

Third E-HOD Advisory Board meeting,

31st August 2015, 16:00 – 18:00 h

Lyon

Summary of proceedings

SUMMARY

The E-HOD advisory board meeting was held in the Lyon Convention Centre, one day before the SSIEM2015.

Forty-two network members, Industry- and Patient Organizations representatives attended. The meeting afforded the occasion to review and evaluate the objectives and deliverables of E-HOD, better understand all stakeholders' perspectives and to discuss the work to be performed until 15th May 2016 as the EC funding will end.

It is most important for partners to understand their role and the European funding mechanism. We have to focus on our deliverables. In case of any questions or lack of information project partners should seek help immediately from the coordinator or project manager. The immediate tasks are to initiate or continue the support of the ethical approval process of our collaborating partners. The next advisory board will be held before the SSIEM 2016 in Rome.

WELCOME NOTE (HENK BLOM)

COORDINATION AND MANAGEMENT E-HOD (HENK BLOM)

New Partners were welcomed. E-HOD has grown to a large international network with actively collaborating partners in US, Qatar, South America, Australia and Taiwan.

Henk Blom presented scope, objectives and an overview of the management of the project. Current EHOD projects are finalizing the recommended consensus guidelines, the translation of patient information, a systematic review of NBS approaches and the evaluation. Milestones and deliverables were presented.

Activities planned:

- Finalizing Guidelines – no further meetings
- Advisory board meeting - SSIEM2015 and SSIEM2016
- Evaluation Quality data 2015 – 2016
- Training course – 29th February – 2 March 2016 Prague
- Joint Patient – Expert meeting 29th February, Prague
- Evaluation E-HOD
- Amendment #4 and budget transfers
- Support collaborating partners to obtain ethical approval for the registry
- Continuing communication and dissemination

- **Preparation final report!!**

Steering group meetings are planned in December 2015, February 2016 and May 2016.
Final report is expected in July 2016.

DISSEMINATION OF KNOWLEDGE, PATIENT INFORMATION, WEBSITE (VIKTOR KOZICH)

Website www.e-hod.org has been developed, launched and the web-master updates the website on a weekly basis.

The number of visits on the E-HOD website from February – August 2015 are:

- 2733 visits
- 2401 users
- 84.16% bounce rate

Average time spent on the website is 2:38 minutes with an average page views of 2.84 minutes.

Annual reports have been published on the website and can be down-loaded. Publication on NBS has been published as open access. E-HOD leaflets were printed and distributed in the SSIEM 2014 conference bags. Patient information brochures are published and can be down-loaded from the website. Several poster presentations

E-HOD dissemination at conferences:

- SSIEM (Annual meeting of the Society for the Study on Inborn Errors of Metabolism), Barcelona 2013; Lyon 2015
- ERN discussion group, Netherlands, 25-26 Oct 2013;
- RADIZ (Summer school), Zurich, 2013;
- REACT (International Congress on Research of Rare and Orphan Diseases), Basel, 2014;
- 116th Annual meeting of the Japan paediatric society, Hiroshima, April 2013;
- RARE 2013, Montpellier, 28-29 November 2013
- CHAFEA Regional conference, Dec 2-3 2014, Prague
- International Rare Disease Day, Vilnius, 27 February 2015

E-HOD REGISTRY AND FIRST RESULTS (STEFAN KOELKER / FLORIAN GLEICH)

There is an update in the second activity report.

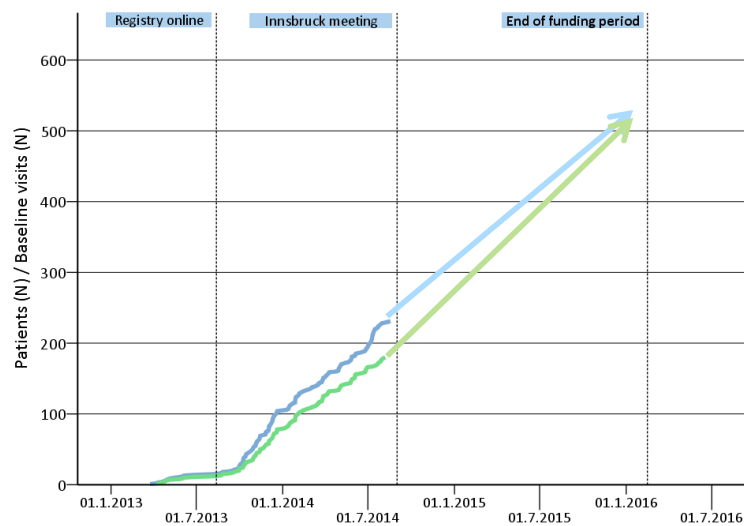
There will be an overview what topics need to be reviewed.

We should at least try to reach our goal of 600 patients before May 2016. Mid April 2016 will be the ultimate date to enter patient data into the registry. Until this date the data will be included in the first analysis of the data. Heidelberg needs time to analyse the quality of the data before the end of the EC funding period (15th May 2016)

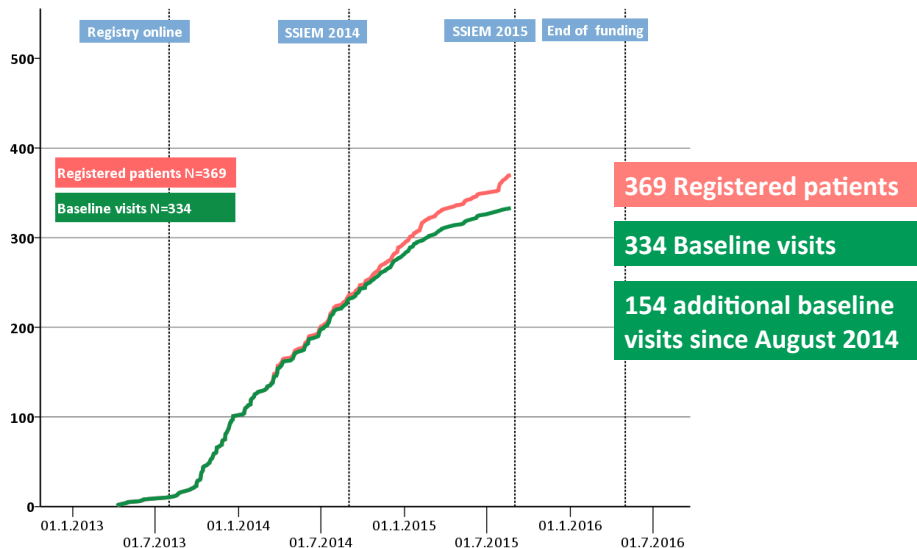
Two remarks:

1. take care we do not register patients more than one time into the register.
2. Enter base-line and regular visits. This will be a challenge, as most (adult) patients don't visit their metabolic specialist on a yearly basis.

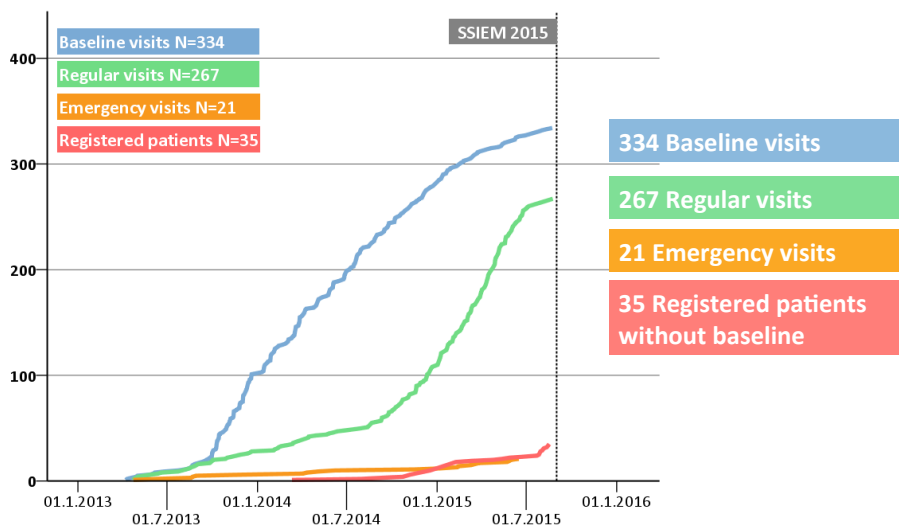
Continuing the trend



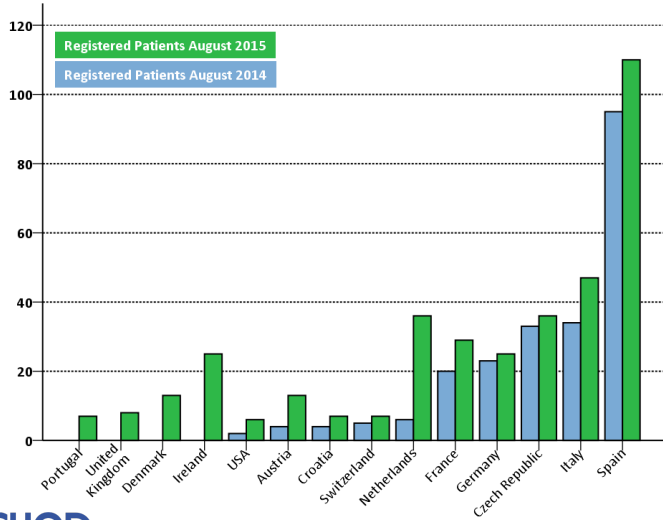
Progress of the registry (as of 24 August 2015)



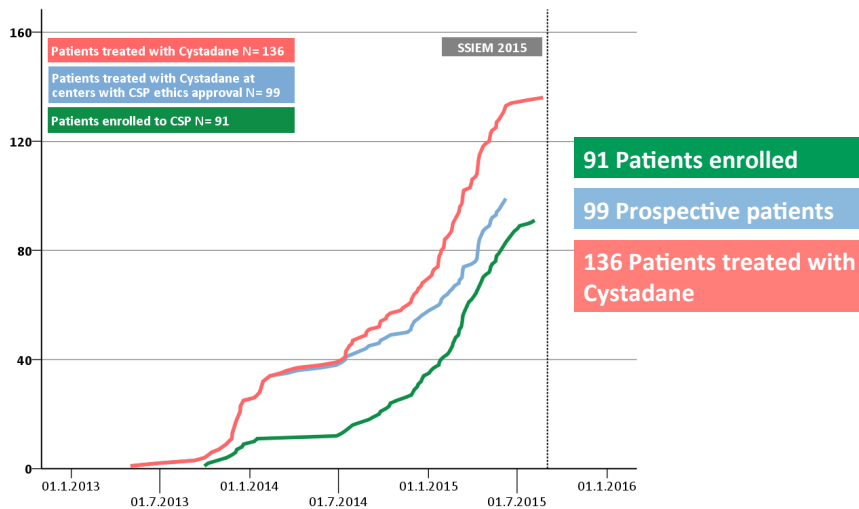
Progress of the registry (as of 24 August 2015)



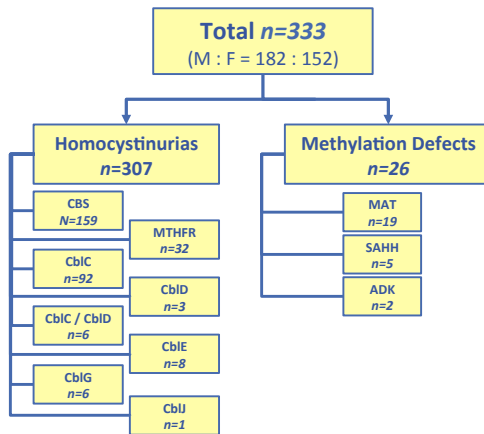
Geographical coverage of the registry



Progress of the Cystadane Surveillance Protocol



Emerging pattern of diseases



Data quality and areas of improvement:

Anthropometric data at birth

Missing Values, Medical History (Form B)		
	Complete data sets	
	Count	% of total
Gestational age	222	67
Weight at birth	205	61
Length at birth	157	47
Head circumference at birth	126	38

Dietary treatment

Missing Values, Dietary Treatment (Form E)		
	Complete data sets	
	Count	% of total
Special diet	527	88
Natural protein	198	79
Calories	112	44
AAM	208	83
Tube feeding	522	87
Fatty acids	514	86

Neuropsychological tests, behaviour and qol

Missing Values, Neuropsychological Development (Form H) and Quality of Life (Form I)		
	Complete data sets	
	Count	% of eligible patients
Behaviour questionnaire	42	16
PedsQL child report	12	15
PedsQL parent report	18	22
WHOQOL	19	13
Neuropsychological test	27	9

EMDA – THE EUROPEAN METABOLIC DISORDER ALLIANCE (MARIKE GROENENDIJK)

VKS, Climb and two other patient organisations have recently set up the European Metabolic Disease Alliance (EMDA). As to date there is a draft website www.eumda.org and the organisation has been launched summer 2015. Patient organisations reps of VKS and Climb have been invited for the

Advisory Board meeting to present the latest developments, but due to circumstances they were not able to attend.

There are two very active support groups on HCU and Methylation defects on Facebook. These groups have connected patients and parents of patients from all over the world with each other. Members can share experiences on treatment; diets and their daily lives with the group. When appropriate E-HOD shares actively information with these groups.

Support group members do follow E-HOD on Facebook and believe this is a very important project to improve health and better care for patients. HCU Network Australia has recently become E-HOD partner and Tara Morrison, founder and vice president of this patient organisation was happy to attend this E-HOD Advisory Board meeting. She was shortly introduced to the group.

Tara and Marike have explored the need for an international patient – expert meet and a draft agenda will be discussed with the RRD-Foundation after this Advisory Board meeting.

NEWBORN SCREENING (MARTINA HUEMER)

Tasks:

1. Guideline development
2. Review of current screening approaches and evaluation of NBS parameters

“Newborn screening for homocystinurias and methylation disorders: systematic review and proposed guidelines” has been published. Orphan Europe generously supported us to make it available as free access publication.

Monkey survey has been sent to all contributors of the registry to review the screening approaches:

1. Survey monkey questionnaire to all contributors to the registry to identify the NBS centres.
2. Questionnaire to NBS centres to identify the general screening approach regarding the homocystinurias
3. Identification of cases not identified by NBS from the registry

GUIDELINE DEVELOPMENT (ANDREW MORRIS, CARLO DIONISIS-VICI, IVO BARIC)

Group 1: CBS Guidelines development (Andrew Morris)

Andrew has presented the scope, purpose and methods of the CBS guideline development. The guideline development group includes individuals from all relevant professional groups (paediatricians, adults group, dieticians, biochemists).

During the 1st meeting of the guideline group questions were agreed and allocated. A list of relevant publications was prepared by Kimberly Chapman and put on the login and pass word protected area of the E-HOD website by Viktor Kozich’s team. Participants reviewed literature and prepared statement for the 2nd guideline meeting. On the 2nd meeting in Amsterdam we discussed and agreed the statements. Systematic methods were used to search for evidence (SIGN and GRADE)

All participants prepared supporting paragraphs based on literature search. During the 3rd meeting supporting paragraphs were agreed.

Completion

Participants have some outstanding tasks to do on supporting paragraphs

References need to be added to some supporting paragraphs and then formatted
Shortened version to be prepared and submitted for publication
Full guideline will be put on the E-HOD website as free access.

Group 2: **Cbl/MTHFR Guidelines development** (Carlo Dionisi-Vici). The semi-final draft, based on the results of the previous meetings held in Rome and Lyon, has been prepared by the two leading teams (Rome and the Zurich) at the end of November and will circulate among all co-authors by the end of the year for a final revision. The idea is to submit the manuscript to OJRD

Group 3: **Methylation Guidelines development** (Ivo Baric). Guidelines are prepared in draft. The group is waiting for 2 manuscripts to be accepted for publication. Goal: ready for publication in the beginning of 2016

Final remarks (Henk Blom)

Next advisory Board meeting will be held in September 2016 before the SSIEM meeting in Rome

List of participants

Persephone Augoustides-Savvopoulou

Ivo Baric

Matthias Baumgartner

Jean-Francois Benoist

Henk Blom

Avihu Boneh

Elisabeth Brachet

Peter Burgard

Kimberly Chapman

Lut De Baere - BOKS

Carlo Dionisi-Vici

Dries Dobbelaere

Ida Vanessa Doederlein Schwartz

Peter Freisinger

Florian Gleich

Marike Groenendijk

Martina Huemer

Mirian Janssen

Pavel Jesina

Stefan Koelker

Viktor Kozich

Cyril Martin – Orphan Europe

Allan Meldgaard Lund

Fanny Mochel

Andrew Morris- CMFT

Tara Morrison – HCU-Network Australia

Elaine Murphy

Tatiela Nalin

Markus Ott - Nutricia

Elisabetta Pasquini

Danijela Petkovic Ramadza

Celine Plisson – RRD-Foundation

Roland Posset – UKL-Heidelberg

Angeles Ruiz Gomez

Manuel Schiff

Anastasia Skouma

Christian Staufner

Beate Szczerbak - Nutricia

Vassili Valayannopoulos

Monique Williams

Barbara Wojcicka-Bartlomiejczyk

Wuh-Liang Wu