

*Final Announcement*

**4th JOINT MEETING  
BELGIAN-DUTCH  
NEUROMUSCULAR  
STUDY GROUP AND  
GERMAN REFERENCE  
CENTER FOR  
NEUROMUSCULAR  
DISEASES, DGNN**



**MAY 25 AND 26, 2018**

**HOTEL KASTEEL BLOEMENDAL  
VAALS, THE NETHERLANDS**

DEDICATED TO PROF. J.M. SCHRÖDER ON THE OCCASION OF HIS 80<sup>TH</sup> BIRTHDAY

## INVITED SPEAKERS:

Enrico Bertini, Rome, Italy | Gisèle Bonne, Paris, France | Sebahattin Cirak, Cologne, Germany | Bouke Hazenberg, Groningen, The Netherlands | Christian Hübner, Jena, Germany | Mario Losen, Maastricht, The Netherlands | Coen Ottenheijm, Amsterdam, The Netherlands | Claudia Sommer, Würzburg, Germany | Vincent Timmerman, Antwerp, Belgium | Peter Van den Bergh, Brussels, Belgium | Marianne de Visser, Amsterdam, The Netherlands

## ORGANIZERS:

Joachim Weis, Aachen, Germany | Peter Van den Bergh, Brussels, Belgium | Kristl G. Claeys, Leuven, Belgium | Karin Faber, Maastricht, The Netherlands | Nathalie Goemans, Leuven, Belgium | Nicolette Notermans, Utrecht, The Netherlands | Werner Stenzel, Berlin, Germany |  
in collaboration with the European Graduate School of Neuroscience (EURON)  
Founding Director: Harry Steinbusch, Maastricht, The Netherlands /  
Director: Gunter Kenis, Maastricht, The Netherlands

## REGISTRATION:

Deadline for registration: April 30<sup>th</sup>, 2018

Online registration via the website:

[http://www.euronschool.eu/page/145/Online\\_registration/](http://www.euronschool.eu/page/145/Online_registration/)

Registration fee: € 150 (including program, lunches, dinner and abstract book). Fee has to be paid via online payment: a link will be send to you via the organization after registration. *Reference: 30974375N/EURON*

## HOTEL ROOMS:

A limited number of rooms have been reserved for participants.

Please book your room directly with the hotel Kasteel Bloemendal

Tel: +31-(0)43-3659800 | [info@bloemendal.valk.nl](mailto:info@bloemendal.valk.nl)

[www.hotelbloemendal.nl/nl](http://www.hotelbloemendal.nl/nl). **Reference: BLO-GF14095**

## OTHER RECOMMENDED HOTELS:

Hotel Ibis Aachen am Marschiertor (centre of Aachen, close to main train station; bus line to Vaals)

Hotel Mercure am Dom (centre of Aachen; bus line to Vaals)

Aquis Grana City Hotel (centre of Aachen; bus line to Vaals)

## CONTACT INFORMATION:

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## PROGRAM

Friday, May 25, 2018

09.30 – 10.30 Registration (coffee/tea)

Welcome

*P. Van den Bergh, J. Weis, K. Claeys*

### Session 1 Motor neuron diseases and Myasthenia

*Chairpersons: N. Goemans, M. De Baets*

10.45 – 11.15 New developments in SMA

*E. Bertini, Rome, Italy*

11.15 – 11.45 Experimental therapies for myasthenia gravis: treatment strategies in the pipeline towards clinical application

*M. Losen, Maastricht, NL*

11.45 – 12:00 Ocular Vestibular Evoked Myogenic Potentials in Myasthenia Gravis

*R.H. de Meel, K.R. Keene, M.R. Tannemaat, J.J. Verschuuren*

12:00 – 12:15 Alpha-spectrin haploinsufficiency due to SPTAN1 nonsense mutations causes a spectrum of juvenile onset dominant hereditary motor neuropathies

*D. Beijer, T. Deconinck, J. De Bleecker, A. Malandrini, M. Zulaica Ijurco, J. Andoni Urtizberea, P. De Jonghe, J. Baets*

12:15 – 12:30 Whole exome sequencing of distal spinal muscular atrophy cases identifies new genotype-phenotype associations

*F. Baas, M. Weterman, F. van Ruisen, A. van der Kooi, J. Hoogendijk, K. van Spaendonck-Zwarts, M. de Visser*

12:30 – 12:45 Matrin 3 pathophysiology in ALS and related disorders

*P. Volkmer, E. Shimbo, H. Guo, A. Yamoah, P. Tripathi, T. Goodarzi, E. Vliet, D. Troost, J. Anink, E. Aronica, P. Arun, A. Hermann, J. Weis, A. Goswami*

12.45 – 13:00 RNA: DNA hybrid (R-loop) mediated double strand breaks (dsbs) and neurodegeneration in ALS-FTD

*A. Yamoah, T. Goodarzi, H. Guo, P. Tripathi, E. Vliet, D. Troost, J. Anink, E. Aronica, A. Hermann, J. Weis, A. Goswami*

13.00 – 14.00 Lunch

Friday afternoon, May 25, 2018

### Session 2 Peripheral neuropathies I

*Chairpersons: C. Sommer, J. De Bleecker*

14.00 – 14.15 J. Michael Schröder: Neuromuscular pathologist

*J. Weis*

14.15 – 14.45 Identification of common molecular determinants of axonal Charcot-Marie-Tooth disease

*V. Timmerman, Antwerp, Belgium*

14.45 – 15.15 Autoimmune neuropathies: novel antigens and future therapies

*P. Van den Bergh, Brussels, Belgium*

15.15 – 15.30 Clinical spectrum of the anti-GQ1b antibody syndrome: a case series of 10 patients

*A. de Bruyn, K.G. Claeys*

- 15.30 – 15.45 Mutant Atlastin-1: Effects on the endoplasmic reticulum/  
nuclear envelope and altered proteostasis  
*I. Katona, S. Tey, H.S. Daimagüler, J.M. Schröder,  
J. Klingelhöfer, A. Goswami, J. Weis*
- 15.45 – 16.00 The diagnostic value of unspecific sensorimotor and  
autonomic symptoms in the follow-up of Val30Met TTR  
mutation carriers  
*M.F. Dohrn, A.P. Sousa, K. Valdez, C. Alves, A. Martins da  
Silva, T. Coelho*

**16.00 – 16.30 Coffee break**

**Session 3      Peripheral neuropathies II**  
*Chairpersons: K. Faber, U. Schara*

- 16.30 – 17.00 Hereditary sensory neuropathies  
*C. Hübner, Jena, Germany*
- 17.00 – 17.30 Small fiber neuropathy and dermal nerve fiber involvement in  
Parkinson´s disease: an update  
*C. Sommer, Würzburg, Germany*
- 17.30 – 18.00 Hereditary Transthyretin-Mediated Amyloidosis (hATTR) with  
polyneuropathy: recent advances in diagnosis and treatment  
*B. Hazenberg, Groningen, NL*
- 18.00 – 18.15 Digit wrinkle scan©: from normative values to its clinical  
applicability in small fiber neuropathy  
*I. Joosten, M. Sopacua, G. De Natris, D.S.H. Bovenkerk,  
R.M.M. Potten, C.G. Faber, I.S.J. Merkies, J.G.J. Hoeijmakers*
- 18.15 – 18.30 Improving assessment of anxiety and depression in small  
fiber neuropathy: the Rasch-transformed Hospital Anxiety  
and Depression Scale (HADS)  
*B. de Greef, B. A. Brouwer, M.H.J. Pruppers, C. Baas,  
J.G.J. Hoeijmakers, C.G. Faber, I.S.J. Merkies*
- 18.30 – 18.45 The applicability of corneal confocal microscopy in small  
fiber neuropathy  
*M. Sopacua, J.G.J. Hoeijmakers, B.T.A. de Greef,  
M.M. Dickman, R.M.M.A Nuijts, I.S.J. Merkies, C.G. Faber*
- 19:30              Dinner**

**Saturday morning, May 26, 2018**

**Session 4      Muscle disorders I**  
*Chairpersons: K. Claeys, W. Stenzel*

- 08.30 – 09.00 Novel classification of LGMD  
*M. de Visser, Amsterdam, NL*
- 09.00 – 09.30 Striated muscle Laminopathies from gene defects to  
pathophysiology mechanisms  
*G. Bonne, Paris, France*
- 09.30 – 09.45 BVES loss-of-function mutations in limb-girdle muscular  
dystrophy 2X with cardiac conduction disorders  
*W. De Ridder, I. Nelson, B. Asselbergh, B. De Paepe,  
M. Beuvin, R. Ben Yaou, C. Masson, A. Boland, J.-F. Deleuze,  
T. Maisonobe, B. Eymard, J. De Bleecker, S. Symoens,  
R. Schindler, T. Brand, K. Johnson, A. Töpf, V. Straub,  
P. De Jonghe, G. Bonne, J. Baets*

- 09.45 – 10.00 Novel findings from a Dutch-Belgian case series of patients with immune-mediated necrotizing myopathy indicate that seronegative patients form a distinctive subgroup  
*J. Lim, A. Rietveld, J.L. De Bleecker, U. A. Badrising, C.G.J. Saris, A.J. van der Kooij, M. de Visser*
- 10.00 – 10.15 Overlap Myositis in Felty Syndrome with mitochondrial affection  
*T. Kendzierski, I. Schneider, T. Kraya, G. Stoltenburg-Didinger, G. Keyßer, S. Zierz*
- 10.15 – 10.30 T cell exhaustion vs. T cell senescence in immune mediated necrotizing myopathy and Inclusion body myopathy  
*S. Knauß, C. Preuße, Y. Allenbach, S. Leonard-Louis, M. Touat, N. Fischer, H. Radbruch, V. Matyash, M. Endres, H.H. Goebel, O. Benveniste, W. Stenzel*
- 10.30 – 10.45 New variant of necklace fibres display peculiar lysosomal structures and mitophagy  
*J.L. Rinnenthal, C. Dittmayer, K. Irlbacher, I. Wacker, R. Schröder, H.H. Goebel, C. Butori, L. Villa, S. Sacconi, W. Stenzel*

**10.45 – 11.15 Coffee break**

**Session 5 Muscle disorders II**  
*Chairpersons: A. van der Kooij, A. Schänzer*

- 11.15 – 11.45 Sarcomere structure and function in nemaline myopathy  
*C. Ottenheijm, Amsterdam, NL*
- 11.45 – 12.15 Genotype-phenotype correlations in congenital myopathies  
*S. Cirak, Cologne, Germany*
- 12.15 – 12.30 Altered muscle spindle function in murine models of muscular dystrophy  
*S. Kröger, S. Rossmannith, C. Haupt, H. Brinkmeier, R. Bittner, L. Gerwin*
- 12.30 – 12.45 Endoplasmic reticulum stress induces Myostatin high molecular weight aggregates and impairs mature Myostatin secretion  
*R. Sachdev, K. Kappes-Horn, L. Paulsen, Y. Duernberger, C. Pleschka, P. Denner, B. Kundu, J. Reimann, I. Vorberg*
- 12.45 – 13.00 The histopathological spectrum of malignant hyperthermia and non-anaesthetic rhabdomyolysis due to RYR1 mutations  
*G.J. Knuijman, N.C. Voermans, I. Bodi, L. Eshuis, M. Lammens, L. Heytens, M. Snoeck, E.-J. Kamsteeg, A. Radunovic, C. Sewry, H. Jungbluth, B. Küsters*
- 13.00 – 13.15 Glycogen Storage Disease Type IV: A Wide Clinical Range of Neuromuscular Phenotypes  
*H. Kölbl, A. Della Marina, O. Kaiser, F. Stehling, J. Weis, A. Abicht, U. Schara*

**13.15 Closure**  
*P. Van den Bergh, J. Weis, K. Claeys*

**13.30 Lunch**

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