European Task Force on Brain and Neurodegenerative Lysosomal Storage Diseases

FONDAZIONE BRAINS FOR BRAIN – ONLUS
c/o Dipartimento di Pediatria di Padova
Via Giustiniani 3 – Padova
C.F.: 92212200288
Iscritta al Registro Prefettizio P.G. Padova, n° 52
Registro Comunale delle associazioni n° 3063

Inherited NeuRoMetabolic Diseases INFORMATION NETWORK

ACRONIM : InNerMeD-I-Network

STARTING DATE: 2013-04-01
DURATION (in months): 30

PRIORITY AREA: Actions under the second objective 'Promote health'

ACTION: Prevention of major and rare diseases

SUB-ACTION: Support for European rare diseases information networks - Setting up of new rare disease registers or rare disease information networks

WHY INNERMED I NETWORK

NeuroMetabolic Disorders (NMDs) represent an important group of Rare Diseases (RDs) constituted by metabolic diseases that show clinical neurologic/cognitive symptoms at any time of the disease progression.
Today, despite a current general lack of awareness of these conditions, active drugs exist for replacement of the missing enzyme. These drugs can, if promptly used in newborn or young children, slow the neurodegeneration process and increase the life expectancies. In addition, thank to pharmacogenetic and genetic advancements, there is the potential for pre-symptomatic and, in many cases, prenatal diagnosis.
Unfortunately, data on NMDs are not only few but also very bad disseminated outside the restricted arena of the experts that are also very disperse and poorly connected. . Lack of awareness of these conditions can lead to delayed diagnosis and start of treatment,

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with consequent tragic results. Increasing awareness is therefore the first crucial step in fighting these conditions.

PROJECT OVERVIEW

InNerMeD-I-network wants to create a network of information targeted on diagnosis and treatment of NMDs based on the collection and exchange of proper information among scientific community, health professionals, patients, patients association and all interested stakeholder. The project aims to increase current knowledge on NMDs and speed up the timely and precise identification of patients to which apply the available treatments. The network will also favor biomedical research, straightening research capacities and fostering innovative therapeutic tools derived from the recent scientific advancements based on biomarkers use and personalised approaches

THE PARTNERS

InNerMeD-I involve a formidable concentration of competences combining partners specific expertises. **The partnership includes both public and private referral clinical centres:**

- **the Brains for Brain Foundation**, coordinator and main leader partner, thanks to its great expertise gathered in the neurological field and its long-standing experience in the exploitation and dissemination of new scientific knowledge and contribution to the implementation of **EU research** activities particularly assures the partners coherence and capacity to share decisions and to put together relevant knowledge and research capacity;
- **the Benzi Foundation,** strategic partner for the management and for the set up of the ICT platform. The Benzi Foundation has extensive experience in the realisation and management of IT tools. It supported TEDDY NoE (FP6 project) and currently supports GRIP (Global Research in Paediatrics, FP7 project started on January 2011) in the realization of e-learning courses.

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- **the Center for Metabolic Disorders at the University of Copenaghen (RH)** covers the whole of Denmark concerning diagnosis, biochemical monitoring, clinical evaluation and treatment of patients with metabolic disorders, and in particular with Lysosomal Storage Disorders;
- **the Center for lysosomal storage disorders at the University of Mainz (UMC-M)** takes care of about 800 patients with lysosomal storage disorders, and participates in clinical trials for enzyme replacement therapy as well as in several EU projects;
- **the University of Zagreb School of Medicine (UZSM)** is the oldest educational institution for medical training and research in the South-East Europe and works actively in biomedical research;
- **the Hospital Sant Joan de Déu (HSJD)** is the Spanish referral center for Neurometabolic diseases and in paediatric care.

To complement these strong scientific groups the partner FGB will participate adding its specific capacities in term of manage scientific EU projects, set-up and implement technical IT instruments such as surveys, e-learning and, in particular, the EU-Orphan platform that will serve as basis for developing the project Platform and Electronic Repository.
Finally the EEM is a (Bio)ethics research centre of Marseille Hospital. It will support the Network in identifying ethical issues and providing recommendations both to health professionals and patients/families. He will also have the role of evaluating the project.

OBJECTIVES

In the NMDs field, it is crucial to suppress the storage as soon as possible before the young brain undergo neurodegeneration and/or secondary events (e.g. brain inflammation, alteration of intracellular trafficking, impairment of autophagy, or oxidative stress) as these manifestations are irreversible.
The general Objective of the proposed Network is to create a critical mass of knowledge to be disseminated in order to:

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• increase awareness on these conditions among physicians and general public since this action can lead to an anticipated diagnosis and, when available, an adequate therapy;
• straighten research capacities and foster innovation in favour of the population affected by inherited NMDs;
• provide practical support for sharing experiences and results;
• circulate knowledge on clinical and experimental approaches for diagnosis and treatment of NMDs and skills in view of empower patients and families as actors of the diseases management.

The Network also aims to apply the criteria for Networks stated in European Health Programmes on Rare Diseases (European Reference Network, ERN) and in the Paediatric Regulation (European Network of Paediatric Research at the European Medicines Agency, Enpr-EMA).

These objectives will be achieved by

- **the harmonization , simplification, validation of the material present in the web regarding the pediatric neurodegenerative diseases to allow the stakeholders to access to material scientifically valuable and reliable**
- **The collection, evaluation and organization in an accessible repository of all the published and unpublished material on clinical features and clinical research projects, evidence based medical data and international pharmacological data and research regarding pediatric neurodegenerative diseases.**
- **The organization and validation of scientific material, the production of scientific documents, guidelines, recommendations, textbooks of reference, referral material to be exploited by stakeholders to promote awareness and strategic plans to be used in the application of the national plans on rare diseases**

METHODS

- NETWORKING: The establishment of an enlarged network will be achieved thanks to main, associated and collaborating partners exclusive specific experience in the area

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of interest of the project (NMD diagnosis and treatment, RDs and ODs, systematic review methodology, clinical studies methodology in small populations, IT setting up and management, etc.) and to their consolidate networking capacity that will allow the involvement of other people and organisations having both interest and competences.

- IT PLATFORM DEVELOPMENT: a publicly available infrastructure assuring user-friendly access and easy exchange of information among users, who do not necessarily need specific technical competences, will be allowed starting from IT instruments. It will include:
  A project web portal based on a Content Management System for publicising 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.
- PRODUCTION OF DOCUMENTS AND RECOMMENDATIONS. All collected and integrated information will be assessed for scientific evidence and assembled in guidelines and recommendations summarising current existing best practices and most recent advancements in NMD diagnosis and treatment, including biomarkers and genetic information.

EXPECTED OUTCOMES

This project will contribute to reduce the gap affecting NMDs in different ways.

- Through the proposed Network a large number of actors operating in a real interdisciplinary manner will be involved with the aim to create, in line with the provisions of Directive 2011/24/EU, a critical mass of competences instead of a dispersed expertise available a multinational level. Knowledge dissemination will increase awareness on NMDs epidemiology speeding up the timely and precise identification of patients to which apply the available treatments.

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- Thanks to partners experience and competence in CTs methodologies in paediatrics and Rare Diseases, a translation of scientific breakthroughs into clinical practice will be achieved in collaboration with public health services and private companies.
- Such knowledge advancement addressed not only to the professionals operating in the specialised or general health system, but also to the entire patient populations will generate social benefit permitting the establishment of a standard of care for patients with NMD. This will ensure equity of access to diagnosis, treatment and care across Europe.

The strength of InNerMed project relies on its capacity of creating a reference tool for further interesting activities to be developed after the project will be ended such as:

1- a unique European network that will facilitate research in the NMD area;
2- the generation of new data on epidemiological and genetic aspects
3- the provision of informative and consultative support to pharmaceutical companies and Health Institutions
4- setting-up a Paediatric Network for developing trial and research