



European
Reference
Network

MetabERN

European Reference Network
for Hereditary Metabolic Disorders

Activity Report MetaBERN October 2018-February 2019

This Activity Report gives an overview of the work that has been done in the Work Packages and Subnetworks from October 2018 to February 2019 (Year 2 of MetabERN)

WORK PACKAGES

Work Package 1 Coordination & Management - Maurizio Scarpa, Cinzia Bellettato, Corine van Lingen (HCP), Lut Debaere & Anne Hugon (Chairs of the Patient Board)

Maurizio Scarpa, Coordinator of MetabERN, accepted the Direction of the Regional Coordination Center for Rare Diseases at the University Hospital in Udine, Italy. This is effective from January 2019. In the last months the Coordination Office (CO) has therefore worked hard on the amendment of the MetabERN grant agreement and related projects making sure that from 1st of March the Regional Coordination Center for Rare Diseases at the University Hospital in Udine, Italy is the new coordinating hospital taking over from Helios Kliniken in Wiesbaden (Germany). "The CO has organized Teleconferences with all the Work Packages and Subnetworks in this period to discuss the deliverables and the progress for year 2.

We have also organized a meeting on the Clinical Patient Management System that took place in January (Groningen, Netherlands). More information on that can be found under Work Package 5 in this report. The meeting on Clinical Guidelines that was planned for February 2019 had to be cancelled due to a low attendance rate. Furthermore, the CO worked in the internal monitoring system as well as started collecting the input for the end-of-year report that needs to be send to CHAFAEA/European Commission by the end of April following a strict template outline. The Annual meeting taking place in April 2019 has also been prepared and the agenda drafted in this period, with the help of a steering committee made up of HCPs and a patient representative.

Work Package 2 Dissemination - Mireia del Toro (HCP), Carlota Pascoal (Single Point of Contact Patient representative)

This Work Package has send out the monthly newsletters, kept the website up to date and managed the Facebook, LinkedIn and Twitter accounts. We would appreciate if all HCPs become followers of these accounts in order to increase our social media footprint. WP-2 has also developed two abstracts on MetabERN for the Word Congress in Orlando that took place in February. We are always looking for input and news for the newsletter so any information on this can be

send to Mireia del Toro and Simon Bond (mdeltoro@vhebron.net and simon.bond@metab.ern-net.eu). This Work Package is also working closely with WP7 on the dissemination of training and education webinars that are being organized.

Work Package 3 Evaluation -Viktor Kozich (HCP), Hanka Dekker (Single Point of Contact Patient representative)

Survey

The WP on Evaluation has finalized the Survey on HCPs needs and expectations that was sent out in the autumn of 2018. We received 61 responses (sometimes more than 1 per MetabERN Health Care Provider); this represents 70% of MetabERN-members that replied to the survey. We are currently analysing the results and we are aiming in having the results of this survey and the one on Patient's needs and expectations published in a peer-reviewed journal.

Relationship with Third Parties

The ERN Working Group on Legal & Ethical issues, relations with Stakeholders which is an ERN-overarching Working group is working with the Board of Member States to set up a Board statement on ERNs & Industry on clarify which activities are allowed (or not) for industry (or other private) support and possibly allowing for the exploration of "joint" funding mechanisms where several private partners could jointly support one (or even several) ERN(s). Since the Member States are the official 'owners' of the ERNs they need to agree and find consensus on this topic.

Monitoring:

The Monitoring and Quality Improvement Working Group has set up 18 indicators that were build following a systematic approach: a) establishing priorities, b) Methodology: Definitions, Indicators numerator/denominator, c) Discussion on the feasibility, utility and appropriateness, d) technical analysis of needed IT resources e) and capacity to develop the info system with the DG SANTE IT services The 18 selected indicators are covering the main areas of work of the ERNs (structure, process and outcomes). Data collection from 2017 and 2018 has started and the deadline is the end of March. The Coordination Office is collecting the data. MetabERN has also started internal monitoring of HCP involvement and engagement, which is measured according to providing input for reports, patient data collection, responses to surveys and other requests from the Coordination office.

Work Package 4 Guidelines/Care Pathways and Standardisation for Medical Care and Transition- Ursula Plöckinger, Carlo Dionis Vici, Maria Luz Couce, Leona Wagner (Single Point of Contact Patient representative)

Work Package 4 organised a meeting for the representatives of each Subnetwork to educate and inform them on how to set up Guidelines using the electronic platform that was designed by Ursula Plöckinger. Unfortunately the meeting had to be cancelled due to a low attendance rate. During the Annual Board meeting there will a parallel session organized for this purpose.

The electronic platform (Google Drive) is available and working and accessible for all those who have the link. If using the platform, make sure to read the manuals and step-by-step Guide and consult the person responsible for Guidelines in your SNW). The standardized forms of CPRs will be available soon.

Specific emphasis was directed to guarantee utmost transparency on the process of setting up CPRs, with respect to the literature used, the persons involved and the quality criteria (according to AGREE II and Grade). Detailed “how to do” as well as necessary forms are available as matrices facilitating the use of the platform. Two groups are active in using the platform for their first CPR.

Work Package 5 Virtual Counseling Terry Derks, Klaus Mohnike (HCP), Jean-Baptiste Farcet (Single Point of Contact Patient representative)

A meeting on the Clinical Patient Management System (CPMS) was organized on the 14th January in Groningen (Netherlands) aimed at 1 HCP per member states represented in MetabERN. The objectives of the meeting were:

1. To familiarise a group of MetabERN-members representing their EU member state with the CPMS;
2. To exchange views on CPMS between different stakeholders, based on experiences, survey outcomes, lessons-learned, steps-to-take and potential future functionalities, in particular for MetabERN.

The meeting had several important outcomes regarding the consent form and the need to have a close look at the enrolment page for patients. The European Commission and the Coordination Office are working together on addressing these issues. The full report of the meeting can be found on our [website](#).

In November 2018 this WP has applied for a grant for the INEA/CEF [TELECOM](#) call in which we made a proposal for the following 3 topics:

- 1) The development and implementation of metabolic data sets to improve the use of CPMS. This is enabled through disease-specific case descriptions with minimal data sets, which are essential to facilitate access to highly specialised knowledge and care for patients suffering from rare and severe diseases;
- 2) The development and implementation of case-oriented e-learning/e-training within MetabERN;
- 3) The generation of interconnectivity in the entire care chain between IEM-patients, primary care physicians, local hospitals and the centres of expertise to prevent metabolic emergencies.

Work Package 6 Research, Translational Activities and Clinical Trials-Maurizio Scarpa, Cinzia Bellettato, Jean-Michel Heard (HCP), Sylvia Sestini (Single Point of Contact Patient representative)

This Work Package has worked on a couple of studies being:

- Review of methods available to cross to blood-brain barrier: manuscript published, to read click [here](#).
- Research capabilities and activities in the network: two surveys have been planned; 1) one addressed to HCPs and 2) one addressed to patients organisations. The survey for HCPs is completed and analysed, a manuscript for publication is in preparation. Survey for patient's organizations is drafted and is also being prepared for publication in conjunction with the HCP survey.
- Clinical research on IMDs:
 - A mapping of on-going trials performed;
 - The survey on patient's perception of clinical trial sent to patient organizations;

- o A questionnaire on on-going clinical trials to be sent to HCPs is in preparation

A representative of this Work Package (Jean-Michel Heard) has participated in several meetings that were aimed at Research.

- EJP-RD: WP14 and WP20 (clinical expert task force);
- RARE2030: coordination of WP6;
- C4C: IMI2 RIA, WP20 on clinical trials;
- The European drug repurposing platform;
- The preparation of proposal: H2020-SC1-2018, 825990 - EU-China Health, (this proposal was unfortunately not granted)

All the information collected is meant to provide a broad description of the research landscape. Together with elements on innovative therapies showing how they are addressed by the clinical trials that are currently performed in the centres, and how they are concretely translated in the clinical practice when treatments have been approved by EMA. This is useful information to identify the challenges and disparities with regard to access to EMA approved medicines within EU Member States.

Title of the Event (if possible with hyperlink to website)	Place	Date	Type of contribution (Oral communication, abstract and poster), etc	Target audience	Estimated Number of participants
RD-ACTION, European Medicines Agency, and DG SANTE Workshop: How ERNs can add value to clinical research in rare	European Medicines Agency, London	29th - 30th May 2018	Oral communication	EMA officers ERN representatives	70
RARE2030 Kick off meeting	Fondation Universitaire EURORDIS Brussels	21-22 January 2019	Oral communication	Eu Parliament Eu Commission Representatives, participants to RARE2030	25
Interdisziplinäres Stoffwechsel-Colloquium Ein Auftrag des NAMSE* - Wir & Sie setzen es um - enge Kooperation zwischen Niederlassung und Zentrum – Auftaktveranstaltung *Nationales Aktionsbündnis für Menschen mit seltenen Erkrankungen	Interdisziplinäres Stoffwechsel-Centrum, Charite, Berlin	28.2.2018	Oral communication	Physicians	15
	Interdisziplinäres	25.4.2018	Oral communication	Physicians	8

Interdisziplinäres Stoffwechsel-Colloquium Splenomegalie – könnte es Morbus Gaucher sein? Was tun?	Stoffwechsel-Centrum, Charite, Berlin		ation		
Interdisziplinäres Stoffwechsel-Colloquium HyperCKämie, Muskelschwäche, Myalgie – kann es eine Glykogenose II (M. Pompe) sein? Was tun?	Interdisziplinäres Stoffwechsel-Centrum, Charite, Berlin	6.6.2018	Oral communication	Physicians	6
Interdisziplinäres Stoffwechsel-Colloquium Mukopolysaccharidosen (MPS) – von leichten Gelenkkontrakturen bis zur Multiorganerkrankung. Es geht nicht nur den Pädiater an	Interdisziplinäres Stoffwechsel-Centrum, Charite, Berlin	26.9.2018	Oral communication	Physicians	12
Interdisziplinäres Stoffwechsel-Colloquium Aminosäurestoffwechselstörungen am Beispiel der Phenylketonurie - eine lebensbegleitende medizinische Betreuung	Interdisziplinäres Stoffwechsel-Centrum, Charite, Berlin	24.10.2018	Oral communication	Physicians and dietitians	18

Work Package 7 Capacity-building and Education-Nadia Belmatoug (HCP), Margaret Guiliani (Single Point of Contact Patient representative)

WP7 has worked on the website by reviewing it and is waiting for the opinion of the representatives of the Subnetworks that is required.

Furthermore, this WP has worked closely with the SSIEM in setting up a programme for a webinar that will take place on Thursday the 25th of April 2019. The announcement can be found on the [website](#). The webinar will include among others a patient testimony. The second [webinar](#) will be held on 23 May (14.30 CET) with involvement of the team of François Eyskens Center of Inherited Metabolic Diseases CEMA UZA (Belgium). From now on, a webinar will be organized every 3 months.

Other work that is in progress is the questionnaire for patient organizations is prepared with collaboration of Margaret Guiliani (the Single Point of Contact Patient Representative for this WP), Lut Debaere and Anne Hugon (Chair and Vice-Chair of the MetabERN Patient Board) which will be disseminated to all Patient Organisations (POs) linked to MetabERN.

Work Package 8 Continuity of Care- Trine Tangeraas, Yngve Thomas Bliksrud (HCP), Anne-Grethe Lauridsen (Single Point of Contact Patient representative)

In this reporting period, WP8 has prepared and sent out a survey on “Diagnostic overview: Basis metabolic tests and enzyme analyses”. Work is in progress on a second survey on newborn screening and genetics. The purpose for the data that is collected by these surveys is that we are planning to publish a paper with the goal to get an overview of the diagnostic situation in Europe and potentially reduce gaps in diagnosis in the field of IMDs. We also aim to create networks to implement diagnostic capacity in countries where diagnosis is not available and to help patients to be better informed about diagnostics within the EU.

Subnetworks

Amino and organic acids-related disorders (AOA)- Henk Blom, Stefan Kölker, Francjan van Spronsen (HCP), Marie Devaux (Single Point of Contact Patient representative)

WP-1 Coordination and Management

- Telephone conferences with WP responsible persons and coordinators of specific tasks of AOA: 18th October 2018, 15th November 2018, 20th December 2018, 17th January 2019, 1st February 2019
- Major topics:
 - Strategies to improve activity of participants to improve collaborations with WPs, other SNWs and ERNs, and patient representatives.
 - Strategy to implement the U-IMD registry to support activities of SNWs.
 - Discuss annual AOA / MetabERN meeting and dissemination.

WP-2 Dissemination

Posters:

- 4th-7th September 2018: SSIEM Conference, Megaron Athens Conference Centre, Athens: *U-IMD: Unified European Registry for Inherited Metabolic Disorders as a patient database for MetabERN*
- 27th September 2018: Night of Research, Heidelberg / Mannheim (Germany): *Establishing of international patient registries for rare inherited metabolic diseases*

Oral presentations:

- 17th April 2018: Kick off meeting of ERN registries, HITEC building, Chafea, Luxemburg: *U-IMD – Unified Registry for Inherited Metabolic Disorders*
- 2nd September 2018: MetabERN meeting, Megaron Athens Conference Centre, Athens (Greece):
The U-IMD registry
- 13th November 2018: Annual E-IMD Members Meeting, Brussels:
U-IMD and collaboration with MetabERN and ERKNet
- 13th November 2018: Annual E-IMD Members Meeting, Brussels:
Evidence-based recommendations for isovaleric aciduria
- 13th November 2018: Annual E-IMD Members Meeting, Brussels:
Organ transplantation in individuals with urea cycle disorders and classic organic acidurias
- 13th November 2018: Annual E-IMD Members Meeting, Brussels:
Advantages and limitations of available nosologies for IMDs

Webpage:

Launching a webpage (<https://u-imd.org/>), to inform about the activity,

presentation of results of the U-IMD project.

WP-4 Guidelines

The following work has been done in this area:

1. **Organ transplantation for individuals with organic acidurias and urea cycle disorders:** Project partners have met during the SSIEM conference in Athens (4th September) to identify a strategy for describing and evaluating current practice of organ transplantation in these disease groups in Europe. A tentative list of research questions and variables has been established prior to the meeting.
2. **Recommendations for isovaleric aciduria:** First version was presented and discussed during the E-IMD Members Meeting (Brussels, 13th November 2018)
3. **Recommendations for methylmalonic and propionic aciduria:** Revision of the published recommendations
4. **Recommendations for phenylketonuria:** As part of the dissemination of the first European guidelines we have launched a project to write a “cook book” of the guidelines with practical help for all involved professionals caring for PKU patients and the patients themselves.
5. **Recommendations for tyrosinemia type 1:** Establishing a group of experts to work on the recommendations.

WP-6 Research, Translational Activity and Clinical Trials

- Establishing the U-IMD registry: This is the first IMD registry covering all (more than 1,000) known IMDs. It also includes about 30 IMDs with renal manifestation that will be studied in collaboration with the ERKNet consortium. The ERKNet and U-IMD registries are fully interoperable to facilitate collaboration on this specific topic. A beta version of the new U-IMD registry has been produced (Jan 2019).
- Natural history study for S-adenosylhomocysteine hydrolase deficiency: Literature search and collection of additional relevant information, identification of physicians who follow patients with this disease.

WP-7 Capacity building and Training

Provision of independent and professional education and training for healthcare professionals in collaboration with the *Recordati Rare Diseases Fondation d'entreprise* (<http://www.rrd-foundation.org/en/content/courses>):

1. Mitochondrial medicine 30 years on: state of the art, Nice, France, 11-13 April 2019
2. Inherited vitamin-responsive disorders: to the rescue of the neurodegenerative disorders, Paris, France, 27-29 June 2018
3. Modern management of inherited metabolic disorders, Rio de Janeiro, Brasil, 18-20 October 2018

WP-9 Patient Empowerment

Marie Devaux has disseminated information about a “Dietician Training”: a generic training (3/4 hours) that was developed in English to present to dietic students what are AOA IMDs with PKU specificities. It is essentially a visual support (very little text), based on an interactive format and the “learn by doing” principle. The first experiences in Toulouse were very positive. The Patient Representatives in Cyprus PRs are interested in reproducing this initiative.

Peroxisomal Disorders (PD)- Marc Engelen, François Eyskens (HCP), Jean-Baptiste Farcet (Single Point of Contact Patient representative)

WP-2 Dissemination

Title of the Event (if possible with hyperlink to website)	Place	Date	Type of contribution (Oral communicat ion, abstract and poster), etc	Target audience	Estim ated Num ber of partic ipants
Neonatal screening	UZA	January 14 and 18 2019	oral	Midwives, nurses	107
BOKS info	Brussels	February 2 2019	Oral: MetabERN and organisation of the care or rare metabolic disorders in Belgium	Patients and parents	100
Staff Pediatrics	UZA	February 22 2019	Red flags and awareness for peroxisomal disorders, in particular Zellweger spectrum dis	Trainees Child neurologists Pedatric staff	35

WP-4 Guidelines

Marc Engelen is working on a GL for X-ALD and François Eyskens on the Zellweger spectrum disorders

WP-6 Research

- CLSI guidelines on screening for X-ALD; guidelines for diagnosis and treatment of Fabry disease, screening for Gaucher disease in high risk population of patients with unexplained splenomegaly
- Clinical trial: Unigal (Protalix) for Fabry Bridge study

Carbohydrate, fatty acid oxidation and ketone bodies disorders (C-FAO)-Carlo Dionisi, Terry Derks, Ute Spielerkötter (HCP), Anne Hugon (Single Point of Contact Patient representative)

WP1 Coordination and Management

Attendance 4th Conference on European Reference Networks in Brussels on Nov 21-22

WP-2 Dissemination: Coordinated by the UMCG, we are half way with our Priority Setting Partnership for liver glycogen storage diseases. For more information, see www.igsdpsp.com.

WP-5 Virtual Counseling

Terry Derks has participated in the writing of the grant proposal for the 2018 CEF Telecom Call eHealth (CEF-TC-2018-4) and the organisation and co-chairing of the CPMS meeting in January (see WP-5 for more information)

WP-7 Capacity Building and Training

- Start of the PoLiMeR (Polymers in the Liver: Metabolism and Regulation) consortium Horizon 2020 - Research and Innovation Framework Programme (call: H2020-MSCA-ITN-2018).
- Colleagues are asked to disseminate this [Summer School Pediatrics](#) opportunity with biomedical students in their faculty. Dates for 2019 will be announced soon.

Lysosomal Storage Disorders (LSD)- Ans van der Ploeg, Giancarlo Parenti, Dominique Germain (HCP), Nuno Marques (Single Point of Contact Patient representative)

WP-1 Coordination and Management

This Subnetwork organised a successful first meeting of the LSD subnetwork (November 3rd, Rotterdam, The Netherlands).

WP-2 Dissemination and Teaching activities

- Continuing the keeping up to date of the MetabERN website and social media channels;
- Development of a MetabERN brochure;
- Poster presentation of the LSD Subnetwork on the 15th WORLD Symposium meeting, Orlando, 4-8 February 2019.

WP-4 Guidelines and Care Pathways and Standardisation, Transition

- A draft of Pompe disease guidelines has been finalized and is awaiting internal evaluation;
- Guidelines for NPC have been developed by a working group coordinated by Dr. Hiwot;
- A new working group for alpha-mannosidosis (coordinated by Prof. Hennermann) is now starting its activities;
- Proposals to select two new diseases to work on have been put forward and examined at the Rotterdam's Subnetwork meeting.

WP6-Research, Translational Activities and Clinical Trials

During the LSD meeting in Rotterdam, five research projects were pitched in which we can collaborate.

WP7- Capacity Building and Training

The results from survey on teaching activities per HCP were collected and put on the MetabERN website.

Pyruvate metabolism, Krebs Cycle defects, Mitochondrial oxidative phosphorylation disorders, disorders of thiamine transport and metabolism (PM-MD)- Shamima Rahman, Manuel Schiff, Enrico Bertini (HCP), Javier Perex Minguez (Single Point of Contact Patient representative)

No updated information has been given by this SNW for the moment.

Congenital disorders of glycosylation and disorders of

intracellular trafficking (CDG)- Peter Witters, Pascale de Lonlay, Tomas Honzik (HCP), Rita Francisco (Single Point of Contact Patient representative)

WP-2 Dissemination

- Scientific meeting EUROCDG organized by Gert Matthijs on 20 -21 March 2019 in Paris
- The 1st French CDG meeting with participation of the MetabERN leaders of CDG Subnetwork will be at the 22 March 2019 (see below the program) with a collaboration of patient Organizations: Rita Francisco (Portuguese CDG organisation) and French organizations
- Organization of the meeting in Barcelona on the 19 July 2019 (Families and Research)
- Retrospective paper on PMM2-CDG
<https://www.ncbi.nlm.nih.gov/pubmed/30293989>

WP-4 Guidelines

A first e-mail send to all MetabERN HCP's requesting them to work on the second guideline on the disease PMI-CDG.

WP-7 Capacity-building and training

- Elaboration of the first case report that will be broadcasted on the MetabERN education website
- Preparing slides and courses for WP7

Disorders of Neuromodulators and Other Small Molecules (NOMS) - Angeles Garcia Cazorla, Thomas Opladen, Eliane Sardh (HCP), Lut Debaere (Single Point of Contact Patient representative)

Title of the Event (if possible with hyperlink to website)	Place	Date	Type of contribution (Oral communication, abstract and poster), etc	Target audience	Estimated Number of participants
ANNUAL MEETING CIBERER (SPANISH NETWORK OF BIOMEDICAL RESEARCH IN RARE DISEASES) https://www.ciberer.es/xii-reunion-anual-ciberer	El Escorial, Madrid, Spain	12,13,14 March 2019	Oral "Cambio de paradigma en las enfermedades de los neurotransmisores: de los defectos bioquímicos de las monoaminas a la fisiopatología de la sinapsis" "Research in metabERN"	Members of the CIBERER, researchers	300
	Hospital	8	Oral	Families	150

II CONGRESO PARA FAMILIAS Y PROFESIONALES EN GLUT-1 http://www.aglutar.org/	Niño Jesús, Madrid	March 2019	"Aspectos clínicos poco descritos del déficit de GLUT1"		
6th SYMPOSIUM OF PEDIATRIC MOVEMENT DISORDERS https://www.childmovdisorderssymposium.com/	Hospital Sant Joan de Déu, Barcelona	7-8 February 2019	Oral Synaptic Metabolism: a new approach to inborn errors of neurotransmission	Physicians, researchers	300
EPNET General Assembly Meeting	Rotterdam	26-27th October 2018	Discussion EPNET / MetabERN collaboration	Physicians, researchers	150

WP-4 Guidelines BH4 and porphyria guidelines are going on. The BH4 guidelines could be finished during 2019

WP-6 Research:

- Research projects ongoing I-NTD: CSF proteomic studies in NT deficiencies; iPSc development in TH and BH4 defects, Brain Mapping.
- Grant applications EJP RD application on feb2019 (synaptic vesicle disorders)
- Purines research (animal model Lesch-Nyhan, ADSL new drug (patent), clinical trial in France)
- Phase1 study i acute intermittent porphyria (siRNA givosiran, published Feb 6 2019 in NEJM).
- Drug-drug interaction study in acute porphyria (givosiran), finalized, not yet published
- 3 clinical trials going on in acute porphyrias (ALN-AS1-002, EXPLORE and ENVISION)
- Glut-1: new drug under development, clinical-biochemical-response to treatment study CIBERER, Spain; APP for ketogenic diet monitoring, Santiago de Compostela, Spain); "voice characterization" in GLUT-1 (Hospital Sant Joan de Déu, Barcelona)

WP-7 Teaching courses:

- Paediatric Movement Disorders Course, Barcelona Feb 2019
- Yearly: iNTD meeting ; Ketogenic diet courses
- MDs and Students TRAINING: DU in metabolic diseases (France), Online Master in Metabolic Diseases (Santiago de Compostela, www.mastermh.es), Fellowship in Neurometabolic Diseases (online and practical training in 2019, Barcelona)
- GLUT-1 patients, researchers and physicians meeting. March 8, Madrid

WP-9 Patient Empowerment

This SNW is actively working with several patients' associations: Patients' associations: Lesch-Nyhan action French association, Spanish and Italian Lesch-Nyhan association, European Lesch-Nyhan association, AFMAP (porphyrias in France), RMP (porphyrias in Sweden), IPPN, the International Porphyria Patients Network (<http://www.ippn.info/>), DeNeu (Spanish NT), SSDAH German Association, German NT Association, AsGLUTdiece (GLUT-1 Spanish)

Patient Board

Representatives of the Steering Committee and Patient Board of MetabERN had a fruitful meeting in Brussels on 12 January 2019. During this meeting several important steps were taken in organising the patient representation side of our network. The participants set up an Action plan with clear priorities for the coming years, made a Calendar with important upcoming events and meetings.

Patient Board -Steering Committee January to March 2019

Activity

- ✓ 3 teleconferences with the Steering Committee
- ✓ 1 Patient Board (PB) meeting - MetabERN Patient Steering Committee and the Patient Executive

During the meeting we have worked on:

1. Patient governance structure
 2. Role of patient board and board bodies
 3. Working method
 - The general governance of the Patient Board was discussed and finalized.
 - Specific duties and responsibilities were clarified and generally approved.
 - Patient Executive Committee provided feedback on the Patient Action Plan 2019: Several key action points and responsible persons for their implementation were identified
 - The updated Patient Executive Committee represented a good opportunity to clarify the interaction between
 - Identify potential challenges of Single Points of Contact (SPOCs) and Work Package leaders that were standing in the way of collaboration
- ✓ The Patient Executive Committee carries the voice of all patient representatives involved in the Subnetworks (SNW) and Work Packages (WP) to the MetabERN healthcare professionals.

Patient Board bodies: The Steering Committee

Role:

- Develop an annual action plan with patient-focused outcomes
- Supervise the work in progress of the Patient Executive Committee



Lut Debaere



Anne Hugon



Hanka Dekker



Sylvia Sestini



Marie Balanca



Colin Brown



Rita Francisco

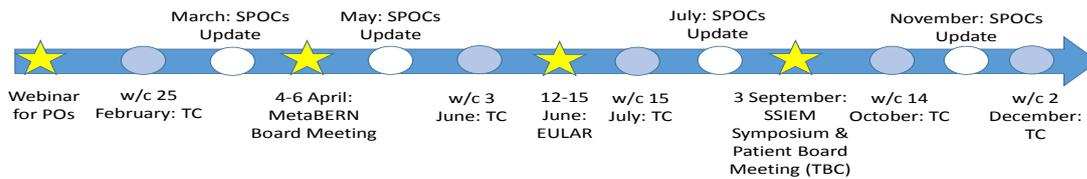


New Action Plan 4 Priorities defined

- Improve awareness of rare metabolic diseases and the role of MetabERN / WP2- Dissemination
- Improve MetabERN website content / WP2- Dissemination

- . Develop patient empowerment tools / WP9 Patient Empowerment
- . Foster access to cross-border healthcare services / WP2- Dissemination

The time Line



KEY ACTION POINTS

The Patient Board initiated the activities of the action plan recorded in January. MetabERN considers patient empowerment as one of the key features of patient-centered healthcare. Collaboration with patient organisations is instrumental for the design and delivery of programs and activities.

- . **Following the Governance of PB:**
 - o Refine and share list of all MetabERN patient organizations
 - o Verification of Letter of endorsement from patient organizations,
 - o The involvement and activity of the members.
 - o Relaunch if necessary, before opening to new members
- . **To insure a better communication inside the PB**
 - o Create/share a calendar with all upcoming events and activities (meetings, TCs, Reports)
 - o Create a social media calendar
 - o Make a planning/agenda to share activity reports every 8 weeks
 - o Reports form for SPOCS
 - o Setup WhatsApp Group for Steering Committee members
- . **Analyses of the website** to visibly redefine the place of Patients. The objective is allowing any sick person to provide the right information at the best place
 - o To provide support in the website revision and creation of a "Patient section"
 - o Include an editorial article developed by patient groups into the MetabERN newsletter
- . **Projets Started**
 - o Shooting of video to develop testimonials for website "Patient story"
 - o An outline has been developed for a patient leaflet explaining what MetabERN is from a patient's perspective
 - o Revise Dropbox to make sure that it is compliant with GDPR (develop a data protection policy, create secure files that contain all sensitive data)
 - o Patient input on clinical trials survey and Revision of the patient needs survey

MetabERN Board Meeting (4 April, Frankfurt)

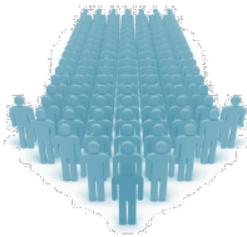
- ✓ Planning the organisation of the agenda of a face-to-face meeting for the Patient Executive Committee members attending the MetabERN

They set up a list of roles for the Patient Board of MetabERN:

- Advise on overall planning, assessment and evaluation of MetabERN activities and initiatives

- Contribute to the development and dissemination of patient information, policy, good practices, care pathways and guidelines
- Contribute to research by helping healthcare professionals to define areas that are important to patients and their families
- Provide support in the identification of other patient organisations to join the MetabERN Patient Board
- Share link to MetabERN website and circulate newsletter
- Contribute to editorial articles included in the newsletter

The Patient Executive Committee: Single Points of Contact (SPOCs)



The role of SPOCs is to:

- Be the entry point for the MetabERN healthcare professionals that need to interact with patient representatives
- Provide input on the patients action plan
- Interact with other SPOCs from the different subnetworks or Work Packages about ongoing and upcoming projects that may require their contributions
- Regularly report to the Steering Committee and inform them about the work in progress within the subnetworks or Work Packages
- Inform the Steering Committee about relevant events and achievements to be included in the newsletter

MetabERN Patient Governance Structure

Patient Board 1 Patient Representative per Patient Organisation			ERN
Steering Committee	Patient Executive Committee		ERN Coordinator Maurizio Scarpa MD, PHD
	Single Points of Contact for Subnetworks	Single Points of Contact for Work Packages	
Chair Lut De Baere	AOA Marie Devaux	WP 1 Lut De Baere	Vice Coordinator Stéfan Kölker, MD
Vice-Chair Anne Hugon	C-FAO Anne Hugon	WP 2 Carlota Pascoal	Vice Coordinator Ans Van Der Ploeg
Medical liaison Dr. Christina Lampe	LSD Nuno Marques	WP 3 Hanka Dekker	Staff Project Manager Cinzia Maria Bellettato Stakeholder Manager Corine Van Lingen Project Manager for the Patient Board Simon Bond
Hanka Dekker	PD Jean-Baptist Farcet	WP 4 Leona Wagner	
Colin Brown	PM-MD Javier Perez Minguez	WP5 Jean-Baptist Farcet	
Rita Francisco	CDG Rita Fransisco	WP 6 Sylvia Sestini	
Sylvia Sestini	NOMS Lut De Baere	WP 7 Margaret Guiliani	
Marie Devaux		WP 8 Anne-Grethe Lauridson	