

Programme Summary

Monday 29 September 2008

15.00 – 18.00	Hilton Hotel	Registration and setting up of posters
18.00	Discovery Museum	Welcome Ceremony & Reception

Tuesday 30 September 2008

07.00 – 08.15	Hilton Hotel	Registration and setting up posters
08.30 – 09.00	Sage Hall 1	Opening of Congress 2008
09.00 – 10.30	Sage Hall 1	Invited lectures - D.I. 1-3 New Insights into the pathogenesis of facioscapulohumeral muscular dystrophy, myotonic dystrophy and other dominant muscular dystrophies
10:30-11:00	Sage	Morning Tea & Coffee
11:00-12:00	Sage Hall 1	Invited lectures - D.I. 4-5 New Insights into the pathogenesis of facioscapulohumeral muscular dystrophy, myotonic dystrophy and other dominant muscular dystrophies
12:00-13:00	Sage Hall 1	Oral presentations - G.O. 1-3 Insights into new muscle diseases
13:00-14:30	Sage Barbour Room	Meeting of Editorial Board of Neuromuscular Disorders
	Hilton Hotel	Lunch & Exhibition
14:30-15:30	Hilton Hotel	Parallel sessions 1-7 Chaired poster discussion session 1
	Poster Area 1	Posters 1 - D.P.1 01 – 13 Facioscapulohumeral muscular dystrophy
	Poster Area 2	Posters 2 - G.P.1 01 – 14 Collagen VI, laminopathies and neuropathies
	Poster Area 3	Posters 3 - G.P.2 01 – 12 Alpha-dystroglycan CMD including animal models
	Poster Area 4	Posters 4 - T.P.1 01 – 14 Clinical assessment tools
	Poster Area 6A	Posters 5 - D.P.2 01 – 14 Clinical studies in myotonic dystrophy, myotonias and other channelopathies
	Poster Area 5A	Posters 6 - M.P.1 01 – 16 Inherited and acquired myasthenias
	Poster Area 5B	Posters 7 - G.P.3 01 – 15 Mitochondrial diseases
15.30 - 16.00	Hilton Hotel	Poster Viewing session 1: presenters of all posters discussed on Tuesday 30th September to be available for poster viewing
16.00 - 16.30	Hilton Hotel	Afternoon Tea & Coffee and Exhibition
16:30 - 17:00	Hilton Hotel	Poster Viewing session 2 : presenters of all posters discussed on Wednesday 1st October to be available for poster viewing
17.00 - 18.00	Hilton Hotel	Parallel sessions (8-13) Chaired poster discussion session 2
	Poster Area 1	Posters 8 - T.P.2 01 – 16 Antisense and cellular approaches to therapy development

Full Programme

Monday 29 September 2008

12.30-16.00	Sage Seminar Room	WMS Executive Board Meeting with lunch
15:00-18:00	Hilton Hotel	Registration and setting up of posters
18:00	Discovery Museum	Welcome Ceremony & Reception

Tuesday 30 September 2008

07:00-08:15	Hilton Hotel	Registration and setting up posters
08:30-09:00	Sage Hall 1	Opening of Congress 2008
09:00-10:30	Sage Hall 1	New Insights into the pathogenesis of facioscapulohumeral muscular dystrophy, myotonic dystrophy and other dominant muscular dystrophies; Invited lectures (D.I. 1-3) Chairpersons: K. Bushby, V. Dubowitz
	D.I.1	The Jacobsen Memorial Lecture sponsored by the FSH Society Genetic and epigenetic studies of FSHD <u>S.M. van der Maarel</u>
	D.I.2	Facioscapulohumeral muscular dystrophy: Transition from a mendelian trait to a complex genetic disease? <u>R. Tupler</u>
	D.I.3	A contribution of "junk DNA" and transcription factors to the molecular mechanism of FSHD <u>A. Belayew</u>
10:30-11:00	Sage	Morning Tea & Coffee
11:00-12:00	Sage Hall 1	New Insights into the pathogenesis of facioscapulohumeral muscular dystrophy, myotonic dystrophy and other dominant muscular dystrophies; Invited lectures (D.I. 4-5) Chairpersons: K. Flanigan
	D.I.4	Myotonic dystrophy: Can mechanistic insight translate into therapeutic success? <u>C.A. Thornton</u>
	D.I.5	Update in laminopathies: phenotypes, pathogenesis and therapies <u>N. Lévy</u>
12:00-13:00	Sage Hall 1	Insights into new muscle diseases; Oral presentations (G.O. 1-3) Chairpersons: H. Topaloglu, C. Wallgren-Pettersson
	G.O.1	The phenotype of myofibrillar myopathy associated with p.W2710X mutation in filamin C: A study of 31 German patients <u>R.A. Kley; J. Kirschner; K. Eger; D. Fischer; A. Huebner; M. Vorgerd</u>
	G.O.2	Proteomic identification of the LIM domain protein FHL1 as the gene-product mutated in reducing body myopathy <u>J. Schessl; Y. Zou; M.J. McGrath; B.S. Cowling; B.S. Maiti; S.S. Chin; C. Sewry; R. Battini; Y. Hu; D.L. Cottle; M. Rosenblatt; L. Spruce; A. Ganguly; J. Kirschner; A.R. Judkins; J.A. Golden; H.H. Goebel; A.L. Taratuto; F. Muntoni; K.M. Flanigan; C.A. Mitchell; C.G. Bönnemann</u>

Full Programme

G.O.3		Mutations in contactin-1, a neuronal cell adhesion molecule expressed at the neuromuscular junction, cause a novel form of congenital lethal myopathy A.G. Compton; D.E. Albrecht; S.T. Cooper; D. Mowat; K.J. Jones; N. Yang; J. Seto; B. Ranscht; M. Bahlo; S.C. Froehner; <u>K.N. North</u>
13.00-14:30	Sage Barbour Room	Meeting of Editorial Board of Neuromuscular Disorders.
13.00-14:30	Hilton Hotel	Lunch & Exhibition
14:30-15.30	Hilton Hotel	Poster session 1: parallel sessions (1-7)
	Poster Area 1	Posters 1 - Facioscapulohumeral muscular dystrophy Chairpersons: B. Eymard, B. Udd
D.P.1.01		Clinical features of facioscapulohumeral muscular dystrophy in Czech population <u>S. Vohanka</u> ; J. Sedlackova; L. Fajkusova; J. Bednarik
D.P.1.02		A robust tool to quantify disability in patients affected by Facioscapulohumeral Muscular Dystrophy <u>C. Lamperti</u> ; G. Fabbri; F. Greco; M. Servida; L. Vercelli; C. Fiorillo; C. Borsato; M. Cao; P. Cudia; R. Frusciante; R. Di Leo; L. Volpi; R. D'Amico; E. Pastorello; L. Ricciardi; G. Galluzzi; G. Siciliano; A. Muzio; G. D'Angelo; C. Rodolico; L. Morandi; G. Tomelleri; C. Trevisan; C. Angelini; L. Santoro; E. Ricci; L. Palmucci; M. Moggio; R.G. Tupler
D.P.1.03		Trying to understand the clinical variability in FSHD <u>P. Arashiro</u> ; I. Eisenberg; A.T. Kho; A. Cerqueira; M. Canovas; R. Pavanello; L.M. Kunkel; M. Zatz
D.P.1.04		Size and number of D4Z4 alleles play a role in FSHD phenotype G. Fabbri; C. Fiorillo; C. Borsato; <u>F. Greco</u> ; E. Bonifazi; L. Vercelli; L. Palmucci; G. Tomelleri; C. Angelini; L. Santoro; R. Tupler
D.P.1.05		D4Z4 repeat number and skeletal muscle changes in FSHD <u>Y.K. Hayashi</u> ; K. Goto; A. Ohkuma; S. Noguchi; I. Nonaka; I. Nishino
D.P.1.06		Gene expression analysis in MRI positive FSHD muscles <u>M. Pescatori</u> ; G. Tasca; R. Frusciante; M. Mirabella; M. Rossi; E. Iannaccone; A. Broccolini; E. Ricci
D.P.1.07		Lower limb muscles MRI findings in patients with 4q35-linked facio-scapulo-limb, type 2 muscular dystrophy (FSLD2) (or a facioscapuloperoneal dystrophy) <u>V.M. Kazakov</u> ; D.I. Rudenko; V.O. Kolynin; A.V. Pozdnyakov
D.P.1.08		Morphological pattern of muscle biopsy in a large cohort of FSHD patients <u>V. Crugnola</u> ; V. Ghiaroni; P. Ciscato; M. Servida; A. Prella; M. Sciacco; F. Tiberio; S. Borsa; R.G. Tupler; N. Bresolin; M. Moggio; C. Lamperti
D.P.1.09		Study of DUX4 and DUX4c in facioscapulohumeral muscular dystrophy <u>A. Tassin</u> ; C. Vanderplanck; E. Anseau; S. Cloet; M. Barro; D. Laoudj-Chenivresse; M.C. Dabauvalle; M. Wehnert; Y.W. Chen; A. Belayew; F. Coppée
D.P.1.10		Structural and functional characterization of muscle fibres in the novel mouse model of facioscapulohumeral muscular dystrophy V. Sancisi; E. Germinario; S. Peron; <u>V. Ghiaroni</u> ; E. Morini; D. Danieli-Betto; R.G. Tupler
D.P.1.11		Saethre-Chotzen syndrome: A severe case of Facioscapulohumeral Muscular Dystrophy (FSHD) in a patient S. Monges; M. Torrado; J. Castaño; A.L. Taratuto; C.A. Bacino; E.D. Corona; <u>F. Lubieniecki</u> ; <u>A.L. Rosa</u>

Full Programme

- D.P.1.12 FSHD and Williams syndrome: The occurrence of two genetic disorders due to heterozygous DNA fragment deletions in the same patient
G.L. Vita; M. Romano; S. Sinicropi; L. Colantone; A. Toscano; G. Vita; C. Rodolico
- D.P.1.13 Isolated paraspinal myopathy with bent spine syndrome: A distinct subtype of adult facioscapulohumeral muscular dystrophy
M.K. Kottlors; G.M. Meng; F.X.G. Glocker
- Poster Area 2** **Posters 2 - Collagen VI, laminopathies and neuropathies**
Chairpersons: V. Allamand, C. Matsuda
- G.P.1.01 Redefining the clinical spectrum of collagen VI disorders
A. Nadeau; M. Kinali; M. Main; C. Jimenez-Mallebrera; B. North; E. Clement; A.Y. Manzur; S.A. Robb; F. Muntoni
- G.P.1.02 Irich congenital muscular dystrophy (UCMD): Report on eight patients
A. Nalini
- G.P.1.03 Important variability in clinical severity in a family with Col VI-related myopathy: Potential implication of digenism?
F. Chapon; C. Gartioux; C. Ledeuil; L. Demay; L. Brinas; D. Herlicoviez; S. Allouche; A. Ferreira; F. Leturcq; P. Richard; V. Allamand; G. Bonne
- G.P.1.04 Design of a novel array-CGH to explore allelic and genetic heterogeneity in COLVI related myopathies
M. Bovolenta; E. Martoni; M. Fabris; S. Fini; A. Urciolo; P. Sabatelli; E. Mercuri; E. Bertin; P. Bernardi; P. Bonaldo; L. Merlini; A. Ferlini; F. Gualandi
- G.P.1.05 siRNA mediated allele specific selective silencing of a dominant negative COL6A3 mutation causing UCMD
Y. Zou; J. Schessler; C. Bonnemann
- G.P.1.06 Altered interaction of mutant lamin A and barrier to autointegration factor (BAF)
C. Matsuda; K. Kameyama; I. Nishino; Y.K. Hayashi
- G.P.1.07 Hauptmann-Thannhauser muscular dystrophy - what is it ?
H.H. Goebel; C.G. Bönemann; M. de Visser
- G.P.1.08 Myopathy and radiculopathy in Marfan syndrome
N.C. Voermans; S. Pillen; M.J. Zwarts; M. Lammens; J. Timmermans; I.M. Rooij; B.C.J. Hamel; B.G.M. van Engelen
- G.P.1.09 Nuclear changes in skeletal muscles of AD-EDMD/LGMD1B
Y.E. Park; Y.K. Hayashi; K. Goto; S. Noguchi; I. Nonaka; I. Nishino
- G.P.1.10 Clinicopathological and genetic study of CMT2
E. Shugaiv; B. Senegin; E. Battaloglu; P. Serdaroglu; F. Deymeer; M.A. Akalin; Y. Parman
- G.P.1.11 Mitofusin gene mutations (MFN 2) cause a severe CMT2A with diaphragm paresis involvement
J. Colomer; A. Nascimento; F. Palau; A. Abeledo; C. Llarena; H. Galvez; C. Ortez; M. Pons
- G.P.1.12 Hereditary Motor Sensory Neuropathy Type 1A (HMSN1A) with superadded inflammatory polyneuropathy in two children
A. Desurkar; J.P. Lin; W. Jan; S. Al-Sarraj; H. Jungbluth; E. Wraige
- G.P.1.13 A new locus for distal hereditary motor neuropathy maps to chromosome 16p
E. Brusse; D. Majoor-Krakauer; B.M. De Graaf; G.H. Visser; A.J.W. Boon; B.A. Oostra; A.M. Bertoli-Avella

Full Programme

- G.P.1.14 A novel form of severe childhood autosomal recessive sensory neuropathy associated with optic atrophy and deafness maps to chromosome 8 q24.22-term
M. Srouf; M.I. Shevell; B. Brais
- Poster Area 3** **Posters 3 - Alpha-dystroglycan CMD including animal models**
Chairpersons: B. Talim, M. Brockington
- G.P.2.01 Frequency of different forms of congenital muscular dystrophies in a referral center
B. Talim; Z. Akcoren; G. Haliloglu; D. Orhan; S. Gucer; G. Kale; H. Topaloglu
- G.P.2.02 Genotype-phenotype correlations in congenital muscular dystrophies with defective glycosylation of dystroglycan: A multicentric Italian study
S.M. Messina; F.M.S. Santorelli; M.M. Mora; C.B. Bruno; E.P. Pegoraro; A.P. Pini; T.M. Mongini; A.D. D'Amico; M.P. Pane; R.B. Biancheri; A.B. Berardinelli; A.T. Toscano; L.M. Morandi; M.I. Moroni; G.P.C. Comi; C. Uggetti; C.P.T. Trevisan; L.F. Farina; A.P. Pichiecchio; C.S. Scuderi; A.R. Ruggieri; E.B. Bertini; E.M. Mercuri
- G.P.2.03 Clinical and molecular characterization of 12 patients with defective α -dystroglycan glycosylation
I. Moroni; S. Saredi; A. Ruggieri; C. Pantaleoni; L. Morandi; M. Mora
- G.P.2.04 Molecular study of Lissencephaly type II : 52 families
C. Bouchet-Seraphin; L. Devisme; S. Vuillaumier-Barrot; M. Gonzales; M. Chelbi; C. Le Bizec; F. Encha-Razavi; N. Seta; SOFFOET
- G.P.2.05 Two new foetal cases of syndrome of Walker-Warburg related to LARGE gene
S. Vuillaumier-Barrot; L. Devisme; C. Bouchet-Seraphin; P. Loget; E. Charluteau; A. Eude-Caye; O. Boute; C. Fallet-Bianco; M.J. Perez; B. Gilbert-Dussardier; M. Gonzales; F. Encha-Razavi; N. Seta
- G.P.2.06 Walker-Warburg syndrome with POMT1 mutations can be associated with cleft lip and cleft palate
J. Vajsar; B. Baskin; K. Swoboda; D.W. Biggar; H. Schachter; P.N. Ray
- G.P.2.07 Alpha-dystroglycanopathy in an Italian patient due to large intragenic and single nucleotide deletions in the POMGnT1 gene
S. Saredi; A. Ruggieri; C. Pantaleoni; S. D'Arrigo; E. Mottarelli; F. Blasevich; L. Morandi; I. Moroni; M. Mora
- G.P.2.08 LARGE overexpression in transgenic mice: Implications for therapeutic interventions in muscular dystrophy
M. Brockington; S. Torelli; S. Cirak; S.C. Brown; D.J. Wells; F. Muntoni
- G.P.2.09 Glycosylation of α -dystroglycan in cultured cells and its restoration by glycosyltransferase
F. Saito; Y. Arai; H. Hagiwara; T. Shimizu; K. Matsumura
- G.P.2.10 Characterisation of the brain and eye phenotype of the FKRП knock-down mouse
M.R. Ackroyd; L. Skordis; M. Kaluarachchi; F. Muntoni; S.C. Brown
- G.P.2.11 Developmental defects in a zebrafish model for muscular dystrophies associated with the loss of FKRП
P. Thornhill; D. Bassett; H. Lochmüller; K. Bushby; V. Straub

Full Programme

- G.P.2.12 Generation of a model mouse for Fukuyama congenital muscular dystrophy carrying a retrotransposal insertion in the 3' UTR in the fukutin gene: Therapeutic benefit of enhanced dystroglycan glycosylation to dystroglycanopathy
T. Toda; M. Kanagawa; A. Nishimoto; T. Chiyonobu; S. Takeda
- Poster Area 4 Posters 4 - Clinical assessment tools**
Chairpersons: J. Florence, M. Eagle
- T.P.1.01 Use of the six minute walk test as an endpoint in clinical trials for neuromuscular diseases
J.M. Florence; A. van der Ploeg; P.R. Clemens; D.M. Escolar; P. Laforet; B. Rosenbloom; M. Wasserstein; A. Skrinar; A. Pestronk; J.E. Mayhew
- T.P.1.02 The 6 minute walk test (6MWT) as a clinical trial outcome measure in Duchenne/Becker Muscular Dystrophy (DMD/BMD)
C.M. McDonald; E.K. Henricson; J.J. Han; A.R. Nicorici; R.T. Abresch; L.A. Atkinson; A.L. Reha; G.L. Elfring; L.L. Miller
- T.P.1.03 Gait pattern assessment over 200m in Duchenne muscular dystrophy (DMD)
P.Y. Jeannet; R. Ganea; V. Spehrs-Ciaffi; N.M. Goemans; K. Aminian; A. Paraschiv-Ionescu
- T.P.1.04 The falling factors of children with DMD
I. Alemdaroglu; Ö. Yilmaz; A. Karaduman; H. Topaloglu
- T.P.1.05 Is a reduced muscle test sufficient to give an overall impression of muscle function in spinal muscle atrophy II?
U. Werlauff
- T.P.1.06 Reliability of novice vs. expert tester scoring for the modified Hammersmith functional motor scale for spinal muscular atrophy
K.J. Krosschell; E. Dinsmore; C.K. Ingram; J.A. Maczulski; M. Pietsch; C.B. Scott
- T.P.1.07 Egen Klassifikation revisited in SMA
B.F. Steffensen; A. Mayhew; A. Aloysius; M. Eagle; E. Mercuri; S. Messina; E. Mazzone; A. Nadeau; M. Main; E. Scott; U. Werlauff; B. Werge; A.M. Glanzmann; F. Muntoni
- T.P.1.08 Validation of the expanded Hammersmith functional motor scale in SMA type II and III
A.M. Glanzman; J.M. O'Hagen; M.P. McDermott; W.F. Martens; P.A. Ryan; J.F. Flickinger; J. Quigley; S. Riley; J. Montes; W.K. Chung; L. Deng; B.T. Darras; D.C. De Vivo; P. Kaufmann; R.S. Finkel
- T.P.1.09 Correlation of hand-held myometry with alternative methods of assessment of muscle strength and function in DMD patients
C. Marchesi; M. Main; M. Kinali; F. Muntoni
- T.P.1.10 Reliability and validity of measuring foot and ankle muscle strength in very young children
K.J. Rose; J. Burns; R.A. Ouvrier; M.M. Ryan; K.N. North
- T.P.1.11 Respiratory muscle endurance in children
J.L. Heraghty; J. Henderson; T.N. Hilliard; A. Majumdar; P.J. Fleming
- T.P.1.12 Breathing pattern in neuromuscular disorders
M. Romei; M.G. D'Angelo; A. Lo Mauro; S. Bonato; S. Gandossini; A.C. Turconi; G.P. Comi; E. Marchi; A. Pedotti; N. Bresolin; A. Aliverti

Full Programme

- T.P.1.13 Evaluation of a mechanical stretching device for patients with trismus and neuromuscular disorders
A.K. Kroksmark; B. Johansson Cahlin; Å. Mårtensson; B. Ahlborg
- T.P.1.14 Interventions for increasing ankle flexibility in patients with neuromuscular disease: A Cochrane systematic review
K.J. Rose; J. Burns; K.N. North; D.M. Wheeler
- Poster Area 6A** **Posters 5 - Clinical studies in myotonic dystrophy, myotonias and other channelopathies**
Chairpersons: B. van Engelen, M. Hanna
- D.P.2.01 Genotype and phenotype studies of Myotonic Dystrophy 1 (DM1) in Hungarian patients
A. Herczegfalvi; H. Piko; H. Merkli; R. Horvath; V. Karcagi
- D.P.2.02 Development of a nurse led multidisciplinary clinic for myotonic dystrophy type 1
C. Harling; M. Guglieria; G. Bell; J. Bourke; M. McCallum; M. Eagle; W.K. Stewart; P. Chinnery; V. Straub; K. Bushby
- D.P.2.03 Clinical Follow up in a large cohort of patients affected by Myotonic Dystrophy type 1
C. Harling; M. Guglieri; G. Bell; J. Bourke; M. McCallum; M. Eagle; W.K. Stewart; P. Chinnery; V. Straub; K. Bushby
- D.P.2.04 Measuring quality of life in myotonic dystrophy type 1: A pilot study using the Individualized Neuromuscular Quality of Life questionnaire (INQoL)
K.A. LaDonna; W.J. Koopman; S.L. Venance
- D.P.2.05 Perceived functioning and disability in subjects with myotonic dystrophy
K.M.E. Kierkegaard; A. Tollback; L. Edstrom
- D.P.2.06 Correlation between measures of muscle mass, strength, function and quality of life (QOL) in patients with myotonic dystrophy type 1 (DM-1): Implications for clinical trials
S. Pandya; N. Dilek; W. Martens; C. Quinn; C. Thornton; R.T. Moxley
- D.P.2.07 Risk of arrhythmia in type 1 myotonic dystrophy: The role of clinical and genetic variables in a large cohort of patients
P. Cudia; P. Bernasconi; R. Chioldelli; A. Martini; E. Canioni; S. Romaggi; G. Ferrari; L. Morandi
- D.P.2.08 Strong association between myotonic dystrophy type 2 (DM2) and frequency of auto-immune diseases and autoantibody formation
A. van de Logt; A.A. Tieleman; A.A. den Broeder; B.G.M. van Engelen
- D.P.2.09 Non-genomic effects of sex hormones on CLC-1 may contribute to gender differences in myotonia congenita
D. Fialho; D.M. Kullmann; M.G. Hanna; S. Schorge
- D.P.2.10 Electrophysiological studies in a mouse model of Schwartz-Jampel syndrome
A. Echaniz-Laguna; F. Rene; C. Marcel; M. Stum; B. Fontaine; J.P. Loeffler; S. Nicole
- D.P.2.11 A case of acquired neuromyotonia in the Netherlands case description and diagnostic work-up
H.J.G. Dieks; B.G.M. van Engelen
- D.P.2.12 Episodic ataxia type 1 in identical twins
T.D. Graves; S.M. Zuberi; H. Morris; S. Schorge; D.M. Kullmann; M.G. Hanna

Full Programme

- D.P.2.13 Neonatal hypotonia can be a sodium channelopathy
E. Matthews; A. Guet; M. Mayer; S. Vicart; S. Pemble; D. Sternberg; B. Fontaine; M.G. Hanna
- Poster Area 5A Posters 6 - Inherited and acquired myasthenias**
Chairpersons: J. Colomer, J.P. Bouchard
- M.P.1.01 Molecular characterization of congenital myasthenic syndromes in Southern Brazil
V.M. Mihaylova; R. Scola; J.S. Mueller; P. Lorenzoni; B. Gervini; L. Werneck; A. Huebner; A. Abicht; H. Lochmueller
- M.P.1.02 Congenital myasthenia syndromes - eight northern Portuguese patients
M.M.A. Santos; T.P.M. Coelho; J.M.L. Lima; V. Mihaylova; J. Muller; H. Lochmuller
- M.P.1.03 DOK7 mutations presenting as a limb girdle muscular dystrophy in French Canadians
M. Srouf; V. Bolduc; J.P.L. Bouchard; D. Brunet; J. Mathieu; B. Brais
- M.P.1.04 The effect of dok-7 on acetylcholine receptor clustering in C2C12 Cells
H. Spearman; J.A. Cossins; S. Maxwell; C. Slater; J. Newsom-Davis; D. Beeson; J. Palace; A. Vincent
- M.P.1.05 Successful long-term acetylcholinesterase inhibitor therapy in patients affected by Rapsyn (RAPSN) mutation early onset phenotype
A. Nascimento; J. Colomer; M. Pineda; J. Mihaylova; L. Turon; A. Garcia - Ribes; C. Ortez; J. Muller; C. Jiménez -Mallabrera; H. Lochmüller
- M.P.1.06 A heterozygous Rapsyn (RAPSN) gene mutation (N88K) and (S201R), is a cause of progressive Limb Girdle Myasthenia
J. Colomer; A. Nascimento; J.S. Müller; A. Abicht; H. Lochmüller; L. Turón; C. Ortez; D. Pascual-Vaca
- M.P.1.07 Down regulation of ColQ by RNA interference as an alternative therapy in Myasthenia
J. Kenyon; D. Beeson; D. Annane; B. Eymard
- M.P.1.08 Allele-specific RNA interference to target a model of slow-channel congenital myasthenic syndrome
G. McClorey; D. Beeson; M. Wood
- M.P.1.09 Marked heterogeneity of epidemiological studies of anti-acetylcholine receptor antibody positive myasthenia gravis
A.S. Fitzpatrick; C. Cardwell; P. McCarron; J.P. McConville
- M.P.1.10 Anti-MuSK antibodies are not associated with prolonged pure ocular symptoms
G. Sirin; V. Yilmaz; Y. Parman; P. Serdaroglu-Oflazer; G. Saruhan-Direskeneli; F. Deymeer
- M.P.1.11 Myasthenia gravis: Alternative reasons for unsatisfactory outcome evidenced by a prospective study
M. Dunand; F.X. Borruat; S. Botez; T. Kuntzer
- M.P.1.12 Determinants of quality of life in adult Myasthenia Gravis
R. Seyedsadjadi; M. Rose; J. Weinman; S. Pandya; C. Jackson; D. Sanders; J. Kissel; N.A. Muscle Study Group
- M.P.1.13 What is the mechanism of weakness and fatigability in myasthenia?
M.J. Titulaer; P. Masrori Shamakha; P.W. Wirtz; J.J.G. Verschuuren; J.G. van Dijk
- M.P.1.14 Myasthenia gravis: Value of muscle biopsy taken during thymectomy
J. Zamecnik; D. Vesely; B. Jakubicka; J. Pitha; J. Schutzner; R. Mazanec

Full Programme

- M.P.1.15 Lambert Eaton Myaesthenic Syndrome in an adolescent with delayed onset of unilateral ptosis
A. Majumdar; L. Lingappa; F. O'Callaghan; A. Oware
- M.P.1.16 Lambert Eaton myasthenic syndrome and myasthenia gravis in the same Thai patient
C. Dejthevaporn; R. Witoonpanich
- Poster Area 5B** **Posters 7 - Mitochondrial diseases**
Chairpersons: M. Tulinius, R. Taylor
- G.P.3.01 Clinical and histopathological features in muscle biopsies of children with suspected mitochondrial disease
B. Koksai; B. Talim; Z. Akcoren; D. Orhan; S. Gucer; G. Kale; H. Topaloglu
- G.P.3.02 Diagnostic relevance of myohistological examination in patients with mitochondrial disorders
P. Tacik; M. Deschauer; S. Zierz
- G.P.3.03 A histochemical and molecular genetic investigation of the selective, extraocular muscle involvement in chronic progressive external ophthalmoplegia
R.W. Taylor; L.C. Greaves; K.J. Krishnan; J. Kerin; M.J. Barron; E.L. Blakely; P.G. Griffiths; D.M. Turnbull
- G.P.3.04 Clinical spectrum of cytochrome-c-oxidase deficiency in children
B. Koksai; B. Talim; G. Haliloglu; G. Kale; H. Topaloglu
- G.P.3.05 Autosomal dominant progressive external ophthalmoplegia: Report of a Thai family
R. Witoonpanich; P. Jindahra; P. Lertrit; Z. Wang; Y. Goto; S. Phudhichareonrat
- G.P.3.06 A patient with two mitochondrial DNA mutations causing PEO and LHON
A.R. Moslem; A. Melberg; O. Palm; R. Raininko; E. Stålberg; A. Oldfors
- G.P.3.07 High levels of mitochondrial DNA deletions in extraocular muscles of aged persons
T. Klopstock; C. Lau; A. Bender; J. Müller-Höcker
- G.P.3.08 Atypical myofiber changes of a childhood mitochondrial DNA depletion syndrome with a novel Thymidine Kinase 2 gene mutation
J. Collins; L.Y. Tang; P. Morehart; A. Myatt-Norman; L. Miles; L.J.C. Wong; B. Wong
- G.P.3.09 Mutation in PDHA1 gene mimicking a Guillain-Barré-Syndrome
V. Haug; R. Horvath; R. Horvath; J. Kühr; R. Korinthenberg; J. Kirschner
- G.P.3.10 Clinical and genetic characterization of two brothers with a similar mitochondrial DNA deletion syndrome
J.P.L. Bouchard; P.G. Gould; J. Girouard; N. Dupré; D. Brunet
- G.P.3.11 Novel ETFDH mutations and normal CoQ10 level in Taiwanese patients with multiple acyl-CoA dehydrogenase deficiency
W.C. Liang; A. Ohkuma; Y.K. Hayashi; L.C. Lopez; M. Hirano; I. Nonaka; S. Noguchi; Y.J. Jong; I. Nishino
- G.P.3.12 Riboflavin-Responsive multiple Acyl-CoA dehydrogenation deficiency (MADD-RR): Clinical, biochemical, molecular genetic and 31 P-MRS studies
A. Toscano; I. Nishino; B. Garavaglia; O. Musumeci; R. Lodi; B. Barbiroli; W.C. Liang; G. Vita
- G.P.3.13 Supplementation studies in primary human muscle cells with mtDNA depletion caused by mutations in the DGUOK and POLG1 genes
S. Bulst; A. Abicht; C. Thirion; H. Lochmüller; R. Horvath

Full Programme

G.P.3.14	Comparative human mitochondrial genome analysis using the Affymetrix MitoChip v2 and conventional cycle sequencing <i>A.J. Duncan; M.G. Sweeney; E. Stern; R. Taylor; C. Woodward; M.B. Davis; M.G. Hanna; S. Rahman</i>
G.P.3.15	Waking the sleeping giant; habitual physical inactivity in people with mitochondrial disease <i>M.I. Trenell; S. Apabhai; D.M. Turnbull</i>
15:30-16:00	All Poster Areas Poster Viewing session 1: presenters of all posters discussed on Tuesday 30th September to be available for poster viewing
16:00-16:30	Hilton Hotel Afternoon Tea & Coffee and Exhibition
16:30-17:00	All Poster Areas Poster Viewing session 2: presenters of all posters discussed on Wednesday 1st October to be available for poster viewing
17:00-18:00	All Poster Areas Poster session 2: parallel sessions (8-13)
	Poster Area 1 Posters 8 - Antisense and cellular approaches to therapy development Chairpersons: J Verschuuren, A. Aartsma-Rus
T.P.2.01	Antisense oligomer design: Targeting and assay systems <i>S.D. Wilton; C. Mitrpant; P.L. Meloni; A.M. Adams; S. Fletcher</i>
T.P.2.02	Development of antisense oligonucleotides for dystrophin exon skipping based on target site accessibility predicted by dynamic co-transcriptional pre-mRNA secondary structure analysis <i>Z.A.D. Pramono; D.K.B. Wee; X. Qianbin; P.S. Lai; W.C. Yee</i>
T.P.2.03	Characterisation of a complex dystrophin mutation: Assume nothing when designing exon skipping strategies <i>S. Fletcher; M.R. Davis; H.R. Madden; S.D. Wilton</i>
T.P.2.04	Optimization of antisense-mediated exon skipping in mouse models for Duchenne muscular dystrophy <i>A. Aartsma-Rus; H.A. Heemskerk; C.L. de Winter; M. Van Putten; S. De Kimpe; J.C.T. van Deutekom; G.J.B. van Ommen</i>
T.P.2.05	Direct comparison of 2'O-Methyl and PMO AONs for exon skipping in preclinical model of in DMD <i>H.A. Heemskerk; C.L. de Winter; S.J. de Kimpe; P. van Kuik-Romeijn; N. Heuvelmans; G.J. Platenburg; G.J.B. van Ommen; J.C.T. van Deutekom; A. Aartsma-Rus</i>
T.P.2.06	Modulation of small mutations in dystrophin "skippable" exons: In vitro studies to identify the optimal PS-AONs <i>P. Spitali; M. Fabris; S. Falzarano; P. Sabatelli; M. Bovolenta; M. Neri; E. Martoni; E. Bassi; S. Tuffery-Giraud; M. Claustres; J.M. Cuisset; F. Gualandi; P. Rimessi; A. Ferlini</i>
T.P.2.07	The systemic administration of a low dose of 2OMePS-AON combined with novel cationic polymethylmethacrylate nanoparticles induces the rescue of dystrophin expression in the mdx murine model <i>P. Rimessi; P. Sabatelli; M. Fabris; P. Braghetta; E. Bassi; P. Spitali; G. Vattermi; G. Tomelleri; L. Mari; D. Perrone; A. Medici; M. Neri; M. Bovolenta; E. Martoni; N. Maraldi; P. Bonaldo; F. Gualandi; L. Merlini; L. Tondelli; K. Sparnacci; A. Caputo; M. Laus; A. Ferlini</i>
T.P.2.08	Dystrophin expression after systemic delivery of morpholino antisense oligonucleotide in mdx mouse: A dose-response analysis <i>A. Malerba; G. Dickson; I. Graham</i>

Full Programme

- T.P.2.09 Induced exon skipping in normal and mdx muscle
S. Fletcher; J.P. Steinhaus; C. Mitropant; P.L. Meloni; S.D. Wilton
- T.P.2.10 Restoration of dystrophin expression in mdx mouse by peptide-conjugated antisense oligonucleotides
H.F. Yin; Y.Q. Seow; H.M. Moulton; P.L. Iversen; J.K. Boutilier; M.J.A. Wood
- T.P.2.11 A model of human muscle regeneration in vivo to test potential therapies for DMD
C. Adkin; J. Meng; V. Arechevala-Gomez; J. Morgan; F. Muntoni
- T.P.2.12 Pax3/Pax7 transcriptional activity is required both in vitro and in vivo for muscle differentiation
L. Boldrin; C.A. Collins; V.F. Gnocchi; P.S. Zammit; J.E. Morgan
- T.P.2.13 Murine embryonic stem cells injected into mdx mouse — In vivo myogenic capacity and immunogenic reaction
D. Ayub-Guerrieri; P.C.M. Martins; P.C.G. Onofre; V.F. Lopes; L.V. Pereira; C.M.C. Mori; M. Vainzof
- T.P.2.14 Overexpression of MyoD1 in human synovial stem cells facilitates their myogenic differentiation in vitro
J. Meng; J.E. Morgan
- T.P.2.15 Treatment of spinal muscular atrophy by transplantation of embryonic (ES) derived neural stem cells
S. Corti; M. Nizzardo; M. Nardini; C. Donadoni; F. Fortunato; N. Bresolin; G.P. Comi
- Poster Area 2 Posters 9 - SMA and ALS**
Chairpersons: E. Bertini, P. Kaufmann
- G.P.4.01 Is assessment of respiratory function a good outcome measure for SMA II and III?
M. Guglieri; M. Eagle; M. McCallum; K. Bushby; V. Straub
- G.P.4.02 Neurogenic atrophy of bulbar muscles in patients with SMA type 2
C. Sixt; C. Lindberg; M. Nordin
- G.P.4.03 The international SMA parent survey: Therapeutic and clinical trial considerations
R.S. Finkel; E. Bertini
- G.P.4.04 PTC Therapeutics identifies compounds that modulate SMN protein expression at the post-transcriptional level
S. Paushkin; A. Dakka; L. Khandker; R. Kaushal; H. Qi; M. Woll; J. Hedrick; A. Mollin; Y.M. Moon; M. Weetall; E. Welch
- G.P.4.05 Early intervention with a semi-elemental, limited protein, low fat enteral formula (SF) improves survival and growth parameters in children with Spinal Muscular Atrophy (SMA) 1 and severe SMA 2
K. Rao; B.L. Wong
- G.P.4.06 Daily salbutamol in young patients with SMA type II
M.P. Pane; S.S. Staccioli; S.M. Messina; A.D. D'Amico; M.P. Pelliccioni; E.M. Mazzone; M.C. Cuttini; P.A. Alfieri; R.B. Battini; M.M. Main; F.M. Muntoni; E.B. Bertini; M.V. Villanova; E.M. Mercuri
- G.P.4.07 A preliminary study on the relationship between VDR gene polymorphisms and risk for childhood SMA disease
M. Stavarachi; P. Apostol; D. Cimponeriu; M. Toma; N. Butoianu; L. Gavrilă

Full Programme

- G.P.4.08 Determination of the SMN1 and SMN2 copy number based on real-time PCR in Hungarian families
M. Nagymihaly; V. Karcagi; A. Herczegfalvi; L. Timar
- G.P.4.09 The association between three genetic polymorphisms in homocysteine pathway and SMA phenotype
P. Apostol; D. Cimponeriu; M. Stavarachi; M. Toma; I. Radu; D. Usurelu; N. Butoianu; S. Magureanu; L. Gavrila
- G.P.4.10 Analysis of histone modification and expression changes of SMN gene after HDAC inhibitor treatment
E. Zapletalova; M. Hlavna; L. Fajkusova
- G.P.4.11 Fugu rubripes survival motor neuron gene promoter activity in neuronal and non-neuronal mammalian cells
P. Kathirvel; W.P. Yu; C.C. Lim; B. Venkatesh; P.S. Lai; W.C. Yee
- G.P.4.12 Translational readthrough modulates SMN stability: Potential for aminoglycosides as an SMA therapy
C.R. Heier; S.M. Hammond; R.G. Gogliotti; C.J. DiDonato
- G.P.4.13 A co-inducer of the heat shock response ameliorates disease in a mouse model of SBMA
N. Nirmalanathan; J.R.T. Dick; A.R. La Spada; L. Greensmith; M.G. Hanna
- G.P.4.14 In ALS, Viral-Dysmetabolic mechanisms acting via Neuronal-Nurturing Cells (NNC's) could portend replacement therapy and should be sought by multi-tissue screening for rev-transcriptase (RT) and viral tracks
W.K. Engel
- G.P.4.15 Sensory nerve conduction studies in patients with progressive state of amyotrophic lateral sclerosis
T.M. Makino; C.W. Watanabe
- Poster Area 3** **Posters 10 - Autosomal dominant diseases including caveolin-3 disorders and MFM**
Chairpersons: Z. Argov, A. van der Kooi
- D.P.3.01 Immunohistochemical and ultrastructural findings in myofibrillar myopathies
K.G. Claeys; M. Fardeau; D.O. Fürst; P.F.M. van der Ven; A. Behin; G. Brochier; O. Dubourg; B. Eymard; G. Faulkner; C. Guidy; R. Kley; T. Maisonobe; L. Manere; P. Richard; R. Schröder; T. Stojkovic; T. Suominen; K. Tolksdorf; P. Vicart; B. Udd; T. Voit; G. Stoltenburg
- D.P.3.02 Desminopathies: what can we learn from a long term follow-up?
A. Behin; T. Stojkovic; K. Claeys; K. Wahbi; D. Duboc; H.M. Becane; O. Dubourg; T. Maisonobe; G. Stoltenburg; M. Fardeau; P. Richard; B. Goudeau; P. Vicart; B. Eymard
- D.P.3.03 Inflammatory myopathy in scapulo-ilio-peroneal atrophy with cardiopathy in two unrelated families is associated with the desmin mutation Asn342Asp
A.J. van der Kooi; M.M.J. Kraak; P.F. Ippel; A.F. Vrancken; J.E. Hoogendijk; A. van den Wijngaard; J.P. van Tintelen; M. de Visser; F.G.I. Jennekens; J.D.H. Jongbloed
- D.P.3.04 Inclusion body myopathy with Paget's disease and frontotemporal dementia (IBMPFD): Extending the clinical features in a large pedigree
R. Barresi; T.D. Miller; A.P. Jackson; R. Charlton; J. Stone; V. Straub
- D.P.3.05 Molecular pathogenesis of hereditary inclusion body myopathies
S. Krause; N. Garcia-Angarita; A. Aleo; S. Hinderlich; M.C. Walter; S. Mitrani-Rosenbaum; T. Hoppe; H. Lochmuller

Full Programme

- D.P.3.06 Amyloidogenesis in a mouse model of DMRV/hIBM
M.C. Malicdan; S. Noguchi; Y. Hayashi; I. Nonaka; I. Nishino
- D.P.3.07 Welander distal myopathy: The evasive gene
P. Hackman; S. Hollo; H. Luque; M. Tokola; J. Kere; L. Edstrom; G. Ahlberg; B. Udd
- D.P.3.08 Physiotherapeutic description of an uncommon form of severe autosomal dominant limb girdle muscular dystrophy (LGMD) with early respiratory failure
M. Kånåhols; K. Dahlbom
- D.P.3.09 Caveolinopathy - new mutations and additional symptoms
A. Aboumoussa; J. Hoogendijk; R. Barresi; R. Charlton; R. Herrmann; T. Voit; J. Hudson; M. Roberts; D. Hilton-Jones; M. Eagle; K. Bushby; V. Straub
- D.P.3.10 Partial caveolin 3 deficiency in acquired rippling muscle disease
M. Mirabella; R. Charlton; E.M. Valente; S. Petrini; A. D'Amico; M. Roberts; E. Ricci; F. De Benedetti; R. Barresi; E. Bertini; V. Straub
- D.P.3.11 Influences of caveolin-3 mutations on canonical signaling pathways
E. Brauers; K. Reiss; M. Esser; J. Weis; A. Krüttgen
- D.P.3.12 Autosomal Recessive Ala93Thr mutation of Caveolin 3 gene: a new family
F. Magri; C. Lamperti; D. Ronchi; E. Fassone; N. Grimoldi; M. Moggio; N. Bresolin; G.P. Comi
- D.P.3.13 Morphological, stem cell and myosin abnormalities in the cav-3^{-/-} and mdx dystrophic embryo reveal an embryonic basis for muscular dystrophy
J. Smith; D. Merrick; L.K.J. Stadler; D. Larner
- D.P.3.14 A small-molecule inhibitor targeting transforming growth factor- β type I receptor kinase ameliorates muscular atrophy in a mouse model of caveolin-3-deficient muscular dystrophy
Y. Ohsawa; T. Okada; A. Kuga; S. Hayashi; T. Murakami; Y. Sunada
- Poster Area 4** **Posters 11 - Inflammatory diseases of muscle and nerve**
Chairpersons: V. Askanas, F. Mastaglia
- G.P.5.01 Interleukin (IL)-1 β induces accumulation of β -amyloid in skeletal muscle: Distinct interactions between inflammatory and degenerative pathomechanisms in sporadic inclusion body myositis
J. Schmidt; K. Barthel; A. Wrede; M. Salajegheh; M. Baehr; M.C. Dalakas
- G.P.5.02 Upregulation of α B-crystallin interrelates with APP and precedes accumulation of β -amyloid in the muscle of sporadic inclusion body myositis (sIBM)
I.E. Muth; K. Barthel; M. Baehr; M.C. Dalakas; J. Schmidt
- G.P.5.03 Pro-inflammatory cell-stress in primary muscle cell cultures of patients with sporadic inclusion body myositis compared to controls
K. Barthel; M. Baer; M.C. Dalakas; J. Schmidt
- G.P.5.04 ANT1 expression and RAGE-NF- κ B pathway in sporadic inclusion body myositis
C. Rodolico; V. Cianci; V. Macaione; M. Aguenouz; A. Mazzeo; A. Ciranni; N. Lanzano; M.G. de Pasquale; G.L. Vita; A. Toscano; G. Vita
- G.P.5.05 Transient overexpression of the Rho family exchange factor GEFT stimulates myogenic differentiation of inclusion-body myositis (IBM) mesoangioblasts
R. Morosetti; C. Gliubizzi; A. Broccolini; T. Gidaro; P.A. Tonali; M. Liu; E. Ricci; M. Mirabell

Full Programme

- G.P.5.06 HLA alleles and MHC haplotypes in sporadic inclusion body myositis: Frequencies and phenotypic correlations
M. Needham; A. Scott; F. Christiansen; I. James; A. Corbett; T. Day; L. Kiers; N. Laing; R. Allcock; F.L. Mastaglia
- G.P.5.07 Possible prognostic factors in the clinical course of patients with inclusion body myositis
F.M.E. Cox; J.J.G. Verschuuren; A.R. Wintzen; U.A. Badrising
- G.P.5.08 The prognosis of anti-synthetase syndrome is related to the lung involvement
R. Stanciu; Z. Amoura; M. Guiguet; A. Rigolet; L. Musset; F. Capron; D. Touitou; P. Cacoub; J.C. Piette; S. Herson; O. Benveniste
- G.P.5.09 Severe necrotising myopathy and cardiomyopathy with anti-signal recognition peptide antibodies
E. Matthews; M. Parton; R.L. Humbel; P. Elliot; C. Parry; J. Holton; M.G. Hanna
- G.P.5.10 Role of regulatory T cells in a new mouse model of experimental autoimmune myositis
S. Solly; Y. Allenbach; S. Grégoire; O. Dubourg; B. Salomon; G. Butler-Browne; L. Musset; S. Herson; D. Klatzmann; O. Benveniste
- G.P.5.11 Calf myositis or focal polyarteritis nodosa: a single entity?
T. Lebouvier; A. Magot; A. Masseur; P. Marcorelles; Y. Péréon; J.M. Mussini
- G.P.5.12 Decorin and lumican are differentially expressed in canine masticatory muscle myositis
O. Paciello; F. Trapani; S. Papparella
- G.P.5.13 Scapuloperoneal neuropathy responsive to intravenous immunoglobulin
N.J. McCathie; R. Quinlivan; M. James
- G.P.5.14 Treatment of hyposialylation in human and murine DMRV/hIBM culture cells with various kinds of sialic acid derivatives and its precursors
S. Noguchi; M.C.V. Malicdan; I. Nishino
- Poster Area 6B** **Posters 12 - Imaging**
Chairpersons: I. Illa, M. Kinali
- G.P.6.01 Establishing the parameters for clinical trials of antisense oligonucleotide therapy in Duchenne muscular dystrophy
M. Kinali; V. Arechavala-Gomez; L. Feng; A. Glover; M. Guglieri; H. Jungbluth; H. Roper; R.M. Quinlivan; D. Hunt; A.M. Manzur; A. Henderson; J. Gosalakkal; K. Hollingsworth; J. Allsop; E. Mercuri; J. Morgan; C. Sewry; V. Straub; K. Bushby; M. Rutherford; F. Muntoni
- G.P.6.02 MRI in Duchenne muscular dystrophy: Tracking progression
P. Garrod; K.G. Hollingsworth; B. Aribisala; K. Bushby; V. Straub
- G.P.6.03 Specific CT scanner muscle pattern helps to differentiate contractural lamin A/C and collagen VI related myopathies
N. Deconinck; E. Dion; A. Ferreira; B. Eymard; P. Richard; V. Allamand; R. Benyaou; G. Bonne; T. Stojkovic
- G.P.6.04 Dynamic muscle ultrasound detects fibrillations
S. Pillen; M. Nienhuis; I. Arts; N. Van Alfen; G. Drost; M. Zwarts
- G.P.6.05 Test/retest reliability and machine/machine correlation of dual energy x-ray absorptiometry (DEXA) measurements in patients with myotonic dystrophy type 1 & 2 (DM-1 and DM-2)
S. Pandya; N. Dilek; W. Martens; C. Thornton; R.T. Moxle

Full Programme

- G.P.6.06 Assessing murine dysferlin-deficient muscular dystrophy using intravascular contrast agents in 7T MRI
A.V. Vieweger; M.O. Obst; V.G. Gross; S.M. Müller; J.S. Steinbrin; B.M. Misselwitz; S.S. Spuler; M.G. Gutberlet; L.L. Lüdemann; S.S. Schmidt
- G.P.6.07 Simultaneous assessment of muscle perfusion and metabolic function in mice combining ¹H-NMR imaging and ³¹P-NMR spectroscopy in vivo
C. Baligand; C. Wary; J. Ménard; D. Bertoldi; E. Giacomini; P.G. Carlier
- G.P.6.08 Noninvasive evaluation of necrotic change in canine X-linked muscular dystrophy in Japan (CXMDJ) by fat-suppressed T2-weighted imaging
M.K. Kobayashi; A.N. Nakamura; D.H. Hasegawa; M.F. Fujita; H.O. Orima; S.T. Takeda
- G.P.6.09 Skeletal muscle characterization in golden retriever muscular dystrophy dogs by a nuclear magnetic resonance imaging longitudinal study
J.-L. Thibaud; D. Bertoldi; I. Barthélémy; S. Fleury; A. Monnet; S. Blot; P.G. Carlier
- Poster Area 5C** **Posters 13 - Dystrophin: gene and structural analysis and related proteins**
Chairpersons: J.C. Kaplan, L. Dux
- G.P.7.01 Combinatorial DHPLC analyses to identify point mutations in the dystrophin gene in 144 DMD/BMD patients
A. Trimarco; A. Torella; A. Cuomo; M.T. Bassi; M. Fanin; G. Galluzzi; C. Minetti; L. Politano; V. Nigro
- G.P.7.02 Comprehensive genetic analysis and clinical follow-up findings in 203 DMD patients
R. Virgilio; F. Magri; R. Del bo; S. Ghezzi; S. Tedeschi; M.G. D'Angelo; D. Coviello; A. Prele; A. Bordoni; M. Sciacco; C. Lamperti; S. Corti; Y. Torrente; M. Moggio; N. Bresolin; G.P. Comi
- G.P.7.03 Identification of Polyadenylated (PolyA+) transcripts within the dystrophin gene with a high density microarray
M. Bovolenta; P. Spitali; M. Fabris; M. Neri; E. Martoni; S. Falzarano; E. Bassi; P. Rimessi; F. Gualandi; A. Ferlini
- G.P.7.04 High Density-Comparative Genomic Hybridization (HD-CGH) array as a tool to detect deep intronic mutations in the dystrophin gene
M. Bovolenta; S. Fini; M. Neri; M. Fabris; E. Martoni; E. Bassi; P. Spitali; S. Falzarano; C. TrabANELLI; A. Venturoli; E. Ashton; S. Abbs; F. Muntoni; P. Rimessi; F. Gualandi; A. Ferlini
- G.P.7.05 Becker muscular dystrophy with a stop codon mutation in the 5' of the dystrophin gene
F. Magri; R. Del Bo; F. Fortunato; S. Ghezzi; R. Cagliani; M. Sironi; M.G. D'Angelo; V. Crugnola; M. Moggio; N. Bresolin; G.P. Comi
- G.P.7.06 Dystrophin gene analysis in Hungarian Duchenne/Becker Muscular Dystrophy families – Detection of carrier status in symptomatic and asymptomatic female relatives
V. Karcagi; V. Vancso; H. Piko; B. Nagy; Z. Ban; A. Herczegfalvi
- G.P.7.07 In vitro dissection of the pathogenic mechanisms of muscle fibrosis in Duchenne muscular dystrophy
S. Zanotti; S. Saredi; C. Cappelletti; A. Ruggieri; F. Blasevich; P. Bernasconi; L. Morandi; M. Mora
- G.P.7.08 Dystrotelins and the core function of dystrophins and dystrobrevins
R.G. Roberts

Full Programme

- G.P.7.09 A third syntrophin binding site: Higher isoform diversity of alpha-dystrobrevin in non-rodents
S.V. Boehm; H. Jin; R.G. Roberts
- G.P.7.10 Myostatin interacts with syndecan-4 and PKC-alpha in skeletal muscle
A. Keller - Pinter; L. Mendler; L. Dux

Wednesday 1 October 2008

- 08:00-09:30 Sage Hall 1 Advances in the understanding and treatment of myasthenic disorders; Invited lectures (M.I. 1-3)
Chairpersons: D. Hilton-Jones, F. Deymeer**
- M.I.1 Congenital myasthenias - an overview
A.G. Engel
- M.I.2 Congenital myasthenic syndromes and the formation of the neuromuscular junction
D.M.W. Beeson
- M.I.3 Congenital myasthenic syndromes - diagnosis and therapy
H.K.M. Lochmuller
- 09:30-10:00 Sage Morning Tea & Coffee
- 10:00-11:00 Sage Hall 1 Advances in the understanding and treatment of myasthenic disorders; Invited lectures (M.I. 4-6)
Chairpersons: A. Emslie Smith, P. Van der Bergh**
- M.I.4 Autoimmune Myasthenia: Clinical aspects
A. Evoli
- M.I.5 Auto antibody mediated effector mechanisms in Myasthenia Gravis and its animal model: about antibodies and anchor proteins
M. De Baets
- M.I.6 What we can learn from animal models of autoimmune myasthenia gravis
G.D. Shelton
- 11:00-12:00 Sage Hall 1 Congenital myasthenia and dysferlin; Oral Presentations (G.O.4-6)
Chairpersons: C. Bönneman, A. Toscana**
- G.O.4 Neuromuscular junction formation in Dok-7 deficient zebrafish embryos
J.S. Müller; P. Thornhill; K. Bushby; V. Straub; H. Lochmuller
- G.O.5 Partial functionality of a mini-dysferlin molecule identified in a patient affected with moderately severe primary dysferlinopathy
M. Krahn; N. Wein; W. Lostal; N. Bourg-Alibert; K. Nguyen; S. Courier; C. Vial; V. Labelle; D. De Petris; A. Borges; M.G. Mattei; C. Roudaut; K. Miyake; P. McNeil; P. Cau; F. Leturcq; M. Bartoli; N. Levy; I. Richard
- G.O.6 Sj1 dystrophic mice express large amount of human muscle proteins following systemic delivery of human adipose-derived stem cells
N.M. Vieira; C.R. Bueno Junior; V. Brandalise; E. Zucconi; M. Secco; M.D. Carvalho; M.F. Suzuki; P. Bartolini; P.C. Brum; M. Vainzof; M. Zatz
- 12.00-13.30 Hilton Hotel Lunch & Exhibition

Full Programme

13.30-14.30	Hilton Hotel	Poster session 3: parallel sessions (14-20)
	Poster Area 1	Posters 14 - Applications of animal models of disease <i>Chairpersons: H. Amthor, E. Hardeman</i>
G.P.8.01		Functional muscle, histological and biomarker analysis in the mdx mouse <i>M. Van Putten; C.L. de Winter; J.C.T. van Deutekom; G.J.B. van Ommen; A. Aartsma-Rus</i>
G.P.8.02		Humanizing mouse glycosylation to build a better mdx model for DMD <i>K. Chandrasekharan; J.H. Yoon; M. Camboni; J.R. Mendell; Z. Sahenk; P. Janssen; A. Varki; P.T. Martin</i>
G.P.8.03		Generation of double mutant mouse for the genes dystrophin and large: A new model for neuromuscular diseases <i>P.C.M. Martins; D. Ayub-Guerrieri; V.F. Lopes; P.C.G. Onofre; C.M.C. Mori; M. Vainzof</i>
G.P.8.04		Disrupted Src signalling in dystrophin-deficient mdx muscle cells responding to stress <i>G.M. Smythe</i>
G.P.8.05		Effect of hematopoietic prostaglandin D synthase inhibitor on bupivacaine hydrochloride-induced muscular necrosis <i>M. Hayashi; K. Aritake; I. Mohri; Y. Sato; H. Suzuki; M. Masaki; E.K. Mitamura; Y. Urade</i>
G.P.8.06		Treatment with a soluble Activin type IIB receptor increases muscle size and strength of dystrophin deficient muscle <i>J.L. Lachey; E.E. Pistilli; S. Bogdanovich; A.E. Pullen; R.S. Pearsall; T.S. Khurana; J. Seehra</i>
G.P.8.07		Increased oxidative metabolism in mdx muscle treated by a combination of exon skipping and myostatin blockade <i>C. Hourdé; S. Marie; E. Mouisel; L. Garcia; J. Dumonceaux; H. Amthor</i>
G.P.8.08		Codon optimisation of microdystrophin results in improvements in expression and physiological outcome in the mdx mouse following AAV8 gene transfer <i>H. Foster; D.J. Wells; C. Trollet; T. Athanasopoulos; I. Graham; K. Foster; J.G. Dickson</i>
G.P.8.09		Evaluating the potential of AAV8 mediated intravenous transfer of myostatin propeptide to ameliorate the muscle pathology in mdx mouse <i>K. Foster; I. Graham; H. Foster; C. Trollet; P. Yaworsky; F. Walsh; P. Sharp; D. Wells; G. Dickson</i>
G.P.8.10		Immunosuppressive regimen fails to improve dystrophic phenotype and impairs muscle function in the dog model of Duchenne muscular dystrophy <i>I. Barthélémy; A. Uriarte; J.L. Thibaud; S. Blot</i>
G.P.8.11		Steroid treatment causes deterioration of myocardial function in the delta-sarcoglycan deficient mouse model for dilated cardiomyopathy <i>R. Bauer; G.A. Mac Gowan; A. Blain; K. Bushby; V.W. Straub</i>
G.P.8.12		Mannosidase I inhibition rescues the human α -sarcoglycan R77C recurrent mutation <i>I. Richard; M. Bartoli; E. Gicquel; L. Barrault; T. Soheili; M. Malissen; B. Malissen; B. Udd; O. Danos</i>
G.P.8.13		Accelerometry, a new tool to assess gait quality in dystrophin-deficient dogs <i>I. Barthélémy; E. Barrey; J.L. Thibaud; A. Uriarte; S. Blot; J.Y. Hogrel</i>

Full Programme

Poster Area 2

Posters 15 - Congenital myopathies I

Chairpersons: C. Sewry, H. Goebel

- G.P.9.01 A mutant skeletal muscle α -actin gene, fused to enhanced green fluorescent protein (EGFP) produces a unique myopathic mouse model
K.J. Nowak; G. Ravenscroft; C. Jackaman; E.M. Lim; C.A. Sewry; A. Potter; S. Squire; R. Fisher; E. Baker; J.J. Feng; S. Marston; V. Fabian; P.J. Morling; A.J. Bakker; L.M. Griffiths; J. Papadimitriou; K.E. Davies; N.G. Laing
- G.P.9.02 Expression of cardiac α -actin spares extraocular muscles in skeletal muscle α -actin diseases - determination of cardiac α -actin by MRM mass spectrometry
G. Ravenscroft; S.M.J. Colley; K.R. Walker; S. Clement; S. Bringans; R. Lipscombe; V. Fabian; N.G. Laing; K.J. Nowak
- G.P.9.03 Characterization of the nebulin promoter and detection of brain-specific transcripts
S.A. Ranta; V.L. Lehtokari; E. Nuutinen; D. Brudzewsky; C. Wallgren-Pettersson; K. Pelin
- G.P.9.04 Development of the multiplex ligation-dependent probe amplification (MLPA) method for identifying large scale mutations in the nebulin gene
M. Lunkka-Hytonen; V.L. Lehtokari; K. Pelin; D. Brudzewsky; C. Wallgren-Pettersson
- G.P.9.05 Pathomechanism of SIL1 mutated Marinesco-Sjögren syndrome
M. Okada; S. Noguchi; M.C. Malicdan; I. Nonaka; Y.K. Hayashi; I. Nishino
- G.P.9.06 Marinesco Sjögren syndrome: Correlation of nuclear changes to mutations in BAP/SIL1
J.M. Schröder; J. Senderek; J. Weis
- G.P.9.07 Congenital fibre type disproportion and non compaction cardiomyopathy associated with insulin resistance
D. Diodato; S. Sampaolo; A. Varone; G. Limongelli; M. Simonetti; P. Calabrò; R. Calabrò; G. Di Iorio
- G.P.9.08 Severe cardiomyopathy in hyaline body myopathy: Ten years of follow-up
A.N. Yuceyar; L. Baysal; O.E. Ozbay; A.S. Kocaman; H. Karasoy
- G.P.9.09 Autophagic vacuolar myopathy in a female patient
M. Ohlsson; C. Lindberg; A. Oldfors
- G.P.9.10 A case of fibro-dysplasia ossificans progressiva with a novel mutation (G356D) of the activin receptor type 1 gene (ACVR1(ALK2))
H. Furuya; K. Ikezoe; N. Fujii; J.I. Kira; H. Arahata; Y. Fukumaki
- G.P.9.11 Histopathological findings in Vici syndrome
V.M. McClelland; O. Miller; W. Jan; L. Amaya; A. Buj-Bello; V. Biancalana; M. Bitoun; I. Bodi; D.M. Ruddy; S. Mohammed; H. Jungbluth
- G.P.9.12 Novel myosin heavy chain immunohistochemical double staining for the diagnostic routine assessment of fiber types
O. Raheem; S. Huovinen; T. Suominen; H. Haapasalo; B. Udd
- G.P.9.13 Expressions of FOXO1 and FOXO3a in skeletal muscle atrophy associated with neuromuscular diseases
A. Ishii; N. Ohkoshi; A. Tamaoka
- G.P.9.14 Long term effects of botulinum toxin treatment on muscle morphology in patients with cerebral palsy
J. Patrick; A. Roberts; C.A. Sewry

Full Programme

- Poster Area 3** **Posters 16 – Dysferlinopathy**
Chairpersons: S. Spuler, V. Nigro
- G.P.10.01 Dysferlinopathies in Southern Italy
A. Palladino; S. Aurino; M.R. Cecio; G. Piluso; L. Passamano; V. Nigro; L. Politano
- G.P.10.02 Proteomics identification of differentially expressed proteins in the muscle of dysferlin myopathy patients
C. De La Torre; I. Illa; G. Faulkner; R. Robles-Cedeño; R. Dominguez-Perles; E. Gallardo
- G.P.10.03 Quantification of dysferlin in monocytes: A useful tool for the detection of patients and carriers of dysferlinopathy
N. De Luna; E. Gallardo; R. Rojas-Garcia; R. Dominguez-Perles; J. Diaz-Manera; C. De La Torre; P. Gallano; I. Illa
- G.P.10.04 Is complement inhibition a potential therapy in dysferlin-deficient muscular dystrophy?
V. Schöwel; J. Zabojszcza; S. Schmidt; S. Spuler
- G.P.10.05 Preclinical drug trials investigating potential treatments for dysferlin deficiency
M.A. Hornsey; L. Klinge; S.H. Laval; R. Barresi; H. Lochmuller; V. Straub; K. Bushby
- G.P.10.06 Abnormal T-tubule morphology in dysferlin deficient muscle
L. Klinge; C. Sewry; S. Laval; M.A. Hornsey; Y. Chiu; V. Straub; R. Barresi; H. Lochmüller; K. Bushby
- G.P.10.07 Role of inflammasome in the pathogenesis of Dysferlin deficiency
R. Rawat; E.P. Hoffman; K. Nagaraju
- G.P.10.08 Increased lysosomes trafficking and poly(ADP-ribose)polymerase-1 expression in dysferlin myopathy: Implications in muscle fiber necrosis
R. Dominguez-Perles; N. De Luna; C. De la Torre; S.T. Cooper; R. Robles-Cedeño; R. Rojas-Garcia; J.A. Diaz-Manera; I. Illa; E. Gallardo
- G.P.10.09 Mitochondrial dysfunction in dysferlinopathy
J.L. Murphy; R. Charlton; R. Barresi; K.M. Bushby; R.W. Taylor; D.M. Turnbull
- G.P.10.10 Dysferlin does not play an essential role in skeletal myoblast fusion
Y. Chiu; S.H. Laval; H. Lochmuller; M.A. Hornsey; L. Klinge; V. Straub; K. Bushby; R. Barresi
- G.P.10.11 T-CAP, the gene responsible for LGMD2G, may interact with dysferlin
M. Cacciottolo; V. Saccone; G. Numitone; G. Piluso; S. Aurino; V. Nigro
- G.P.10.12 Dissociated localization of caveolin-3 and dysferlin during muscle regeneration after cardiotoxin injury
A.K. Kuga; Y.O. Ohsawa; Y.S. Sunada
- G.P.10.13 Evidence of ferlin mediated membrane shedding in muscle cells highlights the existence of muscle cell exosomes, myosomes
U. Ramachandran; K. Saleki; G. Marlow; R. Bashir
- Poster Area 5C** **Posters 17 - Clinical trial infrastructure**
Chairperson: T. Sejersen
- T.P.3.01 Interactive, web-based system facilitates multi-center clinical research operations
R. Buchsbaum; J. Montes; J.L.P. Thompson; P. Kaufmann
- T.P.3.02 The ICC: Working to facilitate the conduct of fast, efficient and effective clinical trials in SMA
C. Joyce

Full Programme

- T.P.3.03 TREAT-NMD- Activity 7: Accelerate preclinical phase of new therapeutic treatment development
R. Willmann; M.A. Rüegg; R. Fairclough; K.E. Davies; S. Possekel; T. Meier
- T.P.3.04 EuroBioBank for the development and management of supranational biobanks to support research on neuromuscular disorders
S.K. Baumeister; A.M. Bodin; F. Bignami; H. Lochmueller
- T.P.3.05 TREAT-NMD global patients' registries: A unified global source of information about patients with neuromuscular diseases
V. Humbertclaude; S. Tuffery-Giraud; D. Hamroun; F.O. Desmet; S. Baumeister; M. Lalande; G. Collod-Beroud; H. Lochmüller; M. Claustres; C. Beroud
- T.P.3.06 The TREAT NMD registry of outcome measures for neuromuscular disease - an introduction
J.M. Auld; R. Seyedsadjadi; M. Rose
- T.P.3.07 TREAT-NMD work on standards of diagnosis and care of NMDs
T.S. Sejersen
- T.P.3.08 The TREAT-NMD Clinical Trials Coordination Centre (CTCC)
A. Stanescu; J. Kirschner; C. Marx; A. Pohl; A. Tassoni; S. Geismann; H. Maier-Lenz; R. Korinthenberg
- Posrer Area 4 Posters 18 - Myotonic dystrophy I and II, and OPMD**
Chairpersons: G. Vita, G. Butler-Browne
- D.P.4.01 Endocrine and metabolic disorders in myotonic dystrophy type 1
M.N. Cardoso; E. Santos; A. Carvalho; T. Coelho
- D.P.4.02 Dysphagia in Myotonic Dystrophy type 2
A.A. Tieleman; J. van Vliet; S. Knuijt; B.J.M. de Swart; R. Ensink; B.G.M. van Engelen
- D.P.4.03 Sleep disorder and sleep-related breathing impairment in myotonic dystrophy
C.W. Watanabe; M.H. Higaki; T.M. Makino
- D.P.4.04 Cerebral white matter affection in myotonic dystrophy type 1 and 2 - a diffusion-tensor-imaging study at 3T -
M. Minnerop; J.C. Schoene-Bake; S. Mirbach; C. Helmstaedter; B. Weber; M. Tittgemeyer; T. Klockgether; C. Kornblum
- D.P.4.05 CTG expansions in relation to symptoms and muscle histopathology in DM1 patients
G.S. Butler-Browne; M. Lindstrom; A. Klein; T. Ansved; V. Mouly; L.E. Thornell
- D.P.4.06 No correlation of increase in size of ribonuclear inclusions to type II fibre atrophy in myotonic dystrophy type 2 over time
R. Cardani; R. Perbellini; E. Mancinelli; G. Meola
- D.P.4.07 In vitro study of DM1 primary myotubes
E. Loro; A. Botta; C. Catalli; V. Romeo; F. Rinaldi; C. Angelini; L. Vergani
- D.P.4.08 P16 triggers premature senescence of congenital DM1 myoblasts
G.S. Butler-Browne; A. Bigot; E. Gasnie; V. Mouly; D. Furling
- D.P.4.09 Defective mRNA in myotonic dystrophy accumulates at the periphery of nuclear splicing speckles
I. Holt; S. Mittal; D. Furling; G.S. Butler-Browne; J.D. Brook; G.E. Morris

Full Programme

- D.P.4.10 Muscblind-like proteins: Similarities and differences in normal and myotonic dystrophy muscle
I. Holt; D. Furling; M. Fardeai; C.A. Sewry; V. Jacquemin; G.S. Butler-Browne; J.D. Brook; G.E. Morris
- D.P.4.11 Expression and siRNA targeting of PABPN1 as a model for oculopharyngeal muscular dystrophy (OPMD)
C. Trollet; J. Benstead; I. Graham; R. Yáñez; M. Antoniou; G. Dickson
- D.P.4.12 Phenotype and genotype study of oculopharyngeal muscle dystrophy in patients from southern Spain
C. Paradas; M. Cabrera; J. Jiménez; E. Rivas; R. García; J. Bautista; C. Márquez
- Poster Area 6A** **Posters 19 - Pompe Disease**
Chairpersons: P. van Doorn, E. Wraith
- G.P.11.01 Alglucosidase alfa in infants and children with Pompe disease
E. Wraith; B. Byrne; W.L. Hwu; N. Leslie; H. Mandel; M. Nicolino; P.S. Kishnani
- G.P.11.02 Initial efficacy of enzyme replacement therapy for a patient with childhood-onset Pompe disease
K. Ishigaki; T. Murakami; T. Nakanishi; K. Shishikura; H. Suzuki; Y. Hirayama; M. Osawa
- G.P.11.03 Variable response to enzyme replacement therapy in children and adults with Pompe disease: Need for identification of prognostic factors
N.A.M. Van der Beek; C.I. Van Capelle; J.M. De Vries; A.J.J. Reuser; P.A. Van Doorn; A.T. Van der Ploeg
- G.P.11.04 A follow up observation of enzyme replacement therapy in Pompe disease
A. Dubrovsky; J.C. Corderi; H. Amartino; A. Calle
- G.P.11.05 The pharmacological chaperone AT2220 increases trafficking, processing, and cellular activity of acid alpha-glucosidase and is a potential new treatment for Pompe disease
A.C. Powe; J.F. Flanagan; R. Khanna; X. Wu; W. Liang; R. Dhulipala; T. Yin; K. Tang; R. Soska; L. Pellegrino; S. Shao; E.R. Benjamin; K.J. Valenzano; B.A. Wustman; D.J. Lockhart; H.V. Do
- G.P.11.06 Pharmacological chaperone therapy for Pompe disease: Phase 1 clinical trials and ex vivo response study results
C.W. Pine; H. Nafar; D.J. Palling; A.E. Slonim; A. Pestronk; B.J. Byrne; P. Kishnani; B. Ranes; C.J. Kissling; R. Schwab; S. Connolly; S. Sitaraman; J. Castelli; F. Insinga; R. Lazauskas; M.C. Furlow; H.V. Do; D.J. Lockhart; B.A. Wustman
- G.P.11.07 Impairment of the endosomal/lysosomal system may contribute to muscle wasting in Pompe disease by altering the trafficking and processing of membrane stabilizing proteins
B.A. Wustman; T. Voit; A.M. Cuervo; A.C. Massey; U. Bandyopadhyay; A.C. Powe, Jr.; K. Cheng; H.V. Do; D.J. Lockhart
- G.P.11.08 Cytoplasmic body with acid phosphatase activity - Hallmark of adult-onset Pompe disease on muscle pathology
I. Nishino; Y. Oya; K. Monma; S. Noguchi; Y.K. Hayashi; I. Nonaka
- G.P.11.09 Evaluation of optoelectronic plethysmography (OEP) in assessment of respiratory function in patients with type II glycogenosis (adult form)
G. Remiche; A. Lo Mauro; A. Aliverti; P. Tarsia; G.P. Comi; N. Bresolin; M.G. D'Angelo

Full Programme

G.P.11.10 Frequent pulmonary dysfunction in patients with late-onset Pompe disease necessitates careful evaluation during routine follow-up
N.A.M. Van der Beek; C.I. Van Capelle; K. Van der Velden; W.C.J. Hop; H. Stam; A.T. Van der Ploeg; P.A. Van Doorn

G.P.11.11 Phenotypes of Pompe disease siblings
D. de Castro; K. Laloui; S. Sacconi; V. Doppler; C. Payan; F. Zagnoli; P. Petiot; D. Orlikowski; A. Magot; C. Caillaud; P. Laforêt

G.P.11.12 Tongue weakness in Pompe disease
A. Dubrovsky; J.C. Corderi

Poster Area 6B **Posters 20 - Dystrophinopathy - clinical observations** **Chairpersons: G. Nigro, T. Voit**

G.P.12.01 Epidemiology of the dystrophinopathies in the Netherlands
J.C. van den Bergen; C.S.M. Straathof; R.F. Pangalila; R. Broekgaarden; W.C.G. Overweg-Plandsoen; A.M. Aartsma-rus; H.B. Ginjaar; E. Bakker; G.J.B. van Ommen; J.J.G. Verschuuren

G.P.12.02 Asymmetric hypertrophy and contractures in an adult male due to somatic mosaicism for a DMD stop mutation
E. Graham; A. Ahmed; L. Kane; S. Cooke; C. Longman; W. Stewart; R.K.H. Petty

G.P.12.03 Marked hemi-atrophy in a manifesting carrier of Duchenne muscular dystrophy—Possible role of skewed X-inactivation
S. Rajakulendran; S. Farmer; T. Yousry; E. Ashton; S. Abbs; J. Holton; M.G. Hanna; E. Matthews

G.P.12.04 Clinical features, particularly those of the central nervous system of patients with Becker muscular dystrophy, including autopsied cases
K. Adach; H. Kawa; M. Sait; S. Kashiwa; N. Kaga; T. Sano

G.P.12.05 Gastrostomy tube feeding in patients with severe Duchenne muscular dystrophy
H. Tana; H. Kon; T. Takahas; M. Yoshioka; H. Onoder; K. Ishida

G.P.12.06 Resting energy expenditure and nutritional inadequacy in Duchenne muscular dystrophy
H. Komaki; M. Shimizu; E. Nakagawa; M. Yoshimura; Y. Ohya; T. Fujisaki; Y. Saida; C. Kubota; S. Itoh; R. Shimazaki; K. Sato; T. Ishikawa; H. Mochizuki; T. Takanoha; M. Konagaya; T. Mlyazaki; K. Tatara

G.P.12.07 Cardiac involvement in manifesting female carriers of Duchenne and Becker muscular dystrophy
M.K. Al-raqad; A. Aboumoussa; M. Guglieri; J.P. Bourke; K. Bushby

G.P.12.08 Clinico-pathological characteristics of the Becker muscular dystrophy with rimmed vacuole
K. Momma; S. Noguchi; Y.K. Hayashi; K. Motoyoshi; K. Kamakura; I. Nonaka; I. Nishino

G.P.12.09 Audit of Duchenne muscular dystrophy in southwest England
N. Loh; K. Mahmood

G.P.12.10 Delayed development and learning difficulties as a predominant symptom in female carriers of Duchenne and Becker muscular dystrophy
M. Guglieri; A. Aboumoussa; M. Eagle; J. Bourke; V. Straub; K. Bushby

G.P.12.11 Creatine kinase and transaminases in Duchenne muscular dystrophy
P.R. Fequiere; B.L. Wong; P.S. Horn

Full Programme

- G.P.12.12 Predictive factors for progressive foot deformity in non ambulant boys with Duchenne muscular dystrophy
C. Nicholson; M. Main; M. Kinali; F. Muntoni; E. Mercuri
- 14.30-15.30 Hilton Hotel Poster Area 1** **Poster session 4: parallel sessions (21-26)**
Posters 21 - Congenital myopathies II
Chairpersons: A. Oldfors, J. Nonaka
- G.P.13.01 A family with Trismus pseudocamptodactyly and nemaline rods on muscle biopsy. Expanding the nemaline myopathy phenotype ?
A. Majumdar; S. Betmouni; P. Lunt
- G.P.13.02 Functional study of mutated beta-tropomyosin causing nemaline myopathy, cap myopathy and distal arthrogyrosis
M. Marttila; E. Nuutinen; D. Brudzewsky; S. Ollila; K. Donner; K. Pelin; C. Wallgren-Pettersson
- G.P.13.03 Mechanisms of rod formation in disease
A.V. Vandebrouk; A.D. Domazetovska; S.T.C. Cooper; B.I. Ilkovski; K.N.N. North
- G.P.13.04 Genetic and ultrastructural findings in Selenoprotein N1-related congenital myopathies
R. Cagliani; E. Fruguglietti; A. Berardinelli; M.G. D'Angelo; A. Prella; K. Gorni; S. Orcesi; C. Lamperti; E. Signaroldi; R. Tupler; M. Moggio; G.P. Comi
- G.P.13.05 Investigating the pathophysiology of SEPN1-related myopathy using gene expression microarrays
N.F. Clarke; S. Nadaud; M. Rederstorff; A. Lescure; A. Krol; P. Guicheney
- G.P.13.06 Distal myopathy in multi-minicore disease
S. Okahashi; I. Nonaka; S. Wu; C.A.M. Ibarra; S. Shalaby; Y.K. Hayashi; S. Noguchi; I. Nishino
- G.P.13.07 Late-onset axial myopathy with cores due to a novel dominant mutation in the skeletal muscle ryanodine receptor (RYR1) gene
H. Jungbluth; S. Lillis; H. Zhou; S. Abbs; M. Swash; F. Muntoni
- G.P.13.08 Benign phenotype in compound heterozygosity autosomal recessive central core disease: Case report
L.J.M. Negrão; A. Matos; O. Rebelo; A. Geraldo; R. Santos
- G.P.13.09 Identification of a point mutation in the skeletal muscle ryanodine receptor gene associated in the homozygous state to central core disease
G. Melli; L. Colleoni; P. Bernasconi; S. Romaggi; V. Tegazzin; R. Mantegazza; L. Morandi
- G.P.13.10 High frequency of polymorphisms in the RYR1 gene in Brazilian patients with centronuclear myopathy
L.U. Yamamoto; L.S. Maia; D. Ayub-Guerrieri; P.C.G. Onofre; V.F. Lopes; D. Zilberztajn; P.C.M. Martins; A.S. Senkevics; A.L.F. Santos; K. Sell; M. Zatz; H.C. Silva; J. Gurgel-Gianneti; M. Vainzof
- G.P.13.11 Autosomal centronuclear myopathy in a Turkish family
H. Karasoy; A.N. Yuceyar; O.E. Ozbay; A. Gokcay
- G.P.13.12 Congenital myotonic dystrophy and myotubular myopathy may be differentiated by type 2C fibers and peripheral halos
C. Fujimura; S. Noguchi; N. Minami; I. Nonaka; Y.K. Hayashi; I. Nishino
- G.P.13.13 Identification and characterization of a novel congenital myopathy
J.J. Dowling; K. Majczenko; M. Blaivas; M. Burmeister

Full Programme

- G.P.13.14 A new form of congenital myopathy with severe fibre immaturity
H. Amthor; S. Quijano-Roy; M. Mayer; K. Maincent; B. Estournet; J. Bataille; A. Essid; A. Lombes; P. Guicheney; M. Fardeau; K. Claeys; C. Rambaud; A. Gelot; G. Stoltenburg-Didinger
- G.P.13.15 Diagnosis and outcome of hypotonia in infancy
G.P. Ambegaonkar; S. Shah; H. Roper
- Poster Area 2** **Posters 22 - Pharmacological studies in the mdx mouse**
Chairpersons: U. Ruegg, R. Baressi
- T.P.4.01 Identification and characterization of small molecules for the treatment of Duchenne muscular dystrophy
W. Friesen; S. Acharjee; J. Zhuo; R. Baiazitov; S. Lee; Y.C. Moon; H.L. Sweeney; E. Welch
- T.P.4.02 Evaluation of novel compounds for upregulation of utrophin in animal models of Duchenne muscular dystrophy therapy
R.J. Fairclough; A. Potter; D. Powell; S. Squire; M. Bland; A. Bareja; J. Tinsley; K.E. Davies
- T.P.4.03 Diltiazem and verapamil protect dystrophin-deficient muscle fibers of mdx mice from degeneration: Potential role in calcium buffering and sarcolemmal stability
H. Santo Neto; C.Y. Matsumura; M.J. Marques
- T.P.4.04 Tamoxifen improves the structure and the function of skeletal muscle in mdx mice
O.M. Dorchies; J. Reutenauer; O. Vuadens; S.A. Comyn; U.T. Ruegg
- T.P.4.05 Urocortins improve skeletal muscle structure and function of mdx mouse via a cyclic AMP/PKA dependent pathway
J. Reutenauer; O.M. Dorchies; F.X. Boittin; O. Patthey-Vuadens; U.T. Ruegg
- T.P.4.06 Green tea polyphenols and pentoxifylline stimulate dystrophic myotube formation and maturation in primary cultures
O.M. Dorchies; U.T. Ruegg
- T.P.4.07 Green tea polyphenols as potential treatment of Duchenne muscular dystrophy
U.T. Ruegg; O.M. Dorchies; J. Reutenauer; O. Vuadens; E. Roulet
- T.P.4.08 In-vivo direct and indirect myocardial effects of captopril in the mdx mouse
R. Bauer; V.W. Straub; A. Blain; K. Bushby; G.A. Mac Gowan
- T.P.4.09 Left ventricular remodeling after steroid therapy in the mdx mouse
R. Bauer; G.A. Mac Gowan; A. Blain; K. Bushby; V.W. Straub
- T.P.4.10 Melatonin prevents oxidative-stress mediated mitochondrial permeability transition and death via enhancement of reduced pyridine nucleotides and glutathione in mouse skeletal muscle cells
Y. Hibaoui; E. Roulet; U.T. Ruegg
- T.P.4.11 Effect of calpain and proteasome inhibition on calcium-dependent proteolysis and muscle histopathology in the mdx mouse
A. Briguet; M. Erb; I. Courdier-Fruh; P. Barzaghi; G. Santos; H. Herzner; C. Lescop; H. Siendt; M. Henneboehle; P. Weyermann; J. Dubach-Powell; G. Metz; T. Meier; J. Magyar
- T.P.4.12 Carbonic anhydrase inhibitors' therapeutic effects on dystrophin-deficient mouse muscle
C.P. Pertl; L.S. Segalat; H.L. Lochmueller; C.T. Thirion

Full Programme

Poster Area 3

Posters 23 - Autosomal recessive limb-girdle muscular dystrophies

Chairpersons: *I. Ginjaar, K. Wrogemann*

- G.P.14.01 Immunohistochemical analysis of calpain 3: Advantages and limitations in diagnosing LGMD2A
R. Charlton; M. Henderson; J. Richards; J. Hudson; K. Bushby; R. Barresi
- G.P.14.02 MLPA analysis of the CAPN3 gene detects large deletions in LGMD2A patients
I. Ginjaar; S. Tuit; W. Frankhuizen; A. van der Kooij; P. Doorn; D. Sival; E. Bakker
- G.P.14.03 Quantitative analysis of CAPN3 and DMD transcripts: Involvement of nonsense-mediated mRNA decay
J. Sedlackova; K. Stehlikova; M. Hermanova; P. Vondracek; L. Fajkusova
- G.P.14.04 Interactions of Myospryn with M-band Titin and Calpain 3
J. Sarparanta; A. Vihola; S. Marchand; G. Blandin; P. Hackman; E. Ehler; I. Richard; B. Udd
- G.P.14.05 Trim32 is the gene for limb girdle muscular dystrophy type 2H. What do we know about it?
K. Wrogemann; H. Ding; Y. Heng; A. Kania; X. Wu; N. Patel; A. Funk; C. Hirst; S. Krawitz; D. Gietz; M. DelBigio
- G.P.14.06 LGMD2H patients of non Hutterite origin with mutations in TRIM32 gene
V. Saccone; M. Scutifero; A. Palladino; M.G. Di Gregorio; V.M. Ventriglia; G. Piluso; N. Canki-Klain; V. Nigro; L. Politano
- G.P.14.07 Novel mutation in telethonin causing autosomal recessive muscular dystrophy type 2G in a Moldavian patient
M. Olive; A. Shatunov; O. Carmona; J.A. Martinez-Matos; L.G. Goldfarb; I. Ferrer
- G.P.14.08 Sarcoglycanopathies: Clinical and histopathological characteristics in 58 patients
A. Nalini
- G.P.14.09 Molecular genetic testing of the sarcoglycanopathies
D. Routledge; J.A. Hudson; A. Curtis
- G.P.14.10 Myalgia and exercise-induced myoglobinuria is frequent in LGMD 2I
C. Lindberg; C. Sixt; A. Oldfors
- G.P.14.11 Family with unusual muscular dystrophy with predominant scapulo-humero-peroneal distribution and autosomal recessive inheritance.
T. Torbergesen; S. Løseth
- G.P.14.12 Phenotype of three putative novel limb girdle muscular dystrophies (LGMD) - exclusion of all known LGMD loci with microsatellite analysis
M. von der Hagen; M.C. Walter; H. Lochmueller; K.M.D. Bushby; M. Vorgerd; A. Huebner
- G.P.14.13 Phenotypic characterization of two unrelated UK families with X-linked myopathy and an identical mutation in the FHL1 gene
A. Sarkozy; K. Bushby; D. Hilton-Jones; C.F. Dougan; H. Lochmüller; C. Windpassinger; V. Straub
- G.P.14.14 Novel FHL1 mutations in fatal and benign reducing body myopathy
S. Shalaby; Y.K. Hayashi; I. Nonaka; S. Noguchi; I. Nishino
- G.P.14.15 Evaluating the role of the dystroglycan α/β interface in human muscular dystrophies
F. Sciandra; S. Morlacchi; B. Giardina; M. Bozzi; A. Brancaccio

Full Programme

Poster Area 4

Posters 24 - Psychosocial aspects of muscle diseases

Chairpersons: *S. Pandya, B. Steffensen*

- G.P.15.01 Transition to adulthood for young men with DMD
J.S.W. Carpenter; D.W. Abbott; K. Bushby
- G.P.15.02 Transition in neuromuscular conditions
E. Perkins
- G.P.15.03 Neuropsychological profiles of children with DMD in south India
V. Viswanathan; S. Archana Simon; B.R. Lakshmi; M. Sakthivel; G. Aarthy; R. Saravanan
- G.P.15.04 Investigating the effects of family education in disabled children in Turkey
A. Karaduman; Ö. Yilmaz; Y. Yakut; A. Meriç; H. Kayihan; A. Livanelioglu; Ö. Aras; B. Aras; E. Telci; E. Simsek; A. Mutlu; E. Açık; I. Alemdaroglu; N. Yagli; H. Topaloglu
- G.P.15.05 The comparison of quality of life in children with cerebral palsy and neuromuscular diseases
A. Karaduman; Ö. Yilmaz; H. Tüzün; M.K. Günel; B. Aras; A. Mutlu; T. Tarsuslu; Ö. Aras; L. Eker; H. Topaloglu;
- G.P.15.06 On-line psychological support for parents of children and teenagers with neuromuscular disease
I. Amayra; E. Lazaro; J.F. Lopez; A. De la Cruz Beldarrain
- G.P.15.07 On-line psychological support for children and teenagers with neuromuscular disease
I. Amayra; A. De La Cruz Beldarrain; J.F. Lopez; E. Lazaro
- G.P.15.08 Shifting questions: a new protocol for taking the medical history, facilitating an early and accurate diagnosis for muscular dystrophy patients and modifying the clinical course
E.B.S. Meirelles; T.F. Soares; A.A. Castro; G.S.C. Lins; C.T.S. Castro; A.J. Godoy
- G.P.15.09 Consent, choice and children in research: Exploring decision making by parents of children with Duchenne muscular dystrophy considering participation in genetic research projects
R.A. Henderson; T. Shakespeare; K. Bushby; S. Woods
- G.P.15.10 Screening for signs of myopathy associated with drugs of abuse and hepatitis C infection in patients admitted for detoxification treatment
J. Reimann; H. Kölsch; C.G. Schütz
- G.P.15.11 Inclusive medical education to improve early diagnosis: The model of muscular dystrophies
G.S.C. Lins; A.A. Castro; T.F. Soares; E.B.S. Meirelles; C.T.S. Castro; A.J. Godoy
- G.P.15.12 Neuromuscular disease recognition rising awareness in Latvia
Z. Krumina; I. Grinfelde; N. Pronina; R. Lugovska; B. Lace

Poster Area 6B

Posters 25 - Pharmacological treatment of Duchenne muscular dystrophy

Chairpersons: *B. Griggs, E. Mercuri*

- T.P.5.01 Phase 2b Study of PTC124 in Duchenne/Becker muscular dystrophy (DMD/BMD): Demographic and other baseline data
L.A. Atkinson; A.L. Reha; G.L. Elfring; R. Finkel; B. Wong; K. Flanigan; C.M. McDonald; K. Bushby; T. Voit; G. Spinella; V. Cwik; L. Miller

Full Programme

- T.P.5.02 Pentoxifylline treatment fails to rescue muscle strength and function deterioration in prednisone-treated Duchenne muscular dystrophy (DMD)
D.M. Escolar; K. Gorni; A.C. Tesi-Rocha; J. Mah; Y. Nevo; A. Korenberg; H. Kolski; T. Bertorini; A. Connolly; N. Kuntz; A. Zimmerman; L. Morgenroth; A. Arrieta; J. Mayhew; J. Florence; L. Nei; F. Hu; E. Henricson; R. Leshner; A. Dubrovsky
- T.P.5.03 Equipose concerning corticosteroid use in boys with Duchenne muscular dystrophy: persistent wide variations in practice
R.C. Griggs; B.E. Herr; M. Eagle; E. McColl; G. Bell; R. Tawil; S. Pandya; M. McDermott; K. Bushby; TREAT-NMD Investigators; MSG Investigators
- T.P.5.04 Randomized, double-blind, controlled study to compare efficacy and tolerability of standard daily prednisone regime with a novel intermittent high dose regime in ambulant boys with Duchenne muscular dystrophy
D. Escolar; C. McDonald; A. Korenberg; T. Bertorini; T. Lotze; M. Ryan; P. Clemens; R. Leshner; A. Pestronk; L. Morgenroth; A. Arrieta; E. Henricson; J. Mayhew; J. Florence; T. Duong; L. Nei; F. Hu; A.C. Tesi-Rocha; A. Connolly
- T.P.5.05 Benefits of initiating prednisone treatment in non-ambulatory patients with Duchenne Muscular Dystrophy(DMD)
S. Pandya; D. Fox; K. Fox; C. Westfield; Y. Su; K. Campbell; D. Guntrum; E. Ciafaloni; R.T. Moxley
- T.P.5.06 Two excited Duchenne muscular dystrophy brothers: The late use of steroids
A. Brassanini; C.A. Melo e Souza; A.J. Godoy; M.B. Magario
- T.P.5.07 Long bone fractures and bone healing in boys with Duchenne muscular dystrophy treated with daily, long-term Deflazacort
D. Biggar; V.A. Harris; B. Alman; J. Vajsar
- T.P.5.08 Vertebral fractures in corticosteroid-treated boys with Duchenne muscular dystrophy
G.P. Ambegaonkar; M. Kinali; F. Muntoni; S.A. Robb; A.Y. Manzur
- T.P.5.09 Carvedilol can reduce cardiac events in Duchenne muscular dystrophy
T. Matsumura; T. Tamura; S. Kuru; Y. Kikuchi; M. Kawai
- T.P.5.10 Growth hormone may improve growth, neuromuscular and pulmonary function in Duchenne muscular dystrophy
M.M. Rutter; S.R. Rose; M. Kalra; B.L. Wong
- T.P.5.11 Idiopathic Juvenile Osteoporosis: An important cause of gait deterioration in childhood
B.G. McCullagh; P. Tomlin; M.Z. Mughal; C. De Goede; R. O'Connor
- Poster Area 5C** **Posters 26 - Glycogen storage disorders**
Chairpersons: C. Navarro, J. Howell
- G.P.16.01 The Pompe registry: Tracking Pompe disease symptoms in a broad patient population
B. Byrne; P.S. Kishnani; L. Case; L. Merlini; W. Müller-Felber; A. Van der Ploeg
- G.P.16.02 Results of a survey examining neuromuscular specialists' and neurologists' ability to suspect Pompe disease
D. Marsden; W. Harris
- G.P.16.03 A clinical and genetic study of 2 patients with myopathy and phosphorylase kinase deficiency (glycogenosis type VIII)
A. Echaniz-Laguna; H.O. Akman; M. Mohr; C. Tranchant; I. Maire; S. di Mauro

Full Programme

- G.P.16.04 Branching enzyme deficiency should be considered in the differential diagnosis of severe congenital hypotonia
A.L. Taratuto; H.O. Akman; M. Saccoliti; M. Riudavets; N. Arakaki; M. Fernández; L. Mesa; G. Sevelever; H. Goebel; S. Di Mauro
- G.P.16.05 McArdle disease. Molecular study of the myophosphorylase gene (PYGM) in a series of 120 patients
I. Viéitez; S. Teijeira; B. San Millán; J.M. Fernández; S. Miranda; B. Eymard; P. Laforêt; T. Stojkovic; A. Nadaj; C. Navarro
- G.P.16.06 Investigation of possible treatment regimes for McArdle's disease using the sheep model of the disease
J.M. Howell; R. Quinlivan; C. Sewry
- G.P.16.07 Biochemical and molecular genetic identification of patients with phosphoglycerate mutase deficiency
P.R. Joshi; M. Knape; S. Zierz; M. Deschauer
- G.P.16.08 Two novel mutations associated with Muscle Phosphoglycerate Mutase (PGAM) deficiency
O. Musumeci; A. Toscano; A. Naini; J. Vissing; N. Lanzano; S. DiMauro
- G.P.16.09 Phosphoglucomutase deficiency: A rare glycogen storage disease with an adult onset
T. Stojkovic; M.O. Rolland; M. Hezode; C. Wary; P. Carlier; F. Petit; M. Piraud; J. Wissing; B. Eymard; P. Laforet
- G.P.16.10 Danon disease: A novel donor site LAMP2 mutation causing alternative transcripts
C. Di Blasi; L. Jarre; M. Mora

Thursday 2 October 2008

- 09:00-10:30** **Sage Hall 1** **Therapeutic advances in neuromuscular disorders;
Invited lectures (T.I. 1-3)
Chairpersons: S. Braun, N. Goemans**
- T.I.1 Translational research in neuromuscular diseases - where do we stand?
V. Straub
- T.I.2 Preclinical drug trial efforts for muscular dystrophy: Methods and end points
K. Nagaraju
- T.I.3 The rationale for immunosuppressive treatment in DMD
R. Korinthenberg
- 10:30-11:00** Sage Morning Tea & Coffee
- 11:00-12:00** **Sage Hall 1** **Therapeutic advances in neuromuscular disorders;
Invited lectures (T.I. 4-5)
Chairpersons: F. Muntoni, M. Zatz**
- T.I.4 Splice manipulation therapies: Opportunities and challenges
S.D. Wilton; S. Fletcher
- T.I.5 Re-establishment of dystrophin expression by exon skipping and/or trans-splicing approaches
L. Garcia
- 12.00.13.00** **Sage Hall 1** **WMS General Assembly - Hall 1, The SageGateshead**

Full Programme

14.00-15.30	Sage Hall 1	Testing therapies in neuromuscular disease; Oral Presentations (T.O.1-5) Chairpersons: I. Nishino, D. Selcen
T.O.1		Cyclosporin A as a potential treatment for collagen VI-related muscular dystrophy: A cellular study of mitochondrial dysfunction and its rescue <i>D. Hicks; A.K. Lampe; S.H. Laval; V. Allamand; C. Jimenez-Mallebrera; M. Walter; F. Muntoni; S. Quijano-Roy; P. Richard; V. Straub; H. Lochmuller; K.M.D. Bushby</i>
T.O.2		Omigapil/SNT-317 prevents apoptosis and ameliorates the pathology of laminin-alpha2 deficient muscle dystrophy <i>T. Meier; M. Erb; S. Meinen; P. Barzaghi; M. Rüegg</i>
T.O.3		SNT-MC17/idebenone in Duchenne muscular dystrophy: long-term blinded controlled preclinical study in the mdx mouse followed by a 12 month double-blind randomized controlled trial in humans <i>G.M. Buyse; N. Goemans; G. Van der Mieren; M. Erb; J. D'hooge; P. Herijgers; E. Verbeken; A. Jara; A. Van Den Bergh; I. Courdier-Fruh; P. Barzaghi; M. van den Hauwe; D. Thijs; I.J.M. de Groot; U. Schara; B. Ceulemans; L. Mertens</i>
T.O.4		Safety and efficacy results from a randomized, double-blind, placebo-controlled study of alglucosidase alfa for the treatment of Pompe disease in juveniles and adults <i>P. Laforet; P.R. Clemens; D. Corzo; D. Escolar; J. Florence; A. van der Ploeg; S. Lake; J. Mayhew; A. Pestronk; B. Rosenbloom; A. Skrinar; M. Wasserstein</i>
T.O.5		Oral dexamethasone pulse therapy versus daily prednisolone in subacute inflammatory myopathies: A randomised clinical trial <i>M. De Visser; J. Van de Vlekkert; J.E. Hoogendijk; R.J. De Haan; A. Algra; I. Van der Tweel; W.L. Van der Pol; E.V. Uijtendaal</i>
15.30-16.00	Sage	Afternoon tea & coffee
16.00-18.00	Sage Hall 1	Late Breaking News
LBN01		LGMD2L is caused by mutations in the skeletal muscle highly expressed transmembrane TMEM16E gene <i>V. Bolduc; I. Thiffault; M. Tétreault; M.J. Dicaire; Y. Robitaille; J. Jarry; M.F. Rioux; L. Loisel; J. Mathieu; J.P. Bouchard; B. Brais</i>
LBN02		Mutation in BAG3 Defines Severe Novel Muscular Dystrophy of Childhood <i>Duygu Selcen; Francesco Muntoni; Barbara K. Burton; Elena Pegoraro; Caroline Sewry; Anna V. Bite; Andrew G. Engel</i>
LBN03		Mutations in DNM2 can present as a congenital muscular dystrophy <i>R. Susman; N. Yang; M. Shingde; S. Arbuckle; K. North</i>
LBN04		Massive downregulation of the V-ATPase by hypomorphic alleles of the VMA21 gene causes an autophagic myopathy <i>Iulia Munteanu; Nivetha Ramachandran; Peixiang Wang; Pauline Aubourg; Jennifer Rilstone; Nyrie Israelian; Taline Naranian; Paul Paroutis; Ray Guo; Zhi-Ping Ren; Ichizo Nishino; Brigitte Chabro; Jean-Francois Pellissier; Carlo Minetti; Bjarne Udd; Michel Fardeau; John T. Kissel; Hannu Kalimo; Nicolas Levy; Morris F. Manolson; Cameron A. Ackerley; Berge A. Minassian</i>
LBN05		A new X-linked form of Emery-Dreifuss Muscular Dystrophy is caused by FHL1 gene mutations that lead to abnormal muscle differentiation <i>L. Gueneau; A. Bertrand; J.P. Jais; M.A. Salih; T. Stojkovic; M. Wehnert; S. Saitoh; A. Vershuren; M. Beuvin; E. Lacene; S. Heath; D. Zelenika; T. Voit; B. Eymard; R. Ben Yaou; G. Bonne</i>

Full Programme

LBN06 Direct and simultaneous visualization of D4Z4 arrays on distinct 4qA, 4qB and 10q stretched alleles: implication for FSHD diagnosis and physiopathology
Karin Nguyen; Pierre Walrafen; Catherine Vovan; Rafaëlle Bernard; Aaron Bensimon; Nicolas Lévy

LBN07 New congenital myopathy associated with a mutation in the cardiac myosin-binding protein C gene MYBPC3
Homa Tajsharghi; Trond P. Leren; Leif Brunvand; Hilde M. Dahl; Mar Tulinius; Anders Oldfors

18.00

Prize Giving

Welcome to WMS 14: Geneva, 9-12 September 2009

Handing over the WMS Flag

Close of the 2008 Congress