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on behalf of all InNerMeD partners

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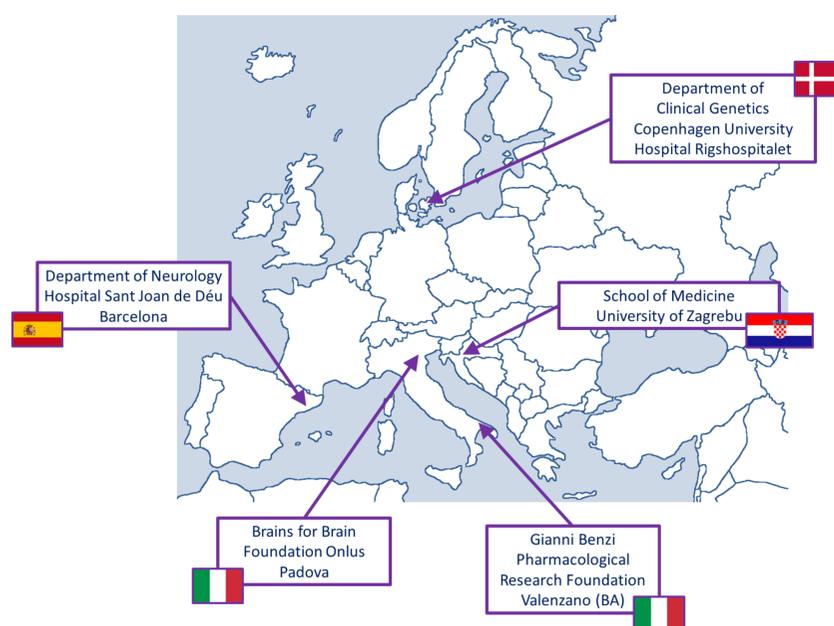
INTRODUCTION

Inherited NeuroMetabolic Diseases (iNMDs) can be defined as genetic disorders of the metabolism affecting at any extent the nervous system. iNMDs are mostly rare and each of them shows clinical heterogeneity; they may affect more than one organ system and can cause significant morbidity and mortality. Many iNMDs are characterised by a precocious involvement of the nervous system that progressively worsens leading to severe mental retardation with devastating consequences and premature death. Today active drugs exist for the therapy of some of these conditions. These drugs can, if promptly used in newborns or young children, slow the neurodegeneration process and increase life expectancy. In addition, thank to pharmacogenetic and genetic advancements, there is the potential for pre-symptomatic and, in many cases, prenatal diagnosis. Unfortunately, data on iNMDs including diagnosis, treatment options, epidemiology, biomedical and pharmacological research are scarce as well as poorly disseminated outside the restricted group of experts that are also inadequately connected.

Inherited Metabolic Disease-Information-Network (**InNerMeD-I-Network**) aims to develop a network of information targeted to the diagnosis and treatment of iNMDs, based on the exchange of information among experts, to collect standardized data and to disseminate validated data among patients and all the interested subjects.

The project is co-funded by the Health Programme of the European Union (Grant Agreement no. 2012 12 12), is guided by Brains for Brain Foundation Onlus and includes 5 European partners which are shown in **Figure 1**.

FIGURE 1



OBJECTIVES

General Objectives of the proposed Network are:

- to increase **knowledge** on iNMDs among physicians, researchers, patients and general public since this action can lead to an early diagnosis and, when available, an appropriate treatment;
- to expand **network activities** among centres of excellence focused on metabolic, neurological and paediatric diseases;
- to straighten **research** capacities and promote innovation in the field of iNMDs;
- to provide practical **support** for sharing experiences and results.

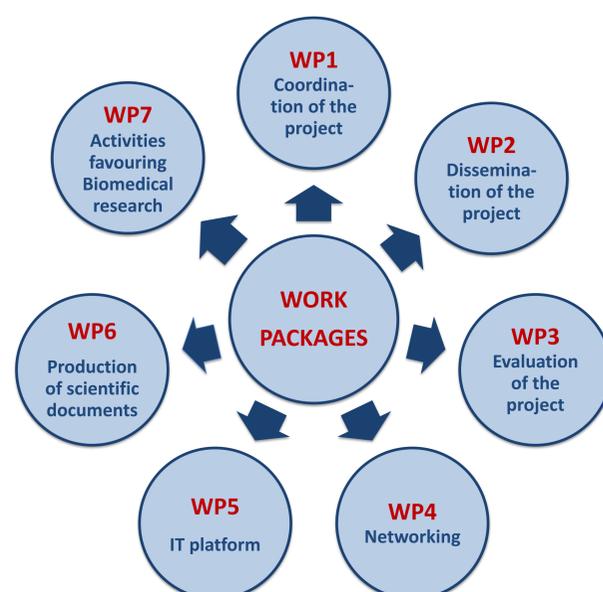
Therefore, through the proposed Network a large number of actors operating in a real interdisciplinary manner, will create a critical mass of competences at multinational level, collecting all the different expertises. The convergency of EU scientific communities will favour for these disabling diseases the implementation of best practices and will lay the foundation to the development of new therapies, otherwise very difficult to carry on in these small and overall young populations.

METHODS AND MEANS

The project is divided into 7 work- packages, as shown in **Figure 2**, and it will entail the following methodological steps:

- 1. NETWORK SET UP.** A core network represented by Partners will be set up and enlarged to include other people and organizations having both interest and competences in the iNMDs field.
- 2. IT PLATFORM DEVELOPMENT.** A publicly available platform assuring user-friendly access and easy exchange of information among users will be developed to connect the node of the Network and to disseminate Network products. It will include:
 - a project web portal based on a Content Management System (CMS), as the main tool for publicizing the project and disseminating its results;
 - an electronic repository to make available all information provided through the project;
 - interactive systems for sharing data and files. IT procedures will be implemented to share data with remote databases and IT sources of information developed within the project.
- 3. PRODUCTION OF DOCUMENTS AND RECOMMENDATIONS.** All collected and integrated information will be assessed for scientific evidence and assembled in guidelines and recommendations summarizing current existing best practices and most recent advancements in NMD diagnosis and treatment. Drafts documents will undergo an appraisal process using the AGREE system (Appraisal of Guidelines for Research & Evaluation), a tool validated for evaluating clinical practice guidelines pertaining to medical care. Finally evidence based conclusions will be disseminated through the platform.

FIGURE 2



CONTACTS

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