

E-IMD

European registry and network for
intoxication type metabolic diseases

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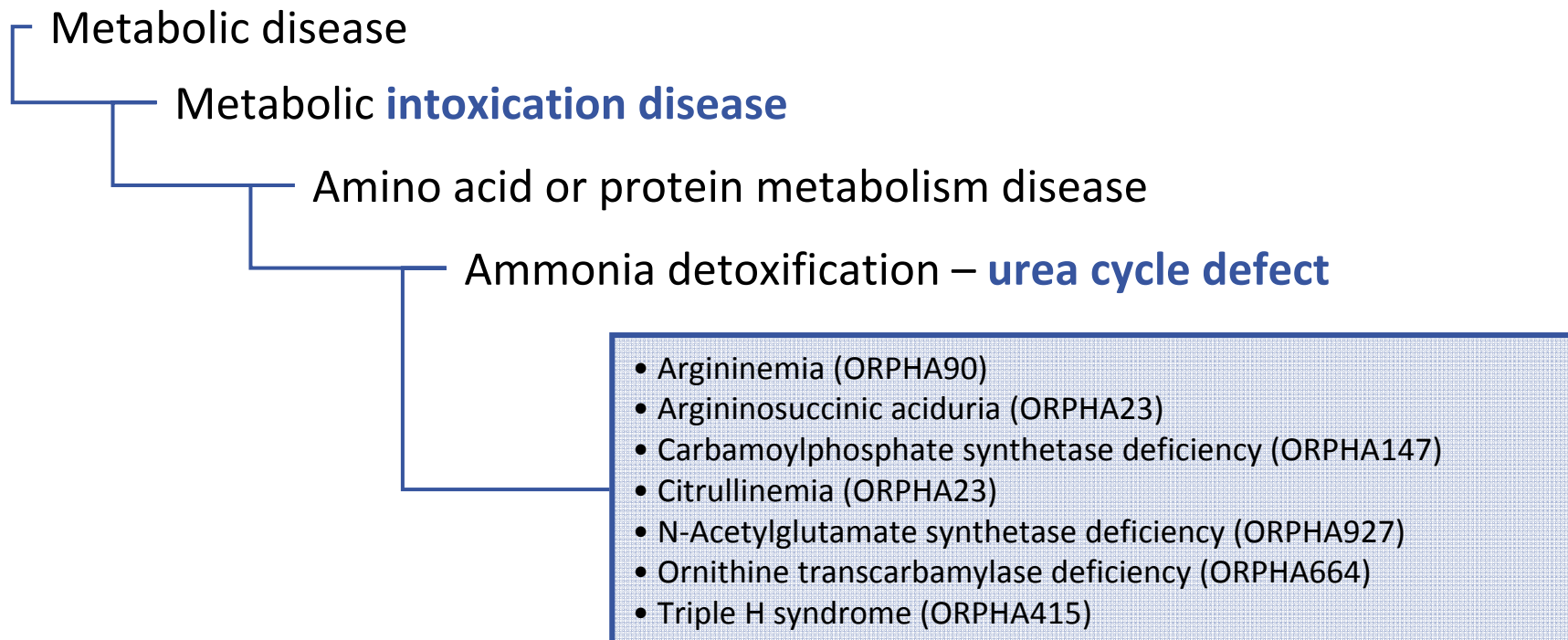


Intoxication type metabolic diseases

- E-IMD focuses on rare inherited metabolic diseases of **amino acid metabolism** resulting in accumulation of **endogenous toxic metabolites**
- The E-IMD disease panel includes **11 diseases** from **2 different subgroups** of intoxication type metabolic diseases:
 - **Urea cycle defects**
 - **Classical and cerebral organic acidurias**

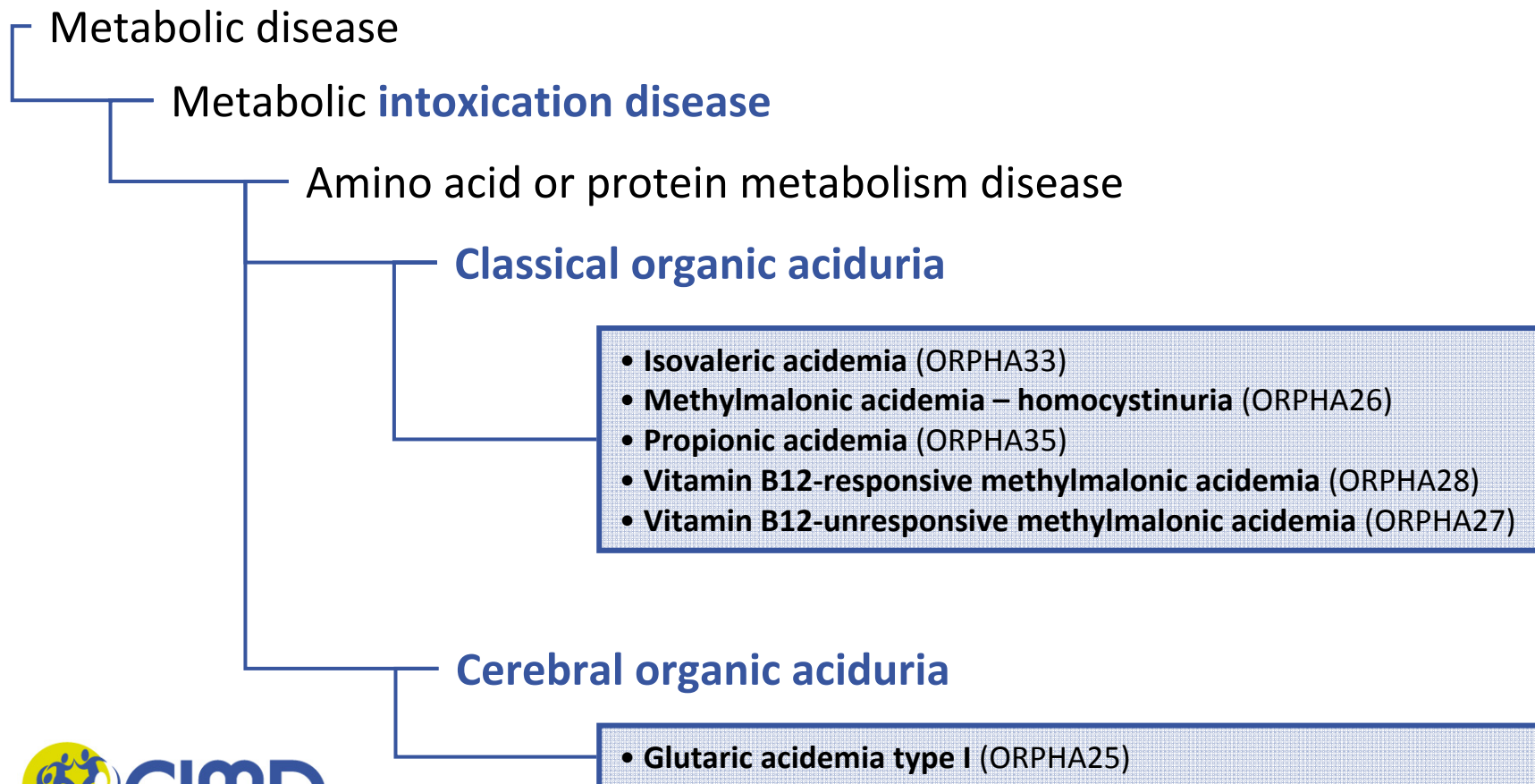
Specification of the rare disease group – 1

Orphanet classification of rare inborn errors of metabolism (www.orpha.net)



Specification of the rare disease group – 2

Orphanet classification of rare inborn errors of metabolism (www.orpha.net)



Specification of the coordinating centre – 1

E-IMD coordinator

University Hospital Heidelberg, Germany

Project leader



Prof. Dr Stefan Kölker, MD

University Children's Hospital, Clinic I

Head, Division of Inherited Metabolic Diseases

Legal representative

Prof. Dr Claus R. Bartram, MD

Dean, Medical Faculty

Specification of the coordinating centre – 2

Medical Faculty Heidelberg

- Oldest medical faculty in Germany (founded in 1386)
- Motto (coined by Vinzenz Czerny): „*In scientia salus*“

University Children's Hospital

- Founded in 1860
- Long-lasting history in the study of rare inherited metabolic diseases
 - One of the largest **metabolic centres** worldwide
- Coordinated large-scale **network projects**
- Accredited **European Training Centre** in Metabolic Diseases (certified by European Academy of Paediatrics in 2003)

Centre for Rare Diseases University Medicine Heidelberg

- Founded in 2010
- Official partner of the „**Kindernetzwerk e.V.**“ (network for children and adolescents with chronic diseases including rare diseases)

Main and specific objectives of the E-IMD

Supporting clinical management and quality of care

General objective

To promote health for children, adolescents and adults with rare **organic acidurias** (OADs) and **urea cycle defects** (UCDs) in Europe.

Specific objectives

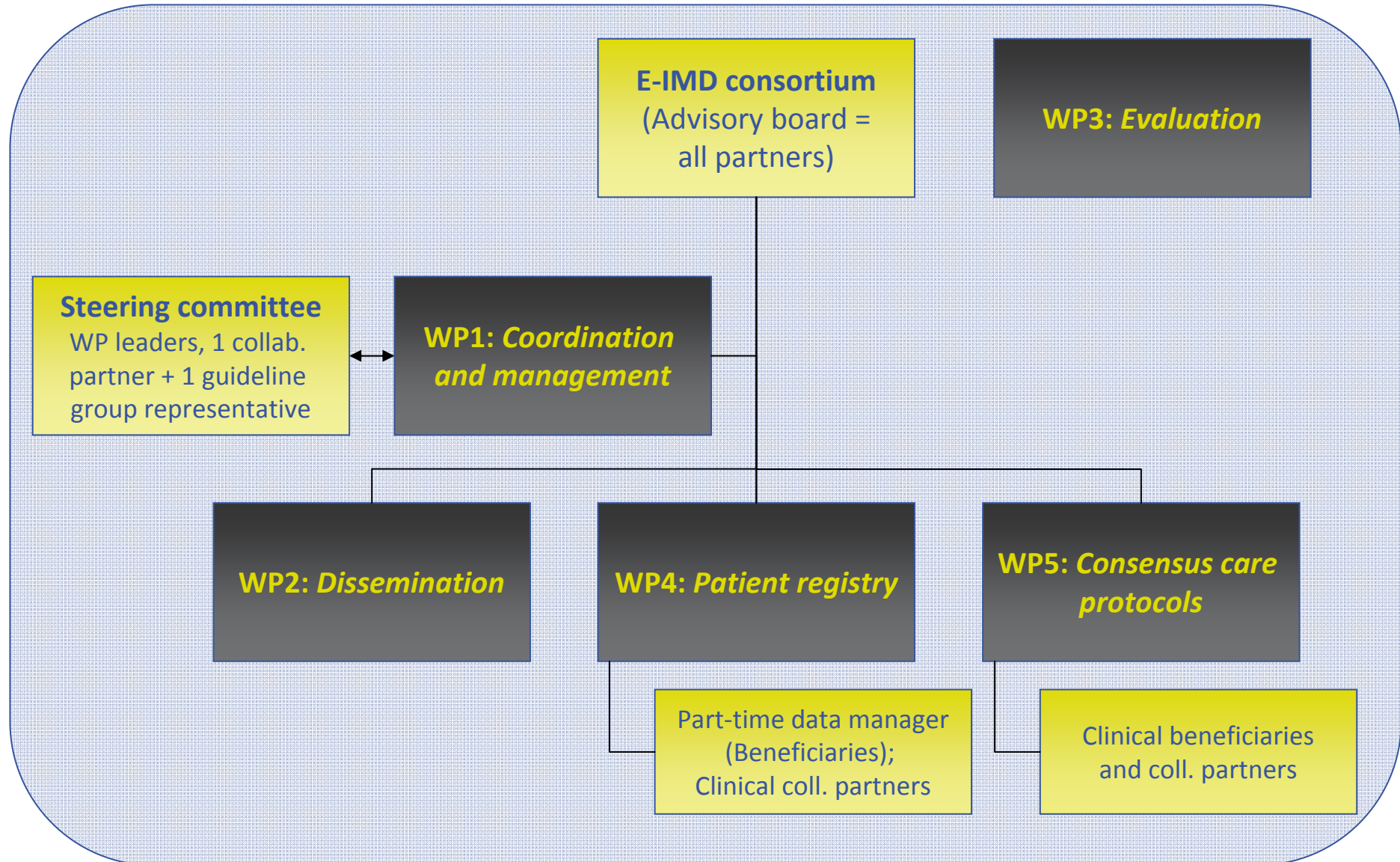
1. European patient registry: to describe the natural history, epidemiology, current diagnostic and therapeutic strategies, to provide information to national and EU healthcare authorities

→ improving knowledge; evaluating current strategies

2. European evidence-based consensus care protocols provided via a website in official EU languages

→ providing structured information for patients, parents and healthcare professionals, improving care, providing a template for national guidelines and patient brochures

E-IMD: Management structure



WP2: E-IMD website



<http://www.e-imd.org/>

WP4: E-IMD patient registry



<https://www.eimd-registry.org/>

(WP2): Training courses



Orphan Europe Academy
E-learning hyperammonemia module
Online by 1 December 2011

Where does the project stand now? – 1

Coverage and development process of the network

1. Start of the project (Jan 1, 2011):

Continents: **1**

Countries: **15**

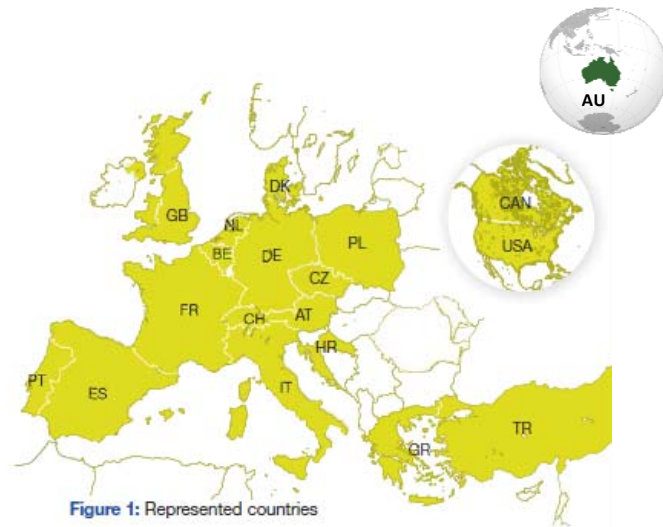
Partners: **27**

a) Coordinator/Associate: **13**

b) Collaborating: **15**

Where does the project stand now? – 1

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2. Today (Oct 26, 2011):

Continents: **3 (+ 200%)**

Countries: **20 (+ 33%)**

Partners: **47 (+ 74%)**

a) Coordinator/Associate: **13**

b) Collaborating: **34 (+ 127%)**

Where does the project stand now? – 2

Dissemination and outreach to healthcare professionals and patient organizations

Patient organizations

- Eurordis, BOKS (Belgium), Kindernetzwerk (Germany), KRIKOS ZOIS (Greece), VKS (Netherlands), CLIMB (United Kingdom)

National and international Societies for Inherited Metabolic Diseases

- International: SSIEM, SIMD
- National: in partner countries

Scientific consortia

- Urea Cycle Disorders Consortium (UCDC) within the Rare Diseases Clinical Research Network (RDCRN), USA

Future perspectives

- Harmonization of E-IMD and UCDC registries → transatlantic collaboration
- Therapeutic development (orphan drugs) and postmarketing disease registry for intoxication type metabolic diseases
- Revision and further improvement of evidence-based guidelines for diagnosis, therapy and long-term management
- Standardization of care for patients with intoxication type metabolic diseases in Europe
- Interdisciplinary, certified training for healthcare professionals