4 FEBRUARY, 2015

OFFICIAL OPENING CERIMONY OF THE CENTER FOR RARE DISEASES AT THE HORST SCHMIDT KLINIK IN WIESBADEN (GERMANY)

Opening ceremony of the Center on 4 February 2015
On Wednesday 4 February, patients affected by rare diseases and their families, together with public authorities, researchers, health professionals, industry representatives and anyone who has a genuine interest in rare diseases celebrated the official opening of the newly established Center for Rare Diseases (CRD) at the Horst Schmidt Klinik in Wiesbaden (Germany).

The Center was established in January 2014 thanks to the fruitful collaboration between two key opinion leaders in the field of Neurodegenerative disorders: **Prof. Maurizio Scarpa MD PhD** from the University of Padova, Italy and **Dr. Christina Lampe MD** from the University of Mainz, Germany. The two of them infact, decided to join their expertise to the Department of Pediatrics at the **Horst Schmidt Klinik (HSK)**, **lead by Prof. Markus Knuf.**
CRD is part of the Children's Hospital of the HSK and collaborates with all other clinics of the HSK needed for the management of the multisystemic and progressive diseases offering a unique opportunity for patients affected by Rare Diseases, in particular those affecting the Central Nervous System. Moreover, the recently established partnership with the HELIOS Group added additional potential to the Center since it now belongs to a German-wide network of 110 acute hospitals and rehabilitation hospitals, 49 ambulatory healthcare centers, five rehabilitation centers and 15 nursing homes. It represents a Certified Center of Excellence under the National Action Plan for Rare diseases in Germany, and being located in Wiesbaden, just 30 km far from Frankfurt, it is strategically positioned not just within the German city network but also within the entire EU area.

M. Scarpa, Director of the CRD and President of the B4B Foundation (B4B), together with **Ralf Engels, Managing Director HSK and Markus Knuf, Director HSK Dep. of Pediatric and Adolescent Medicine** officially opened the ceremony welcoming the participants and briefly summarizing the activities initiated last year that gave rise to the establishment of the CRD. The Center constitutes an extraordinary sign of entrepreneurial activities of clinic and academic units aiming at bringing benefits to the entire community of patients affected by rare diseases and in particular for those populations of paediatric patients with unmet medical needs. 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, 4 millions of which are living in Germany. Among these affected patients, about 50% of cases are children. The difficulty in obtaining the correct diagnosis is the first dramatic problem that patients affected by rare diseases must face and it can take years or even decades to overcome it. In fact, due to the nature of rare diseases, to their complexity and to the scarcity of cases, there is still a deep lack of information and scientific knowledge about such conditions. In particular, there is a great necessity to:

- improve and accelerate diagnosis
- prognosticate disease severity
- predict its progression
- assess therapeutic effectiveness accurately

CRD in Wiesbaden represents a concrete answer to these patients needs since it aims to stimulate an interdisciplinary collaborative effort for a timely and accurate diagnosis. The social relevance of such multidisciplinary and collaborative approach consists in the creation of best conditions for clinicians to work together in the most effective and efficient way in order to produce the best health outcomes for the patients and the entire community.

In such contest, **Prof Scarpa** underlined the important role played **by InNerMeD (Inherited NeuroMetabolic Disease Information Network),** an European project funded by the Executive Agency for Health & Consumers (DG-SANCO) under the Second Programme of Community action in the field of

Health, 2008-2013 to be the first European Network on inherited neurometabolic diseases (iNMDs). The project started on April 2013 with the aim to create a multimedial network of information targeted on research, diagnosis and treatment of iNMDs. The Network will increase the current knowledge on iNMDs, speed up the timely and precise identification of patients, who may benefit of the available (experimental and marketed) treatments, and also favour biomedical research. This will be achieved by setting up, activating, sustaining and enlarging a network that responds to the need of a valid geographic coverage, a multidisciplinary composition and the inclusion of major stakeholders such as health Authorities and patient organization. InNerMeD project, in fact, wants to help primary care doctors, clinicians, patients and their family, patients association and organization, companies to work together and interact with each other producing a critical mass of information which will then fall back positively on the patient.

Prof Scarpa further stressed the importance of a joint effort to tackling rare diseases, including pooling and coordinating of information and scientific resources. He underlined that, in line with the InNerMeD principles, CRD is proud to initiate a joint action to further strengthening intersectoral cooperation inside CRD establishing an enlarged German network of Hospitals, Centers, Scientific Organizations, and many other interested parties wishing to work together develop solutions that address each patient's specific needs ensuring that each patients receive effective and seamless care

The importance of a National action schedule for people with rare diseases was also underlined by **Christophs Nachtigaeller, President of the German national Alliance for Chronic rare**. **Diseases (ACHsE),** who described the role that ACHSE has played since 2004 in Germany, raising awareness and advocating for patients' needs, patient empowerment and recognition of the expert knowledge of people living with rare diseases. Today ACHSE is a very lively network of 120 members, representing approximately 4 million patients and is an active member of the EURORDIS Council of National Alliances and was a key advisor in the EUROPLAN-EURORDIS Advisors team on National Plans. He stressed the importance of a multi-stakeholder collaboration and concerted effort with all stakeholders working together in a patient-centered approach. He really welcomed the opening of the CRD underlining that a such nationally recognised center of excellence, collaborating nationally and internationally with other Organizations and Professional Networks constitutes a cornerstone of the German rare disease plan. Such action in fact, aims to provide patients with the best, targeted care available

The importance of ***EU-Engagement for people with rare diseases*** was correspondingly stressed by **Maria da Graça Carvalho, BEPA, European Commission.** Unfortunately, Due to unexpected and irrevocable work commitments she could not directly participate to the meeting, but she sent a message in which she highlighted the importance of establishing close links and collaboration at local and EU level to stimulate and better coordinate the gathering of expertise and improving healthcare for rare disease patients. She particularly expressed her joy in welcoming the opening of the new CRD in Wiesbaden since, thanks to its attested national and international network capacity provides improved capability for diagnosis and specialised management of rare diseases.

The importance of pulling together expertise was underlined by **David Begley, Vice-President of the Brains for Brain Foundation (B4B, www.brains4brain.ue).** B4B is an international task force composed of outstanding scientists and clinicians leaders in the neurological field grouped together with Biotech companies to create coordinated and organized research projects aimed at the comprehension and cure of rare genetic diseases that affect and seriously damage the brain, in particular the children's one. Dr Begley stressed the importance of a global collaboration in the field of the ***"EU Research***", underlying 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. Due to such great need of cross border collaboration and cooperation, B4B, is pleased to integrated its efforts with the CRD ones and work together as a joint venture to increase scientific research, knowledge dissemination, social and sociomedical assistance, health assistance in the field of rare neurological disorders.

Helmut Hehn, Representative of the German Society for Sclerosis Tuberosa (TSD) discussed the theme "***Rare but not so rare***" describing the life-changing experience of being the father of a child affected by Tuberous sclerosis complex (TSC), a rare genetic disorder that causes non-malignant tumors

to form in many different organs primarily in the brain, eyes, heart, kidney, skin and lungs. He particularly underlined the challenges, and the difficulties that patients and families must faced during the diagnostic pathway. In this scenario he emphasized the important role played by the (TSD) in supporting German patient's needs. Today the TSD has create a strong interaction among the 14 hospitals and the centers associated throughout the country, providing a guiding function in diagnostic and therapeutic care. The organization of annual meetings assures and enhances the research projects & health care information exchange. He expressed his deep gratitude to the CRD for the commitment to the support of TSD projects.

Carmen Kunkel representative of the Society for Mucopolysaccharidosis further underlined the ***"Importance of networks***", especially in the context of such complex and devastating disorders as the Mucopolysaccharidosis (MPS). Indeed, knowledge on these disorders are not widespread and the complexity of these diseases make necessary to have a close network among the different parties. She particularly emphasised the importance of including patient organisations in any collaborative effort aiming to improve conditions of people affected by rare diseases and stated her full support to CRD activities explaining that MPS Society works nationally to ensure that patients' personal needs are met and to assure a good quality of life to people affected by MPS. She also stressed the importance of sharing views on new ways of collaboration that can address the existing gaps in the field of rare diseases and underlined the importance of providing support to families who have children with MPS since the diagnosis of their children is often accompanied by feelings of isolation and uncertainty.

Stefan Wirth, Head of the section "pediatric and neonatology", Director of the Center for Pediatrics and Adolescent Medicine, HELIOS Klinikum Wuppertal introduced the "Helios Network", a private and strong network of 110 clinics which bases it success on the intensive interdisciplinary exchange of knowledge of its employees and the rapid implementation of innovations ensuring patients the best possible care. He underlined the importance of the quality management system not just for avoiding mistakes but also for monitoring and constantly improving the standard procedures of medical treatments. The importance of coordinated actions in the health care system was underlined and Helios-network commitment to collaborate with CRD in providing benefit for individual patients, healthcare systems and entire society was expressed.

Followed two spectacular **Patients' Performances** of girls affected by rare diseases who danced and played violin, during which the participants were emotionally engaged and actively involved to enjoy the spirit of such extraordinary exhibition. Bringing together patients, families, clinicians, scientists and industry partners the performance represented a great opportunity for establishing positive and more closer relationships among patients, specialists and the entire community of people with an interest in rare diseases.

The **Photo exhibition *"Closer than Rare"*** organized by FEDRA (Portugal Rare Disease Federation) with the purpose of impacting the Institutional leaders and the society to the problems of rare diseases further contributed to generate meaningful and positive impact on the community.

The informal feedback given by the participants was very positive throughout. The CRD opening ceremony represented a good example of the new concept of medicine based on the collaboration of excellences to create the critical mass needed to improve the management and the quality of life of patients with rare diseases. Besides the occurrence was very useful for establishing new interactions between the participants and strengthening existing ones. The meeting objective of initiating joint actions aiming at creating a model of intersectoral cooperation that could facilitate the set-up of an enlarged network in the area of rare diseases aimed to improve patient quality of life was achieved.