



European
Reference
Network

MetabERN

European Reference Network
for Hereditary Metabolic Disorders

Activity Report March-May 2018

WORK PACKAGES

Work Package 1-Coordination & Management

Since the start of the Second Year the Coordination Office (CO) has worked on monitoring. The Working Group operating under the ERN-Coordinators Group (ERN-CG) has set up an overarching system for Quality which includes among others indicators on the use of CPMS (number of patients, participation in panels), the composition of the Multidisciplinary Teams (MDT), the number of clinical trials, guidelines and publications written and participated in by the Health Care Provider (HCP). The CO has come up with a few more indicators to add to this list, which will be implemented in the course of the year.

We have also submitted the Grant application for the Second Year grant in April, which has been approved at the time of writing. We were successful in putting some expenses related to the second year (e.g. second board meeting) in the first year financial report. For this reason, we will start with 200.000 Euros and we might be able to pay some for activities of the SNWs/WPs.

The Coordination Office has also asked for input from the Work Packages (WP) and Subnetworks (SNW) in writing a timeline and detailed planning for the Second Year plan. We have received input from 5 of the 8 Work Packages and 2 of the 7 Subnetworks (the deadline was the 22nd of May).

The CO has started writing the plans for the third to fifth year that we are supposed to submit on September 13 of 2018. We will be sending out the template that is filled in by us to the WPs and SNWs for you to review.

Work Package 2- Dissemination

This Work Package has sent out the monthly newsletters, kept the website up to date, managed the Facebook, LinkedIn and Twitter accounts, and developed a template for the Patients newsletter. We would appreciate if all HCPs become followers of these accounts in order to increase our social media footprint. WP-2 has also developed an abstract on MetabERN for the SSIEM meeting.

Work Package 3- Evaluation

The WP on Evaluation has finalized the Patient Survey on needs and expectations and sent it out to all the POs that are dealing with the field of IMDs.

It has also been working on finalizing the survey on the needs and expectations of Health Care Providers, which we are hoping to send out in the next weeks.

This WP has worked with WP-1 on the development of the monitoring requirements that we will be communicating on before the start of the summer period.

Work Package 4- Guidelines/Care Pathways and Standardisation for Medical Care and Transition

The electronic platform (Google Drive) is available and working and accessible for all those who have the link (which has been sent out earlier this year (but can be find here as [well](#)). If using the platform, make sure to read the manuals and step-by-step Guide and consult the person responsible for Guidelines in your WP/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). The platform was discussed at the Rome meeting (December 2017) and presented to all SNWs during a Web-based conference. Detailed "how to do" as well as necessary forms are available as matrices facilitating the use of the platform. Two groups are already active in using the platform for their first CPR.

In February 2018 U. Plöckinger retired – A. Ziagaki has already taking over the CPR project. Due to the EU requirements for guidelines the WP4 group together with Maurizio Scarpa consented on Carlo Dionisi is the one responsible for setting up the GL program. Maria Couce Luz will be dealing with dissemination and evaluation.

Work Package 5-Virtual Counseling

This WP has been working on drafting a MetabERN specific survey in order to find out the real underlying issues with the low usage numbers of the CPMS within MetabERN.

Klaus Mohnike and Terry Derks are working with the European Commission on integrating CPMS with the existing hospital systems.

Work Package 6- Research, Translational Activities and Clinical Trials

This Work Package has been focusing on developing the survey on the access to treatment that will be sent out soon. The results will be used as a basis for a White Paper on this topic.

Work Package 7- Capacity-building and Education

WP-7 has analyzed the results of the survey they held at the start of Year 2 on Training Programmes (internships, training, organisation of training) and drafted a report. It is available in Circa BC under WP-7 for everyone to read.

Work Package 8- Continuity of Care

WP-8 is working on setting up cooperation with Oprhanet and ERNDIM in order to suggests continuity of care/diagnostic tools that should give us an overview/what is possible when patient is moving from 1 country to another. Patients and physicians should be able to see what diagnostic tests they have access to in which country. If that is OK as a report we can continue to make a list in the form of a map showing the diagnostic labs in Europe so people can click on the map to see where to get their analysis.

The Coordinators are also looking into newborn screening with the international Society for Newborn Screening (ISNS). They are planning to do a data collection exercise with the members and MetabERN could tag on to this.

Subnetworks

AOA

PM-MD

The mitochondrial was resend in an attempt to capture as many centres as possible. In the last months the SNW has reconfigured the WP leads. A start has been made to organize monthly webex meetings/telecalls. With regard to classification Rudy and Luisa volunteered to lead on this and will capture existing classification systems, assess pros/cons and determine whether any of these are already suitable or if a new system is needed; we are hoping that this exercise might lead to a publication in due course. Discussions with MMS have been ongoing. For the CPMS there is a plan to submit a patient. A bid has been in a bid for a Recordati Mitochondrial course in 2019, fingers crossed for a positive outcome; Shamima will continue to organise SSIEM Academies for the next year. Niklas and Rudy started mapping out of mito labs across Europe (biochemical and genetic for WP8).

C-FAO

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LSD

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PD

On May 18 2018 the Annual Symposium of Metabolics.be (the Belgian Society for Inherited Metabolic Diseases) took place, which was organized by the reference center of Antwerp (university hospital UZA). Main topic "Organelles Disease Revisited": the clinical and lab diagnosis and treatment was presented by experts of the AMC (The Netherlands): BT Poll Thé, M Engelen and R. Wanders. The collaboration between François Eyskens and Marc Engelen on peroxisomal disorders started.

CDG

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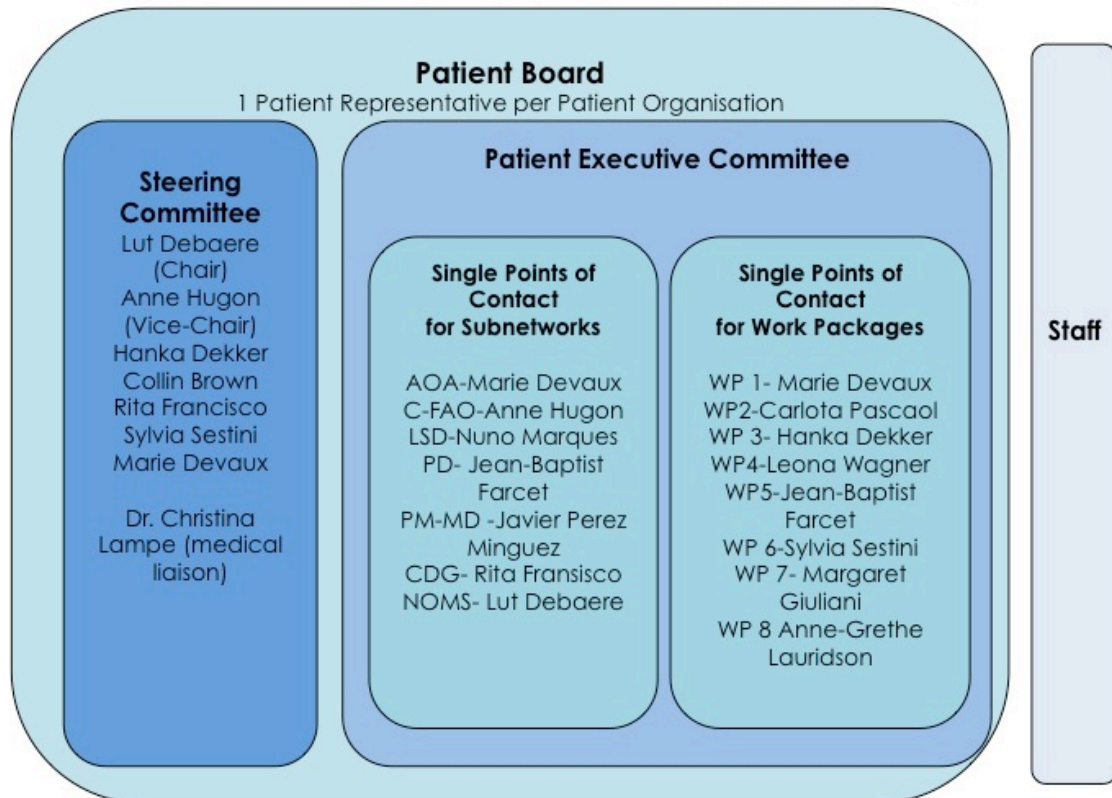
NOMS

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Patient Board

In the last months the Patient Board has been taking shape. There is a patient leaflet, the Governance Terms of Reference and the Action plan for this year have been approved. The structure for patient involvement has been taken shape and can be found below.

MetabERN Patient Governance Structure



In the next couple of weeks the names of the patient representatives who are designated to the different WPs and SNWs will be put in contact with the coordinators and leaders in order to start working together.