

Table Of Content

| | |
|---|---|
| European registry and network for Intoxication type Metabolic Diseases | 2 |
| Summary | 3 |
| Coordinator, Leader contact and partners | 5 |
| Great Ormond Street Hospital for Children NHS trust | 5 |
| Birmingham Children’s Hospital NHS Foundation Trust | 5 |
| Sveučilište u Zagrebu, Medicinski fakultet | 5 |
| Klinisk Genetisk Afdeling Rigshospitalet | 5 |
| Orphan Europe Sarl | 5 |
| Centro de Investigación Biomédica En Red de Enfermedades Raras | 5 |
| Assistance Publique-Hôpitaux de Paris, Hôpital Necker-Enfants Malades | 5 |
| Azienda Ospedaliera di Padova | 5 |
| Academisch Medisch Centrum | 5 |
| Instytut “Pomnik-Centrum Zdrowia Dziecka” | 5 |
| Hospital de São João, EPE | 5 |
| Centre Hospitalier Régional et Universitaire de Lille – CHRU de Lille | 5 |
| Outputs | 8 |
| D02 - Launch website and electronic newsletter (EN) | 8 |
| D01 - E-IMD promotional leaflet (EN) | 8 |
| D03 - Interim report 1 (EN) | 8 |
| D04 - Interim report 2 (EN) | 8 |
| D05 - Training course (Orphan Europe Academy) (EN) | 8 |
| D06 - European patient registry of OAD and UCD cases (EN) | 8 |
| D07 - Annual meetings (EN) | 8 |
| D08 - Final report (EN) | 8 |
| D09 - Consensus care protocols and information brochures (EN) | 8 |
| D10 - Evaluation of process and outcome indicators (EN) | 8 |
| Final report (EN) | 8 |
| Internal Assessment Note (EN) | 8 |

European registry and network for Intoxication type Metabolic Diseases

JA2015 - GPSD [705038]

START DATE: 01/01/2011

END DATE: 01/05/2014

DURATION: 40 month(s)

CURRENT STATUS: Finalised

PROGRAMME TITLE: Second Programme of Community action in the Field of Health 2008-2013

PROGRAMME PRIORITY: -

CALL: Promote Health (Hp-2010)

TOPIC: PROMOTE HEALTH (HP-2010)

EC CONTRIBUTION: 779746 EUR

KEYWORDS: Diagnostic, Health system, Patients registries, Quality evaluation, Rare diseases and disorders

PORTFOLIO: Rare diseases

General objectives

The overall aim of the European registry and network for Intoxication type Metabolic Diseases (E-IMD) is to promote health for individuals affected with rare organic acidurias (OADs) or urea cycle defects (UCDs). E-IMD has two specific objectives:

(1) To establish a European patient registry describing the disease course, epidemiology, diagnostic and therapeutic strategies for OADs and UCDs and to provide information to national and EU healthcare authorities. Anonymised data collection via a web-based password-protected EU registry will be based on routine follow-up parameters in 15 EU countries.

(2) To provide European evidence-based consensus care protocols for patients with OADs and UCDs. Based on the largest available collection of patient data (see objective 1) and a systematic literature search, a European consensus group will describe the best evidence available for the diagnosis and treatment. Consensus care protocols will be translated into official EU languages, provided via the E-IMD website and serve as a template for national guidelines and patient brochures.

Strategic relevance and contribution to the public health programme

E-IMD focuses on patients affected with one of the ten OADs or UCDs. Each of these intoxication type metabolic diseases has an estimated prevalence of 1 in 50,000 to 200,000 newborns within the EU. Given the particularly low number of patients and the complexity of the diseases, no national or regional project in Europe would be able to perform this work. The registry will bring together physicians, carers, scientists, patients and industry from at least 15 European countries. The countries with fewer resources will benefit from those with more resources i.e. access to consensus guidelines in their own language. It will map onto the evolving national rare disease plans and is complementary to existing rare disease networks such as Orphanet, CEMARA and CIBERER. In accordance with the second Health Programme E-IMD promotes health, improves quality of life, and reduces health inequalities for patients with rare IEM. It provides best scientific evidence, empowers patients and their families and strengthens the Community's status in the field of rare IEM.

Methods and means

E-IMD will set up the first web-based password-protected EU registry for OADs and UCDs entering relevant parameters of patients with the 4 most common OAD (methylmalonic, propionic, isovaleric aciduria, and glutaric aciduria type I) and 6 UCD (inherited deficiency of N-acetylglutamate synthase, carbamoylphosphate synthase 1, ornithine transcarbamylase, arginase, argininosuccinate synthase and lyase). This provides a unique basis for improving our knowledge and understanding of the

natural history of these diseases and their impact for patients and their families, and for evaluating patient care and outcome. Differences between centres and countries will be evaluated, and actions taken to address inequalities i.e. provision of advice and consensus care protocols. The registry and systematic literature search are important sources to provide evidence-based up-to-date information and consensus care protocols to patients and their families as well as health care professionals. Consensus care protocols will be developed according to the Scottish Intercollegiate Guidelines Network (SIGN).

Expected outcomes period

The overall intended impact is to improve access to rapid diagnosis and care for patients with OADs and UCDs; the E-IMD philosophy is that all patients in all MS should have an equal right to the best up-to-date care.

E-IMD will help make this happen by

- 1) objectively measuring and evaluating current management strategies and outcome of patients in at least 15 European countries which is an important basis to better understand the natural history of these rare diseases, to optimize current treatment strategies and to initiate cohort clinical studies,
- 2) providing evidence-based and consensus-agreed diagnostic and management protocols which will increase the transparency for patients and experts, will help to harmonise current diagnostic and therapeutic approaches, and will be adapted to specific country infrastructures, and
- 3) empowering patients and patient organisations by providing up-to-date information in their own language.

COORDINATOR



Universitätsklinikum Heidelberg (Universitätsklinikum Heidelberg)

Im Neuenheimer Feld 672
69120 Heidelberg

Germany

PARTNERS



Great Ormond Street Hospital for Children NHS trust

Street: Great Ormond Street
City: UK-WC1 3JH London

Country: United Kingdom



Birmingham Children's Hospital NHS Foundation Trust

Street: Steelhouse Lane
City: UK-B4 6NH Birmingham

Country: United Kingdom



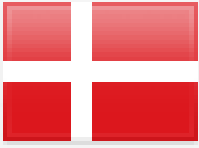
Sveučilište u Zagrebu, Medicinski fakultet

Street: Šalata 3
City: HR-10000 Zagreb

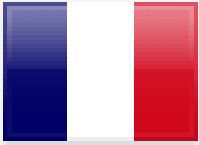
Country: Croatia

Klinisk Genetisk Afdeling Rigshospitalet

Street: Blegdamsvej 9
City: DK-2100 Copenhagen



Country: Denmark



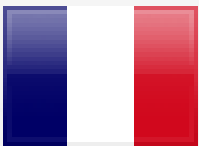
Orphan Europe Sarl
Street: Immeuble "Le Wilson"70, avenue du Général de Gaulle
City: F-92058 Paris La Défense

Country: France



Centro de Investigación Biomédica En Red de Enfermedades Raras
Street: C/ Álvaro de Bazán, 10 Bajo
City: E-46010 Valencia

Country: Spain



Assistance Publique-Hôpitaux de Paris, Hôpital Necker-Enfants
Malades
Street: 149, Rue de Sèvres
City: F-75015 Paris

Country: France



Azienda Ospedaliera di Padova
Street: Via Giustiniani 1
City: I-35128 Padova

Country: Italy



Academisch Medisch Centrum
Street: Meibergdreef 9
City: NL-1005 AZ Amsterdam

Country: Netherlands

Instytut "Pomnik-Centrum Zdrowia Dziecka"



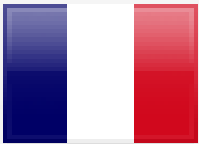
Street: Al. Dzieci Polskich 20
City: PL-04-730 Warsaw

Country: Poland



Hospital de São João, EPE
Street: Alameda Prof Hernâni Monteiro
City: P- 4202-451 Porto

Country: Portugal



Centre Hospitalier Régional et Universitaire de Lille – CHRU de Lille
Street: Rue Eugène Avinée
City: F-59037 Lille

Country: France

D02 - Launch website and electronic newsletter (EN)

Universitätsklinikum Heidelberg

European registry and network for Intoxication type Metabolic Diseases (E-IMD)

Published on: 01/01/2011

http://azorina.cc.cec.eu.int:8082/publications/20101201/20101201d02-00_nw_lgb_ps.pdf

D01 - E-IMD promotional leaflet (EN)

Universitätsklinikum Heidelberg

European registry and network for Intoxication type Metabolic Diseases (E-IMD)

Published on: 01/01/2011

http://azorina.cc.cec.eu.int:8082/publications/20101201/20101201_d1-00_lft_en_ps.pdf

D03 - Interim report 1 (EN)

Universitätsklinikum Heidelberg

European registry and network for Intoxication type Metabolic Diseases (E-IMD)

Published on: 01/01/2011

http://azorina.cc.cec.eu.int:8082/publications/20101201/20101201_d3-00_en_is.pdf

D04 - Interim report 2 (EN)

Universitätsklinikum Heidelberg

European registry and network for Intoxication type Metabolic Diseases (E-IMD)

Published on: 01/01/2011

http://azorina.cc.cec.eu.int:8082/publications/20101201/20101201_d4-00_en_is.pdf

D05 - Training course (Orphan Europe

Academy) (EN)

Universitätsklinikum Heidelberg

European registry and network for Intoxication type Metabolic Diseases (E-IMD)

Published on: 01/01/2011

http://azorina.cc.cec.eu.int:8082/publications/20101201/20101201d05-00_ot_h_gb_ps.pdf

D06 - European patient registry of OAD and UCD cases (EN)

Universitätsklinikum Heidelberg

European registry and network for Intoxication type Metabolic Diseases (E-IMD)

Published on: 01/01/2011

http://azorina.cc.cec.eu.int:8082/publications/20101201/20101201d06-00_ot_h_gb_ps.pdf

D07 - Annual meetings (EN)

Universitätsklinikum Heidelberg

European registry and network for Intoxication type Metabolic Diseases (E-IMD)

Published on: 01/01/2011

http://azorina.cc.cec.eu.int:8082/publications/20101201/20101201d07-00_ot_h_gb_ps.pdf

D08 - Final report (EN)

Universitätsklinikum Heidelberg

European registry and network for Intoxication type Metabolic Diseases (E-IMD)

Published on: 01/01/2011

http://azorina.cc.cec.eu.int:8082/publications/20101201/20101201d08-00_ffr_gb_ps.pdf

D09 - Consensus care protocols and information brochures (EN)

Universitätsklinikum Heidelberg

European registry and network for Intoxication type Metabolic Diseases (E-IMD)

Published on: 01/01/2011

http://azorina.cc.cec.eu.int:8082/publications/20101201/20101201d09-00_lft_gb_ps.pdf

D10 - Evaluation of process and outcome indicators (EN)

Universitätsklinikum Heidelberg

European registry and network for Intoxication type Metabolic Diseases (E-IMD)

Published on: 01/01/2011

http://azorina.cc.cec.eu.int:8082/publications/20101201/20101201d10-00_ev_r_gb_ps.pdf

Final report (EN)

Universitätsklinikum Heidelberg

European registry and network for Intoxication type Metabolic Diseases (E-IMD)

Published on: 01/01/2011

http://azorina.cc.cec.eu.int:8082/publications/20101201/20101201_d08-00_ff_r_gb_ps.pdf

Internal Assessment Note (EN)

Universitätsklinikum Heidelberg

European registry and network for Intoxication type Metabolic Diseases (E-IMD)

Published on: 01/01/2011

<http://azorina.cc.cec.eu.int:8082/publications/20101201/consensus-e-imd-final.pdf>