

TEMPLATE of case description for case database (CDB-IMD)

Version 3, June 2021

Please submit the following information, along with pertinent (positive or negative details), and visual aids where possible (pictures, videos, chromatograms...).

Title of the case:**

Clinician and laboratory specialist (+center):

Name:

Function:

Institute (department+institute):

Country:

Email of corresponding "author":

If article is published, please mention original article:

** Mention official name of disease (OMIM) with introducing title and year of presentation.

Year of presentation is relevant for the understanding of application of certain analysis.

1. Clinical presentation

Please present the medical history in a story like fashion (see prototype OTC case) and consider the following issues: Chief complaint (presenting problem), age of presentation, description of signs and symptoms; pertinent positive and negative symptoms relevant to the case, when did symptoms start and in what order? Progression of the symptoms over time.

2. Medical history

Please present the medical history in a story like fashion (see prototype OTC case) and consider the following issues: sex, age and ethnicity, pertinent prenatal history, labor and delivery, family history, history of hospitalizations or surgeries/ relevant dietary and medication history

3. Results of neonatal screening? (please fill in only in case of positive screening)

- In which country did neonatal screening take place?
- On which test did patient test positive?
- Which biomarkers were used and what was cut-off?
- Was positive screening result confirmed by dedicated tests?

4. Physical examination

What are relevant physical characteristics (weight/ height/ head circumference and please mention percentiles of age) and results of physical examination (neurological, heart, lungs etc).

Are there data of fundoscopic findings. Photographs or videos, if available are welcome. If abnormal, provide a normal for comparison (as description or reference photograph).

5. Differential diagnosis?

- What is your differential diagnosis based on the 1-3? (focused on metabolic disorders!)
- What would be the next step in diagnostics?

6. Imaging data or other functional tests

- Was a CT or MRI or PET-CT performed? Possibility to include CT or MRI that shows abnormality (accompanied by reference picture from healthy individual)

- Neurodiagnostic testing (EEG, EMG, nerve conduction...)
- Hearing testing
- Other functional test

7. Clinical chemistry results

- Basic laboratory results (clinical chemistry, hematology etc) at moment of clinical presentation; which results indicate the IMD?
- Please fit our relevant parameters in this table.
- Mention the age-based reference values
- *Automatic calculation of SI to American units (will be programmed later)*

Table Clinical chemistry results on day of presentation (age; months or years)

Analyte	Body fluid	Result	Reference value (unit: SI-unit and American unit)	Comment

Reference values, if applicable, are age-based.

8. Metabolic screening/metabolomics:

- *which body fluid/analysis/methods*
- *results as numbers and/or chromatograms + interpretation of results by laboratory specialist*

Table Metabolic profiling of relevant metabolites in plasma/urine/CSF at the day of clinical presentation (age of patient is).

Metabolite	Result	Reference value (unit: SI-unit or American unit)	Interpretation

Example with list of metabolites is in excel table.

9. Differential diagnosis based on laboratory results:

- Differential diagnosis (metabolic +/- genetic), based on clinical / investigations / chemistry / metabolic testing
- Next approach to diagnostic testing, or confirmatory testing (enzyme/DNA/ RNA analysis)

10. Enzyme or genetic diagnosis (or: immunohistochemistry/flowcytometry etc)

- Tissue/body fluid
- Analyses (enzyme and method, or DNA testing and method).
- Result/interpretation of results by laboratory specialist

11. What is final diagnosis?

- OMIM name of IEM with number:
- link with OMIM:
- link DDRMD/IEMbase:
- ICIMD classification (see pdf on CDB-IMD site)

12. Treatment

- What was the treatment?
- How was response to treatment monitored (clinically/laboratory)?
- Was there emergency protocol?

13. Clinical course and follow-up

- What is the clinical course under/without treatment
- disease progression (signs and symptoms?)

14. Clinical and biochemical synopsis (i.e. pathway, essential teaching aspects)

- What are the takeaway points that make this an informative teaching case?
- Discuss the case briefly from this aspect.
- Indicate the pathway to explain the findings (NEW!!)

15. Patients' experience (not applicable now)

16. References

- Provide 1-3 relevant references from literature