



IT'S TIME TO STOP WASTING!

9th International Annual Duchenne Conference

Frequently Asked Questions



Holiday Inn
Coram St Bloomsbury London

WELCOME

Thank you for coming to this years International Duchenne Conference organised by Action Duchenne.

We have tried to organise this years Conference to reflect the Aims of our Charity

- 1) To be a leader in supporting innovative research to cure and treat Duchenne
- 2) To work as a partner with organisations providing resources to help to improve the length and Quality of Life of those living with Duchenne
- 3) To be led by parents and young people affected by Duchenne and Becker Muscular Dystrophy

We are pleased to see the ongoing progress of all the Exon Skipping research and clinical trials. Action Duchenne from 2003 has been involved in both funding and supporting this very promising avenue of research. We are delighted to be able to invite Biopharma including GSK, Prosensa, AVI and the members of the MDEX Consortium who are now progressing this work towards drugs for our sons.

Action Duchenne has recently agreed a £186K grant to support a new gene research project with George Dickson's group in London that could benefit all boys and there are other very promising new research projects using AAV technology presenting at the Conference.

Action Duchenne with our partners in the Pilot Trials Now consortium have funded new clinical trials for IGF1 and Sildenafil. These are trials on known drugs that might benefit all boys with DMD. We are looking forward to hearing progress updates.

In the last year there has been further research and now an extensive UK trial organised by John Bourke in Newcastle to investigate the benefits of ACE inhibitors and beta blockers for the heart. These drugs are already recommended for use with Duchenne patients but we need more data on just how well they might work and when they should be administered. We welcome several international researchers in this field to this years Conference.

85% of all our funding goes directly into research or to support young people living with Duchenne. Action Duchenne spent 56p for every £ we raised on cutting edge research or clinical trials and 29p for every £ on Special projects to support young people in school, in community programmes and campaigning for better standards of care

We are proud of the fantastic Include Duchenne project that has worked with children in over 80 schools and has helped many Duchenne youngsters with behaviour and learning difficulties. Young people through our Genius programme have made films and like Tyran produced exciting and ground breaking photography exhibitions in the community.

We are thrilled to have been awarded a £500K grant by the Big Lottery to start up our Takin' Charge project that will produce new innovative materials and support for those young people going through transition to adulthood.

As always the Charity has been lobbying the Governments of the UK, Scotland, NI and Wales to improve research funding and access to specialist medical care. We launched our DMD Centres project that helps families to find special care across the UK www.dmdcentres.org

We have asked some FAQ's of researchers and clinicians to help you to understand the incredible scope and development of Research in all fields. Please contact them and meet with them at the Conference they are always willing to discuss their projects.

Everyone at Action Duchenne hopes you have a great Conference and that you make new friends and meet up with old buddies.

Nick Catlin
CEO Action Duchenne

A big thank you to our Trustees who are all parents, close relatives or are living with Duchenne:

Godfrey Adams, Paul Ackroyd, Divyesh Popat, Dr Karl Bettelheim, Tony Levene, Mark Silverman, Damian Culhane, Paul Chandler, Mary Down, Jayne Evans and John Hastie.

Special Thanks to our expert Scientific Advisory Board

Dame Professor Kay Davies, professor Matthew Wood, Professor Kate Bushby and professor Eric Hoffman.

To our National Advocacy Council members chaired by parent Kathy Weddell and our DMD Registry steering committee Chaired by Dr Steve Abbs

A Big thanks to all our Staff:

Kelly Ferguson, Kate Angus, Esther Derber, Iain Clarke, Diana Ribeiro, Owen Callaghan, Dean Widd, Angela Stringer, Eilidh Macpherson, Janet Hoskin (Decipha), Celine Barry. The invaluable support from Andreina West at PR Artistry, Anton Faulconbridge at Rantmedia, Claire Hersant at Voyant Design, Dan Grasshan and Sarah Hewitt at Give Us the Fish.

Friday 4th from 7.30pm The Action Duchenne Gala dinner is hosted by stand up Comedienne Francesca Martinez in the Turner and Booker rooms. There will be a raffle and celebration of all things Duchenne.

Last year Francesca was commissioned to write another episode of her BBC sitcom series. Francesca toured Australia for two months, with shows in Melbourne, Sydney and Adelaide, and received a special commendation for Best Established Comedian at the Adelaide Fringe Awards. Francesca appeared on Radio Four's Word Of Mouth presented by Michael Rosen. She was invited to be a regular guest on Radio Four's Broadcasting House after successful appearances alongside Jon Snow, Michael Portillo and Robert Fisk. Francesca appeared on Radio 4's Loose Ends with Emma Freud, Clive Anderson and Ardal O'Hanlon. She was one of the main contributors in the BBC2 documentary 'Are You Having A Laugh' which looks at the portrayal of disability on television.

There is an Elephant in the Room

Friday 9.00 – 9.30

Dr Karl Bettelheim Action Duchenne

Karl is a retired research scientist that has a grandson with Duchenne. He is a Trustee at Action Duchenne, a member of the Charities Research sub committee and writes a regular research blog <http://www.actionduchenne.org/karlsresearchupdate> on the Charities website.

What do NHS Reforms mean for patient care? Changing the map for Duchenne

Friday 9.30 – 10.30

Arlene Wilkie, Neurological Alliance

The Neurological Alliance is a campaigning organisation – the collective voice of over 50 brain and spine charities and other key stakeholders in England, Arlene is the Chief Executive of the Alliance. Action Duchenne has recently joined the Neurological Alliance, and is involved in it's joint policy work.

Alastair Kent, Rare Disease UK, (RDUK).

RDUK is the national alliance for people with rare diseases and all who support them. Alastair Kent, is the Chair of RDUK and leads on government liaison and strategic planning. Action Duchenne has been working closely with RDUK over the last two years to lobby for improved research funding for rare diseases and to improve access to patient care.

Eilidh Macpherson, Action Duchenne

Eilidh is the Campaigns and Advocacy Manager for Action Duchenne, responsible for parliamentary advocacy and working with our members to develop policy responses. She's recently been working on the new project www.dmdcentres.org that maps specialist muscle centre service provision across the UK. For more information you can get in touch with her on eilidh@actionduchenne.org.

Biomarkers for clinical trials

Friday 11.00 – 11.30

Dr Sebahattin Cirak UCL

- 1) At what stage is this research?
Discovery Phase
- 2) What are the aims of your study?
The aim of the study is to identify modifier genes for DMD and CoVI related myopathies and biomarkers which can be used to monitor disease progress, drug response and that are in future suitable as surrogate markers for clinical trials.
- 3) Who is funding this research?
The European Union FP7
- 4) If at the stage of a clinical trial who is eligible to participate?
All patients with DMD , BMD, CoVI related myopathy and other muscular dystrophies can participate.
- 5) When will your study be completed?
End of 2012
- 6) Are there any results published?
There first publications published from the Dutch collaborators.
- 7) Will there be any future clinical trials relating to your research and if so when?
We hope that the biomarker can be used in future trials.
- 8) Where can I find out more about your research?
Via the Treat-NMD web side. <http://www.bio-nmd.eu/>

Macrophages: A minimally invasive tool for monitoring collagen VI myopathies, Francesca Gualandi MD, PhD; Rosa Curci PhD; Patrizia Sabatelli BSc; Elena Martoni PhD; Matteo Bovolenta PhD; Mario N. Maraldi PhD; Luciano Merlini MD, Alessandra Ferlini MD, PhD
In Muscle and Nerve, Wiley, vol. 44; iss 1; pp 80-84 (July 2011)

Serum matrix metalloproteinase-9 (MMP-9) as a biomarker for monitoring Duchenne muscular dystrophy (DMD) disease progression, P.A.C. 't Hoen, V.D. Nadarajah, M. van Putten, A. Chaouch, P. Garrood, V. Straub, H.B. Ginjaar, A.M. Aartsma-Rus, G.J.B. van Ommen, J.T. den Dunnen, H. Lochmüller

In Journal of Neuromuscular Disorders, Elsevier, vol. 21, iss 9-10, p. 656 (Oct 2011)

Pharmacogenomics: Role in Medicines Approval and Clinical Use, G. Novelli, P. Borgiani, C. Ciccacci, N. Di Daniele, G. Sirugo, M. Papaluca Amati
In Public Health Genomics, Karger, vol. 13, iss. 5, pp. 284-291 (Oct 2009)

Diversity of human skeletal muscle in health and disease: Contribution of proteomics, Cecilia Gelfia, Michele Vassoa, Paolo Cerretelli
In Journal of Proteomics, Elsevier, vol. 74, iss. 6, pp. 774-795 (May 2011)

Setup for human sera MALDI profiling: The case of rhEPO treatment, Chiara Fania, Michele Vasso, Enrica Torretta, Paul Robach, Gaetano Cairo, Carsten Lundby, Cecilia Gelfi

I. In Electrophoresis, Wiley, Vol 32, iss. 13; pp. 1715-1727 (June 2011)

Regular updates and patient newsletters published at: <http://www.bio-nmd.eu/patient-newsletters/>

Caring for the Heart

Friday 11.00 – 11.30

Dr John Bourke Newcastle

- 1) At what stage is this research?
The 'Duchenne muscular dystrophy heart protection trial' is now actively recruiting at five hospitals in the UK.
- 2) What are the aims of your study?
The 'Heart Protection Trial' aims to find out whether the weakness in heart pumping (cardiomyopathy), which occurs in almost all boys with DMD, can be prevented by taking two commonly used heart medicines daily in combination from the time before there is any evidence of heart involvement.
- 3) Who is funding this research?
The British Heart Foundation
- 4) If at the stage of a clinical trial who is eligible to participate?
Boys with genetically confirmed DMD - aged between 7 and 12 years (less than 13th birthday) - who have not been taking heart therapies already (steroid therapies don't count) and whose hearts are normal [on the basis of a simple heart scan (echocardiogram)] are suitable for the trial. The study requires heart assessments every six months – that is one more heart-clinic visits than normally required for boys with DMD per year.
- 5) When will your study be completed?
The trial is planned to run for 5 years. Boys will be recruited over the next two years and the trial will end when the last boy has been followed up for a minimum of three years.
- 6) Are there any results published?
No results are available at this time, since the study only started in July 2011.
However, some years ago a French trial found that taking just one of the two medicines included in this research (the ACE-inhibitor, perindopril) was beneficial for the heart in boys with DMD. There are good reasons to think that combination therapy (perindopril & the beta blocker, bisoprolol) will offer greater 'heart protection'.
- 7) Will there be any future clinical trials relating to your research and if so when?
Plans for any additional or follow-on trials of heart treatments will depend on the results of this study as they become available.
- 8) Where can I find out more about your research?
Further information about this particular trial can be obtained from Ms G Watson, Trial Manager [email: gillian.watson@ncl.ac.uk; phone: 0191-222 8813; fax: 0191-222 8901]; Dr John Bourke, Chief Investigator for the whole trial (email: john.bourke@nuth.nhs.uk) or from the principle investigator for the trial at which ever of the five hospitals is nearest to you:
 1. Great Ormond Street Foundation Trust, London (Prof F Muntoni :f.muntoni@ich.ucl.ac.uk)
 2. Newcastle upon Tyne NHS Foundation Trust, Newcastle upon Tyne (Dr JP Bourke: john.bourke@nuth.nhs.uk)
 3. John Radcliffe Foundation Trust (Dr S Jayawant:sandeep.jawayant@orh.nhs.uk)
 4. Birmingham Heartlands & Solihull NHS Trust (Dr H Roper:helen.roper@heartofengland.nhs.uk)
 5. Birmingham Children's Hospital NHS Trust (Dr M Smith:martinsmith3@nhs.net)

Introduction to Exon Skipping

Friday 11.30 – 12.00

Professor Steve Wilton Western Australia

- 1) At what stage is this research?
Exon skipping has moved from a concept to clinical trials in little more than a decade. While this is very fast in the standard world of drug development, there is still a long way to go, and the use of antisense oligomers (genetic bandaids) has yet to be shown to make a difference in boys/men with DMD.
- 2) What are the aims of your study?
To develop and refine exon skipping strategies for all DMD mutations that may benefit from this approach, not just commonly encountered deletions in the dystrophin mutation hotspots.
To develop new and innovative approaches to make exon skipping as effective as possible
- 3) Who is funding this research?
National Institutes of Health USA
Muscular Dystrophy Association of USA
Muscular Dystrophy Association of Western Australia
Medical Health Research Infrastructure Fund of Western Australia
National Health and Medical Research Council of Australia
Duchenne Ireland
James and Matthew Foundation
- 4) If at the stage of a clinical trial who is eligible to participate?
The State Government of Western Australia has provided funds to establish a on-stop muscular dystrophy clinic for DMD in anticipation of commencing clinical trials in 2012.
Initial trials will involve non-ambulant patient, to address safety issues, and then we hope to first address any patient whose mutation may be responsive to splice switching intervention. Local boys will be seen first but we hope to transfer materials to other clinics across Australia and the world as resources permit.

5) When will your study be completed?

I wish I knew! It will be an on-going process taking years, especially when we consider this should be regarded as a form of personalized medicine.

6) Are there any results published?

The MDEX trials using a bandaid directed to exon 51 was published in Lancet in July, 2011

Proof-of-concept showing unequivocal improvements in a severe animal model (dko) was published as Goyenville A, et al. (2010) . Molecular Therapy; 18: 198-205.

New work to be presented at the conference is still in press, and describes alternative exon skipping strategies and unexpected trends in bandaid design.

7) Will there be any future clinical trials relating to your research and if so when?

As soon as possible, resources permitting

8) Where can I find out more about your research?

www.anri.org.au

March of the Young men

Friday 11.30 – 12.00

Gordon McClurg

Gordon (32) was born in Belfast and, aged three, was diagnosed with Duchenne Muscular Dystrophy. Throughout this muscle wasting disorder, Gordon has focussed on leading a normal active family life. This has taken him through 14 years of mainstream education and four years of a Business Degree at the University of Northumbria from which he graduated with Honours in 2001. Along the way he has encountered many obstacles to living a normal life and has developed his own techniques for overcoming them. He now lives independently in a purpose designed bungalow with continuous care support from his own team of personal care assistants. Although Gordon has severe physical limitations, he leads an active professional & social life and has established Care Management Services to provide advice, support and Care Services to other young disabled people who wish to live independently.

Care Management Services

Care Management Services supports people from across the UK, with a variety of neuro muscular conditions, to live fully independent lives in their own adapted homes, attend college or university, undertake professional careers, have access to top quality healthcare and an active social life, travelling at home and abroad. Meet CMS Managing Director, Gordon (32 with Duchenne Muscular Dystrophy), at the Conference and book yourself in for his Workshop on 'Independent Living'. Find out more by clicking on the link to the CMS website:- www.caremanagementservices.co.uk

Conference Talk Abstract

March of the Young Men (25 mins)

In recent months, developments in trials of real treatments have become a regular feature of our e-mails, frequently visited websites, professional journals and tabloid press. Still, the advancement towards widely available treatments and a cure is hindered by underfunding from both public and charitable sectors. This does not just arise from the poor economic situation and expenditure cuts but reflects the fact that DMD is not a sexy, newsworthy topic for coverage. Even senior practitioners in neuromuscular and related fields still express reservations about the rationale of investing valuable time and resources in those with a continuing degenerative illness based on the quality of their lives and contribution to society. Underfunding affects access to trials, access to treatment, access to clinical care and management and access to opportunities to live the lives we want. We, the new generation of young men with DMD who have benefitted most from these advances, shoulder a peculiar responsibility to reach the media through our determination and achievements to challenge and reverse this perception and excite investment for those who follow.

Independent Living Workshop (1 Hour)

Many young people with Duchenne and their families lower aspiration and expectation as they approach adulthood, beset by health problems and physical constraints. Lack of information, support and resources are also hindrances keeping young men from reaching their real potential. This workshop deals with setting personal goals and developing strategies and methods to move from adolescence to adulthood and even living independently by making your own choices and following them through. It captures your ideas and ambitions and gives you guidance on how to make them real and where to find support. Look out for the 'Independent Living Action Plan' & have a personal consultation with Gordon McClurg on the Care Management Services Exhibition Stand.

Duchenne Alliance

Friday 12.30 – 1.00

Christine McSherry Jett Foundation USA and Carlo Rago USA

We are a member of the Duchenne Alliance and we are using the Duchenne Dashboard to optimize our funding strategies. The Alliance has a variety of interests that reflect the missions of the members including - but not limited to - basic science, preclinical therapeutics, clinical trials, quality of care, and quality of life. Proposals submitted through the Duchenne Dashboard enjoy several advantages: 1. transparency - the author can determine who sees the submission, 2. thorough review - web 2.0 features allow crowd-sourcing of scientific advisors, 3. rapid response - the Alliance operates in real-time and the author will receive feedback and funding accordingly, 4. single portal - one door provides instant access to all participating Foundations. Submission of NIH grants with scores close to the pay line are strongly encouraged.

<http://duchennealliance.org/>

christine@jettfoundation.org

Improving the design of antisense drugs using peptides

Friday 12.00 – 1.00

Dr Mike Gait and Professor Matthew Wood

- 1) **At what stage is this research?**
My group in Cambridge is involved in basic research to find better ways of delivering antisense drugs, particularly those targeting DMD and other neuromuscular diseases.
- 2) **What are the aims of your study?**
Our aim is to design peptides as covalent conjugates of PMO and other antisense oligonucleotides to enhance their delivery into muscle cells and thus improve their effectiveness.
- 3) **Who is funding this research?**
Our work is funded primarily by the Medical Research Council, but there are also grants awarded to us as part of a consortium from the Wellcome Trust/Health Innovation Challenge Fund as well the AFM (French muscular dystrophies association). We collaborate with the laboratory of Matthew Wood in Oxford and also with several other research groups in the MDEX clinical consortium.
- 4) **If at the stage of a clinical trial who is eligible to participate?**
The work is not yet at the stage of clinical approval.
- 5) **When will your study be completed?**
We hope to continue for some time to develop peptides suitable for use with different diseases and different antisense oligonucleotide types, but we believe that a candidate peptide for clinical development in DMD is within months of being finalised.
- 6) **Are there any results published?**
Matthew Wood's and my lab jointly published a paper recently in Molecular Therapy representing the basis of a peptide lead compound using a mouse model of DMD:
Yin et al. (2011) Molecular Therapy, Volume 19 (7) pages I295-I303.
- 7) **Will there be any future clinical trials relating to your research and if so when?**
We hope that a candidate peptide may become suitable for use in the clinic for DMD treatment as a conjugate to a PMO oligonucleotide within about 2 years, if it passes all the safety tests and if we find it represents a potential improvement over the current type of PMO oligonucleotide used clinically. In this case, our MDEX clinical partners would expect to recruit a few patients for a phase I clinical trial in the UK sometime in 2013.
- 8) **Where can I find out more about your research?**
<http://www2.mrc-lmb.cam.ac.uk/group-leaders/a-to-g/m-gait>

Using AAV Vectors and microdystrophin

Friday 2.00 – 2.30

Professor George Dickson Royal Holloway College London

- 1) **At what stage is this research?**
Our research is at a so-called pre-clinical translational stage of development, but the process of enabling a clinical trial is on the horizon.
- 2) **What are the aims of your study?**
We aim to develop clinically-powerful but safe and non-immunological gene therapy medicine which is wholly optimised and targeted to cure dystrophin deficiency potentially in all DMD patients.
- 3) **Who is funding this research?**
This research is currently funded by MDC and Action Duchenne, but in the past has also been funded by AFM, Duchenne Ireland, and the Daiwa Foundation.
- 4) **If at the stage of a clinical trial who is eligible to participate?**
We are not yet at the clinical trial stage but the therapy should be appropriate for all DMD patients. Initial safety trials would probably initially recruit older boys with DMD for ethical reasons. Ultimately treatment would be delivered at as young an age as safely acceptable.
- 5) **When will your study be completed?**
We have an ongoing programme. Logistically we need to create a clinical translational network for AAV microdystrophin akin to the MDEX network for exon skipping.
- 6) **Are there any results published?**
Yes, we have extensive published work. Searching for "G Dickson" on Pubmed will bring up most of these.
- 7) **Will there be any future clinical trials relating to your research and if so when?**
This question is aligned to Question 5 above. We have an ongoing programme. Logistically we need to create a clinical translational network for AAV microdystrophin akin to the MDEX network for exon skipping. Clinical trials might be in Paris, or London, or Newcastle, or even in the US
- 8) **Where can I find out more about your research?**
Contact Professor Dickson onDirect Tel: 44-(0)1784-443545; Mobile: 07944-137754; Fax:44-(0)1784 414224; Email: g.dickson@rhul.ac.uk. Web site: http://pure.rhul.ac.uk/portal/en/persons/george-dickson_f8b3f42b-1968-4521-8c46-f9250bd7fc63.html

Bone protection for children with DMD treated with corticosteroids

Friday 2.00 – 2.30

Dr Ros Quinliven

Ros has recently published a leaflet for families on this subject that is available at the Conference.

In terms of research, Ros is currently leading a pilot study of a new AFO/footwear combination to stabilise gait in children with DMD. The research is being held in ORLAU, Robert Jones and Agnes Hunt NHS Trust, Oswestry. The trial is funded by the Muscular Dystrophy Campaign and is open for recruitment. To be eligible children need to be toe walking. The trial requires 3 visits over 12 weeks. Please contact Will Bromwich or Nick Emery 01691 404000 if interested.

MDEX Clinical Trial programme and study 28

Friday 2.30 – 3.00

Professor Francesco Muntoni UCL and Great Ormond Street London

- 1) **At what stage is this research?**
 - Exon 51 PMO (MRC/ AVI): phase IIa recently finished, phase IIb planned
 - Exon 51 PMO non ambulant (AVI/ AFM): Planned for 3Q2012
 - Exon 51 20ME (Prosensa / GSK): phase IIb underway
 - Exon 53 PMO (AVI): planning stage
 - Exon 53 PPMO: preclinical optimisation
 - Exon 23 PMO MDX: noninvasive biomarker discovery
- 2) **What are the aims of your study?**

demonstrate that PMO and 20ME antisense oligomers are safe and efficient in restoring dystrophin production in boys with eligible deletions for skipping of exons 51 and also 53

Search for non invasive imaging biomarkers for dystrophin restoration
- 3) **Who is funding this research?**

MRC/ AVI/ GSK-Prosensa/ Wellcome Trust/ AFM/ ICE project/ EU
- 4) **If at the stage of a clinical trial who is eligible to participate?**

At the moment only exon 51 skippable boys (see www.mdex.org.uk for details of deletions)
- 5) **When will your study be completed?**

exon 51 20ME in 3Q 2012
- 6) **Are there any results published?**

Our recently completed PMO exon 51 is published in the Lancet
Exon skipping and dystrophin restoration in patients with Duchenne muscular dystrophy after systemic phosphorodiamidate morpholino oligomer treatment: an open-label, phase 2, dose-escalation study. Cirak S, Arechavala-Gomez V, Guglieri M, Feng L, Torelli S, Anthony K, Abbs S, Garralda ME, Bourke J, Wells DJ, Dickson G, Wood MJ, Wilton SD, Straub V, Kole R, Shrewsbury SB, Sewry C, Morgan JE, Bushby K, Muntoni F. Lancet. 2011 Aug 13;378(9791):595-605
- 7) **Will there be any future clinical trials relating to your research and if so when?**

Yes several trials are planned, see above
- 8) **Where can I find out more about your research?**

www.mdex.org.uk and <http://www.ucl.ac.uk/ich/research-ich/dubowitz/research>

AVI 4658 Clinical Trials

Friday 3.00 – 3.30

Dr Ed Kaye AVI

- 1) **At what stage is this research?**

We are currently in Phase 2.
- 2) **What are the aims of your study?**

The aim of the study is to demonstrate the safety, tolerability, and efficacy of AVI-4658 (Phosphorodiamidate Morpholino Oligomer) for exon 51 skipping.
- 3) **Who is funding this research?**

The research is being funded by AVI BioPharma

- 4) **If at the stage of a clinical trial who is eligible to participate?**
Ambulatory boys between the ages of 7 and 13 years of age, with stable cardiac and pulmonary function, who have a genetic mutation which is amendable to exon 51 skipping.
- 5) **When will your study be completed?**
The study results will be available in the second quarter of next year.
- 6) **Are there any results published?**
The results of a Phase I/2 dose ranging study have been published in Lancet July 25, 2011 Cirak et al.
- 7) **Will there be any future clinical trials relating to your research and if so when?**
Depending on the results of the Phase 2 study, an extension study is planned and a multi-national Phase 3 randomized controlled study beginning in the end of 2012.
- 8) **Where can I find out more about your research?**
You can look at our Website under AVIBioPharma.

Peptide based inhibition of NF-kB

Friday 3.00 – 3.30

Professor Paul Jansen Ohio USA

- 1) **At what stage is this research?**
Advanced pre-clinical stage. We are currently testing pharmacokinetics and toxicity in a dog model
- 2) **What are the aims of your study?**
To test the therapeutic value of NF-kB inhibition via a small peptide in muscular dystrophy
- 3) **Who is funding this research?**
The bulk of the work is funded by an NIH UOI grant
- 4) **If at the stage of a clinical trial who is eligible to participate?**
No trial yet.
- 5) **When will your study be completed?**
At the end of the year all the pre-clinical data will be evaluated
- 6) **Are there any results published?**
Yes, preclinical works have been recently published:

Peterson JM, Kline W, Canan BD, Ricca DJ, Kaspar B, Delfin DA, DiRienzo K, Clemens PR, Robbins PD, Baldwin AS, Flood P, Kaumaya P, Freitas M, Kornegay JN, Mendell JR, Rafael-Fortney JA, Guttridge DC, Janssen PML. Peptide-based inhibition of NF- κ B rescues diaphragm muscle contractile dysfunction in a murine model of Duchenne muscular dystrophy. *Molecular Medicine*, 2011;17:508-515.

Delfin DA, Xu Y, Peterson JM, Guttridge DC, Rafael-Fortney JA, Janssen PML. Improvement of cardiac contractile function by peptide-based inhibition of NF- κ B in the utrophin/dystrophin-deficient murine model of muscular dystrophy. *Journal of Translational Medicine* 2011, 9:68.

- 7) **Will there be any future clinical trials relating to your research and if so when?**
If the dog data shows consistency with all the mouse data, a trial will be initiated.
- 8) **Where can I find out more about your research?**
Other than the published peer reviewed works, we have no public information at this time.

Action of ACE inhibitor perindopril on cardio skeletal muscle in DMD – basic and clinical results

Friday 4.00 – 4.30

Dr Dennis Duboc France

- 1) **At what stage is this research?**
All patients included, two years F UP , end of the study end of November
- 2) **What are the aims of your study?**
to prevent cardio skeletal fibrosis associated to the dystrophic process
- 3) **Who is funding this research?**
Sponsoring , Servier Firma , and french network of “childhood myology pluridisciplinary consultations”
- 4) **If at the stage of a clinical trial who is eligible to participate?**
5 to 8 years old , DMD child
- 5) **When will your study be completed?**
All patients included, two years F UP , end of the study end of November

- 6) Are there any results published?
Cardiac prevention in DMD (J of American College of Cardiology 2005) , Improved survival in DMD(American Heart Journal 2007), situation of the controversial points concerning these treatments (Chabrier , Dev Med Child Neurol. 2010 Nov;52(11):1067-8.)
- 7) Will there be any future clinical trials relating to your research and if so when?
Google , " pub med" , and search at "duboc d" (no homonymus ,select the papers concerning muscle dystrophies)

The Holy Grail of multiple exon-skipping

Friday 4.00 – 4.30

Professor Terry Partridge USA

- 1) At what stage is this research?
In collaboration with Dr Takeda's group in Tokyo, we have shown that it is possible to skip II exons from 45-55.
- 2) What are the aims of your study?
The aim to demonstrate the principle that antisense oligonucleotides can be used to skip several exons so as to generate the optimal shortened dystrophins in a large proportion of DMD boys. Skipping exons 45-55 would treat more than 60% of DMD deletion mutations and produce a dystrophin that is associated with very mild Beckers dystrophy.
- 3) Who is funding this research?
This research is funded by the Foundation to Eradicate Duchenne, NIH and the Department of Defence
- 4) If at the stage of a clinical trial who is eligible to participate?
This is a demonstration in dystrophic dog and the mdx mouse that it is possible, in principle, to skip multiple exons. The precise agents that work on the mouse will not be applicable to man but the work does encourage us to look for similar sequences in man.
- 5) When will your study be completed?
One part is nearly complete and is being prepared for publication
- 6) Are there any results published?
See above. The first part of the work on the dogs was published in 2009 (Yokota, T., et al., Efficacy of systemic morpholino exon-skipping in Duchenne dystrophy dogs. Ann Neurol, 2009.65(6): p. 667-76.)
- 7) Will there be any future clinical trials relating to your research and if so when?
The concepts behind the design of the reagents used in these studies are currently being investigated.
- 8) Where can I find out more about your research?--
Terence A. Partridge Ph.D. FMedSci,
Center for Genetic Medicine Research, Children's National Medical Center, 111 Michigan Avenue, Washington DC 20010, U.S.A.
Tel. 202 476 2192 FAX 202 476 6014
Email tpartridge@cnmcresearch.org

Catena for Duchenne Muscular Dystrophy

Friday 4.30 – 5.00

Dr. Nicholas Coppard Santhera

- 1) At what stage is this research?
Phase 2 randomized controlled clinical study of 12 months duration (DELPHI) completed
Phase 2 open label clinical study of 24 months duration (DELPHI-E) completed
Phase 3 randomized controlled clinical study of 12 months duration (DELOS) ongoing
- 2) What are the aims of your study?
Confirm efficacy of Catena on respiratory function in DMD
- 3) Who is funding this research?
Santhera Pharmaceuticals (Switzerland)
- 4) If at the stage of a clinical trial who is eligible to participate?
DMD patients 10-18 years of age currently not on chronic steroids (ambulatory and mutation status are not relevant)
- 5) When will your study be completed?
DELOS study is still open for enrollment; no definitive end-date known
- 6) Are there any results published?
Data from a preceding phase 2 clinical study with Catena are published: Neuromuscul Disord. 2011 Jun;21(6):396-405
- 7) Will there be any future clinical trials relating to your research and if so when?
tbd
- 8) Where can I find out more about your research?
www.santhera.com and www.clinicaltrials.gov

Revatio for heart Disease in DBMD – Pilot Trials Now clinical trial

Friday 4.30 – 5.00

Dr. Dan Judge USA

- 1) **At what stage is this research?**
This is a clinical trial that is currently enrolling
- 2) **What are the aims of your study?**
To determine whether sildenafil improves cardiac function in DBMD, to determine the effect of this treatment on arm blood flow, and to assess the safety of this medicine in boys and men with DBMD.
- 3) **Who is funding this research?**
Pilot Trials Now
- 4) **If at the stage of a clinical trial who is eligible to participate?**
Inclusion Criteria
People must meet all inclusion criteria in order to participate in this study. These are:
 1. Duchenne or Becker Muscular Dystrophy
 2. Male gender
 3. Age greater than or equal to 15 years
 4. Cardiac dysfunction with ejection fraction less than or equal to 50%
 5. On a stable dose of ACE-inhibitor or angiotensin receptor blocker (ARB) for at least 3 months; beta-adrenergic receptor blockers and glucocorticosteroids are not required but if used, a stable dose for at least 3 months is required.
 6. Ability to provide informed consent
 7. Ability to adhere with study follow-up
 8. Willingness to abstain from food and alcohol for 8 hours prior to blood flow tests
Exclusion Criteria
Anyone meeting any of these exclusion criteria at baseline will be excluded from the study:
 1. Use of nitrates or alpha-adrenergic receptor blockers
 2. Known intolerance or allergy to sildenafil, or a history of any severe allergic or anaphylactic reactions
 3. Any medical or psychosocial condition, which, in the view of the study investigator, makes study participation inadvisable
 4. Known hereditary retinal disorder such as retinitis pigmentosa
 5. History of priapism or conditions that may predispose to priapism such as sickle cell anemia, multiple myeloma, or leukemia
 6. Bleeding disorders
 7. Active tobacco use
 8. Chronic atrial fibrillation or frequent arrhythmia that would result in an irregular pulse
 9. Factors that would preclude obtaining an MRI study – (e.g. implantable pacemaker or cardioverter-defibrillator; body habitus cannot fit into scanner)
 10. Systolic blood pressure (SBP) less than 85 mmHg at baseline evaluation
 11. Chronic kidney disease stages 4 and 5: $GFR < 30 \text{ mL/min/1.73 m}^2$ as determined by serum cystatin C level and the equation $eGFR_{cys} = 76.7 \times (\text{serum cystatin C}^{-1.18})$
 12. Current use of sildenafil.
- 5) **When will your study be completed?**
After 30 participants enroll and complete the 1-year treatment
- 6) **Are there any results published?**
No results have been published yet.
- 7) **Will there be any future clinical trials relating to your research and if so when?**
We anticipate additional larger multi-center trials that will study this medication in the future
- 8) **Where can I find out more about your research?**
<http://clinicaltrials.gov/ct2/show/NCT01168908>
or
<http://www.ncbi.nlm.nih.gov/pubmed/21812510>

IGF-I therapy and muscle function in Duchenne Muscular Dystrophy

Friday 5.00 – 5.30

Dr Meilan Rutter USA

- 1) **At what stage is this research?**
Ongoing. (Recruitment about 80% so far.)
- 2) **What are the aims of your study?**
Primary aim: To determine whether IGF-I therapy improves or preserves muscle function in DMD.
- 3) **Who is funding this research?**
Funded by Charley's Fund, Nash Wicka Foundation, Action Duchenne.
IGF-I medication provided by Ipsen.
- 4) **If at the stage of a clinical trial who is eligible to participate?**
Boys with confirmed Duchenne aged at least 5 years, not in puberty, on daily steroids at least 12 months, and able to walk independently.

- 5) When will your study be completed?
Anticipated mid 2012.
- 6) Are there any results published?
Not yet.
- 7) Will there be any future clinical trials relating to your research and if so when?
This will be determined after assessment of the outcome of this study.
- 8) Where can I find out more about your research?
Action Duchenne, PPMD and Charley's Fund websites.

Early treatment with lisinopril and spironolactone preserves cardiac and skeletal muscle function

Friday 5.00 – 5.30

Professor Paul Jansen Ohio USA

- 1) At what stage is this research?
This research is currently pre-clinical, but because the drugs involved are already and FDA approved, they can already be used clinically. Current bench-top research is ongoing, and a clinical trial is close to start.
- 2) What are the aims of your study?
The overall aims are to explore the mechanisms of how this drug combination is beneficial to learn more about underlying pathways and design further improved treatment strategies.
Aims include quantification of benefit of the drugs separate, and determining whether they can be employed to (partially) reverse dysfunction.
- 3) Who is funding this research?
Initial and ongoing funding from a private foundation: Ballouskies (<http://www.ballouskies.com>)
Currently also funded by a exploratory research grant (to Jill Rafael-Fortney and Paul Janssen) by Parent Project Muscular Dystrophy
- 4) If at the stage of a clinical trial who is eligible to participate?
Current in advanced stages of design
- 5) When will your study be completed?
unknown
- 6) Are there any results published?
Yes, initial results were published in the journal Circulation:

Rafael-Fortney JA*, Chimanji NS, Schill KE, Martin CD, Murray JD, Ganguly R, Stangland JE, Tran T, Xu Y, Canan BD, Mays TA, Delfin DA, Janssen PML*, Raman SV*. Early treatment with lisinopril and spironolactone preserves cardiac and skeletal muscle in Duchenne muscular dystrophy mice. Circulation 2011;124-582-588. PMID: 21768452 *contributed equally.
- 7) Will there be any future clinical trials relating to your research and if so when?
Dr. Raman at Ohio State University is launching a randomized, double-blind clinical trial supported by BallouSkies of an aldosterone antagonist in boys with DMD age 7 years and older whose baseline cardiac MRI shows any fibrosis (scar) and normal ejection fraction (pump function). Enrollment will begin in January, 2012 with results focused on change in heart function and scarring at 1 year to be presented by March, 2013
- 8) Where can I find out more about your research?
<http://medicalcenter.osu.edu/mediaroom/releases/Pages/Heart-Failure-Drugs-for-MD.aspx>

Light at the end of the tunnel

Saturday 9.00 – 9.30

Dame Professor Kay Davies University of Oxford

- 1) At what stage is this research?
Preclinical development of drugs
- 2) What are the aims of your study?
The aim is to increase the levels of the dystrophin related protein, utrophin in muscle.
- 3) Who is funding this research?
Medical Research Council., Muscular Dystrophy Campaign and the Muscular Dystrophy Association, USA.
- 4) If at the stage of a clinical trial who is eligible to participate?
- 5) When will your study be completed?
We will have new candidate drugs by 2012
- 6) Are there any results published?
Only on SMT C1100 showing proof of principle

7) Will there be any future clinical trials relating to your research and if so when?
Yes but in about 2 years time

8) Where can I find out more about your research?
MDC website, MDA website, www.dpag.ox.ac.uk

Clinical Trial protocols are no longer fit for purpose – N=1 provides a radical alternative

Saturday 9.30 – 10.30

Nick Catlin CEO Action Duchenne, Iain Henderson Trustee Action Duchenne

Please refer to the following discussion in neuromuscular disorders: Individual patient (N=1) “Trials” in Duchenne Dystrophy, *Neuromuscular Disorders* 21 (2011) pp.525-526

Dr Annemeike Rus Netherlands

1) At what stage is your research?

I work on antisense-mediated exon skipping, which is currently tested in phase 3 clinical trials by GSK/Prosensa

2) What are the aims of your study?

My work focuses on optimization of exon skipping in animal models (comparing different dosing regimes, maintenance regimes etc), on using combination therapies aiming to restore dystrophin and preserve muscle quality. I also try to elucidate how much dystrophin is needed to improve pathology in animal models and in collaboration with my colleague Peter 't Hoen am trying to find serum biomarkers that allow therapeutic monitoring (so muscle biopsies become obsolete).

3) Who is funding this research?

The Dutch Duchenne Parent project, the Prinses Beatrix Foundation (the Netherlands), the Dutch Organisation of Scientific Research

4) If at the stage of a clinical trial who is eligible to participate?

For the trials performed by GSK/Prosensa patients who need exon 51 skipping to restore the reading frame are eligible. More eligibility criteria can be found on <http://www.clinicaltrials.gov/ct2/show/NCT01254019?term=GSK2402968&rank=1>

5) When will your study be completed?

The GSK/Prosensa trial should be complete in 2013.

Our ongoing mouse studies will be completed in 2012, but new studies to further optimize exon skipping (based on the outcome of mouse studies and human studies) will be needed.

6) Are there any results published?

Much of the work in mouse is published in scientific journals like *Neuromuscular Disorders*, *Molecular Therapy*, *Journal of Gene Medicine*.

The work in clinical trials has been published in the *New England Journal of Medicine* in 2007 and 2011.

7) Will there be any future clinical trials relating to your research and if so when?

Trials for exon 51 are currently ongoing coordinated by GSK (a phase 3 placebo controlled trial, a trial in non-ambulant patients and a trial comparing different dosing regimes). A trial for exon 44 skipping is ongoing coordinated by Prosensa Therapeutics. Prosensa is planning trials for exon 45 and 53 skipping in 2012.

8) Where can I find out more about your research?

My own research: www.dmd.nl/gt - there is a special section for patients and parents where things are explained in simple terms.
The clinical trials: www.prosensa.eu.

Targeting therapeutic molecules to mitochondria

Saturday 11.00 – 11.30

Dr Mike Murphy Cambridge

1) At what stage is this research?

Work on targeting antioxidants to mitochondria has shown efficacy in a number of animal studies and in human trials. These approaches have yet to be tested in muscular dystrophy.

2) What are the aims of your study?

The aim is to see if mitochondria may be a useful therapeutic target in muscular dystrophy.

3) Who is funding this research?

Work in my lab is funded by the MRC.

4) If at the stage of a clinical trial who is eligible to participate?

N/A.

5) When will your study be completed?

N/A

6) Are there any results published?

There are no studies published on muscular dystrophy with mitochondria protective agents as yet.

7) Will there be any future clinical trials relating to your research and if so when?
An aim of attendance at the conference is to explore this possibility.

8) Where can I find out more about your research?
See: <http://www.mrc-mbu.cam.ac.uk/>

Biglycan as a therapy for Duchenne

Saturday 11.30 – 12.00

Dr Justin Fallon USA

- 1) At what stage is this research?
A clinical candidate has been selected and we are performing IND-enabling studies.
- 2) What are the aims of your study?
We aim to test the efficacy of biglycan in DMD patients. Biglycan could potentially treat all forms of DMD, regardless of mutation.
- 3) Who is funding this research?
National Institutes of Health, USA; Private Investors.
Previous support from PPMD (USA), Charley's Fund, Muscular Dystrophy Association
- 4) If at the stage of a clinical trial who is eligible to participate?
Not yet at clinical trial.
- 5) When will your study be completed?
Current stage will be complete in two years
- 6) Are there any results published?
Yes (see below)
- 7) Will there be any future clinical trials relating to your research and if so when?
Our goal is to test biglycan therapy in clinical trials. We are aiming to initiate testing in three years or less.
- 8) Where can I find out more about your research?
Publications:
Amenta et al., Proc Natl. Acad. Sciences (USA), 2011. PubMed ID 21187385
Mercado et al., FASEB Journal, 2006. PubMed ID 16807372
<http://www.tivorsan.com/>

AAV-based mRNA therapies for Duchenne muscular dystrophy

Saturday 12.00 – 12.30

Dr Luis Garcia France

- 1) At what stage is this research?
Preclinical studies
- 2) What are the aims of your study?
Identify new therapeutic avenues
- 3) Who is funding this research?
University Pierre et Marie Curie
CNRS
Inserm
AFM
AMM & DPP-F (ICE since 2007)
DPP-NL (2012)
- 4) If at the stage of a clinical trial who is eligible to participate?
- 5) When will your study be completed?
A clinical trial with AAV(U7) will be proposed soon by AFM-Genethon-Institute of Myology (Paris – France)
- 6) Are there any results published?
Not yet concerning trials
- 7) Will there be any future clinical trials relating to your research and if so when?
A clinical trial with AAV(U7) will be proposed soon by AFM-Genethon-Institute of Myology (Paris – France)
- 8) Where can I find out more about your research?
Web site of the institute of myology
<http://www.institut-myologie.org/>

Enabling DMD clinical trials with large-scale production of AAV vectors

Saturday 12.30 – 1.00

Dr Rob Kotin NIH USA

- 1) **At what stage is this research?**
We're using the exon-skipping rAAV-U7smOPT in the GRMD model. Several groups participate in this study: NIH, Univ of Pennsylvania, and Myology Institute (Paris)
- 2) **What are the aims of your study?**
To establish routes of administration, dosage, biodistribution, efficacy.
- 3) **Who is funding this research?**
The Division of Intramural Research (DIR) of the National Heart, Lung, and Blood Institute of the National Institutes of Health and the International Collaborative Effort (ICE) for Duchenne muscular dystrophy. The ICE consists of the Parent Project (France) and the Association Monegasque Contre Les Myopathies.
- 4) **If at the stage of a clinical trial who is eligible to participate?**
If the pre-clinical endpoints are met, then the DIR-NHLBI may support a clinical trial in Bethesda, MD (USA). Eligibility will depend on the design of the exon-skipping vector.
- 5) **When will your study be completed?**
We are hopeful that the pre-clinical study will end within 18 mos.
- 6) **Are there any results published?**
Bish, et al. Mol Ther 2011 in press
- 7) **Will there be any future clinical trials relating to your research and if so when?**
- 8) **Where can I find out more about your research?**
kotinr@nhlbi.nih.gov

Update on SMT C1100 a utrophin upregulator for Duchenne

Saturday 12.30 – 1.00

Dr Jon Tinsley Summit

- 1) **At what stage is this research?**
Summit's drug candidate to treat DMD, SMT C1100, has reached the stage of Phase I clinical trials. SMT C1100 is a potential disease modifying treatment that will benefit all boys with DMD, regardless of their specific genetic mutation. It is an orally available small molecule that works by increasing levels of utrophin to compensate for the missing dystrophin to restore healthy muscle function. It is anticipated that this approach will also be complementary to the other therapeutic approaches currently in development
- 2) **What are the aims of your study?**
SMT C1100 is a powder and it needs to be formulated into an oral product (the formulation) so that it is suitable to give to patients. A Phase I clinical trial is planned that will test the formulation of SMT C1100 in healthy volunteers to assess how much of the active compound is absorbed into the blood stream. The formulation will be suitable for use by all age group and so appropriate for use in any subsequent patient trials.
- 3) **Who is funding this research?**
New funding is required to conduct the Phase I study in healthy volunteers and Summit is actively seeking both commercial and charitable partners to help support the progression of SMT C1100.
- 4) **If at the stage of a clinical trial who is eligible to participate?**
As indicated, the Phase I study will be conducted in healthy volunteers.
- 5) **When will your study be completed?**
Completion of the study will depend on when the necessary funding is secured. Once the funding is in place, the Phase I trial in healthy volunteers could be completed within twelve months.
- 6) **Are there any results published?**
Yes, a compelling preclinical data package on SMT C1100 was recently published in conjunction with Dame Professor Kay Davies in the peer reviewed scientific journal PLoS ONE. It is the most comprehensive set of data made publically available for any potential DMD treatment. In summary the results show that SMT C1100: increases utrophin protein in dystrophin deficient muscle cells from DMD patients to levels expected to be of significant therapeutic benefit; significantly increases the amount of utrophin in the mdx model; improves whole muscle function in study that is surrogate to the 6 minute walk test, a primary endpoint in human clinical trials; and reduces muscle degeneration, fibrosis and chronic inflammation. A copy of the PLoS paper is available at: www.plosone.org/article/info%3Adoi%2F10.1371%2Fjournal.pone.0019189
- 7) **Will there be any future clinical trials relating to your research and if so when?**
Future clinical trials in patients will depend on the outcome of the Phase I study in healthy volunteers. If this is successful, Summit would anticipate that SMT C1100 could then start to be evaluated in DMD patients within twelve months of completing that Phase I study.

These future clinical trials are still at the planning stage but they could take place in either Europe or the US or both. The location of the Phase I healthy volunteer study will not influence where subsequent patient trials are held.

8) **Where can I find out more about your research?**

More information about Summit's DMD programme can be the programmes section of our company website, www.summitplc.com

Learning and Behaviour problems in Duchenne – Include Duchenne project

Saturday 2.00 – 3.00

Dr James Poysky USA

1) **At what stage is this research?**

We have collected data from over 1200 patient families with DMD from around the world. We are still in the process of analyzing the data, but we have some preliminary results.

2) **What are the aims of your study?**

The aims of our study are to collect information on behavior and learning concerns in DMD, understand what the most common problems in these areas are, and examine the implications that these findings have for clinical care. Preliminary results indicate that 33% of families express concern regarding behavior problems in DMD, and 36% express concerns regarding learning problems. Only a third of those who express concern have received a formal behavior or learning diagnosis by a specialist. These findings will be discussed in the context of the Care Recommendations that were published in Lancet Neurology in 2010.

3) **Who is funding this research?**

Our data is being collected through the DuchenneConnect website (duchenneconnect.org), which is a program of Parent Project Muscular Dystrophy and TREAT NMD. This is a patient registry that collects a broad range of data on many aspects of DMD, including current physical functioning, medical care, genetic mutation, etc., that will be used in research studies. The curators of the patient registry keep information confidential, but will release certain portions of it to researchers. Any information used by researchers will be "de-identified", which means that the researchers will not know the names or contact information of those who participate.

4) **If at the stage of a clinical trial who is eligible to participate?**

ALL families currently or previously affected by DMD are encouraged to go online to duchenneconnect.org, register, and enter their data - even if they do not have concerns about learning or behavior.

5) **When will your study be completed?**

This study is ongoing.

6) **Are there any results published?**

We are in the process of analyzing the data and should submit for publication in a few months.

7) **Will there be any future clinical trials relating to your research and if so when?**

Unknown.

8) **Where can I find out more about your research?**

Go to duchenneconnect.org

Include Duchenne Research project

Saturday 2.00 – 3.00

Janet Hoskin

1) **At what stage is this research?**

The Include Duchenne Project has formed part of my PhD at the Centre for Child Research University of Swansea which will be submitted in December 2011. I hope to publish results from my project with Professor Angela Fawcett.

2) **What are the aims of your study?**

My study has looked at the behaviour and learning of children with Duchenne muscular dystrophy and I have designed an online programme called Decipha which children with Duchenne have followed with their schools and families. Results showed significant improvement in those children who followed Decipha.

- Do children with Duchenne have symptoms similar to dyslexia?
- Do they present with verbal developmental delay?
- Do they have a shared learning profile?
- Are they at high risk of socio-emotional problems and ADHD?
- Will a 36 week online intervention programme benefit children with Duchenne
- Which children will benefit most/least?

3) **Who is funding this research?**

Action Duchenne funded this research through successful funding bids from the Big Lottery and Children in Need.

4) **If at the stage of a clinical trial who is eligible to participate?**

This research project has now ended. However, due to the success of the project, Action Duchenne is continuing to fund a small number of places each year for children with Duchenne to have full learning assessments and follow the 36-week online literacy programme called Decipha. If you have a child who is over the age of 6 years and you are concerned about his/her learning or behaviour then contact the Action Duchenne office or Janet – janet@decipha.org to be placed on a waiting list.

- 5) **When will your study be completed?**
It has been completed.
- 6) **Are there any results published?**
Not as yet, I hope to publish my results with my supervisor Professor Angela Fawcett over the next 6 months.
- 7) **Will there be any future clinical trials relating to your research and if so when?**
Professor Veronica Hinton of Columbia University has published prolifically on learning and behaviour in Duchenne. Dr Jos Hendriksen in the Netherlands has also conducted several large scale studies on both reading and behaviour in Duchenne.
- 8) **Where can I find out more about your research?**
You can read more about the Include Duchenne project and watch a short film at www.actionduchenne.org or at www.decipha.org.

A review of Prosensa's exon skipping programme

Saturday 2.00 – 2.30

Dr Giles Champion Prosensa

- 1) **At what stage is this research?**
The research is at different stages varying from phase III clinical trials for GSK2403968/PRO051 to discovery research for PRO052 and PRO055. PRO044 is in phase I/II trials and PRO045 and PRO053 are planned to start clinical trials next year.
- 2) **What are the aims of your study?**
To understand the safety and efficacy of treating boys with Duchenne Muscular Dystrophy with antisense oligonucleotides designed to skip exons according to genetic mutation
- 3) **Who is funding this research?**
Venture Capital, GSK, Patient Organisations
- 4) **If at the stage of a clinical trial who is eligible to participate?**
Varies according to programme – see details on clinicaltrials.gov
- 5) **When will your study be completed?**
Again varies according to programme.
- 6) **Are there any results published?**
Local and systemic effects of treatment published in the New England Journal of Medicine
Van Deutekom JC, et al. N Engl J Med. 2007;357(26):2677–86. AND Goemans NM, et al. N Engl J Med. 2011;364(16):1513–22.
- 7) **Will there be any future clinical trials relating to your research and if so when?**
Future trials with PRO-44, PRO045 and PRO053 in 2012
- 8) **Where can I find out more about your research?**
www.prosensa.com

Personalised medicines for Duchenne

Saturday 2.30 – 3.00

Padraig Wright GSK

- 1) **At what stage is this research?**
Phase III
- 2) **What are the aims of your study?**
Phase III
- 3) **Who is funding this research?**
GlaxoSmithKline
- 4) **If at the stage of a clinical trial who is eligible to participate?**
Boys with Duchenne caused by mutations amenable to treatment by skipping Exon 51; boys must be ambulant and aged >5 years
- 5) **When will your study be completed?**
Always a bit nervous about this because studies always take longer than we anticipate. I expect study will be completed during 2nd half of 2013
- 6) **Are there any results published?**
2 papers have been published in NEJM n engl j med 357;26 www.nejm.org 2686 december 27, 2007 Also, data from 12 boys who have received treatment for 2 years will be presented at WMS next week
- 7) **Will there be any future clinical trials relating to your research and if so when?**
Yes, we will move to non-ambulant boys/young men and then (depending on results from current studies) we will move to boys with 'early symptoms' or who are currently not diagnosed - you will appreciate this is not straightforward but the logic is that the earlier treatment commences the better the outcome

Stem cells in DMD, what can animal models tell us and what are their limitations?

Saturday 3.00 – 3.30

Professor Terry Partridge Washington USA

- 1) **At what stage is this research?**
We are carefully analysing muscle growth and regeneration in normal and mdx mice
- 2) **What are the aims of your study?**
We are trying to produce a set of measurements that define the way in which muscle grows and regenerates in the mdx mouse so that we can precisely interpret the effects of various treatments on the the course of disease in this animal model. We are also making comparisons with what we can learn about these processes in DMD boys so that we can identify those features of the mouse disease that are comparable with human disease and, equally importantly, those features that are not relevant to DMD.
- 3) **Who is funding this research?**
This research is funded by the Foundation to Eradicate Duchenne, the Department of Defence and currently by the MDA.
- 4) **If at the stage of a clinical trial who is eligible to participate?**
N/A
- 5) **When will your study be completed?**
One part is nearly complete and is being prepared for publication
- 6) **Are there any results published?**
See above
- 7) **Will there be any future clinical trials relating to your research and if so when?**
The outcome of this work is currently being applied to pre-clinical testing of a number of anti-inflammatory drugs and to choosing the best exons for skipping with antisense reagents.
- 8) **Where can I find out more about your research?**
Terence A. Partridge Ph.D. FMedSci,
Center for Genetic Medicine Research,
Children's National Medical Center,
111 Michigan Avenue,
Washington DC 20010,
U.S.A.
Tel. 202 476 2192
FAX 202 476 6014
Email tpartridge@cnmcresearch.org

Stem Cells for DMD

Saturday 3.00 – 3.30

Dr Jenny Morgan

- 1) **At what stage is this research?**
Still at the basic research stage.
- 2) **What are the aims of your study?**
To determine a stem cell that can be easily prepared, retain its stem cell characteristics after being grown in tissue culture, can be delivered systemically and give rise to large amounts of muscle and to more functional muscle stem cells.
- 3) **Who is funding this research?**
At present, the Muscular Dystrophy Campaign, ICE (International Collaborative Effort for Duchenne), Wellcome Trust, Medical Research Council.
- 4) **If at the stage of a clinical trial who is eligible to participate?**
Not yet at this stage.
- 5) **When will your study be completed?**
Current work finishes in 2013.
- 6) **Are there any results published?**
Yes – see links below for details.
- 7) **Will there be any future clinical trials relating to your research and if so when?**
Hopefully, but not immediately.
- 8) **Where can I find out more about your research?**
<http://www.ucl.ac.uk/ich/research-ich/dubowitz>
<https://iris.ucl.ac.uk/research/personal/index?upi=JMORG92>
http://www.cnmd.ac.uk/Dubowitz_Neuromuscular_Centre

Takin' Charge – living a full life

Saturday 3.00 – 3.30

Dr Jes Rahbek Denmark

- 1) **At what stage is this research?**
The study is a follow-up study. The cohort – all Danish persons with DMD – have received regular medical, physiotherapeutical and psycho-social check-ups once or twice a year since 1971.
The cohort I will present is born between 1961 and 1991. I have visited them every five years starting in 2002. From October to December I will visit 108 men in their homes and ask them 250 questions of medical, social and psychological character and about quality of life.
- 2) **What are the aims of your study?**
I want to describe the first Danish adult generation of people with DMD and their life in general.
- 3) **Who is funding this research?**
The Danish NMD patient organization, Muskelsvindfonden, and the Danish National Rehabilitation Centre for NMD.
- 4) **If at the stage of a clinical trial who is eligible to participate?**
NA
- 5) **When will your study be completed?**
It will continue for many years – until DMD treatment is complete.
- 6) **Are there any results published?**
Yes: Rahbek J, Werge B, Madsen A, Marquardt J, Steffensen BF, Jeppesen J
Adult life with Duchenne muscular dystrophy: Observations among an emerging and unforeseen patient population. Pediatric Rehabilitation 2005; 8: 17-28.
- 7) **Will there be any future clinical trials relating to your research and if so when?**
Our study is a follow-up study describing the need for CARE programs after intervention, e.g. clinical trials.
- 8) **Where can I find out more about your research?**
At our website www.rcfm.dk or by contacting us Jes Rahbek jera@rcfm.dk

A new generation of drugs to treat Duchenne

Saturday 4.00 – 5.00

Chair Francesco Muntoni plus panel of speakers from the conference

Please submit your questions from any sessions of the conference to Professor Muntoni and presenters

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