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.

Cécile Acquaviva     Lyon     France
Matthias Baumgartner  Zurich  Switzerland
Bodo Beck             Cologne  Germany
Michael Beck          Mainz     Germany
Jörgen Bierau Maastricht The Netherlands
Olaf Bodamer Boston USA
Annet Bosch Amsterdam The Netherlands
Oliver Braissant Lausanne Switzerland
David Cassiman Leuven Belgium
Tom de Koning Groningen The Netherlands
Terry Derks Amsterdam The Netherlands
Robert Desnick New York USA
Daria Diodato Rome Italy
Carlo Dionisi-Vici Rome Italy
Areeg El-Gharbawy Durham USA
Francesco Emma Rome Italy
Marc Engelen Amsterdam The Netherlands
Roberto Giugliani Porto Alegre Brazil
Andrea Gropman Washington USA
Sarah Grünert Freiburg Germany
Johannes Häberle Zurich Switzerland
Georg Hoffmann Heidelberg Germany
Carla Hollak Amsterdam The Netherlands
Martina Huemer Zurich Switzerland
Jaak Jaekken Leuven Belgium
Mirian Janssen Nijmegen The Netherlands
Heinz Jungbluth London United Kingdom
Richard Kelley Baltimore USA
Femke Klouwer Amsterdam The Netherlands
Ina Knerr Dublin Republic of Ireland
Stefan Koelker Heidelberg Germany
Viktor Kožich Prague Czech Republic
Ilja Kubisch Chemnitz Germany
Robin Lachmann London United Kingdom
Min Ae Lee-Kirsch Dresden Germany
Nicola Longo Salt Lake City USA
Diego Martinelli Rome Italy
Philippa Mills London United Kingdom
Fanny Mochel Paris France
Eva Morava Rochester USA
Thomas Opladen Heidelberg Germany
Marzia Pasquali Salt Lake City USA
Phillip L. Pearl  Boston  USA
Holger Prokisch  Munich  Germany
Antonia Ribes  Barcelona  Spain
René Santer  Hamburg  Germany
Jörn Oliver Sass  Rheinbach  Germany
Maurizio Scarpa  Padua  Italy
Tom Schirris  Nijmegen  The Netherlands
Andreas Schulze  Toronto  Canada
Bernd Schwahn  Manchester  United Kingdom
Guenter Schwarz  Cologne  Germany
Denise Slenter  Maastricht  The Netherlands
Hans Spelbrink  Nijmegen  The Netherlands
Antonella Spinazzola  London  United Kingdom
Robert Steinfeld  Zurich  Switzerland
Sylvia Stockler  Vancouver  Canada
Ulrich Stölzel  Chemnitz  Germany
Davide Tonduti  Milan  Italy
Frederic Tort  Barcelona  Spain
Karin Tuschl  London  United Kingdom
Bert van den Heuvel  Nijmegen  The Netherlands
Clara van Karnebeek  Nijmegen  The Netherlands
Francjan van Spronsen  Groningen  The Netherlands
Frederic Vaz  Amsterdam  The Netherlands
Nanda Verhoeven-Duif  Utrecht  The Netherlands
Christine Vianey-Saban  Lyon  France
Mirjam Wamelink  Amsterdam  The Netherlands
Ron A. Wevers  Nijmegen  The Netherlands
Saskia Wortmann  Salzburg  Austria

**Executive Editorial Board**
Nenad Blau, Zurich, Switzerland (Chair)
Calos Ferreira, Washington, USA
Clara van Karnebeek, Amsterdam, The Netherlands
Katrin Õunap, Tartu, Estonia (SSIEM representative)
Wyeth Wasserman, Vancouver, Canada

**Bioinformatics Programmer**
Brittany Hewitson, Vancouver, Canada
Database QC
Benedikt Dechow, Zurich, Switzerland

July, 2021