
Coordonnateur du Groupe :

Dries Dobbelaere

Membres :

Dr Nadia BELMATOUG : nadia.belmatoug@aphp.fr

Pr Pascale DE LONLAY : pascale.delonlay@aphp.fr

Dr Manuel SCHIFF : manuel.schiff@aphp.fr

Dr Alice KUSTER : alice.kuster@chu-nantes.fr

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Marie DEVAUX (Association Feux Follets) : m.devaux@phenylcetonurie.org

Sandy COURAPIED : Sandy.courapied@chru-lille.fr

Azza KHEMIRI : azza.khemiri@aphp.fr

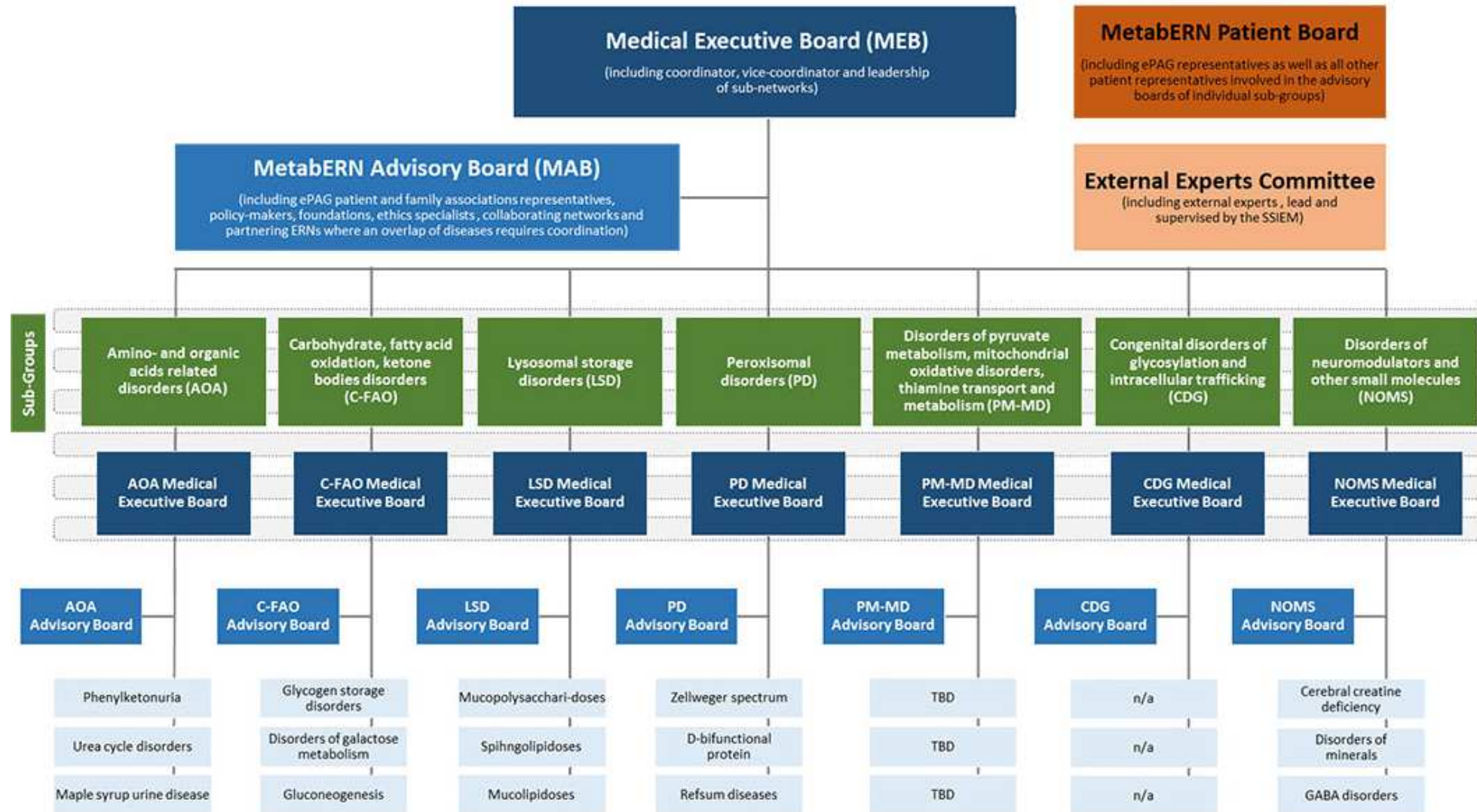
Missions

- Participation des CRM/R à MetabErn / Work Packages
 - Projets européens / Participation à des appels à projet
 - Nouvelle labellisation ERN
 - E-IMD
-

METABERN BOARD

MetabERN Board

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



GOVERNANCE STRUCTURE

MetabERN	Coordinator
<p>COORDINATOR</p> <p>Maurizio Scarpa HSK Wiesbaden</p>	<p>The Network Coordinator is the legal representative of MetabERN. The Coordinator, assisted by the Medical Executive Board, supports and facilitates coordination within and outside of the network.</p> <ul style="list-style-type: none">• Coordinate activities within the network and with core network coordinators• Act as contact point for European Commission and external stakeholders
<p>VICE-COORDINATORS</p> <p>Stefan Kölker University Clinic Heidelberg, DE</p> <p>Eva M Morava-Kozicz UZ Leuven, BE</p> <p>Ans van der Ploeg Erasmus UMC Rotterdam, NL</p>	<p>The Vice-Coordinators support the Coordinator, and, in the event of the Coordinator's absence, the Vice-Coordinators will carry out the Coordinator's duties on his/her behalf.</p> <p>The Vice Coordinators will be generally responsible for:</p> <ol style="list-style-type: none">a) the coordination of the HCPs,b) the coordination of the Patients Associations and stakeholdersc) the coordination of the Communication/ Information Technology Platform.

GOVERNANCE STRUCTURE

Medical Executive Board	<ul style="list-style-type: none">• Coordinating body consisting of network coordinator, the 3 Vice-coordinators, the chairs of the 7 subnetworks and the leadership of the 8 WPs• MEB holds the strategic responsibility for activities of the network and ensure its long-term sustainability.• Will meet frequently (virtually via web and in person) to plan and follow up on the MetabERN activities.• Reports its activities to the MetabERN Board during the annual Board meetings.• MEB consults on a regular basis the MetabERN's Patient Board and Advisory Board to provide input into the planning and implementation of activities.
Advisory Board	<ul style="list-style-type: none">• It consists of representatives of the ePAG patient and family associations, foundations, policy-makers, representatives from collaborating networks and other relevant stakeholders.
Patients Board	<ul style="list-style-type: none">• Representation of all patient and family associations involved in MetabERN activities. It is an independent body which consults, oversees and evaluates the work of MetabERN from the patients' perspective. It works in close cooperation with the MEB.
External Experts Board (TBD)	<ul style="list-style-type: none">• Independent review committee• Consists of external experts and family representatives who are not actively involved in MetabERN – such as SSIEM

7 SUBNETWORKS

Sub-Group	Coordinator(s)
Amino and organic acids-related disorders (AOA)	Henk Blom Stefan Kölker Francjan van Spronsen
Disorder of pyruvate metabolism, Krebs cycle defects, mitochondrial oxidative phosphorylation disorders, disorders of thiamine transport and metabolism (PM-MD)	Enrico Bertini Shamina Rahman Manuel Schiff
Carbohydrate, fatty acid oxidation and ketone bodies disorders (C-FAO)	Carlo Dionisi Terry Derks Ute Spieterkötter
Lysosomal storage disorders (LSD)	Ans van der Ploeg Giancarlo Parenti Dominique Germain
Peroxisomal disorders (PD)	Bwee-Tien Poll The Linda De Meirleir François Eyskens
Congenital disorders of glycosylation and disorders of intracellular trafficking (CDG)	Eva Morava Pascale de Lonlay Thomas Honzik
Disorders of Neuromodulators and Other Small Molecules (NOMS)	Angela Garcia Cazorla Thomas Opladen Eliane Sardh

AOA SUBNETWORK - Members

Country	HCP	Responsible MetabERN member	
Belgium	CHU Liège	François-Guillaume FG	Debray
Belgium	Metabolic Center ULB-VUB	Linda	de Meirleir
Belgium	University Hospital of Antwerp UZA	François	Eyskens
Belgium	BOKS (PO)	Lut	de Baere
Croatia	University Hospital Center Zagreb	Ivo	Baric
Czech Republic	General University Hospital in Prague-GUH (Všeobecná f	Viktor	Kozich
Denmark	Copenhagen University Hospital, Rigshospitalet	Allan Meldgaard	Lund
France	Assistance Publique-Hôpitaux de Paris- University Hospi	Pascale	de Lonlay
France	Lyon University Hospital	Nathalie Guffon	Guffon
France	University Hospital of Lille	Dries	Dobbelaere
France	University Hospital of Marseille, Assistance Publique, Hc	Brigitte	Chabrol
Germany	Charité-Universitätsmedizin Berlin	Ursula	Plöckinger
Germany	Hannover Medical School	Anihb Martin	Das
Germany	Medical Center University of Freiburg	Ute	Spiekerkoetter
Germany	Muenster University Hospital	Frank	Rutsch
Germany	University Hospital Heidelberg	Stefan	Kölker
Germany	University Medical Center Hamburg-Eppendorf, Universi	Kurt	Ullrich
Germany	University Medical Center Mainz	Julia	Hennermann
Italy	Azienda Ospedaliera Universitaria "Federico II", Napoli	Giancarlo	Parenti
Italy	Meyer Children's Hospital	Maria Alice	Donati
Italy	San Paolo Hospital, ASST Santi Paolo e Carlo, Milano	Graziella	Cefalo
Italy	University Hospital of Padova	Alberto	Burlina
Italy	Azienda Ospedaliera Università Integrata Verona	Alessandro	Salviati
Lithuania	Vilnius University Hospital Santariskiu klinikos	Algirdas	Utkus
Netherlands	Academic Medical Center Amsterdam	Annet	Bosch
Netherlands	Erasmus MC, University Medical Center, Rotterdam	Ans	van der Ploeg
Netherlands	University of Groningen, University Medical Center of G	Terry G. J.	Derks
Norway	Oslo University Hospital	Trine	Tangeraaas
Poland	University Hospital Krakow	Beata	Kieć-Wilk
Portugal	Centro Hospitalar do Porto, EPE (CHP)	Esmeralda	Martins
Portugal	Centro Hospitalar do Sao Joao, E.P.E.	Esmeralda Maria	Rodrigues Silva
Portugal	Centro Hospitalar Lisboa Norte, EPE	Ana Maria	Simões Mendes Gaspar
Slovenia	University Medical Centre Ljubljana	Mojca	Zerjav Tansek
Spain	Complejo Hospitalario Universitario de Santiago de Com	Maria Luz	Couce-Pico
Spain	Hospital Universitario Cruces, Barakaldo	Luis	Aldámiz-Echevarría Azuara
United Kindom	Birmingham Children's Hospital NHS Foundation Trust	Suresh	Vijay
United Kindom	Great Ormond Street Hospital NHS Foundaton Trust, LO	Anupam	Chakrapani
United Kindom	University Hospitals Bristol NHS TRUST, Bristol Children'	Efstathia	Chronopoulou
<i>n=15</i>	<i>n=38</i>		

FRENCH MEMBERS

- **Dominique Germain**
Centre de référence Généo pour la Maladie de Fabry et les pathologies héréditaires métaboliques du tissu conjonctif
- **Nadia Belmatoug**
Reference Centre for Lysosomal Diseases (Centre de Référence des Maladies Lysosomales).
- **Nathalie Guffon**
Reference Centre of Inherited Metabolic Disorder
- **Philippe Labrune**
Centre de référence des maladies héréditaires du métabolisme hépatique
- **Laurent Gouya**
Centre de Référence Maladies Rares CRMR Porphyries / CRMR Porphyrias
- **Pascal de Lonlay**
Centre de référence des maladies héréditaires du métabolisme de l'enfant et l'adulte (Reference centre for inherited metabolic diseases of children and adults)
- **Manuel Schiff**
National Centre of expertise for Inborn Errors of Metabolism
- **Dries Dobbelaere**
Center of expertise of inherited metabolic disease
- **Brigitte Chabrol**
Center of expertise of inherited metabolic disease

FRENCH ASSOCIATION

- VML
- Feux follets
- AFG

Enquête pour les associations

Connaissez-vous le réseau MetabERN ?
Quels sont vos besoins et attentes pour MetabERN?

Date limite : 13 Juillet 2018

<https://www.surveymonkey.com/r/BC9Z8G7>

AOA SUBNETWORK - Vertical structure

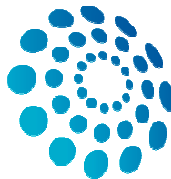
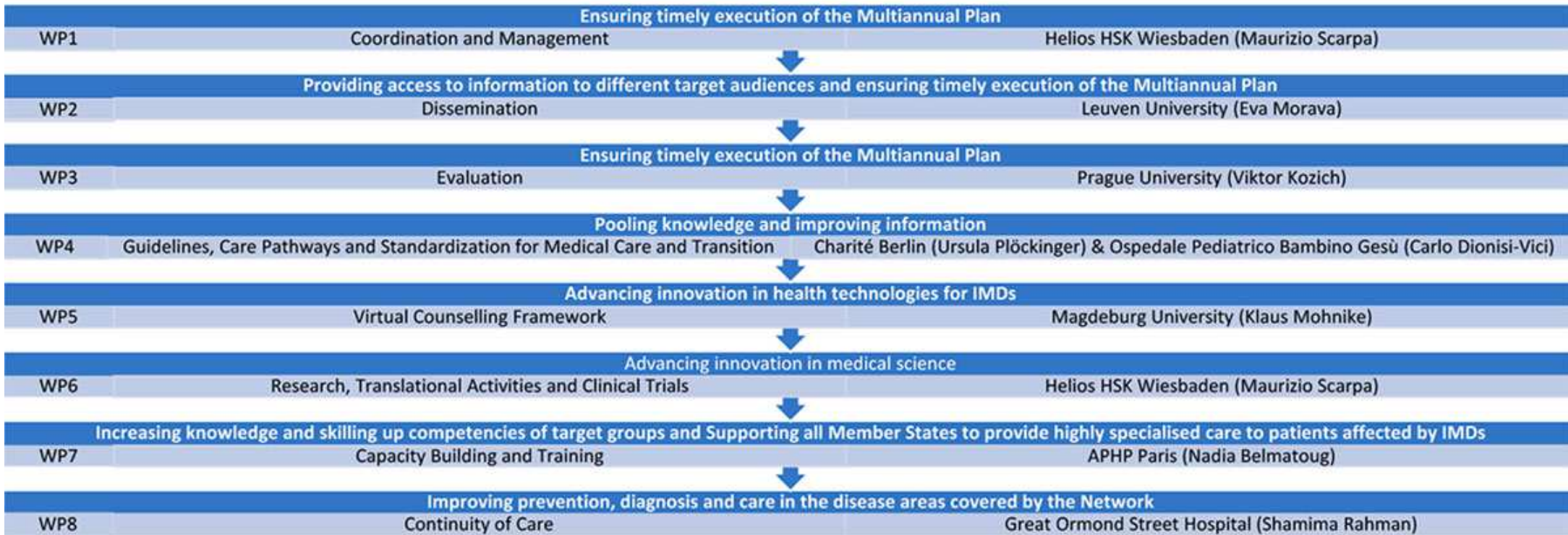
List of planned activities for year 1

Activity no.	Disease	Topic	Responsible AOA members
1	All AOA-related disorders	Create a list of AOA-specific disorders	Santra, Pierre, Kölker
2	PKU	Evaluation and implementation of European recommendations	van Spronsen
3	Tyrosinemia type I	Evidence-base recommendations	van Spronsen, de Laet, Blikrud, Das
4	Tyrosinemia type I	Long-term safety study of orfadin treatment (SOBI-NTBC-005)	Sanchez Pintos, Couce-Pico
5	Alkaptonuria	Evaluation of NTBC treatment	Gomez
6	Classical homocystinuria	Evidence-based recommendations	Blom
7	Isovaleric aciduria	Evidence-based recommendations	Dobbelaere
8	Glutaric aciduria type I	Newborn screening, long-term outcome study	Kölker

WORK PACKAGES – OVERALL LEAD - Horizontal dimension

Workpackage	Topic	Lead
WP1	Coordination and management	Scarpa
WP2	Dissemination	Morava
WP3	Evaluation	Kozich
WP4	Guidelines, Care Pathways and Standardization for Medical Care and Transition	Plöckinger, Dionisi-Vici
WP5	Virtual Counselling Framework	Mohnike
WP6	Research, Translational Activities and Clinical Trials	Scarpa
WP7	Capacity building and training	Belmatoug
WP8	Continuity of Care	Rahman

METABERN SPECIFIC, MEASURABLE, ATTAINABLE, REALISTIC, TIMELY (SMART) OBJECTIVES



European
Reference
Network

MetabERN

European Reference Network
for Hereditary Metabolic Disorders

WORK PACKAGES – RESPONSIBLE AOA MEMBERS

Link between horizontal and vertical dimension

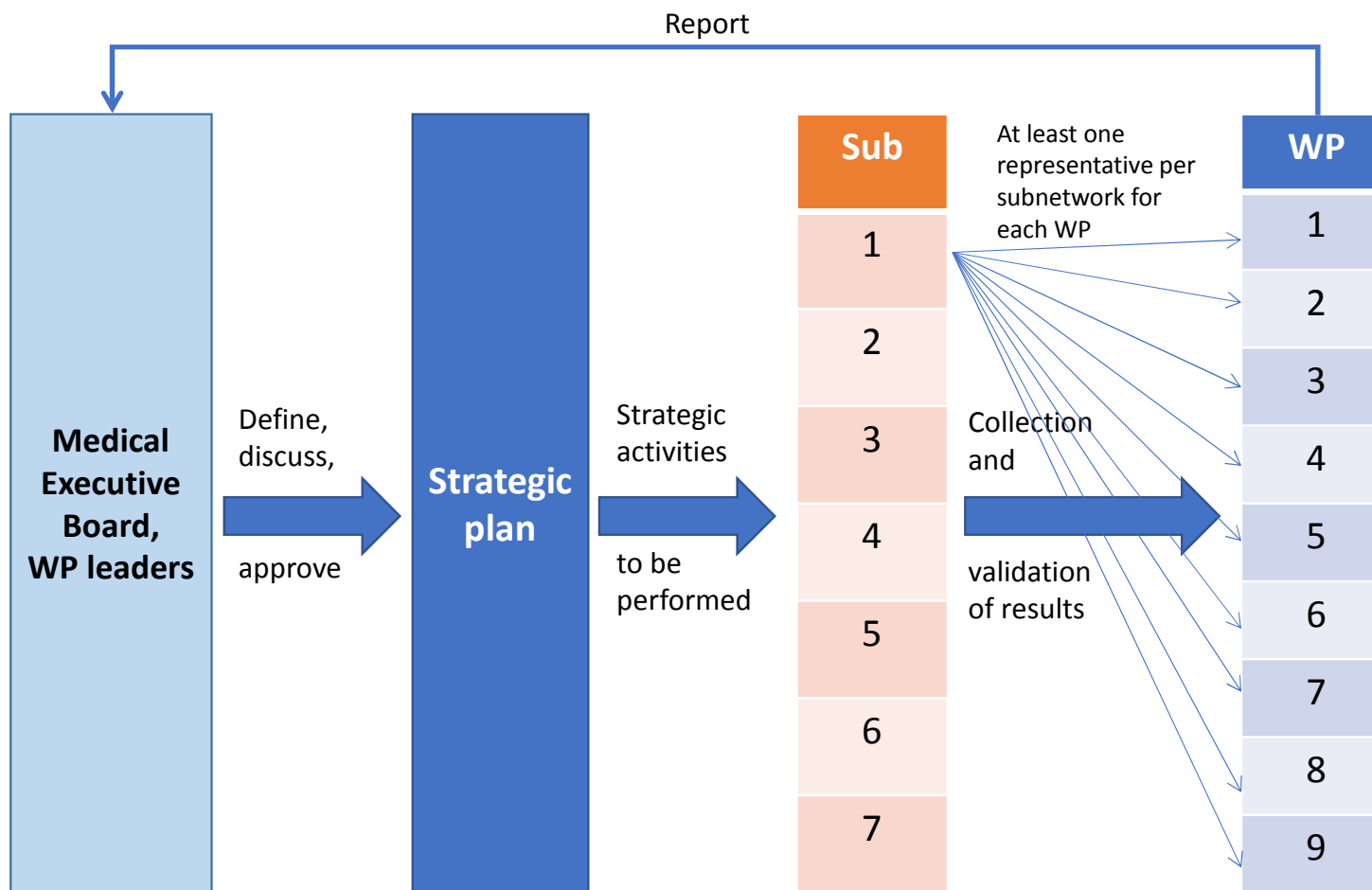
Workpackage	Topic	Lead
WP1	Coordination and management	Kölker
WP2	Dissemination	van Spronsen
WP3	Evaluation	Viktor Kozich
WP4	Guidelines, Care Pathways and Standardization for Medical Care and Transition	Santra, Cefalo
WP5	Virtual Counselling Framework	Rutsch
WP6	Research, Translational Activities and Clinical Trials	Gomes
WP7	Capacity building and training	Dobbelaere
WP8	Continuity of Care	Blikrud

PROJETS EUROPÉENS / PARTICIPATION À DES APPELS À PROJET

- Including prevention and screening
- New disease diagnosis
- Guidelines and SOPs
- Quality of life
- Ethics and transparency
- Registries and outcome measures
- Patient empowerment
- Education and training

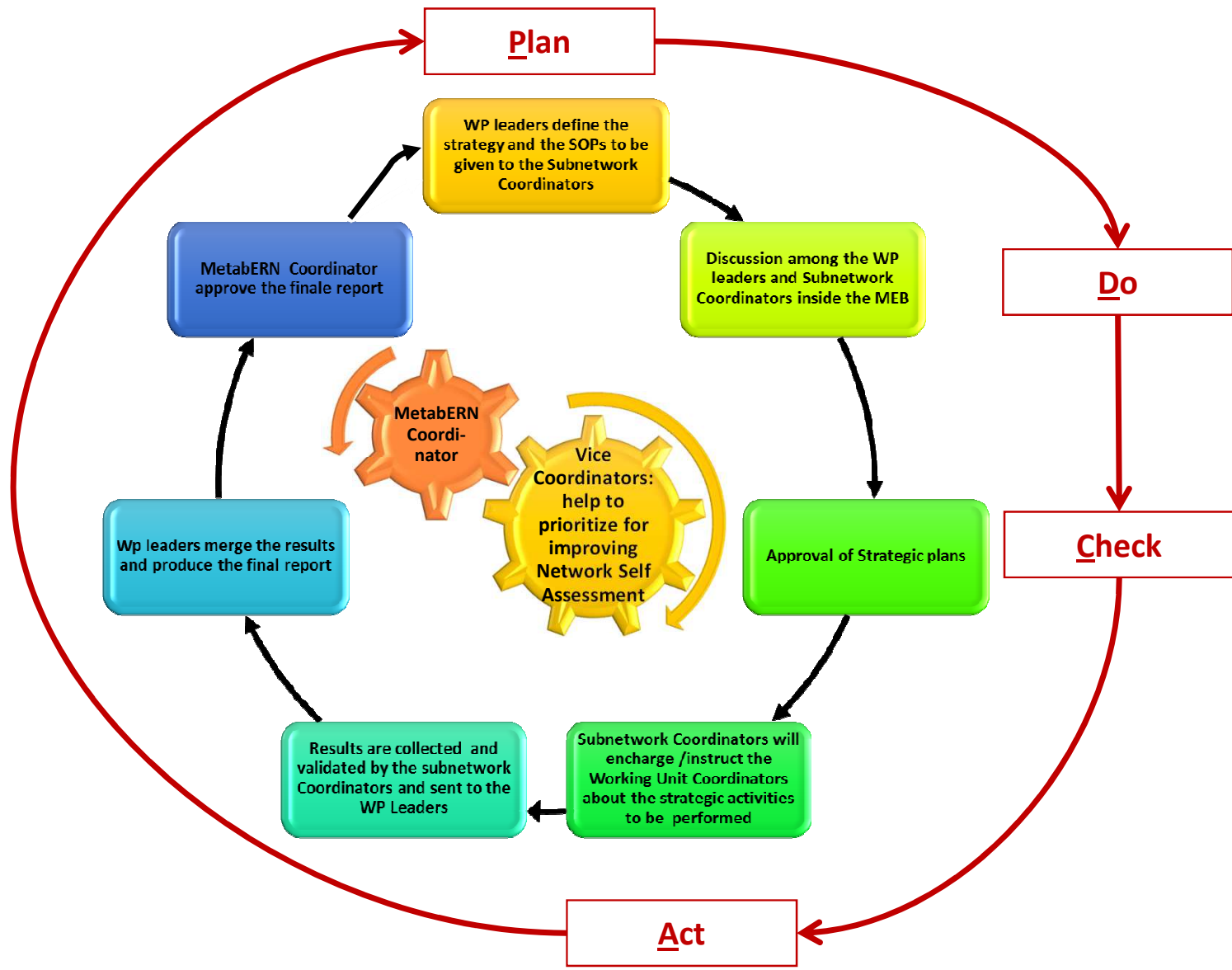
<http://metab.ern-net.eu/>

WORKING STRUCTURE



MEETINGS

- Annual MetabERN Advisory board member's meeting at Frankfurt
- Monthly telephone conferences for AOA members
- Other subnetworks.... ?



NEXT LABELIZATION

... 2019... ?

E-IMD

- **E-IMD member's board meeting** : 13 Novembre 2018 – Bruxelles
- E-IMD website
- E-IMD publications
- E-IMD guidelines
- E-IMD partnership with pharmaceutical companies
- E-IMD newsletters

<https://www.eimd-registry.org>

