were not reviewed by external stakeholders.
4. **The presentation was clear.** This guideline provides specific and clear recommendations, including indemnifying key recommendations and clear recommendations of management options.
5. **Applicability (Modified*).** Implementation tools for treatment, monitoring, and surveillance were included and clear. Implications of recommendations for health systems were not discussed. Monitoring and auditing plans for the outcomes of these recommendations were not addressed in this guideline.

6. **Editorial independence.** At least partial funding was received from the industry; There is no description or declaration specifically addressing how this funding influenced guideline development.

Summary of the AGREE II domain scores given by the appraisers (averaged between the two appraisers):

Domain	score
Scope and purpose	21/21
Stakeholder involvement	11.5/21
Rigour of development	32.5/56
Clarity of presentation	19.5/21
Applicability	15.5/28
Editorial independence	8/14