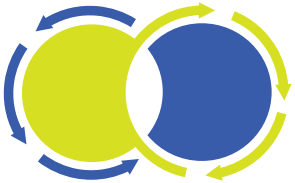


EHOD

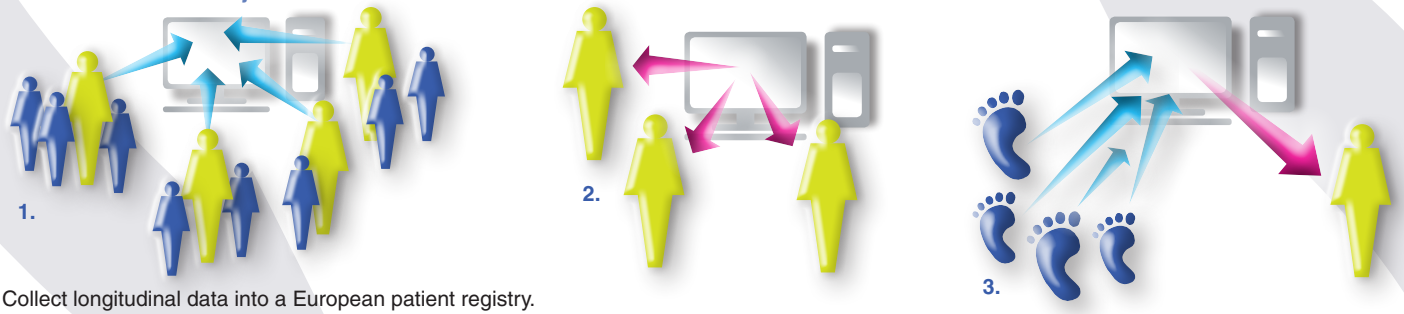
European Network and Registry for
Homocystinurias and Methylation Defects

www.e-hod.org

What is E-HOD?

E-HOD is the acronym for the European network and registry for homocystinurias, methylation defects and folate defects.
Its aim is to improve the health of children, adolescents and adults affected with these rare disorders.

The network has three major activities



1. Collect longitudinal data into a European patient registry.
2. Develop European evidence-based consensus diagnostic and clinical care protocols.
3. Evaluate the different newborn screening programmes for homocystinurias in member states and produce a position paper.

E-HOD extends the existing E-IMD (www.e-imd.org) project on urea cycle defects and organic acidurias by using the existing registry technology and network and extending the disease spectrum to 26 diseases.

About homocystinurias, methylation and folate defects

Homocystinurias, methylation and folate defects are groups of rare intoxication type inborn errors of metabolism with overlapping phenotype. The main types are:

Homocystinurias

- Cystathionine beta-synthase deficiency (CBS)
- Methylenetetrahydrofolate reducase deficiency (MTHFR)
- Combined homocystinuria and methylmalonic aciduria type C (CblC)
- Combined homocystinuria and methylmalonic aciduria type D (CblD)
- Methionine synthase reductase deficiency (CblE)
- Combined homocystinuria and methylmalonic aciduria type F (CblF)
- Methionine synthase deficiency (CblG)
- Combined homocystinuria and methylmalonic aciduria type J (CblJ)

Methylation defects

- Methionine adenosyltransferase deficiency I/III (MAT)
- Glycine N-methyltransferase deficiency (GNMT)
- S-Adenosylhomocysteine hydrolase deficiency (SAHH)
- Adenosine kinase deficiency (ADK)

Folate defects

- Methylenetetrahydrofolate dehydrogenase deficiency (MTHFD)
- Hereditary folate malabsorption (GFT)
- Formiminotransferase deficiency (FTCD)

About 1500 patients with CBS are known in the EU.
Other homocystinurias and methylation defects are very rare; about 100 patients in the EU. Individuals may present in infancy and risk reduced quality of life and life expectancy.

Patients are rare: experts are rare

- Because homocystinurias, methylation and folate defects are rare, centres often base care on their own limited experience and no evidence-based guidelines exist. Therefore, knowledge, experience and expertise need to be shared and centralised at an international level.
- Patients' access to diagnosis, newborn screening programmes, expert care and treatment varies within Europe. Information, treatment options and where to obtain appropriate care needs to be made readily available to patients.

The network

E-HOD has more than 30 partners from 21 countries. It has established links between healthcare professionals, patients' representatives and industry within Europe and globally. E-HOD will continue to expand its network by inviting new members in particular from underrepresented countries.

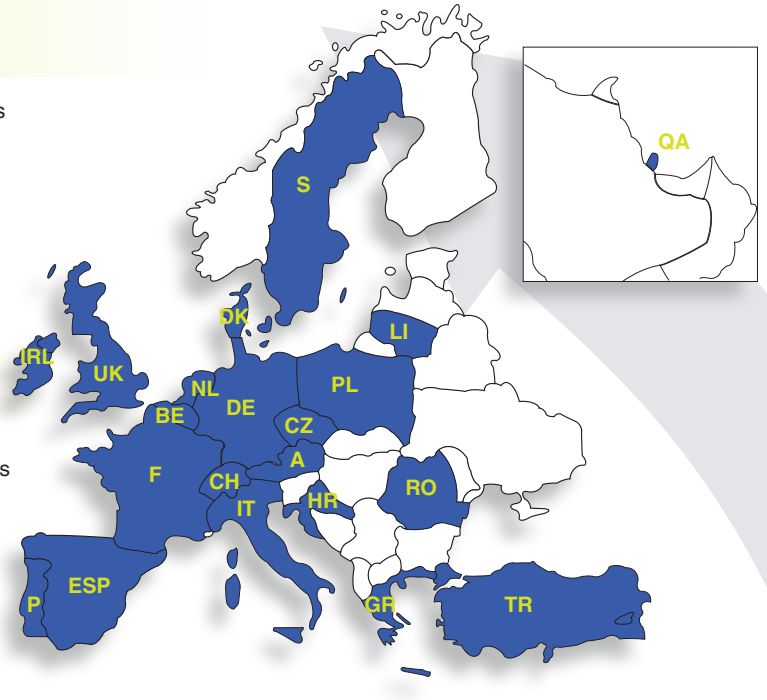


Figure: countries represented on the E-HOD network

Expected outcomes

The overall intended impact is to improve access to rapid diagnosis and optimal care for patients with homocystinurias, methylation defects and folate defects; the E-HOD philosophy is that all patients in Europe should have an equal right to the best up-to-date care.

E-HOD will help make this happen by

1. Objectively measuring and evaluating current management strategies and outcome of patients
2. Providing evidence-based and consensus-agreed diagnostic and management protocols
3. Evaluating the newborn screening programmes on homocystinurias in Europe
4. Empowering patients and patient organisations by providing up-to-date information in their own language
5. Improving sustainability of the registry and network by clustering rare inborn errors of metabolism onto one platform and collaborating with other European and international initiatives.

Getting involved

Please contact the coordinator: Henk Blom (h.blom@vumc.nl) and the project manager Marike Groenendijk (marikegroenendijk@me.com)

Coordinator and associated partners

Organisation legal name	Contact person	City	Country
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Landeskrankenhaus Bregenz	Martina Hümer	Bregenz	Austria
Sveučlište u Zagrebu, Medicinski fakultet	Ivo Baric	Zagreb	Croatia
Univerzita Karlova v Praze, 1.lékařská fakulta	Viktor Kožich	Prague	Czech Republic
Orphan Europe	Samantha Parker	Paris La Defense	France
Assistance Publique - Hôpitaux de Paris	Hélène Ogier de Baulny	Paris	France
Universitätsklinikum Heidelberg	Stefan Kölker	Heidelberg	Germany
Children‘s University Hospital	Ellen Crushell	Dublin	Ireland
Ospedale Pediatrico Bambino Gesù	Carlo Dionisi-Vici	Rome	Italy
UMC St Radboud	Mirian Janssen	Nijmegen	Netherlands
Faculdade de Farmacia da Universidade de Lisboa	Isabel Tavares de Almeida	Lisboa	Portugal
Centro de Investigacion Biomedica en Red de Enfermedades Raras (CIBERER)	Maria Luz Couce	Santiago de Compostela	Spain
The University of Manchester	Andrew Morris	Manchester	United Kingdom

Collaborating partners (as of August 1, 2013)

Organisation name	Contact person	City	Country
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Klinisk Genetisk Afdeling Rigshospitalet	Allan Meldgaard Lund	Copenhagen	Denmark
Hôpital Jeanne de Flandre CHRU	Dries Dobbelaere	Lille	France
Hôpital Necker Enfants Malade	Vassili Valayannopoulos	Paris	France
Universitätsklinikum Freiburg	Ute Spiekerkötter	Freiburg	Germany
Thessaloniki University A, Pediatric Department-Metabolic Laboratory	Persephone Augoustides Savvopoulou	Thessaloniki	Greece
Azienda Ospedaliera di Padova	Alberto Burlina	Padova	Italy
National Centre for Rare Disease Instituto Superiore di Sanita EPIRARE	Domenica Taruscio	Rome	Italy
Kaunas University of Medicine	Birute Burnyte	Kaunas	Lithuania
Volwassenen en Kinderen met Stofwisselingsziekten (VKS)	Hanka Meutgeert	Zwolle	Netherlands
Academisch Medisch Centrum	Fritz Wijburg	Amsterdam	Netherlands
Instytut Pomnik - Centrum Zdrowia	Jolanta Sykut-Cegielska	Warsaw	Poland
Hospital de Sao Joao, EPE	Elisa Leao Teles	Porto	Portugal
Hamad Medical Corporation	Tawfeg Ben-Omran	Doha	Qatar
Institute for Mother and Child Care „Alfred Rusescu“	Paula Avram	Bucharest	Romania
Karolinska Institutet	Anna Wedell	Stockholm	Sweden
Kinderspital Zürich, Universitäts-Kinderkliniken, Eleonoren-Stiftung	Matthias Baumgartner	Zürich	Switzerland
Istanbul University, Children‘s Hospital, Department of Nutrition and Metabolism	Mubeccel Demirkol	Istanbul	Turkey
Great Ormond Street Hospital	Stephanie Grünewald	London	UK
University of Newcastle TREAT-NMD	Kate Bushby	Newcastle	UK
Royal Hospital for Sick Children	Bernd Schwahn	Glasgow	UK
Royal Hospital for Sick Children	Siobhan O‘Sullivan	Belfast	UK
Birmingham Children‘s Hospital NHS Foundation Trust	Anupam Chakrapani	Birmingham	UK
Climb, National Information Centre for Metabolic Diseases	Steve Hannigan	Crewe	UK

This promotional leaflet arises from the project E-HOD (EAHC 2012 12 02) which has received funding from the European Union, in the framework of the Health Programme.

The EU is required by its founding treaty to ensure that human health is protected as part of all its policies, and to work with the EU countries to improve public health, prevent human illness and eliminate sources of danger to physical and mental health. The EU's strategy for protecting and improving human health is chiefly implemented through Commission health programmes.

For more information:

http://ec.europa.eu/health/programme/policy/index_en.htm

www.e-hod.org

European Commission

Executive Agency for Health and Consumers