Rare neurological diseases to be a European research priority

For the first time key stakeholders have officially agreed on the need to improve support for research into rare neurological diseases.

During a conference in the European Parliament, co-organized by the Brains For Brain Foundation, the European Brain Council and the LSD Patient Organisation Collaborative, researchers, scientists, doctors, representatives of patient organizations and pharmaceutical companies, regional, national and EU decision-makers voiced their support for a united approach to face challenges caused by rare neurological diseases.

Participants at the conference on 'Rare neurological diseases of childhood' endorsed a declaration of principles launching a new initiative to increase research into rare neurological diseases and improve their early diagnosis and treatment.

The event follows a European Commission announcement that it will earmark more than €100 million for research and innovation on rare diseases in 2011 as part of the 7th Framework Programme for research. This is the largest single investment in rare diseases so far made by the EU.

The need to develop effective treatments of neurodegenerative diseases was emphasized by Professor Frits Wijburg, of the Academic Medical Center in Amsterdam, who said: 'The last decades have seen major advances in the diagnosis of rare progressive brain diseases in children. In stark contrast, there is yet no therapy for most of these disorders. This can change by an international collaborative effort of devoted scientists'.

In a keynote speech concluding the event, Professor Timothy Cox of the University of Cambridge added: 'Inherited brain diseases are an immense challenge for consorted action in Europe: discoveries in medical science now offer unique opportunities to treat those affected and address the tragic societal burden they represent'.

New drug may allow early intervention in cystic fibrosis

Cystic fibrosis patients with normal to mildly impaired lung function may benefit from a new investigational drug designed to help prevent formation of the sticky mucus that is a hallmark of the disease, according to researchers involved in a phase 3 clinical trial of the drug. The placebo-controlled trial included 352 patients with cystic fibrosis.

The investigational medication denufosol can be given early in the disease, and may help delay the progression of lung disease in these patients, the researchers found.

Denufosol is an ion channel regulator which increases chloride secretion, inhibits sodium absorption and increases the beat frequency of the cilia to clear mucus. These effects combine to enhance airway hydration and help clear mucus.

'Although the lungs of children with cystic fibrosis are thought to be normal at birth, studies have demonstrated significant lung damage that occurs early in life in children suffering from cystic fibrosis,' said lead investigator Dr Frank Accurso, professor of pediatrics, University of Colorado School of Medicine, Denver.

He continued: 'Many patients continue to suffer progressive loss of lung function despite treatment of complications. Because denufosol can be used early in life, it offers hope for delaying or preventing the progressive changes that lead to irreversible airflow obstruction in cystic fibrosis patients.'

Accurso FJ, Moss RB, Wilmott RW, Anbar RD, Schaberg AE, Durham TA, Ramsey BW; the TIGER-1 Investigator Study Group (2010) Denufosol tetrasodium in patients with cystic fibrosis and normal to mildly impaired lung function. *Am J Respir Crit Care Med* Dec 17 [Epub ahead of print]

Nano-measurement of troponin predicts heart failure decline

Traditional risk prediction models for heart failure have relied on measurements of pro-B-type natriuretic peptide (BNP), but studies have provided inconsistent and often inaccurate results.

The introduction of highly sensitive troponin assays has improved accuracy and allowed the detection of even small concentration changes.

Xue et al (2011) assessed the prognostic value of a high-sensitive assay (within the ng/litre range) in patients admitted to hospital with heart failure. The investigation, part of the Veteran Affairs Effects of Therapy study, was performed at the San Diego Veteran Affairs Medical Center in California, USA, in which 144 patients with acute heart failure were followed from admission to 90 days post-discharge.

Troponin I and BNP levels were checked on admission, discharge, and up to four consecutive days during hospitalization. Of the 144 patients 38 reached the study's primary end point of mortality or heart failure-related readmission and 22 had died by 90 days.

Using the new assay, troponin measurements could be quantified in more than 99% of serum samples taken from all patients in the study.

Levels in the higher quartile ranges were significantly associated with increased risk of mortality and readmission; patients with increasing levels during treatment also had higher mortality rates than those with stable or decreasing levels. The associations with troponin were statistically significant, while those with BNP were not.

Xue Y, Clopton P, Peacock WF, Maisel AS (2011) Serial changes in high-sensitive troponin I predict outcome in patients with decompensated heart failure. *Eur J Heart Fail* **13**(1): 37–42