The JOURNEY of living with a RARE DISEASE in 2030

MetabERN: how we share, how we care
Maurizio Scarpa, Coordinator on behalf of all the MetabERN Members
Udine University Hospital, Italy

• Increase awareness amongst general public and Health care professionals, health authorities and relevant stakeholders on inherited metabolic disease (IMDs)
• Disseminate of new information, scientific publications and revision of existing health-related information

• Facilitate and harmonise the newborn screening in EU
• Use of the Unified-IMD registry to study the natural history of asymptomatic and symptomatic state of diseases

• Enhance speedy diagnosis by promotion of diagnostic expert consultation (CPMS) and training sessions
• Improve access to the best diagnostics (biochemical/genetic) and to genome-wide genetic investigations through active collaboration with Orphanet and ERNDIM

• Revise/develop of Care pathways & Guidelines
• Facilitate Clinical Trials/Research (defining research priorities and disseminating research-related information)
• Create big databases with patients data
• Improve the Regulatory process for medicines
• Participate in the production of White papers and Recommendations

• Assess the effectiveness and efficiency of clinical processes
• Develop a common programme on Transition from childhood to adulthood

Recent achievements to start the journey:
• A dedicated group on patient empowerment has been established
• The Patient Board governance structure is up and running including the Patient Executive Committee (PEC). The PEC is the voice of all patient representatives involved in the subnetworks and Work Packages in order to improve their involvement and enable their feedback to be heard by HCPs.
• Patients have been involved in addressing their specific needs, preferences and priorities (Survey, feedback systems, etc.)

Education and Training needs

Social framework

Inclusiveness

Patient Empowerment

ALL WITH ONE GOAL IN MIND: OUR PATIENTS!

Project co-funded/funded by the EU Health Programme 2014-2020