Online Inherited Metabolic Diseases (OIMD)

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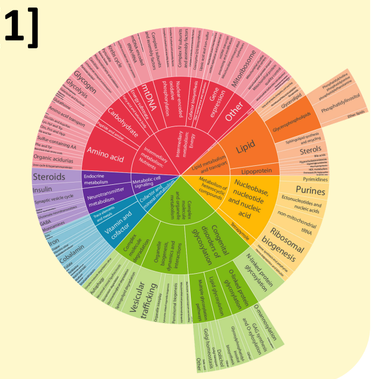
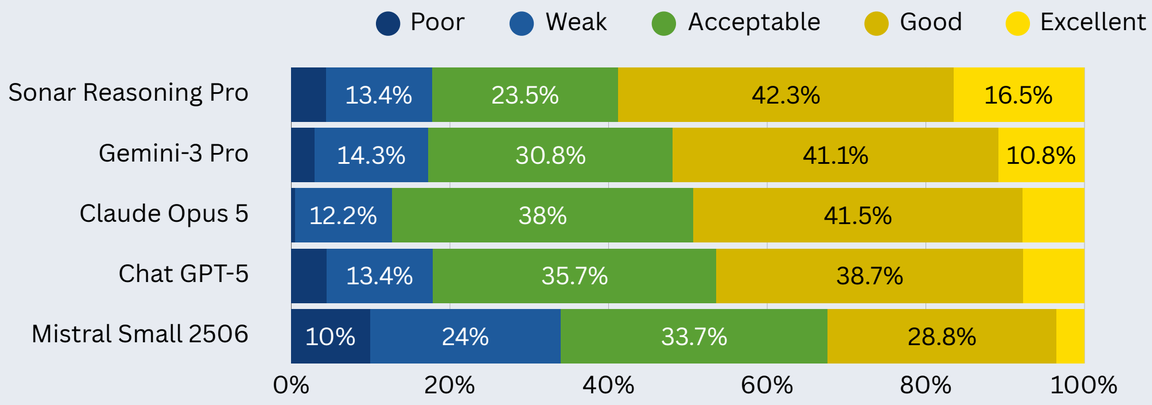
The international classification of inherited metabolic diseases (ICIMD) [1]

- **Hierarchical**, **international system** covering ~1500 **inherited metabolic diseases (IMD)**
- Endorsed by all **major IMD experts** worldwide
- Includes **key genes**, **biochemical pathways** and **mechanisms**



Rating Large Language Model (LLM) output

Comparison of **satisfaction levels** among **METABERN** members (n = 28) who participated in an online survey. Outputs from various LLMs were queried with the **same prompt** to ICIMD records. Participants rated the outputs on a 5-point Likert scale from **poor (1)** to **excellent (5)**. The number of independent ratings per LLM output ranged from 371 (Mistral Small 2506) to 406 (Gemini-3 Pro). No model was clearly preferred across participants.



Concept of OIMD

- **Aim**: Provide online easy-access information on all inherited metabolic diseases
- **Basis**: International classification of inherited metabolic diseases (ICIMD) [1]
- **Concept:** hierarchical **ICIMD structure** with automated **LLM-based content** generation, iterative scientific **validation**, and expert-driven quality assurance.

Hierachical structure from ICIMD [1] **01**

Data: Prompt Chat GPT5.0, syntax postprocessing and integration of HPO terms **02**

LLM generated record: Awaiting review

Author Submission: Experts propose changes in the LLM based records **03**

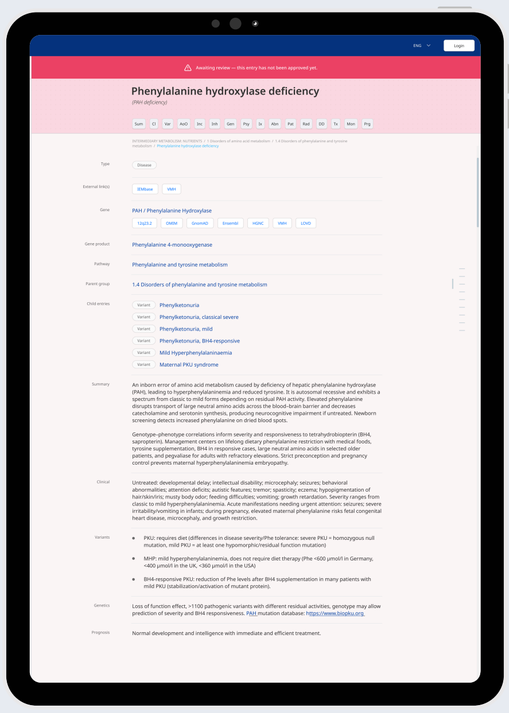
Record currently edited

Review: Record in review by senior experts **04**

Accepted Record: Reviewed and published

Content of OIMD

- Diseases
  - Groups
  - Phenotypes
  - Diseases
    - Variants
- Genes
- Gene Products
- Pathways
- Multimers



Example Entry of the Diseases Record Phenylalanine hydroxylase deficiency.

User roles in OIMD

Current records are publicly available, with a signup process granting four user roles.

Basic User: Discuss records

Author: Propose changes in records

Reviewer: review and publish records

Editor: Restructure hierarchy

Access to oimd.org

Interact and contribute to expand the knowledge forum.

OnlineIMD

[1] Ferreira CR, et al. An international classification of inherited metabolic disorders (ICIMD). J Inherit Metab Dis. 2021 Jan;44(1):164-177. doi: 10.1002/jimd.12348.

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