



**European
Reference
Network**

for rare or low prevalence
complex diseases

 **Network**
Hereditary Metabolic
Disorders (MetabERN)

SCIENTIFIC REPORT



MetabERN

Annual Meeting with SSIEM

3 September 2018
Athens - GREECE



AGENDA

3rd September 2018

9:00-9.05	Opening remarks: Update on the situation of the Network <i>Maurizio Scarpa</i>
9.05-9.20	The patients' activity update <i>Lut de Baere</i>
9.20-9.45	Discussion on how patient representatives and WP/subnetwork coordinators might best work together
9.45: -10:00	The U-IMD Registry
10.00-10.15	Flashes on the Work Packages and Subnetworks achievements
10.15-11:00	Plans for the next 3 years and Internal Monitoring
11:00-11:30	Financial plan and potential projects to be funded
11.30-12.00	General discussion and wrap up
12.00	End of the meeting

MetabERN Kick-off Meeting - Participants

About 60 participants attended the meeting. Importantly, this year, among the participants there were also 14 Patients Representatives (PRs).

Unfortunately, due to personal and unexpected circumstances Maurizio Scarpa (the MetabERN coordinator) could not participate in the meeting and the discussion was therefore led by the two Vice-coordinators: Ans van der Ploeg (NL) and Stefan Koelker (DE). Cinzia Maria Bellettato (MetabERN project Manager) presented the slides on behalf of Maurizio. The presence, the general atmosphere was friendly, free-thinking and productive.

Opening remarks: Update on the situation of the Network

Ans and Stefan, opened the meeting and welcomed the participants. Cinzia then gave an overall presentation of the agenda of the meeting highlighting the importance of creating a collaborative team both at medical and patients level.

She announced that a new wave of technical assessments and audits is being carried out by the Andalusian Agency for Healthcare Quality (ACSA). /?? are the MetabERN HCPs undergoing technical assessment of their applications.

The importance of having all the members actively participating to the activities of the network in a responsible way and the need to use the CPMS was particularly stressed. Every HCP should be listed at least in a task of one of the WP.

Metab ERN has to justify and report on the multiple activities that take place so it is important to stress that we all have to participate in an active way to the different tasks as well as answering to mails and surveys. This will also be monitored as a quality measure for all HCPs. The surveys are very important tools for us when it comes to data collection and information gathering on which we base our future activities.

The patients' activity update

Ms. Lut De Baere, President of the Belgian Patient Organization for Rare Metabolic Diseases and chair of the MetabERN Patients Board highlighted the importance of ensuring the largest possible patient involvement in the network.

In doing so several challenges need to be addressed including:

- Managing and prioritisation of ideas
- Identification and use of adequate communication tools: Face to face meetings, TC, Webex etc.
- Setting of proper time-window for the discussion: Weekend ? Evening ?
- Finance

She also highlighted that it is sometime not easy to work with PRs because of

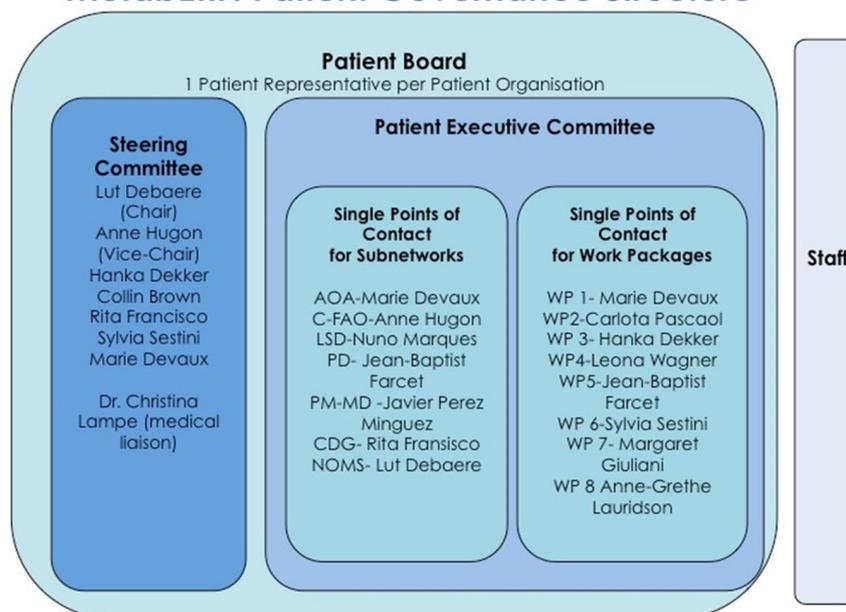
- the different level of experience;
- the different level of knowledge;
- the different culture, languages and time zones;
- Presence of paid and voluntary PRs,;
- Presence of small and big POs.

MetabERN Patience Governance

To achieve its goal and to facilitate patient's participation a governance structure had been defined described in detail.

All the PRs are being involved in the different activities of the MetabERN and in particular they will participate in the governance, ethics, care, research, and evaluation of the MetabERN. Names of the patient representatives who are designated to the different WPs and SNWs have been put in contact with the coordinators and leaders in order to start working together.

MetabERN Patient Governance Structure



- The establishment of a proactive collaboration with Eurordis: one of the major sponsor of the concept of ERNs
- The Collaboration with the 6 representative of the European Patient Advocacy Groups (ePAGs) elected by Eurordis
- The identification and mapping of all the known patient groups active in the area of rare inherited metabolic diseases (we have a database that gives an overview of the existing and known POs: 366 POs have been identified and divided per countries and subnetworks)
- The production of a list of international, national and regional family associations to engage with us.
- Sending out of a formal invitation letter to join the network.
- The sending of a letter to miss Theresa May, the Prime Minister of the United Kingdom, and UK government and to PM inside the EU Parliament regarding Brexit to ensure that a solution is found so that the UK healthcare providers can continue to participate in the ERNs and all EU patients can benefit from a cross border collaboration.

Activities

Activities that are being undertaken by the Steering Committee (SC) include:

- Understanding the needs of the patients
- Enlarging the participation of PRs
- Facilitating the collaboration with Subnetwork-coordinators and Work Package leaders
- Starting the organization of Disease Specific Units
- Collecting patients data from Patient Representatives (PRs)
- Empowering patients in terms of diagnosis and research

Projects 2018-2019 Projects that have been or are being developed by the SC include

- Facilitate the connection between SPOCs PRs (Single Points of Contact Patient Representatives) and resp. Leaders of SNW & WP (done)
- Leaflet (done)
- Travel list (proceeding)
- Knowledge mapped (tbc)
- Patient info on USB (tbc)
- QR bracelet or necklace (emergency) (tbc)

Activities 2018-2019

- Provision of advice
- Definition of Governance & Strategy of MetabERN
- Implementation of the Website
- Mapping of the expertise in EU to help patients to get much needed information about centres of expertise as well as stimulate doctors and caregivers to develop expertise further for the benefit of the patient.
- Proceeding with the list of International, European, National and umbrella organisations and organization of a Matrix file
- Engagement of POs to join the MetabERN
- Facilitate the inclusion of PRs in the SNWs & WPs
- Collaboration with Eurordis & ePAG members
- Development and sharing of a Patient Survey to all POs in 7 languages (data entry closed)
- Development and sharing of a Patient Newsletter (1st is sent out)
- Definition of an Action Plan

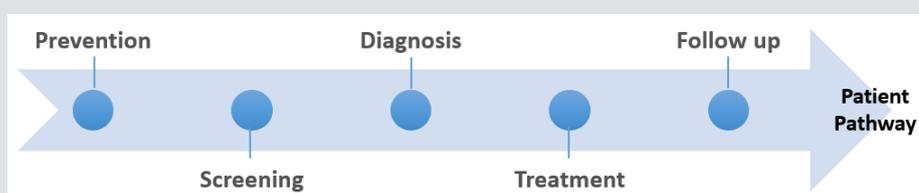
Lut concluded her talk highlighting that MetabERN is a unique opportunity to positively impact the life of patients and the management of their diseases. PRs will be actively involved in the different activities of the network. We have a formal proactive collaboration with EURORDIS to facilitate the collaboration. The Network provides an opportunity to understand and address the significant differences in the treatment and care for metabolic patients across Europe.

Next steps

- Increase the visibility of MetabERN on the website
- Facilitate Networking
- Completing the term of Reference
- Produce benefits for the patients via PRs and POs

Discussion on how patient representatives and WP/Subnetwork coordinators might best work together

Cinzia discussed strategies for Collaborating with patients to improve patient outcomes. Analysing the patients pathway the following idea for collaborating were proposed



Prevention and Screening

- Increase Awareness amongst general public and Health care professionals
- Dissemination of information on metabolic conditions (meeting, conferences, publication, websites, action toward health authorities, etc.
- Produce news and review existing health-related information

Screening and Diagnosis

- Shorten delay in diagnosis by promotion of:
 - MDT diagnostic expert consultation (CPMS)
 - Patient pathway monitoring (tracer patient method)
 - Access to specific diagnostic tools (genetics/ metabolites/enzymatic tests)

Diagnosis Treatment and Follow up

- Revision/development of Care pathways & Guidelines (GLs)
- Facilitation of Clinical Trials/research (defining research priorities and disseminating research-related information)
- Data collection
- Facilitation of the Regulatory process for medicines
- Participation in the production of White papers and Recommendations

Treatment/Follow Up

- Participation and provision of inputs and perspectives on
 - Education of patients
 - Instruction to GPs following the patients
 - Transition from childhood to adulthood
 - QoL surveys and outcomes measurement
 - Training of delegates

Critical Issues for the Forging of Effective Partnerships

It was highlighted that a **strong commitment and active participation is expected from all MetabERN Members**

The following issue have been listed as crucial for and effective partnerships

- Transparent information exchange
- Clear Management of relationships/Governance
- Ensure clarity about patients' roles as health team partners

Crucial issues

- Resources and funding
- Language and communication
- Culture and tradition of tokenism (the fact of doing something only to show that we are following rules or doing what is expected or seen to be fair, and not because we really believe it is the right thing to do)

Discussion

Patients Survey aimed to identify patient's needs and expectations with regard to MetabERN activities:

Cinzia highlighted the fact that although being a very general and easy survey taking only 10 mins, numbers of responders are still quite low. It cost a huge effort translating the questionnaires in different languages and more resources will be necessary to manually merge the results for a global analysis of all the data... Is it still convenient to translate the surveys in so many languages? Is it clear that we should try to get

higher participation, especially from the less responsive countries, do we need a better/different strategy?

Patients and HCPs are not a completely separate entity: they will be asked to interact with each other, work together and collaborate for the benefit of the patients. To this aim from now on PRs (preferably the SPOC) will be invited to take part in those TC that are pertinent/related to the WP/SNW of interest.

The U-IMD Registry

Stefan Kölker

U-IMD is the acronym for the Unified European Registry for Inherited Metabolic Disorders. The overall aim of this project is to promote health for children, adolescents and adults affected by rare Inherited Metabolic Disorders (IMDs) enabling and empowering patients, wherever they live, to access the necessary expertise and services and promoting research on IMDs and the development of safe and efficacious new treatments

The specific main objectives of the project are:

- Creation of a new IMD registry (U-IMD) as official registry of MetabERN
- Update of existing IMD registries for the inclusion of the same data elements (starting with iNTD, later on E-IMD and E-HOD)
- Developing a standard for minimal core data sets shared by the MetabERN/U-IMD and the European Rare Kidney Disease Reference Network (ERKNet) registry.

USERS For various reasons, including data protection, data quality, plausibility and case ascertainment as well as the challenge of administering large quantities of user accounts, access to the registry itself will not be direct for affected individuals with IMDs but mediated by the responsible treating clinics. However, patients can receive a print copy of their own data via the metabolic centre in charge, and the data model contains tools for capturing patient perspective that can be used by the patients directly. Such module for capturing the patient's perspective includes for example the PedsQL quality of life assessment, which already exists as validated version in many languages and was chosen because of this feature. Moreover U-IMD is explicitly designed as a learning system capable of integrating additional tools as needed including tools that are more sensitive to variance resulting from social and cultural factors. Patients have a say in this through their patient advocacy groups that are organized in MetabERN and therefore are part of the U-IMD management structure. U-IMD will be used by physicians treating patients with rare inherited metabolic disorders. Considering the diverse nature of the heterogeneous etiological and clinical spectrum of IMDs to be covered, in order to allow the comparison among key parameters intra- and inter-individually as well as across different diseases and disease groups, and to guarantee interoperability with other registries committed to the same standards the following solution have been adopted:

- Limitation of data collection to a minimal set of common data elements (CDEs)
- Use of controlled dictionaries for the description of the clinical phenotype and medication, as well as standardized tools for assessment of development, quality of life, and patient perspective.

NOSOLOGY USED BY THE U-IMD REGISTRY: U-IMD beneficiaries finally decided on employing the proposed nosology of the IEMbase (Ferreira et al. Genet Med 2018). Major arguments for this decision:

- High level of actuality and availability
- Satisfies the need for clear case definitions
- Existing linkages to MIM Codes, Orphanet Codes and HMDB Codes
- Prospect of cooperation between U-IMD, MetabERN and the IEM Database

ACCESS: U-IMD is a web-based patient registry. U-IMD will be accessible via the internet using password-protected user accounts and encrypted data transfer between server and client.

HOW TO JOIN U-IMD / HOW TO SUBMIT PATIENT DATA

- Contact the Coordinator (Prof. Stefan Kölker) with the expression of interest by email; for non-Members of MetabERN: your application will be evaluated by the U-IMD Steering Committee
- Sign the U-IMD letter of agreement
- Using the template approved by IRB at the University Hospital Heidelberg, prepare and submit ethics application for U-IMD to your local IRB (respecting national/local standards).
- Receive personalized access and start data entry

MetabERN is proud to provide multidisciplinary collaborations that include the leading experts in the field of inherited metabolic diseases. Besides their expertise MetabERN experts bring their already existing contacts to European scientific societies and to relevant stakeholders into the network.

More info at <https://u-imd.org/>

Flashes on the Work Packages and Subnetwork achievements

Coordination & Management-

Main activities consisted in:

- Setting up of an efficient governance system
- Organization of meetings/TC/ webinar to enable a smooth running of the cooperation, coordination, management and communication
- Submission of the Grant application for the Second Year grant in April (200.000 Euros)
- Submission of the First year Report on March (deadline 30/04/2018)
- Writing Plans for the 3rd to 5th year (Deadline September 13 of 2018)
- Close cooperation with other ERNs to collectively address the potential overlap of diseases and the designing of common strategies

It was also highlighted that MetabERN has the endorsement of SSIEM and that on going work consist in facilitating the exchange of knowledge and the establishment of specific training programmes

WHAT IS ASKED FOR: More help and responsiveness and filling in the surveys that are sent out!

Dissemination

The following items have all been set up, sent out and kept up to date since January 2018:

- Website (www.metab.ern-net.eu) with implementation of patient-oriented section;
- Social media channels (Twitter, Facebook, LinkedIn) ;
- Monthly newsletters;
- A patient brochure has been produced in English and French;
- A Press release was released in March 2017 and several publications in general and peer reviewed papers have been produced;
- Several Posters have been exhibited (among them 2 for the SSIEM);
- At least 36 Oral Presentations have been given.

WHAT IS BEING DONE:

- Paper on ERN as 24 ERNs group (this issue will be further discussed with Luca Sangiorgi (Coordinator of the BONE ERN) but should be ready by the end of the year;
- Sponsorship of meetings: e.g. International Mito Patients (IMP) workshop in the second half of this year.

WHAT CAN YOU DO:

- More collaboration;
- More followers for the social media and communication channel;
- More publications;
- We need to boost the use of the Logo and production of publication;
- We need courses and workshops to be endorsed by MetabERN;
- We need to develop SOPs (Standard Operating Procedures) to regulate the relationship and the collaboration with companies. In relation to information and organisation of courses two different situations were discussed: 1. Courses and Workshops that are private from the industry in one hand
2. Courses from different HCP or associations that are supported by the companies. We need to find a final agreement on this.

HOW TO CITE METABERN:

The following digitule ?? could be used for acknowledging MetabERN

- MetabERN can be included as Author's affiliation after the HCP information
- *Or at the end..This work is part of the activity of the European Reference Network for Rare Hereditary Metabolic Disorders (METABERN) - Project ID No 739543.*
- *If there is funding: MetabERN is partly co-funded by the European Union in the framework of the Third Health Programme "ERN-2016 - Framework Partnership Agreement 2017-2021.*

Research

- Several surveys have been and are going to be produced:
- **Access to treatment:** a survey for HCPs has been shared and is still open, another for patients will be sent out soon. The results will be used as a basis for a White Paper on this topic.
- **Clinical Trials;** surveys are under development
- Cinzia highlighted the lack of compliance: If we keep going with the trend of a low response rate we are not sure how we are going to be able to develop the White Paper so it is really important to spare a couple of minutes to answer the surveys.
- Participants and Vice-coordinators raise the issue that we are sending out too many surveys and that they are too long and too articulated. There should be a TC or meeting of different SNW and WP coordinators to prioritize the type and order of surveys.
- To partially solve the inconvenience a PDF version of the online survey will be shared with the participants in order to provide them an idea of the whole content.
- **WHAT IS ASKED:**

Education & Capacity Building

- More collaboration
- More active participation in the Development and revision process of the surveys

Available teaching and training programmes have been identified and are reported in the [educational session of the MetabERN website](#). This sub-site is dedicated to sharing information and documentation linked to training, education and capacity building.

Programme for the young generation are being discussed: to this aim it would be necessary to interact with DG research and DG education to see whether we can design a special curriculum for RDs to have a new medical generation.

Programme on the transition from childhood to adulthood is also being discussed (in collaboration with SSIEM and with the WP4)

Production of videos and materials oriented to increase awareness on all RDs is also being taken into consideration, but the following issue need to be addressed:

- Targets
- Languages
- Interaction with companies (E-G: Agilis Biotherapeutics offers to join efforts to raise awareness of AADC deficiency)

WHAT IS ASKED:

- Definition of SOPs to regulate the relationship with companies and third parties for educational issues as part of a global document that includes all issues regarded in other WP. Different models were proposed but it needs an important discussion regarding legal and ethical issues.
- Collaboration between WP4 and WP7 on the transition programme

Virtual Counselling/Clinical Patient Management System (CPMS)

Cinzia highlighted the fact that we do not have the possibility to proceed with traditional consultation system so we really need this secure system: the CPMS is the only way.

It was highlighted that **with the money from the CEF TELECOM CALLS 2018** we could hire an engineer/consultant to help and expand the use of CPMS in particular and e-Health generally and be able to transfer data in a easier way,

Klaus Mohnike and Terry Derks are working with the European Commission on integrating CPMS with the existing hospital systems. A Chili-Radiology pilot solution aimed to overcome the problems for data exchange between hospital IT and CPMS is under development.

We will need to create Protocols on how we do second opinion: hospital will need to be more engaged and be reimbursed for the use of the CPMS

WHAT IS ASKED:

- Implement the Use of the CPMS for the consultations
- HCPs need to be more active as the use of CPMS is also a quality measure of the activity of the ERN.

Clinical Pathways/Guidelines

Online accessible information platform on GLs and clinical pathway recommendations (CPR) is available. It contains the documentation necessary to produce GLs and CPRs.

GLs and CPRs already available for a number of selected IMDs.

Each disease-specific Subnetwork is working on developing GL/CPRs for the diseases they selected.

Maurizio is now the chair of the WG on Knowledge generation: Training, education, capacity building, GLs. This WG is guided by Enrique Terol (European Commission)

In the following weeks a survey on perception and use of the existing Guidelines and clinical pathways recommendations will be drafted.

Another survey on existing transition programmes per HCP to see whether members have a transition plan in their hospital is planned and will be developed soon. In this regards **a transition working group composed** by Nathalie Weinhold, Graziella Cefalo and Martina Huemer (as external collaborator responsible for gathering the data) on this will be established....(But there is another group led by Nadia..or it's the same?)

Diagnosis

Comprehensive understanding of what kind of diagnostic tests are currently available in Europe is under development: to this aim a detailed list of diagnostic technologies and services certified in each laboratory will be created will needs to be filled in by HCPs

A start has been made in setting up cooperation with ERNDIM and Orphanet to use functionalities from existing databases, in order to create an updated database/directory on metabolites and enzymatic tests performed in each lab in the member-labs of MetabERN.

The mitochondrial survey was resend in an attempt to capture as many centres as possible.

Evaluation and Monitoring

The working group of the Coordinators Group that has been working on producing monitoring indicators for all the 24 ERNs have presented the 18 indicators they have produced and will start testing them with real data in November.

The indicators were briefly introduced and it was highlighted that the use of the CPMS is an important one!

We have implemented the list of 18 indicators with some specific MetabERN-specific internal indicators and we are planning to start our own internal monitoring soon. HCPs will be evaluated using these indicators and in case of a low non-compliance rate can eventually be invited to step back if they not complying with MetabERN quality standards.

More info and the detailed list of the criteria will be shared in the next weeks.

Patient Involvement

In the 3.5 y plan of a new WP (WP9) has been included, It is specifically aimed to the Patient empowerment.

Plans for the next 3 years and Internal Monitoring

It was underlined that the Workplan for the 3rd - 5th year foresees:

For year 3:

- Continuation of data collection and the distribution of surveys and questionnaires
- Creation of specific WGs on selected topics (GLs, NBS, Research Treatment and CT, Patients empowerment etc.)
- Planning of Brainstorming meetings

For year 4:

- Further Elaboration of major outcomes and implementation of data collection (through surveys, CPMS etc)
- Planning and organization of Brainstorming meetings for the wrapping up of activities of the specific WGs, SNW, WPs
- Further elaboration of major outcomes and production of (internal and external) reports

For year 5:

- Production of documents (GL and CPR White Papers, consensus papers, recommendations, Reports, etc) to be presented at and discussed with and possibly adopted by the EC

The following table was briefly discussed focusing on the priorities, methods and foreseen achievements

WP	Priorities	3° -4° y (2019-2020-2021)	5° y (2021-2022)
WP 2	Providing access to information for different target audiences and ensuring timely execution of the Multiannual Plan	Dissemination activities and organization/sponsorship of brainstorming thematic meetings	<ul style="list-style-type: none"> • comprehensive document about the impact of information diffusion on health care, which might be relevant to EU policy • European Parliamentary Meeting on access to care • Organization of an EU Metabolic awareness Day
WP 3	<ul style="list-style-type: none"> • Facilitating activity and compliance of HCPs with the MetabERN goals and vision • Monitoring 	Survey , Progress Report, Evaluation & Monitoring of HCO and POs satisfaction,	<ul style="list-style-type: none"> • Successful running of the Network
WP 4	Development of pts pathways/cross border pathways consensus program for the transition and diffusion of GLs CPR s	Survey, Brainstorming thematic meeting	<ul style="list-style-type: none"> • Consensus paper on the status of the transition programme in IMDs, • GL and CPRs
WP 5	Usage of core service platforms (ECP, UIMD and CPMS);	Facilitate usage of the CPMS by Operational Helpdesk ; CEF	<ul style="list-style-type: none"> • Implementation & optimization of ECP and CPMS use, Recommendations for creation of cross-border MDTs
WP 6	Mapping and critical analysis of research activities on orphan IMDs and of the translation of their output for patient benefit	Submission of proposals for grants programmes Generation of data/ U-IMD Survey	<ul style="list-style-type: none"> • Position paper written by expert group with recommendations for financial support • working group on new clinical trial designs • White book on innovative therapies for IMDs
WP 7	<ul style="list-style-type: none"> • Implement Therapeutic Educ. Program • Exchange teaching & training courses & MetabERN extension into countries currently not represented 	Collaboration with Salzurb Sim. Center and SSIEM, universities	<ul style="list-style-type: none"> • Well-trained medical professionals & empowered pts • Introduction of RDs in the curriculum that every medical student should • White paper
WP 8	Promotion of quality assurance, NBS and prevention programmes	Brainstorming thematic meeting, surveys and internet searches, cooperation ERNDIM	<ul style="list-style-type: none"> • Quality Assurance schemes, NBS in Europe,
WP 9	<ul style="list-style-type: none"> • Pt-centred healthcare • Pts involv. & integration in SNW & WP • Patient- centred evaluation system • Adequacy of industrial therap. strategies 	Survey, Analysis of patient's expectations Brainstorming meeting	<ul style="list-style-type: none"> • Consensus on general recommendations for patient-centred healthcare • White paper with recommendations

Below the 3-5 years plans are discussed in more detail for the different WPs

**WP 1:
Coordination and
management**

WP1 will keep its focus on the maintenance of efficient and effective project management.

Continuous monitoring of the progress of both Work Packages and Subnetworks will be by evaluating the three-month reports that feed the annual report. An overall progress report will be disseminated (as done in year 2) in order to share the activities with other HCPs, the general public and the patients. The organisation of Board meetings involving all stakeholders will be crucial to ensure information spreading and more importantly to create a sense of community and joint effort. By creating synergies with other ERNs, we will be able deal with overlaps in diseases for patient's benefit. Meetings with other ERNs/RD Connect/EC will be crucial for sharing knowledge and best practices and identifying common strategies to avoid duplication of efforts, harmonise and integrate expertise, and impact of EU policy on rare diseases. As in previous years, external subcontractors will be identified to support the management of the Advisory Board and the organisation of the Board Meeting.

**WP2
Dissemination**

WP2 will continue to implement series of dissemination activities (presentations at conferences, scientific and lay published papers, production of press articles, patient support group education and conferences, newsletters, videos, educational web presentations, social media channels and website).

Patient involvement in all dissemination and awareness activities will be crucial. This WP will also ensure the liaison with EU policy makers and relevant stakeholders by targeted communication.

WP3 Evaluation

WP3 will ensure transparency on possible conflicts of interest and the official adoption of a Conflict of Interest policy.

The WP will continue the work on assessing the needs of HCPs by conducting semi-structured phone interviews and online surveys. Similarly patient perception of MetabERN activities and patient-centeredness will be mapped and evaluated.

An internal monitoring questionnaire was developed in the first 2 years and will be used to monitor the performance of the full members, as required by MetabERN. Potential discrepancies will be used to optimize performance.

The persistence of discrepancies in the activities of single HCPs will be discussed for a possible exclusion.

**WP4 Guidelines,
Care Pathways
and
Standardization
for Medical Care
and Transition**

WP 4 will keep focusing on the development of patient pathways/cross border pathways, in close cooperation with the ERN-CG Working Group that will be set up on this topic in the course of 2018. The Network Coordinator will be co-chairing the Guideline Education and training Group (under the ERN-CG).

Two HCPs from MetabERN WP4 will also actively participate in the working group that will develop a methodology, a framework and a tool to ensure a harmonised development of care pathways.

Furthermore, this WP will - together with WP-8- focus on writing a paper on disease specific transition and make sure that this is incorporated in every guidelines/patient pathway that will be produced within MetabERN

WP5

WP5 will cooperate in developing an intellectual property policy that will apply ERN-wide.

With regard to CPMS, WP5 will optimize the virtual second opinion, using this system by increasing the use of CPMS within MetabERN. This will be made possible thanks to the CEF-TELCOM grants that have been awarded to MetabERN and which will be used to facilitate the use of CPMS in all the HCPs.

The use of CPMS is an indicator present in the internal monitoring schema within MetabERN and will also be feature in the ERN-wide monitoring scheme (number of patients entered into CPMS and number of panel case reviews). The exact number will be established by the Working group later in year 2. Continued input and support from the IT Helpdesk is expected.

With regard to multi-disciplinary teams (MDTs), this WP will implement recommendations on the composition of MDTs across borders.

A new E-health project regarding the digitalization and management/follow up of patients will start in collaboration with a software developer developing specific web based programs.

WP6 Research

WP 6 will pursue the efforts to create networks of experts to identify and submit proposals for grants. Research in the field of protein delivery through the blood brain barrier (BBB) has been intense for more than 10 years, with limited positive outcome. Following the identification of partnerships with research institutions, this WP will form an expert working group to produce a critical analysis of these approaches and recommendations for financial support. A comprehensive overview of clinical research activity in the MetabERN network will be provided, resulting in a white paper on “Innovative therapies in IMDs” to be published at the end of year 5. Following studies performed in year 1 and 2 on research activity in the network and access to OMPs, studies in years 3-5 will produce a detailed analysis of clinical research in the network, an analysis of the use of U-IMD for the description of disease natural history and clinical research, and a study of the economical models of innovative therapies for IMDs. These activities will be completed with studies on the “adequacy of industrial strategies for innovative therapy development with patient’s expectations” and with a roadmap to new clinical trials. These 7 complementary studies will be put together and accompanied of a critical analysis of the conclusions to form the “white book” on innovative therapies in IMDs.

WP7 Education and Training

WP7 will work on establishing a Therapeutic Education Programme (TEP). This programme is aimed at supporting collaboration between HCPs and patients to better understand their disease, treatment options and disease monitoring. It will start with finding consensus on the outcome domains for the TEP. Training providers will then be found before implementing the TEP.

In order to expand and align the specialised knowledge of HCPs to ensure high quality of care, this WP will draft a white paper that sums up the best practices, recommendations, minimum requirements and the outcome domains in order to be able to set up a specialised programme for HCPs.

In order to better support underequipped centres WP7 will perform a mapping exercise with regard to the currently available training and teaching programmes in the underequipped centres in order to identify their needs. Best practice programmes will then be disseminated to support these centres.

In order to expand the scope of MetabERN to third countries, an exchange programme will be set up basing itself on the before mentioned mapping exercise.

A training program will be designed in collaboration with the ERN-CG WG on Guidelines and training to attract and form new medical professionals on rare IMDs.

WP8 Continuity of care

In order to be able to roll out the quality assurance schemes this WP will find consensus on this topic in cooperation with ERNDIM and Orphanet before rolling out this scheme within MetabERN used laboratories.

The minimum set of IMDs will be screened to be able to set up a framework on this and have an overview of the current situation of tests offered and numbers that are screened.

A mapping exercise will be undertaken on currently available PND/PGD by surveys and internet searches. This will ensure a comprehensive overview of measures for prenatal and pre-implantation diagnostics. Adult metabolic centres will be mapped to get a better picture of the currently available expertise of diagnostics using the same methods as mentioned above.

In order to improve the transition of seamless care into adult life in cooperation, WP8 will cooperate with WP-4 in writing a disease specific transition consensus paper. The content of this paper will be used in the incorporation of transition in each patient pathway developed under WP-4.

WP9: Patient Empowerment

This WP is newly established in order to create a structured approach in patients' empowerment to effectively improve the establishment of patient-centred healthcare by active involvement and the integration of patients in our programs. The activities of this WP will form an arch over the other 8 WPs and by means of a questionnaire proposed to patient organisation, and/or to HCPs, this WP will acquire key information on patient profile and lifestyle; HCP's behaviour; patients' and HCPs environment, etc. Collected information will be analysed and used to promote the development and implementation of policies, strategies and healthcare services that empower patients to be involved in the decision-making and management of their condition according to their preference, whilst raising awareness about their rights and responsibilities. This knowledge will be shared and put into practice by the other WPs. With regard to the adequacy of industrial strategies for innovative therapies this WP will use the knowledge from WP-6 and the white paper that will be developed by them to improve the strategies where possible.

Patients asked if this WPs has been included for the interest of the EC or of the patients. They were assured that this is totally for the interest of the patients since in such way they can officially report to the EC the activities and achievements

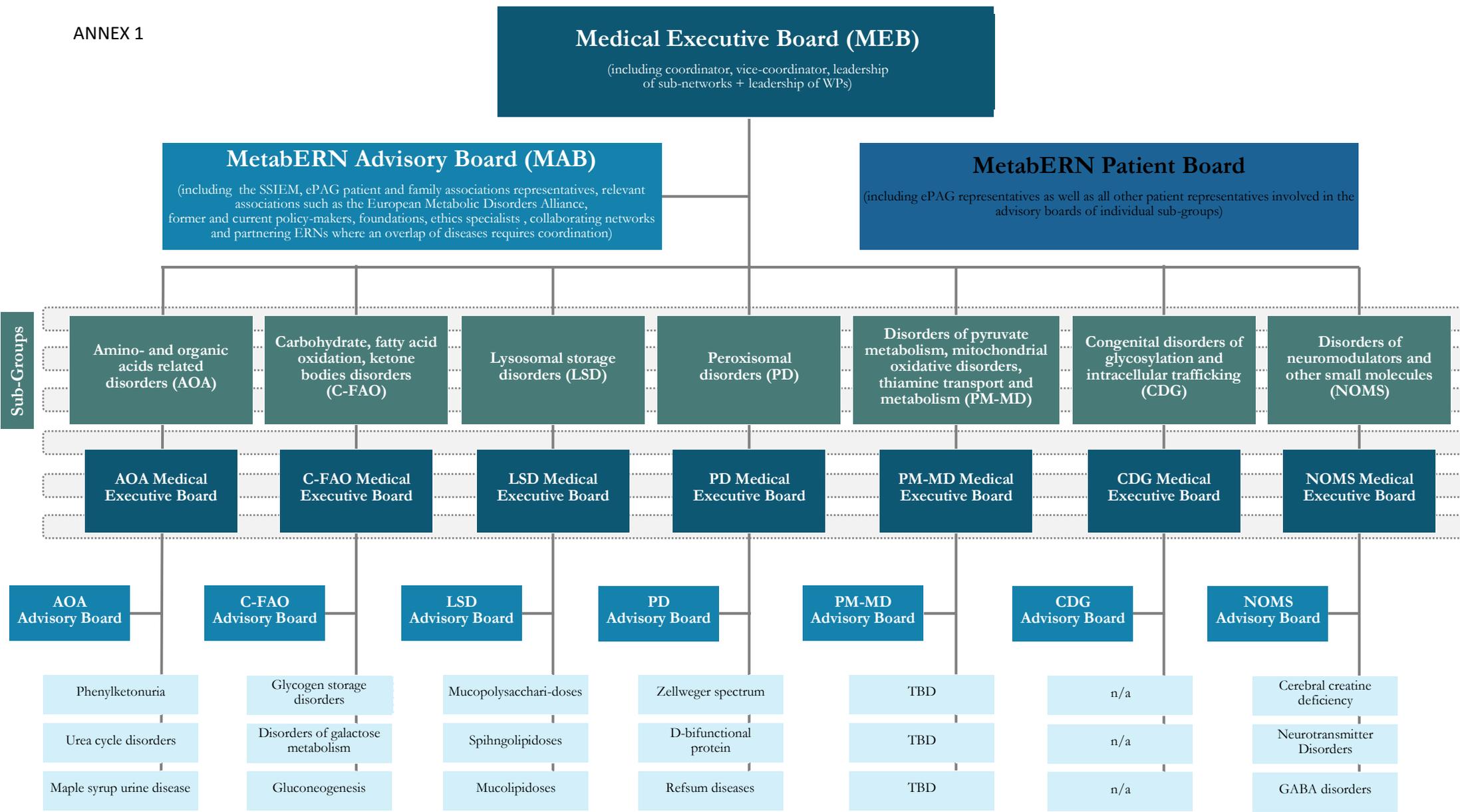
Financial plan and potential projects to be funded

Due to the absence of Maurizio this topic was not discussed

MetabERN Board

(General assembly of representatives from all HCPs, patient groups and other third party stakeholders involved in the activities of the MetabERN)

ANNEX 1



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