EUROPEAN PARLIAMENT

BRAINS FOR BRAIN

SCIENTIFIC REPORT

PAVING THE WAY FOR A COMPETITIVE AND DYNAMIC EU KNOWLEDGE ECONOMY: THE WAY FORWARD IN RARE DISEASES

26 November 2013, 16.00-18.00
EP Roundtable Room, room ASP A3E2
Hosted by AmaliaSartori and Maria da GraçaCarvalho

HSK
Dr. Horst Schmidt Klinik

UNIVERSITÀ DEGLI STUDI DI PADOVA

CASA SOLLIEVO DELLA SOFFERENZA
SAN GIOVANNI ROTONDO

INNERMED
Inherited NeuroMetabolic Diseases Information NETWORK

EXECUTIVE SUMMARY

Executive Summary

Background Rare pediatric neurometabolic diseases (RPNMDs) represent a major public health concern in European societies.

To date 5,000 to 8,000 distinct rare diseases have been identified, affecting between 5% and 8% of the population in total. In other words, it means that between 24 and 36 million people are affected by rare diseases across all 27 EU member states. Among these affected patients, about 12-18 millions (50% of cases) are children mainly affected by RPNDs. To date, there are about 300inherited disorders resulting in brain damage in children, including the Lysosomal Storage Disorders (LSDs). Lysosomal storage diseases (LSDs) are genetic disorders caused by specific enzyme deficiencies resulting in multi-systemic disease affecting different body organs or systems and typically manifesting progressive neurological, renal, cardiovascular, gastro-intestinal, muscular-skeletal, ophthalmological and respiratory symptoms. LSDs are mostly associated with high morbidity and mortality rates hence representing a significant burden on patients, their families, the health care system, and the society. Most LSDs patients, in fact, begin showing signs of the disease early in childhood, often in the first year or two of life.CNS pathology in particular causes mental retardation and progressive neurodegeneration that ultimately ends in early death of these young patients. Indeed 30% of affected children die before the age of 5 with devastating consequences on families and high costs for society.

Unsolved Problems Because of LSDs low-prevalence, there is often lack of information about these conditions and it remains difficult to find clinicians knowledgeable about the diagnosis, management and treatment of these conditions. Getting a correct diagnosis is one of the major challenges for affected children: it can take several years from the time of first symptoms to the one of disease recognition and initial treatment. During this long and frustrating period, families frequently seek advice from numerous doctors and consult different specialists often receiving diverse incorrect diagnoses. Such consequent delay in getting the right diagnosis has important implications for patients treatment since today effective drugs for replacement of the missing enzyme for many of these disorders exist. In fact, if promptly used in newborn or young children, these drugs, can slow the pathological process of neurodegeneration and increase the life expectancy of these patients. It is therefore of paramount importance to diagnose the condition as soon as possible.

Besides the need for early diagnosis, and new research programmes to develop new treatments able to cross the blood brain barrier (BBB), an overriding problem is represented by the replacement of a generation of scientists and physicians who contributed to the advancement of knowledge and available therapies for specific rare diseases, who have reached pension age and are close to retirement. A number of laboratories and clinics have been already closed and many will face the threat of closure in the next 5 years.

The Brains for Brain Foundation believes that medical advances can be achieved in a more active way if we capitalize on our knowledge and talents and strengthen the link between doctorate education, policy activities and investments in research in this field. This could help address the

disconnect between societal needs of children and the curricula of European biomedical researcher and healthcare professionals.

To this aim the Brains for Brain Foundation, in line with the core principles established by "Towards a Maastricht for Research" (The Maastricht for Research Manifesto, published by MEP Amalia Sartori and MEP Luigi Berlinguer in June 2013), has created a network of Universities and Scientific Societies to start a doctorate programme aimed at furthering the knowledge on neurometabolic diseases amongst young physicians and scientists in order to establish an European Network of specialized experts and maintain excellence in Europe.

The challenge of this European collaborative effort lies in addressing the gaps in research in the field of rare paediatric diseases and limiting the brain drain created through the lack of career opportunities in this field in the EU. The development of a new form of cooperation across EU and the establishment of a new generation of educated specialists competent in RPNMDs diagnosis and management represents the first step towards the achievement of a fair, equitable and equal health care system, particularly in regard of patients with RPNMDs.

The European Roadmap : On 16 October 2013 MEP Sartori and MEP Berlinguer launched their manifesto[1] "European Research Area: a Maastricht for Research", which aims to rally the European scientific community and institutions. One of the main key objectives consists in seeking consensus on a series of actions and initiatives that will enable the completion of the European Research Area. Building on the political momentum gained by this initiative, **the B4B Foundation organized t**he roundtable*"Paving the way for a competitive and dynamic EU knowledge economy: the way forward in rare diseases"*hosted by MEP Amalia Sartori and MEP Maria Graça da Carvalho. The meeting was aimed at increasing the awareness of the EU Commission about the need of funding for pregraduates and postgraduates on LSDs and for their sustainability to ensure the progress of research and therapy development. The main objective of the meeting was to coordinate the collaborative effort and possibly find new ways of collaboration that will identify strategy to sustain the project for a new PhD programme on LSDs as pilot project of the Maastricht for Research Programme that which will be launched on 2014.If successful the proposed model of cross-sector cooperation to overcome the gaps in research in the area of RNMPDs in EU could be then replicated in other disease areas and sectors.

The meeting brought together top level representation of all the interested DGs and relevant stakeholders to share views on how to address the gaps in research in the area of rare neurometabolic diseases affecting children.

The general atmosphere was amiable, free-thinking and productive: all participants were highly motivated and overall there was a strong sense of collaboration and of excitement. The outcome of the workshop was very encouraging and positive. It was generally agreed that overcoming barriers in health research and reversing the brain drain should be made political priorities at European level. This will require action from policy makers, hospital administrators, healthcare providers, patient groups and citizens across Europe.

[1] The "European Research Area: a Maastricht for Research" Manifesto paves the way for an ERA Framework Directive. It also identifies a number of proposals to overcome the following problems: 1) Low level of investment and human resources, 2) Fragmentation and low coordination, 3) Knowledge application, 4) Research infrastructures, and 5) Research Careers and Mobility.

The B4B roundtable has rallied multi-stakeholder partners to take the first step to seek endorsement for the set-up of a European PhD Programme in the area of rare neurometabolic paediatric diseases. Stakeholders from 20 European organisations active in the field of health have signed the B4B Manifesto to maximise investments in health research and advance care for children with rare neurometabolic diseases.

AGENDA

Registrations 15.30

Introduction to the manifesto 16.00

MEP Amalia Sartori
MEP Maria da Graça Carvalho
Maurizio Scarpa, Brains for Brain Foundation
David Begley, Brains for Brain Foundation

Videos 16.30

MEP Emma McClarkin
MEP Luigi Berlinguer
MEP Gianni Pittella - Written speech

Presentations by panellists 16.40

Jaroslaw Waligóra, Policy Officer, DG SANCO
Mary Baker, President, European Brain Council
Nathalie Moll, Secretary General, EuropaBio
Michel Goldman, Executive Director, Innovative Medicines Initiative

Mario Roccaro, Policy Officer, DG EAC

Planned contributions 17.15

MEP Oreste Rossi
MEP Angelika Werthmann
Ruxandra Draghia-Akli, Director,Health Directorate, DG Research
Roberto Bertollini, Representative to the EU, WHO
Frank Stehr, NCL-Stiftung
Paul De Raeve, Secretary General, European Federation of Nursing Associations
Duccio Bonifazi, Benzi Foundation
Vassili Valayannopoulos, Chair, SSIEM Education and Training Advisory Committee
Volkmar Gieselmann, Chairman, European Study Group on Lysosomal Diseases

Panel Debate moderated by MEP Maria da Graça Carvalho 17.30

Signature of the Manifesto 17.50

Opening and Introduction to the Manifesto

Dr M. Scarpa, President of the B4B Foundation, officially opened the meeting welcoming the participants and briefly summarizing the activities, initiated last summer, that gave rise to extraordinary achievements such as the production of the manifesto "Towards a Maastricht for Research" developed by Amalia Sartori, *MEP - Chairwoman ITRE Committee* and Luigi Berlinguer, *MEP - Former Italian Minister for Research by Research Europe* with the support of universities and research organisations, which was officially presented to the European Commission on 16 October 2013, and the B4B Manifesto entitled "Investing in Brains to deliver research value in the EU".

Dr Scarpa underlined the importance of working together on steps to bridge the gaps in research in the area of RNMD and presented the reasons underpinning the B4B Manifesto which puts the foundations of a multi-disciplinary PhD, bringing together patients, families, clinicians, scientists and industry partners to advance medical research and overcome the "Blood-Brain Barrier".

He stressed the concept of the importance of the European Research Area (ERA) as an ideal environment in which to create a new PhD and Post-Docprogram to complement what has been already proposed in order to maintain and possibly implement the number of research centers. In particular the initiative aims to:

- address the gaps of knowledge in RNMPDs (since there are a lots of gaps between first symptoms and the diagnosis)
- create a new generation of physicians ready to identify the disorders as soon as possible in order to promptly start the therapy
- cross bridge the cooperation to create synergies and a better cooperation among academic, carers, biotech companies and all the stakeholders interested in RNMPDs
- meet all patients health needs.

It was highlighted that patients represent the first and ultimate goal in this action.
Since a general consensus on the important benefits deriving from the creation of a new generation of educated physician working in network was achieved, it was decided to launch the PhD pilot program inside the Maastricht for Research Program.

David Begley, Vice-President of the B4B Foundation, briefly introduced himself and underlined the importance of a global collaboration in the field of Rare Diseases in EU. He underlined that not just the diseases are rare, but also the clinicians who treat them and understand them are rare and widely dispersed in EU. Each EU country has, in fact, a very limited number of physicians who are expert in rare diseases and this makes rare diseases extremely difficult to diagnose and manage. There is a big need of cross border collaboration and cooperation. The establishment of a global EU networking is the key for the institution of profitable interactions among patients and families associations, clinicians and scientists of different disciplines including pediatricians, neurologists, rheumatologists, cardiologists, surgeons, experts in metabolic medicine, neuroscientists, experts in cerebro-vascular diseases, cell and molecular biologists, geneticists and pharmacists. It is in fact quite clear that being RPNMDs systemic diseases, many are the specialist involved in their management and the establishment of an EU multidisciplinary team of experts working in network and specifically focused on RDs can improve patients outcomes bringing important benefits to the EU society. He also explained that what B4B is suggesting is that the establishment of a PhD program focused on a transnational action and on the exchange of knowledge is fundamental for the development of new effective strategy for crossing the Brain Blood Barrier and of new therapies capable of arresting the neurodegenerative processes, typical of RPNMDs, with important benefit and improvement for RD health care system. He also underlined that in order to cope with the challenges ahead we need to harmonise and strengthen our EU training schemes and to enhance

mobility of researchers through the creation of an European Scientist Passport and through the fostering of a flexible cross border mobility and employment facilities.

MEP Amalia Sartori, Chairwoman of the Parliament's Industry, Research and Energy (ITRE) Committee, opened the discussion with a description of the B4B manifesto as a practical way of responding to a number of gaps in the scientific, knowledge sharing, professional training and healthcare delivery when it comes to the field of rare neurological disorders. She explained that the Brains for Brain (B4B) initiative was actively seeking solutions to identified challenges and with that, Mrs Sartori became the first MEP during the session to sign the manifesto to support patients, families and Europe's bright, young researchers.

MEP Maria da Graça Carvalhon emphasised her support for the B4B Manifesto as a relevant action on rare diseases to speed up results for patients. The promotion and the coordination of Research is extremely important for health care system, and particularly for Rare Diseases, since they represents the tools by which speed up the achievements of results in research and benefits for patients. She also mentioned that within Horizon 2020 the area of health is specified as a priority societal challenge, with rare diseases given particular attention.

Videos

Some Recorded videos of prestigious panellists who could not directly participate to the meeting but wanted to send a message and express their support to this initiative and joy in supporting the B4B effort , were presented.

Emma McClarkin, Conservative MEP for the East Midlands, welcomed the initiative highlighting the importance of working together sharing the knowledge, especially in the area of RPNMDs. She expressed her gratitude to the B4B Foundation for having organized a such important meeting and reaffirmed her commitment to make every necessary effort to accelerate the proposed achievement .

Luigi Berlinguer, Group of the Progressive Alliance of Socialists and Democrats in the European Parliament, congratulated the B4B Foundation for the program and for its Manifesto which goes in the same direction of the Maastricht for Research Manifesto. He expressed his sincere gratitude highlighting that Europe cannot leave without research. He emphasized that EU must be based on Research and underlined the importance of Research and Innovation for the European society. Research is a priority and concrete action is required. A new generation of educated PhD students constitutes the new strength for innovation in Research. EU represents the real environment for Research, for mobility and for the establishment of new interconnections, collaborative inter-sectorial and international relationships among all researchers, Institutions, infrastructures, organizations and associations, companies and all stakeholders interested in the field of RPNMDs.

MEP Gianni Pittella –Vice President of the EU Parliament, provided the following written speech:

"Dear ladies and gentleman, dear friends,
I am really glad to extend a heartfelt greeting to all of you. Furthermore I would like to extend a warm thanks to my colleagues present today and to the Brains for Brain Foundation for having organized

this outstanding initiative. It is an important arena aiming to foster and support the research in the area of rare and neurometabolic diseases affecting children. The European Union and this roundtable share the same conception of the human dignity, of cooperation and solidarity within and between our Member States. A public health system that needs to be adequate, accessible to all and that moves with the times represents one of the major commitments of the European institutions, as it is one of the major rights of citizens. Only research is able to pave the way for a constant progress and for a better future. Moreover when it comes to children, the fight against rare diseases acquires a greater importance. The rarity, the exiguity of cases must not be an alibi to welch this moral commitment. Every person, every suffering child challenges the social and collective conscience of a continent that has been the cradle of civilization and of the contemporary science. The European Union can only function if it remains sympathetic and fraternal. Therefore we must grant to the scientific research all the economic potential, all the know-how, and all the social and institutional networks that represent the real prosperity of our community. The European flag goes beyond the mere economic cooperation or geographic boundaries. It was based on democratic principles and aims to assure a stable welfare for all citizens.
I extend my warmest wishes for this interesting occasion, and l sincerely compliment all the presents and all scientists and researchers for your hard work."

Patricia Reilly, Cabinet Member of Maire Geoghegan-Quinn, responsible for research, highlighted the importance of the cooperation at international level to stimulate and better coordinate research maximizing its related outcomes, particularly in the field of RPNMDs. To this aim, the IRDi research consortium consists of member companies, charities and governmental and non-governmental organisations that work together towards meeting the rare-disease-treatment objective goal. She anticipated that 200 new therapies for rare diseases will be developed by the year 2020 and therefore it's necessary to find the means to diagnose most of them. She observed that, due to the fragmentation of knowledge, it can take years before a patients affected by rare diseases get diagnosed. Partnerships and collaboration between private and public actors are crucial.

MEP Oreste Rossi, who had been the Shadow Rapporteur on the patients' rights directive in crossborder healthcare then asked that more funds be dedicated to exchange of best practices that can result in better diagnosis and treatment.

Jaroslaw Waligóra, Policy Officer, DG SANCO ,highlighted the fact that none of the EU Countries is able to provide sufficient complete support to patients affected by rare diseases. It is therefore crucial to facilitate mobility of expertise and develop, share and spread information, knowledge and best practice to foster the development of the rare diseases diagnosis and treatment. He also stressed the importance of Rare Diseases registries that have a crucial utility not just for addressing basic research needs but also patients' needs.

The main challenges consist of fragmentation, lack of interoperability, increasing regulatory requirements, and lack of sustainability and the EU community is pleased to contribute to the development of a crosstalk across the funded projects about Rare diseases in order to connect each single effort avoiding duplication or overlaps between projects. She also draw attention to the importance of the adoption of national plans and strategies in rare diseases in each Member States and to the necessity of creating Research infrastructure for rare diseases and increase clinicians training opportunities.

Mary Baker, President of the European Brain Council, expressed her pride and joy for having B4B as EBC member. She emphasized how hard is to find clinicians able to make the right diagnosis and capable of offering concrete help and also psychological support to families. Not just to make the diagnosis is hard, but also being diagnosed with a rare disease is terribly hard. She observed that getting diagnosed with a rare diseases is a thing that changes the life of the entire family, and the problem is not just to get families informed but it's also a matter of how to do it. It is crucial to foster the establishment of a cross-sectorial environment in which should take place inter-professional collaborative teams composed by health-care professionals and patient organizations cooperatively working together in a common collaborative effort.

Nathalie Moll, Secretary General of EuropaBio, commended the initiative since it aimed to overcome the "silos" that impede collaborations between the private and public sector and across sectors.
She underlined that rare diseases are life-threatening or chronically debilitating conditions that affect no more than 5 in 10,000 people in the EU. Are between 5,000 and 8,000 the disease so far estimated, 50% of which affect children. Although most of these rare diseases are genetic, serious, chronic and debilitating, patients and their families should not feel isolated by their condition. Cross borders collaboration can help patients, families and carers find common fair, equitable and equal solutions across EU and remind them they are not alone.

Michel Goldman, Chairman of the Innovative Medicines Initiative mentioned opportunities to secure EU funding through Horizon 2020, through measures earmarked for personalised medicine as well as IMI calls. He also stressed the importance of setting-up new types of clinical and economic models in order to facilitate patients' access to treatments. He then highlighted the important role of the coordination across borders and the promotion of partnerships between private and public actors.

Mario Roccaro, from DG Education illustrated other financial tools fostering excellence through cross border and cross sector mobility. In particular he illustrated the new Marie Sklodowska-Curie Actions (MSCA) programme under Horizon 2020 explaining that it is a research fellowship program which aims to support researchers at different stages of their career. MSCA are open to all domains of research and innovation, from basic research to market take-up and innovation services. Fellowships are awarded by the European Commission in various scientific disciplines within the People programme (FP7) to provide European researchers with the opportunity to gain experience abroad and in the private sector, completing their training skills or disciplines critical to their careers.

MEP Angelika Werthmann stated her full support to the initiative and stressed the importance of providing support to families who have children with rare neurometabolic diseases since the diagnosis of their children is often accompanied by feelings of isolation and uncertainty.

This point was also elaborated by **Roberto Bertollini**, WHO Representative to the EU, who highlighted the role of the WHO in working with Member States to raise awareness of the need to improve knowledge-sharing as a mean to improve standards of care across the European Region, in particular about LSDs since they represent ideal model for more common diseases. He stressed the fact that it is crucial to address Research and Development in the proper way allowing the advance of a more scientific culture since this represents the only strategy for preventing the dispersion of patients and families.

Frank Stehr, NCL-Stiftung, underlined that among neurometabolic disorders LSDs, Neuronal Ceroid Lipofuscinosis (NCL), also called Batten disease, is the most common neurodegenerative disease of childhood and is inevitably fatal. The gradual stages of the suffering are blindness, dementia, epilepsy, loss of speech, paralysis and complete helplessness. It therefore represents a severe worldwide problem. There is a crucial need of therapy orientated Research and innovative clinical oriented or translational basic science projects, which can contribute to find a cure for juvenile NCL. He also observed that it is indispensable to have enough funding to assure careers in research field, in order to keep PhD students motivated and engaged.

Paul de Raeve, Secretary-General of the European Federation of Nurses Associations, representing more than one million nurses at European level, emphasised the need to include patient organisations and the healthcare professionals who are closer to the patients in any collaborative effort aiming to advance medical treatment. Nurses, in particular, provide 80% of direct patient care. He stressed that it is essential to invest in processes that transfer knowledge to action increasing investment in human capital through better education and skills.

Fedele (Duccio) Bonifazi, Benzi Foundation, highlighted the importance of increasing knowledge and awareness about Rare Diseases and introduced the Project "Inherited Neuro Metabolic Disease Information Network" (InNerMeD-I-Network, 2012 12 12, Second Health Programme 2008-2013), an European project, started on the 1 April of 2013, aimed to create a multimedial network of information targeted on research, diagnosis and treatment of inherited neurometabolic diseases and based on the collection and exchange of validated information among scientific communities, health professionals, patients, patient associations and all relevant stakeholders. He underlined the importance of paying close attention to rare diseases and related drug development processes taking particularly into consideration regulatory and law legal tools. It is fundamental to boost drug development and favouring access to promising orphan drugs and promoting activities to deliver concrete answers to patients' needs.

Vassili Valayannopoulos, member of the Society for the Study of Inborn Errors of Metabolism Committee, briefly introduced the Society explaining that it aims to foster the study of inherited metabolic disorders and related topics promoting exchange of ideas between professionals working in different fields. He then both highlighted the need for holistic strategies aiming to enable translational research from the laboratory to the clinical practice. In particular, it was stressed the importance of earmarking budget to attract young biomedical scientists to the area of RPNMDs.

Panel Debate and Signature of the Manifesto

It was generally agreed that overcoming barriers in health research and reversing the brain drain should be made political priorities at European level. This will require action from policy makers, hospital administrators, healthcare providers, patient groups and citizens across Europe. The B4B roundtable has rallied multi-stakeholder partners to take the first step to seek endorsement for the set-up of a European PhD Programme in the area of rare neurometabolic paediatric diseases.

Stakeholders from 20 European organisations active in the field of health have signed the B4B Manifesto to maximise investments in health research and advance care for children with rare neurometabolic diseases.

ASSESSMENT OF THE RESULTS, CONTRIBUTION TO THE FUTURE DIRECTION OF THE FIELD, OUTCOMES

The informal feedback given by the participants was very positive throughout. The outcome of the workshop was very encouraging and positive. The meeting was in fact very useful for establishing new interactions between the participants and strengthening existing ones. The Workshop objectives of initiating joint action aiming to create a model of intersectoral cooperation that could facilitate the set-up of a European PhD Programme in the area of RPNMDs were achieved. The meeting represented a great opportunity for facilitating the collaborative effort and possibly find new ways of collaboration that will identify strategy to sustain the establishment of a new PhD programme on LSDs as pilot project of the Maastricht for Research Programme that which will be launched on 2014. If successful, the proposed model of cross-sector cooperation to overcome the gaps in research in the area of RNMPDs in EU could be then replicated in other disease areas and sectors. The workshop clearly confirmed the fact that LSDs provide excellent pilot models to further develop the understanding of RPNMDs and to early identification and diagnosis of RPNMDs. The most effective way of achieving this aim is through the establishment of a combined multidisciplinary approach involving clinicians, basic scientists and biotech companies and Family Associations.
B4B Roundtable represents an important step forward the establishment of a fruitful EU cross border collaboration and cooperation to raise awareness about RPNMDs and keep them on the health-care agenda. Although rare diseases are by definition individually rare, collectively they affect millions of people worldwide. A joint effort to tackling rare diseases, including pooling and coordinating of financial and scientific resources, is essential to ensure that children affected by RPNMDs are given the priority they deserve and that they needs are met.

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For more information:
B4B Manifesto: http://www.brains4brain.eu/site/assets/files/B4B_Manifesto.pdf
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