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TREAT-NMD newsletter no. 47
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welcome

Welcome to the latest TREAT-NMD newsletter. This week's edition features a job opportunity in Newcastle and some interesting news from some of the national registries that partner with us in the TREAT-NMD global patient registries for DMD and SMA. Next week the TREAT-NMD partners are all coming together in Budapest for our fourth Governing Board meeting, so look out for a meeting report in an upcoming newsletter!

As always, we hope you enjoy the newsletter and look forward to hearing your comments - write to info@treat-nmd.eu with anything you'd like to say. Feel free to forward this message to anybody you think might find it of interest, or invite them to sign up to receive the newsletter by visiting our website. Back-issues of this newsletter can be found on our website at http://www.treat-nmd.eu/patients/news/ezine-archive/

Best wishes,

Katie, Volker, Steve, Emma, Rachel and Samantha: the Newcastle TREAT-NMD team

at a glance...

Job opportunity in Newcastle: 4-year clinical research training post

4-year clinical research training post
Institute of Human Genetics, Newcastle University

An exciting opportunity has arisen within the Institute of Human Genetics in Newcastle for a four year clinical research training post funded by the Wellcome Trust in translational medicine as it relates to inherited neuromuscular diseases. This is a growing field, and the Neuromuscular team at the Institute of Human Genetics in Newcastle is involved both in ongoing trials in muscular dystrophy as well as more generally via the TREAT-NMD Network of Excellence for Neuromuscular Disorders and the MRC centre for Neuromuscular Diseases.

Note: candidates must be medically qualified, eligible for GMC registration and in or recently having completed a UK recognised clinical training pathway.

The successful candidate will follow a taught course in techniques related to translational medicine for one year, much of it delivered by international pharmaceutical experts, before embarking on a three year research position carrying out a PhD in the field of translational research. The post would be ideal for trainees in Neurology, Paediatric Neurology or Genetics.

Informal enquiries can be made to kate.bushby@ncl.ac.uk, volker.straub@ncl.ac.uk or hanns.lochmuller@ncl.ac.uk, but please note we will be out of office from 2nd-8th Feb at the TREAT-NMD governing board meeting.

The closing date is 15th February.
For full details see http://www15.i-grasp.com/fe/tpl_newcastle02.asp?newms=%20jj&id=27020

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Update from the SMA registry in the USA

The International Spinal Muscular Atrophy Patient Registry in Indiana is a US-based initiative that is working with TREAT-NMD to provide data to the TREAT-NMD Global SMA registry. All patients registered in the registry will have their (encrypted) data forwarded to the TREAT-NMD Global Registry, which enables them to be considered for future international clinical trials.

If you are are located in the USA or in a country where there is no TREAT-NMD national registry (see http://www.treat-nmd.eu/nationalregistries) you can sign up to the Indiana registry, which recently launched a new website that allows full online interaction.

The International SMA Registry is currently helping to recruit for several US-based studies, including:

Pilot Study of Biomarkers for Spinal Muscular Atrophy (SMA) (BforSMA): The purpose of BforSMA is to identify potential biomarkers (measures) that can be used to evaluate SMA disease severity. A

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"biomarker" is something that can be measured to assess disease and treatments (cholesterol level is a biomarker for heart health).
The Role of Motor Unit Number Estimation (MUNE) in Adults with Spinal Muscular Atrophy: MUNE is a research study that follows the course of motor neuron loss in adults with SMA using the electrophysiological technique of motor unit number estimation (MUNE).
Prospective Controlled Trial of Valproic Acid in Ambulant Adults with Spinal Muscular Atrophy (VALIANT SMA) Study: VALIANT SMA is a research study to assess the efficacy of oral Valproic Acid (VPA) in adult patients with SMA.
Outcome and Effect of Pregnancy in Spinal Muscular Atrophy: This is a research study of the effects of pregnancy and delivery on adult women with SMA.
Phase I/II Trial of Valproic Acid and Carnitine in Infants with Spinal Muscular Atrophy Type I (CARNI-VAL Type I): Carni-Val Type I is a research study to evaluate the combination of Valproic Acid (VPA) and L-Carnitine for the treatment of SMA in infants with SMA type I. These clinical trials are being conducted at several universities across the United States and one location in Canada.
To find out more about all of the open studies the SMA Registry is helping recruit for, please visit https://smaregistry.iu.edu/Participant/researchStudies.asp. If you are interested in any of the studies listed, you can join the Registry and request to have your contact information forwarded to the researchers conducting that study.

About the Registry:
The International Spinal Muscular Atrophy Patient Registry (the Registry) was founded in 1986 at Indiana University. The Registry connects patients and families interested in participating in research and researchers interested in studying SMA. The Registry contains information from over 1,600 families and over 1,700 individuals with SMA from all over the world and continues to grow. The Registry has helped recruit participants for clinical trials and has provided data for important SMA research studies. The Registry helps centralize information on this rare genetic disease, provides families a way to learn about research studies and provides researchers a way to find research participants.
Individuals and families affected by SMA are invited to join the Registry. Participants are asked to complete questionnaires about the symptoms, treatment, medications, and other experiences with SMA.
Participant information is stored in a secure database. Researchers who are interested in studying SMA can request two types of data from the Registry, de-identified information and identifiable information. De-identified information does not contain any names or personal identifiers, and can be given to researchers without having to contact Registry families. Identifiable information includes information that can identify you and will never be released without getting your written permission to do so. Identifiable information includes data such as names and dates of birth. Some researchers may also request contact with families to obtain specific information or to request participation in a research study. In these instances, the Registry will contact each potential participant to ask if they are willing to share their identifiable information for a research project.

The SMA Registry is supported by the Patient Advisory Group of the International Coordinating Committee for SMA Clinical Trials which includes Families of SMA, Fight SMA, Muscular Dystrophy Association, SMA Foundation, and other SMA advocacy groups.

To sign up, visit https://smaregistry.iu.edu

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UK Duchenne patient registry can now accept Irish patients

The TREAT-NMD global registry for Duchenne muscular dystrophy (DMD) collects data from patients via many different national registries across the world, all of which feed their data into the TREAT-NMD global database (see http://www.treat-nmd.eu/nationalregistries). Other TREAT-NMD national registries collect data for patients with spinal muscular atrophy (SMA), and registries for other conditions are in preparation.

In the UK, the national registry for DMD that feeds information to the TREAT-NMD global registry is available online at www.dmdregistry.org. It is run by the patient support group Action Duchenne. The UK registry is pleased to announce that it can now also accept registrations from patients in Ireland. The registry acts as a database for patients diagnosed with Duchenne or Becker Muscular Dystrophy, and because of the precise genetic and medical information it contains, volunteers can be drawn from the registry for clinical research trials. There are approximately 150-200 boys in Ireland with DMD who can now be included in the registry, following the collaborative working of Duchenne Ireland, Action Duchenne and the genetics laboratories in London and Dublin. The initiative is also supported by Muscular Dystrophy Ireland. Details of all those registered remain anonymous and all data is kept confidential.

The registry will facilitate research by collecting relevant data and making it available for specified research projects. Professionals will be able to use the information provided to understand the disease better to develop treatments. It will also strengthen contact between health professionals, researchers and patients and provide a means of feedback of best practice in patient care.

Patients or their parents can join the registry by filling in their details online at www.dmdregistry.org. More information on the steps to follow can be found on the website itself. More information about the TREAT-NMD patient registries initiative in general can be found at http://www.treat-nmd.eu/patientregistries.

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Press release from AVI BioPharma on proof-of-principle study of AVI-4568

TREAT-NMD is in discussions with a range of pharmaceutical companies regarding the possibility of using the TREAT-NMD infrastructure to accelerate clinical trials. In addition to this, several TREAT-NMD partners are involved with trials in Duchenne muscular dystrophy that are already ongoing, including those run by PTC Therapeutics, Prosensa and AVI BioPharma.

AVI BioPharma recently released the following press release on its phase I proof-of-principle trial of the exon skipping product AVI-4658.

AVI BioPharma Announces Successful Clinical Trial of AVI-4658 for Treatment of Duchenne Muscular Dystrophy by Exon Skipping

PORTLAND, OR - January 21, 2009 - AVI BioPharma, Inc. (NASDAQ: AVII), a developer of RNA-based drugs, today announced results from a Phase 1 trial of its drug candidate AVI-4658 for the treatment of Duchenne muscular dystrophy (DMD) by exon skipping. Biopsy data showed that injection of the drug into the muscles of a series of DMD patients successfully induced dystrophin production in each patient.

The proof of principle, single dose escalation study tested the effect of an intramuscular (IM) injection of AVI-4658 in boys with DMD. Each patient received an injection of 0.09 or 0.9 mg of AVI-4658 into the exterior digitorum brevis muscle of one foot and an injection of saline as placebo into the corresponding muscle of the opposite foot to provide an internal treatment comparison. Three to four weeks later, each injected muscle was biopsied and examined for evidence of dystrophin production. Results demonstrated that injection of AVI-4658 elicited dystrophin production in a dose dependent manner in all treated patients. Further, the drug was well tolerated, with no significant detectable drug-related adverse events.

The clinical study was performed in the UK by members of the MDEX Consortium led by Professor Francesco Muntoni. Professor Muntoni commented, "As a clinician and scientist, I am very pleased by these findings and the prospects they offer for the potential treatment of this serious, life threatening condition. Biopsies from muscles injected with the higher dose of test drug showed an unequivocal, widespread and robust response in terms of number of dystrophin positive muscle fibers. We will publish these exciting data in a peer-reviewed journal in due course."

Patients with DMD have a very low capacity to make dystrophin. In general, and in this study, DMD patients have less than 5% dystrophin positive muscle fibers prior to treatment.

"These are promising data which support the continued development of AVI-4658 as a potential exon skipping treatment for DMD and are highly competitive with data disclosed by other companies working in this field," said Leslie Hudson, Ph.D., President and Chief Executive Officer of AVI BioPharma. "A multi-dose, dose escalation trial to examine the efficacy of the drug candidate following systemic administration (IV) -- also in collaboration with the MDEX Consortium -- was opened in December 2008 and our collaborators have begun the work up of the first cohort of DMD patients prior to dosing."

DMD is an incurable muscle-wasting disease associated with errors in the gene that makes dystrophin, a protein that plays a key structural role in muscle fiber function. The drug candidate AVI-4658 is designed to skip exon 51 of the dystrophin gene, allowing for restoration of the reading frame in the mRNA sequence. Restoration of dystrophin production achieved by skipping this exon may improve or significantly slow the disease process, thus prolonging and improving the quality of life for the affected patient population. It is important to note that different mutations in the dystrophin gene require different oligonucleotide drugs. In principle, approximately 80% of all DMD patients could be treated with exon-skipping drugs. AVI-4658, and the four related exon-skipping drugs under development in AVI BioPharma could be used to treat more than half of this population -- if they prove to be effective -- with a potential market value of approximately $2.0 billion. AVI BioPharma was granted orphan drug designation for AVI-4658 by the U.S. Food and Drug Administration in November 2007 and by the European Medicines Agency (EMEA) in December 2008.

The IM injection trial was funded by the Department of Health (UK) and conducted by the members of the MDEX Consortium, led by Professor Muntoni at Imperial College Healthcare NHS Trust facilities. Imperial College London is the sponsor for the trial, with AVI BioPharma serving as its clinical development collaborator.

For more information on AVI see www.avibio.com. For more information on the MDEX consortium see www.mdex.org.uk.

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