



IEMbase - Inborn Errors of Metabolism Knowledgebase

URL: <http://www.iembase.org>

Version 2.0.0

Number of disorders: 1596

Number of clinical and biochemical signs and symptoms: 3679

IEMbase is freely available and offers:

- Easy browsing through IEMs
- Simple search for any IEM or term
- Mini-expert system to search for a combination of clinical symptoms and biochemical markers and their deviations - Differential diagnosis (DDx) panel
- Biochemical tests panel
- Gene panel
- Disorder information, incl. external links (e.g., OMIM, Orpha code, HMDB etc.)
- Age-related clinical symptoms
- Age-related biochemical markers
- Gene information, incl. external links
- WikiPathways (not available for all disorders yet)
- Educational documents (PDFs, videos), not available for all disorders yet
- Gamut tables of all issues of Clinical and Biochemical Footprints of Inherited Metabolic Diseases.

A mobile App for iOS and Android is available in Apple's App store and in the Google Play store.

Treatment information will be completed and updated in 2021/22.

Nosology is adapted to the current ICIMD nomenclature.

IEMbase is a non-profit entity, associated with the SIMD and an Executive Editorial Board planning the current and future development of the project, and with a long-term budget of 1.3M\$.

IEMbase Expert Panel

The Expert Panel members curate disorders information in their field of expertise, report new diseases and updates on phenotypes and/or treatments.

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