Briefing Notes

Motor Neurone Disease
Improving Access to Specialist Care

SITraN
Sheffield Institute for Translational Neuroscience

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Motor Neurone Disease (MND)

“There is no worse disease than MND.” - Dame Cicely Saunders OM DBE FRCP, founder of the modern hospice movement.

What is MND?
Motor neurone disease (MND) has first been described by the French neurologist Jean-Martin Charcot in 1869. MND is a term used for a group of diseases which are caused by the death of motor neurons in the brain and spinal cord leading to progressive muscle paralysis. MND is also known as Lou Gehrig’s or Charcot’s disease or Amyotrophic Lateral Sclerosis (ALS), the most common form of MND.

What happens when motor neurones die?
Motor neurones initiate and control our muscle movements. When motor neurones die, the muscles stop working and waste away. MND often starts with walking problems, followed by the loss of arm and hand movements, until swallowing, talking and finally breathing are affected. Patients die from weakness and paralysis of their breathing muscles on average 2 to 3 years after the onset of symptoms. MND is a progressively debilitating and fatal condition. Only 1 out of 10 diagnosed MND patients survives longer than five years. Many doctors regard MND as the worst disease in medicine.

Who has MND?
Any adult can get motor neurone disease at any age. In over 90% of cases the cause for the disease is unknown, only 5-10% of cases are inherited forms linked to certain genes, e.g. SOD1, C9orf72, TDP43, FUS. In the UK over 5,000 people suffer from MND, and 5 people die from MND every day. Worldwide over 750,000 people suffer from MND, men are more often affected than women. The longest known MND survivor is the eminent physicist Stephen Hawking. Other well-known MND patients include Mao Zedong, former England Manager Don Revie and the actor David Niven.

What are the treatment options?
As yet, there is no cure and no effective treatment for MND. Current treatment is based predominantly on symptom management and palliative care e.g. assisting breathing and nutrition and improving other disease related symptoms. At present, only one drug (riluzole) has been proven to slow the progression and modestly extend survival of MND patients by several months. Non-invasive ventilation (NIV) has also been shown to extend survival, as well as improving quality of life for some people with MND who experience problems with breathing.

What are the challenges to finding a treatment?
Due to its generally rapid course from diagnosis to death, the prevalence of MND in the population is only 6-8/100.000. MND is therefore regarded as a relatively rare “orphan disease” and as a consequence, there is limited interest in government or private sector funding for therapy development. This leaves academic institutions with a major role in developing new treatments for MND. Widely now regarded as a disease of ageing, it is expected that more cases of MND will be seen in our ageing population which further stresses the need for an effective treatment.

MND is a complex disease
Today, MND is regarded as a multisystem disorder not purely affecting motor neurones, although these specialised neurones tend to be affected earliest and most severely. At least 15 genes have so far been identified to be associated with MND and in up to 70% of inherited “familial” cases the gene causing the disease is known. What causes motor neurones to die in MND is not yet fully resolved and is best understood in the subtype of MND linked to a faulty SOD1 gene. A defective SOD1 gene is the known cause of 20% of inherited cases and approximately 2% of all MND cases.

For more information on MND visit: www.mndassociation.org or NHS choices: MND.
**TiM**

**Telehealth in Motor Neurone Disease**

**Improving access to specialist care**
People with motor neurone disease (MND) require specialist care which is currently provided by 19 MND Care centres in the UK and is often unavailable in their own community. Due to mobility problems, MND patients often struggle to travel the long distances to the specialist clinics. Moreover, depending on the course of the disease, patients might not need appointments at an MND clinic as often as patients with a more aggressive, faster progressing form of MND who need access to specialist care more frequently.

**How can telehealth improve care in motor neurone disease?**
By monitoring the health and wellbeing of their patients whilst they are at home, and alerting the specialist care team of any changes that require action, the patients will get specialist support as and when they need it. This could improve the patients’ health, improve the quality of life of both patients and their carers, and lead to more effective use of health resources.

**What is the TiM system?**
TiM is a telehealth system developed by MND specialists Dr Chris McDermott and Dr Esther Hobson at the Sheffield Institute for Translational Neuroscience (SITraN) with the input of MND patients and health professionals. The monitoring system will help the MND care team to collect information on their patients’ condition on a weekly basis and keep them up to date on their patients’ needs.

**How does it work?**
The TiM system runs as an Android app on a tablet computer, data is transferred via 3G mobile signal to the internet and stored on a server after analysis. The patients and carers answer a number of standardised questions on a weekly basis providing essential information on their mobility and problems they experience due to their condition. Their answers are automatically analysed and are available to their MND team who are automatically alerted to a change in their condition.

**What is the potential of TiM?**
SITraN’s Dr Chris McDermott, consultant neurologist at the Sheffield MND Care Centre, conceived the idea and developed the system: “We hope that the TiM system will improve the care and support we can offer patients and their carers by enabling us to respond to problems as they arise.”

Emily Goodall, a member of the Sheffield Motor Neurone Disorders Research Advisory Group, who represent patients and their families suffering from MND said: “This app has the potential to make a huge difference to patients with MND, particularly those living in rural areas where travelling to hospital is very difficult and you can feel very isolated.”
The TiM pilot study

Telehealth in Motor Neurone Disease: A single centre, randomised controlled feasibility and pilot study of the use of the TiM telehealth system to deliver highly specialised care in Motor Neurone Disease, at a distance

In order to understand how the TiM system could work a randomised controlled pilot study will be conducted. It will involve 40 patients who are cared for by the Sheffield Motor Neurone Disease care centre and their main informal carer. Half of the participants will use the telehealth system for a minimum of six months and maximum of eighteen months and information will be collected from patients, carers and their care team. This will include collecting clinical outcome measures, health resource use and the opinions and experience of using the system. All participants will continue to receive their usual care.

What is the aim of this pilot study?
It aims to determine the feasibility and acceptability of the telehealth system to patients, carers and their health care providers. If this pilot study is successful a larger study is planned to determine whether the TiM system could be effective in large numbers of patients. The pilot study will help determine how a larger trial could successfully evaluate the clinical and cost-effectiveness of the system and enable further development of the TiM system to be adopted into the care of patients with MND throughout the UK.

Who can take part in the TiM pilot study?
Patients with MND who are under the care of the MND clinic in Sheffield will be invited by letter to participate. Recruitment starts in September 2014 and the study is expected to last 18 months.

Who has funded and supported the TiM system and pilot study?
The TiM pilot study is a non-commercial, portfolio study supported by the UK Dementia and Neurodegenerative Diseases Clinical Research Network (DeNDRoN). It is funded through a National Institute for Health Research (NIHR) Doctoral Research Fellowship grant to Dr Esther Hobson. The TiM system has been developed in collaboration with clinicians at SITraN, the University of Sheffield, Sheffield Teaching Hospitals and an industry partner, Abbott Healthcare Products. Support has also been received from patients and carers, Devices for Dignity (D4D) and the University of Sheffield Telehealth and Care Technologies group. The Motor Neurone Disease Association has also provided funding support.
Telehealth in Motor Neurone Disease
Patients’ well-being is monitored with the help of an android app on a tablet computer.

*Credit: SITraN*

More screen grabs available

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Images can be downloaded from the SITraN image archive at http://sitran.dept.shef.ac.uk/news/picture-archive/clinical-research1/
For high-res images please contact m.feigenbutz@sheffield.ac.uk per email or phone 0114 222 2250.
Non-invasive ventilation in motor neurone disease

What is non-invasive ventilation?
Non-invasive ventilation (NIV) helps to support breathing in people with weak breathing muscles by slightly boosting the air flow into the lungs in tune with the natural breathing pattern. Generally, a portable ventilator is used which pumps filtered air into the person's lungs through a facemask or nasal tubes. A similar form of breathing support is often used for people with sleep apnoea when breathing repeatedly stops during sleep.

Why NIV? – NIV prolongs life for people with MND
As motor neurone disease (MND) progresses and breathing muscles weaken, people with MND can have problems with breathing. This can lead to poor sleep at night with negative effects on energy levels, general health and well-being. Several studies have shown that NIV improves both quality and length of life of people with MND who suffer from breathing problems. NIV is a treatment recommended by NICE for people with MND. The benefits of NIV are often noticeable within the first few days of using.

What is the challenge?
Neurologist and MND specialist Dr Christopher McDermott said: “Although NIV is an effective treatment, up to forty per cent of patients can struggle to use it. We conducted research with people using NIV and their carers, funded by the National Institute for Health Research (Research for Patient Benefit Programme) to understand why some people had difficulty using NIV. We identified that a group of people gave up early on because of practical issues and not fully being aware of the potential benefits of persevering. To put the research findings to good use we invited members of the public with experience of MND and NIV to work with web designers and film makers to create a new information resource, myNIV. The idea being that they could create something that contained all the things they wished they had known about NIV at the start to make it easier.”

Non-invasive ventilation: A typical ventilation machine with face mask and carrier bag.

Non-invasive ventilation: Air is pumped from a ventilator through a face mask into the lungs.
MyNIV
A web resource for people using non-invasive ventilation in MND

My NIV – A bespoke resource for people with MND
The new myNIV web resource mymnd.org.uk/myniv will give people with MND and their carers all the essential information and practical instructions they need to use NIV successfully. The website is very easy to navigate with clear and concise information; favourite pages can be shared or saved, turning the site into a bespoke information resource - myNIV. Instructional videos and step-by-step guides help users to make NIV work for them and tackle some of the problems they might encounter. Moreover, “top tips” from users for users give helpful insights for the everyday use of NIV.

Making NIV work
The myNIV web resource is thematically divided into three sections. The first part “What is NIV” explains the benefits of NIV and how it works with plenty of key information and important points to consider for patients who want to use NIV. The second section “Getting Started” gives practical information on getting to know the NIV machine and using it with ease and confidence, as well as on how to maintain the kit. Finally, the third section “Living with NIV” gives further advice and real-life insights into making NIV work whether at home or on holiday.
The myNIV webresource at http://mymnd.org.uk/myniv has been developed with the help of

References


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Images are available to download from:
SITraN
The Sheffield Institute for Translational Neuroscience

A Centre of Excellence for Research into MND & Neurodegenerative Diseases
SITraN is the first and only European Institute dedicated to and directly linking basic and clinical research into motor neurone disease (MND) and related neurodegenerative diseases. Our aim is to accelerate therapy development and offer new treatments to patients with neurological disorders.

A Unique Research Facility with a Unique History
SITraN is the result of an unprecedented fundraising initiative set in motion by patients and supporters of Professor Pam Shaw for a research institute dedicated to research into MND and related neurodegenerative disorders including Parkinson’s and Alzheimer’s disease. SITraN was officially opened in November 2010 by The Queen and Prince Philip.

Visionary Leadership, Wide-ranging Expertise and Collaborative Spirit
SITraN founder and director Professor Pamela Shaw has attracted and directs a multi-disciplinary team of eminent doctors and scientists working and collaborating in SITraN under one roof. SITraN scientists combine wide-ranging skills and expertise in clinical neurology, pathology, neuroscience, gene therapy, cell and molecular biology, genetics, biochemistry, bioinformatics, imaging including MRI, stem cell technology, disease modelling, pharmacology, drug screening and development, drug trialling, as well as clinical trials and applications.

Translating Research Discoveries into Benefits for Patients
SITraN is devoted to translational neuroscience – the rapid application of scientific research discoveries to the benefit and treatment of patients with neurological disorders. In order to accelerate the pace of therapy development, scientists and clinicians are working together closely with the added input and feedback from MND patients. Basic research is directed towards finding the disease causes and mechanisms, as well as testing the right targets to develop new and more effective treatments for patients.

Direct links to clinical research and patient care
The specialist MND Care and Research Centre for Motor Neurone Disorders established by Prof Shaw in 2003 in Sheffield serves as a focus of excellence for specialist patient care and clinical MND research, as well as a pivotal resource for the scientific research undertaken at SITraN.

Securing MND research excellence for the future
SITraN will provide a long-term international focus for research excellence in neurodegeneration by offering future clinicians and scientists highly specialised training. In addition to PhD training, two new Masters courses in Clinical Neurology and Translational Neuroscience have been established at SITraN with a combined intake of 40 students per year.

SITraN’s success so far:
Since its opening in 2010, SITraN has grown immensely, now employing close to 100 staff, including 10 professors, and 90 postgraduate students. SITraN scientists have produced over 300 original peer-reviewed research publications and attracted over £15M in research funding for neurodegenerative diseases. The gene therapy for SMA and the drug S(+) apomorphine for MND have received “orphan drug designation” and are being further trialled, as is a promising drug for Parkinson’s disease. While more potential therapies are being uncovered at SITraN, a range of assistive technologies are being developed and trialled to ensure better care and quality of life for MND patients.

For more information on SITraN visit our homepage: www.sheffield.ac.uk/sitran
**SITraN - Fast Facts**

**Name:** Sheffield Institute for Translational Neuroscience, SITraN  
**Location:** Sheffield, UK - within the central campus of The University of Sheffield  
**Opened:** November 2010 by The Queen and Prince Philip  
**Founder and Director:** Professor Dame Pamela Shaw DBE FRCP  
**Founding Patrons:** Sheffield Institute Foundation and The University of Sheffield  
**Honorary Patron:** The Duke of Devonshire KCVO CBE DL  
**USP:** a dedicated centre of research into MND and related neurodegenerative diseases, purpose-built to allow unique collaboration of a multi-disciplinary team of clinicians and scientists under one roof to link pure science to clinical research and accelerate therapy development.  
**Purpose:** Translation of scientific discoveries into new therapies for patients  
**Current Staff:** >100 staff, including 30 faculty academic staff  
**Postgraduate Students:** > 40 MSc students in 2013/14, > 50 PhD students since 2010  
**Research funding since 2010:** > £15 mio  
**Publications of SITraN researchers since 2010:** > 300 peer reviewed original research articles  
**SITraN pipeline:** “Orphan drug designation” for ReSagen (SMA), and S(+) apomorphine (MND)  
**Scientific Disciplines:** SITraN hosts 10 Professorships in  
Neurology – Pamela J Shaw; Translational Neuroscience – Mimoun Azzouz;  
Neuropathology – Paul Ince, Steve Wharton; Movement Disorders – Oliver Bandmann;  
Clinical Neuropsychology – Annalena Venneri; Clinical Neurology – Markus Reuber; Cerebrovascular Neurology – Arshad Majid; Machine learning and Computational Biology – Neil Lawrence;  
Bioinformatics and Computational Biology – Winston Hide;  
**Space:** 2800m$^2$ of research laboratories, offices, conference and research training facilities.  
**Pivotal Resources:**  
- Sheffield Care and Research Centre for Motor Neurone Disorders established and directed by Prof Shaw with a clinical database of >1000 MND patients, and biobanks of human material and tissues donated for research.  
- Host to Sheffield Brain Tissue Bank (SBTB) with material from > 1000 donors.  
- Host to Public and Patient Research Advisory Groups for MND and dementia research  
- Host Centre leading the UK Dementia (DeNDRoN) Clinical Studies Group for MND  
- MRC Centre for Developmental and Biomedical Genetics with access to animal models e.g. zebrafish, drosophila (fruit fly)  
- Access to facilities within University of Sheffield and Sheffield Teaching Hospitals  
- National and international collaborations
Profile: Dame Pamela Shaw
Professor of Neurology, Director of SITraN, University of Sheffield

Dedicated to improving the lives of patients with motor neuron disease (MND)
Professor Pamela Shaw has a worldwide reputation as an acclaimed Clinical Neurologist and Neuroscientist who has dedicated her life’s work to the exemplary care of MND patients and the understanding and development of treatments to combat the disease. She is one of the most renowned and experienced specialist consultants for motor neurone disease worldwide and has been at the forefront of MND scientific and clinical research for the last two decades. She has played a crucial role in influencing and implementing policies, guidelines and infrastructures in the UK to improve care for MND patients and put MND on the national healthcare agenda. In addition to numerous scientific awards and accolades, Pam Shaw’s services to Neuroscience were acknowledged by HM The Queen in the New Year’s Honours 2014 with a Damehood.

Promoting MND research and specialist care for MND patients
After her very successful medical and specialist training in neurology, Pam Shaw took up a post as Honorary Consultant Neurologist in 1991 and directed a specialist MND care and research centre in Newcastle. She embarked on a major clinical and laboratory programme of MND research funded by the Wellcome Trust which she transferred to the University of Sheffield in 2000 accepting a Chair in Neurology. In 2003, with support from the MND Association, she established and continues to direct one of the major national Care and Research Centres for Motor Neurone Disorders in Sheffield which serves as a focus of excellence for MND care and clinical research. She continues to lead the systematic development of clinical research nationwide to tackle major problems faced by MND patients where new approaches and new evidence-based guidelines are required.

Putting motor neurone disease (MND) on the agenda - nationwide
Prof Shaw’s influence has ensured that neurodegenerative diseases are included in the NHS Clinical Research Networks Initiative and she has helped to implement MND care guidelines by providing the required evidence through clinical research. Prof Shaw has taken part in more than 15 MND clinical trials and played a key role in promoting into clinical practice the only known MND treatments to prolong survival, riluzole and non-invasive ventilation. For 8 years she has led the Clinical Studies Group for MND within the NIHR Dementia and Neurodegenerative Diseases (DeNDRoN) network and helped to develop a high quality UK clinical research network (UKCRN) linking 19 MND Care and Research Centres and establishing a network of 10 UK centres experienced in MND clinical trials.

SITraN, a research centre to reach the ultimate goal: Finding a cure for MND
The opening of SITraN by Her Majesty The Queen in 2010 was the culmination of over a decade’s work, for which Pam Shaw provided both the overarching vision and the relentless practical driving force. The unique research institute is dedicated to accelerating the therapy development and improving care and quality of life for patients with MND and other neurological disorders. Scientific research undertaken at SITraN is firmly linked to the clinical research at the specialist MND Care and Research Centre in Sheffield which also serves as a pivotal resource.

Professor Shaw has authored more than 350 publications of original research Google Scholar H-index 69), reviews and book chapters and has edited several books on MND. She has supervised the research training of more than 100 individuals from medicine and science including 30 PhD students and since 1983 has generated more than £50 million in research funding.

For more information on Professor Shaw, her research and biography, visit: http://sitran.dept.shef.ac.uk/people/shaw