RARE NEUROLOGICAL DISEASES OF CHILDHOOD

DECLARATION OF PRINCIPLES

The following parties support this initiative: the Brains for Brain Foundation, the European Brain Council, the Lysosomal Storage Disease (LSD) Patient Collaborative, the Veneto Region and members of the pharmaceutical industry.

We have entered a new golden age in which science is increasingly able to inform the treatment and care of patients with rare neurological conditions. Neurological disorders affect the brain, spinal cord and nerves of the central and peripheral nervous systems. While many neurological disorders are common, others are recognized as rare disorders. Rare neurological diseases, the focus of this document, are mainly inherited conditions, the vast majority of which are fatal during childhood.

The time is right for a new initiative

We propose a new initiative to coordinate the efforts of existing groups in Europe with the shared goal of improving the treatment and care of patients with rare neurological disorders. This initiative will unite all interested parties, who are working:

- to increase the visibility, recognition and awareness of rare neurological disorders in order to facilitate early diagnosis of these conditions
- to promote and facilitate partnership and collaboration between physicians, researchers, patient advocates, carers, policy- and decision-makers and industry
- to encourage and support research and the translation of scientific breakthroughs into clinical practice
- to contribute to the establishment of a standard of care for patients with rare neurological disorders which is agreed across Europe, and to ensure equity of access to diagnosis, treatment and care.

We hope that you will join us in this important initiative by adding your signature to those below.

EBC European Brain Council

REGIONE DEL VENETO

LSD PATIENT COLLABORATIVE

BRAINS FOR BRAIN

Background

- According to the World Health Organization (WHO), an estimated 1 in 10 people will be diagnosed with a severe and sometimes rare neurological disorder during their lifetime.[1] Brain disease is responsible for 35% of the total disease burden in Europe and is associated with an overall cost of €386 billion.[2] For many such disorders, early intervention can prevent or reduce long-term morbidity and the associated healthcare costs.

- Today, over 6000 disease-causing genes have been identified, including many that are associated with rare disorders. Although individually uncommon, collectively there are thousands of rare diseases, making them an important group of disorders that affect a large number of patients throughout Europe. Most are genetically inherited. In the European Union (EU), a rare disease is defined as one that affects fewer than 5 people per 10,000.[3] In the context that we have so far identified somewhere in the region of 5000–8000 distinct rare diseases, between 27 and 36 million people in the EU could be affected (6–8% of the population), making the management of these conditions an important issue.[3] The socioeconomic burden of rare diseases has not yet been quantified.

- For these reasons, the EU takes the position that rare diseases are a serious public health concern and should be a priority in EU health and research programmes.[3] The overall EU strategy is to support member states in diagnosing, treating and caring for up to 36 million EU citizens with rare diseases.

- This year we celebrate the 10th anniversary of the introduction of the Orphan Drug Regulation in Europe. This legislation provides incentives to industry to invest in the development of treatments for rare disorders and has had a positive impact on the care of patients with these conditions. The 2008 European Commission Communication[4] and the 2009 Council Recommendations,[3] which called on EU member states to develop and implement comprehensive national plans to address rare diseases by 2013, reflect the growing momentum associated with rare diseases at the EU level.

- The recent announcement by the European Commission and the US National Institutes of Health (part of the US Department of Health and Human Services) of their plan to join forces to focus on research into rare diseases is another sign of Europe's willingness to tackle these diseases seriously.[5] As part of the commitment made by these two institutions, the European Commission announced that it will earmark more than €100 million for research and innovation on rare diseases in 2011. This will be the largest single investment in this research field by the European Commission so far.

- At present, however, there is an imbalance in the funding allocated to research into different types of disease. In the UK, for example, based on data from 2008–2009, it was estimated that rare diseases affect more individuals than cancer, cardiac disease, Alzheimer's disease or diabetes, but receive less research funding than these other disorders.[6] Research funding for rare diseases, which were estimated to affect 3.5 million individuals in the UK, was £3.6 million during this period – about £1 per affected individual, which is considerably lower than the spending of £185 for each of the 2 million individuals with cancer.

Diagnosis

- Diagnosis of rare neurological diseases is made difficult by the lack of knowledge and awareness of these conditions within the general medical community. This leads to delayed diagnosis and often misdiagnosis, as was highlighted in a book on rare diseases produced in 2009 by the European Organization for Rare Diseases (EURORDIS) and co-funded by the European Commission.[7] This publication also notes that patients with rare conditions experience difficulties in accessing services, often as a result of a lack of referral. Raising the awareness of these disorders and increasing their visibility is therefore paramount to speed up diagnosis and to give affected families access to appropriate counselling.

Treatment and care

- The management of diseases and the provision of therapy also represent a social and economic burden. Treatment options for patients with neurological conditions remain limited. Although representing a third of the burden of all diseases in Europe, brain disorders account for only 15% of the direct healthcare costs. This discrepancy is due in part to the fewer available treatments for brain disorders compared with other types of diseases. It may also be due to a shortfall in healthcare provision, which may reflect both suboptimal training of clinicians in brain-related disorders and a shortage of material resources.[2] Although there have been major advances in treatment, for the majority of disorders this is not curative. Management needs to be holistic, multidisciplinary and to include palliative and supportive care. This means that there are benefits in establishing centres of expertise that specialize in the diagnosis, treatment and care of patients with rare neurological diseases.

Research

- The EU is already committed to supporting research into neurological problems and has awarded major grants to study neurological disorders under the FP6 programme. An even stronger effort has been realized under the FP7 programme.[8] We know that by studying the neurological disorders of well-defined, rare, monogenic diseases affecting children we will be able to discover information and develop techniques that will help us to generate new therapies for more common adult disorders, such as Parkinson's and Alzheimer's diseases.

- One particularly important group of rare disorders that impact on the brain during childhood is the lysosomal storage diseases, which affect approximately 1 in every 7000 newborns.[9] There is already a strong community supporting research and scientific developments in this area, which includes physicians, researchers, patient advocates and industry. Our understanding of these conditions has grown dramatically over the past 10–20 years thanks to advances in cell and molecular biology, and we are moving closer to being able to provide therapies for neurological signs and symptoms in these patients. Further research is needed to increase our understanding of the brain and to drive the development of treatments.

Join us in this new initiative

- We advocate working together with the European Parliament, European Commission, European Council and other interested stakeholders to promote the diagnosis, treatment and care of patients with rare neurological diseases throughout Europe, and to support and encourage scientific research in this field.

- We invite you to join us in supporting this new initiative.

[1]WHO. Neurological disorders – public health challenges, 2006, p. 33. http://www.who.int/mental_health/neurology/neurological_disorders_report_web.pdf (Accessed [11 Nov 2010])

[2]Olesen J *et al*. Consensus document on European brain research. *J Neurol Neurosurg Psychiatry* 2006;77:i1–i49.

[3]Council recommendation on action in the field of rare diseases, 2009. http://www.consilium.europa.eu/uedocs/cms_data/docs/pressdata/en/lsa/108383.pdf (Accessed [11 Nov 2010])

[4]Communication from the Commission to the European Parliament, the Council, the European Economic and Social Committee and the Committee of the Regions on Rare Diseases: Europe's challenges, 2008 http://ec.europa.eu/health/ph_threats/non_com/docs/rare_com_en.pdf (Accessed [11 Nov 2010])

[5]News alert 28 October 2010 http://ec.europa.eu/research/index.cfm?pg=newsalert&lg=en&year=2010&na=na-281010 (Accessed [11 Nov 2010])

[6]2009 Annual Report of the Chief Medical Officer, Department of Health. http://www.dh.gov.uk/prod_consum_dh/groups/dh_digitalassets/@dh/@en/@ps/documents/digitalasset/dh_114012.pdf (Accessed [11 Nov 2010])

[7]The voice of 12,000 patients, Feb 2009. http://www.eurordis.org/IMG/pdf/voice_12000_patients/EURORDISCARE_FULLBOOKr.pdf (Accessed [11 Nov 2010])

[8]Tosetti P *et al*. Brain research EU funding 2002–2009. (http://www.eurosfaire.prd.fr/7pc/doc/1275999339_brain_research_eu_funding_2002_2009_en.pdf) (Accessed [11 Nov 2010])

[9]Poorthuis BJ *et al*. The frequency of lysosomal storage diseases in the Netherlands. *Hum Gene*t 1999;105:151–6.

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