

ENMC Impact Report 2019

Our year in highlights

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1 Message from Dr Ellen Sterrenburg, Chair of the Executive Committee

In the history of the ENMC we have seen shifts in the types of topics that are addressed at the workshops we organise. These shifts are indicative of the phase of neuromuscular research and care we are in.

In 2019 we organised eleven workshops on different issues addressed in this impact report and we see a new shift happening. We enter an era in which new drugs for neuromuscular disorders are being tested and becoming available. And with this, we see that topics such as clinical trial readiness and newborn screening are gaining more attention. It is great that the ENMC can contribute to these urgent and complex arising matters!

Looking ahead to 2020, we can report that we have been able to structurally generate extra income with which we can realise ten workshops a year. This is a good prospect, especially given the current speed of (technological) developments and the related high demand for ENMC workshops.

In 2020 I will step down as chair and board member of the ENMC. Therefore, I would like to conclude by thanking all of those involved with the ENMC.

I am very thankful for your expertise, experience, involvement, financing, dedication and enthusiasm, so that the ENMC can achieve its goals and continue to improve the quality of life of people with neuromuscular disorders.

Keep up the good work!

Dr Ellen Sterrenburg,
Chair of the Executive Committee



*The last Executive Committee meeting of Dr Ellen Sterrenburg, which took place on November 14th 2019 at Schiphol airport Amsterdam. **From left to right:** ENMC Research Director Dr Ana Ferreiro, Dr Kate Adcock from Muscular Dystrophy UK, Dr Ingeborg Meijer from the Dutch Muscle Disorder Patient Association, Dr Raffaella Willmann from the Swiss Foundation for Research on Muscle Disease (FSRMM), Dr Alexandre Méjat from AFM-Telethon, France, Dr Anna Ambrosini from Fondazione Telethon Italy, Dr Jes Rahbek from the Danish Muskelsvindfonden, ENMC Managing Director Dr Alexandra Breukel, Chair of the Executive Committee, Dr Ellen Sterrenburg from Prinses Beatrix Spierfonds, The Netherlands, ENMC Operational Manager Mrs Annelies Zittersteijn and vice-chair of the Executive Committee Dr Arpad von Moers from the Deutsche Gesellschaft für Muskelkranke e.V. (DGM).*

2 The mission of the ENMC

More than 25 years ago, a group of scientists and clinicians, together with parents of children affected by neuromuscular conditions, launched the European Neuromuscular Centre (ENMC). They had in mind the ultimate goal to improve diagnosis, accelerate the search for effective treatments and improve the quality of life of people with neuromuscular conditions. To achieve this goal, it was and still is of utmost importance that experts in this field of (ultra) rare

disorders share their knowledge and experience and collaborate in research worldwide. The ENMC encourages and facilitates this collaborative aim through the organisation of small interactive workshops for multidisciplinary groups of researchers, clinicians and persons affected by a neuromuscular condition, a unique concept in the scientific community.

ENMC Mission Statement

The mission of ENMC is to encourage and facilitate communication and collaboration in the field of neuromuscular research with the aim of improving diagnosis and prognosis, finding effective treatments and optimising standards of care to improve the quality of life of people affected by neuromuscular disorders.



“Connecting people”

3 The ENMC workshops in 2019

This year, thirteen workshop applications were submitted to the ENMC. Of these, nine were selected for financing by the ENMC, with realisation in 2019 and 2020. The high number of workshops approved in 2019 (70%) indicates the high quality of the submitted applications given the rigorous peer review performed by the ENMC Research Committee.

3.1 Summary of ENMC workshops held in 2019

In 2019, eleven workshops were organised which all took place in the Courtyard by Marriott hotel, Hoofddorp, the Netherlands. The workshops are listed in the table below.

ENMC workshops in 2019

Workshop no./Date	Topic	Workshop leaders
Workshop no. 240 25–27 January 2019	The involvement of skeletal muscle stem cells in the pathology of muscular dystrophies.	Prof. G. Butler-Browne, Prof. J. Morgan, Prof. F. Muntoni and Prof. K. Patel
Workshop no. 241 15–17 February 2019	Towards a European unifying lab for Kennedy's disease.	Dr M. Pennuto, Prof. L. Greensmith, Dr G. Sorarù and Dr J.P.F. Pradat
Workshop no. 242 1–3 March 2019	Diagnosis and management of Juvenile Myasthenia Gravis.	Dr P. Munot, Dr E. Niks, Dr J. Palace and Dr S. Robb
Workshop no. 243 22–24 March 2019	Developing guidelines for management of reproductive options for families with maternally inherited mtDNA disease.	Prof. J. Poulton, Prof. J. Steffann, Dr J. Burgstaller and Prof. B. McFarland
Workshop no. 244 10–12 May 2019	Newborn screening in Spinal Muscular Atrophy.	Dr E. Tizzano and Prof. L. Servais
Workshop no. 246 24–26 May 2019	Protein Aggregate Myopathies (PAM).	Dr M. Olivé and Dr R. Schröder
Workshop no. 250 6–8 September 2019	Clinical trial readiness in nemaline myopathy caused by mutations in the nebulin and actin (ACTA1) genes.	Prof. L. Servais, Prof. C. Wallgren-Pettersson, Prof. C. Bönnemann and Prof. U. Schara
Workshop no. 247 20–22 September 2019	Muscle Magnetic Imaging: Implementing muscle MRI as a diagnostic tool for rare genetic myopathy cohorts.	Prof. V. Straub, Dr J. Díaz Manera, Dr G. Tasca and Dr J. Warman Chardon
Workshop no. 248 11–13 October 2019	Myotonic dystrophies: molecular approaches for clinical purposes. Framing a European molecular research network.	Dr D. Wansink, Prof. G. Gourdon, Prof. B. van Engelen and Prof. B. Schoser
Workshop no. 249 29 November–1 December 2019	The role of brain dystrophin in muscular dystrophy: Implications for clinical care and translational research.	Dr H. Kan, Prof. F. Muntoni, Ass. Prof. M. Thangarajh and Dr J. Hendriksen
Workshop no. 251 13–15 December 2019	Polyglucosan storage myopathies.	Prof. A. Oldfors, Prof. P. Laforêt, Dr E. Malfatti and Prof. J. Vissing

Note: The workshop number (no.) is given once an application is officially approved and hence dates can be reserved. It does not always precisely reflect the timing of the execution of the workshop.

240th ENMC international workshop: “The involvement of skeletal muscle stem cells in the pathology of muscular dystrophies”

The first workshop of 2019 brought together participants from Australia, France, Germany, India, Italy, the Netherlands, Spain, Switzerland, the United Kingdom and the United States. They determined some intrinsic problems with stem cell function and consequently provided evidence for stem cell involvement in the pathology of major classes of muscular dystrophy and congenital myopathies.

Background

Muscle bulk and strength declines in muscular dystrophies. The ability of stem cells to regenerate muscle in muscular dystrophies is obstructed by the fat and scar tissue. There is evidence that in some muscular dystrophies, the genetic defect may directly affect the stem cells.

Particular pathologies were discussed in detail, different models were investigated and it was explained how the effect of the environment can affect muscle regeneration. The participants viewed possible reasons why some muscles are affected and others are not.

Some outcomes

- Definitions of stem cells, progenitors and differentiated muscle cells were established.
- Forms of muscular dystrophies which may have their origin in stem cells were identified.
- Some conclusions as to why muscles in different types of muscular dystrophy have different pathological features were drawn.



Participants of the 240th ENMC workshop on “The involvement of skeletal muscle stem cells in the pathology of muscular dystrophies”.

Future platforms

Future major research platforms will be developed in a timeline of two years:

- 1 to investigate the role of stem cells in muscle biopsies of patients with different muscular dystrophies;
- 2 to evaluate the role of stem cells and muscle fibres in two animal model systems for muscular dystrophies, i.e. the zebrafish and fruit flies.

The full scientific report of this workshop can be found in *Neuromuscular Disorders*: **September 2019** Volume 29, Issue 9, Pages 704–715.



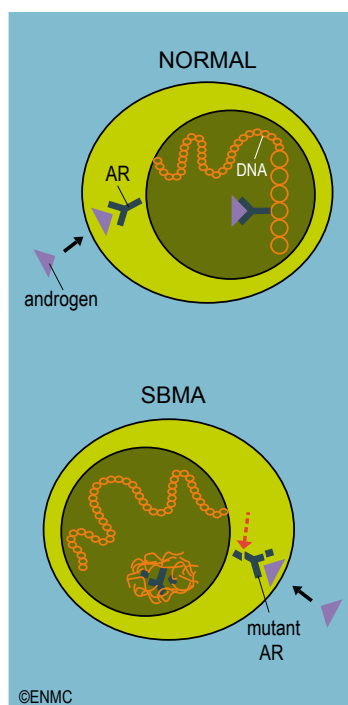
The patient's voice was well covered at this important meeting by Richard Davenport, who is affected by Limb Girdle Muscular Dystrophy, and Alison Stevenson from Muscular Dystrophy UK.

241st ENMC international workshop: "Towards a European unifying lab for Kennedy's disease"

Spinal Bulbar Muscular Atrophy (SBMA) or Kennedy's disease is a rare, adult onset, neuromuscular disease caused by a mutation in the gene encoding for the protein which binds the male hormone androgen. This protein is called androgen receptor (AR) (see illustration on the right).

Scientists and patient representatives from Denmark, France, Germany, Israel, Italy, Spain, the United Kingdom and the United States met to discuss the current understanding of basic disease mechanisms and to share and update the most recent developments in clinical evaluation of patients. The aim was to increase the prospects of developing and testing new treatments that could effectively slow down disease progression.

SBMA is now considered a neuromuscular disorder, since skeletal muscle may be a primary and early site of pathology. Scientists presented findings which indicate that skeletal muscle represents a good target for therapeutic intervention. SBMA is thus a multisystem disorder and a greater understanding of the non-neuronal tissues affected in SBMA is a priority area for research.



European Biobank

A promising approach may be the modulation of AR activity by targeting the synthesis, structure, post-translational modification and degradation of the mutant AR emerged. The participating scientists proposed a European Biobank of tissues from animal models as well as an SBMA patient biobank.

It was proposed that the 6-minute walk test and possibly grip strength could be the most effective and easily performable tools which could constitute reliable outcome measures in upcoming therapeutic trials.

European registry for SBMA

The patient representatives Marco Bertolotti and Gianni Fabris discussed the importance of collaboration between patient associations, clinicians and basic scientists for the advancement of SBMA. The role of patient associations in the dissemination of information was highlighted, for example concerning the importance and need for national registries as a means to update the SBMA patient community in international scientific efforts in SBMA research. All participants agreed on developing an international SBMA registry, gathering clinical data and tissue samples.

242nd ENMC workshop: “Diagnosis and Management of Juvenile Myasthenia Gravis”

Juvenile Myasthenia Gravis (JMG) is a neuromuscular transmission disorder, which results from binding of acquired autoantibodies to components of the neuromuscular junction, resulting in a tiring weakness that affects skeletal muscles, including the swallowing and breathing muscles. Although rare, this condition is treatable, but if JMG is not treated properly, it can be life threatening due to swallowing and respiratory muscle weakness.

An international group of 18 JMG experts from Australia, Denmark, France, Germany, Italy, the Netherlands, Switzerland, the United Kingdom and



Patients and scientists discussing further in the coffee break. From left to right: Mr Gianni Fabris, Dr Gianni Sorarù, Young Scientist Dr Paola Rusmini and Mr Marco Bertolotti.

Next steps

In order to increase scientific and clinical collaborations among groups working in different countries, it was agreed that a large International Conference on SBMA should be organised. The workshop participants highlighted the importance of researchers collaborating with patients' associations in the organisation of the meeting, in order to reinforce the communication of scientific and clinical progress to SBMA patients and families, and to provide the community with the possibility of directly collaborating in the research process.

The full scientific report of this workshop can be found in *Neuromuscular Disorders*: **September 2019** Volume 29, Issue 9, Pages 716–724.

the United States, varying from basic researchers, pediatricians to a patient representative, travelled to The Netherlands to attend this ENMC workshop.

Care of children with JMG

The recently published guidelines for adults with myasthenia gravis are an excellent resource for the treatment of adults, but the scope does not include detail on diagnosis and treatment of juvenile patients. Although most of the current practice for diagnosing and treating JMG has been extrapolated from studies and experiences in adults, there are important differences between juvenile and adult myasthenia

gravis patients, such as exclusion of unrecognised congenital myasthenic syndromes and possibly higher spontaneous remission in JMG. The workshop participants agreed to develop a consensus guideline to optimise early diagnosis and treatment.

Therapies

Various immunosuppressive drugs were also discussed and it was agreed that the best possible choice and timing of introduction needs to balance the risk of long-term neurological deficit against the risk of side effects in young children.

Outcome measures

Various available outcome measures for myasthenia gravis in adults were also discussed. There was agreement about the need for developing specific outcome measures that are clinically meaningful, validated and feasible in young children. Also a national register will be developed to collect longitudinal data on the long-term results and the effects of different drugs/interventions in JMG.

The full scientific report of this workshop can be found in *Neuromuscular Disorders*: **March 2019** Volume 30, Issue 3, Pages 254–264.



Participants of the 242nd ENMC workshop on "Diagnosis and Management of Juvenile Myasthenia Gravis".

243rd ENMC international workshop: “Developing guidelines for management of reproductive options for families with maternally inherited mtDNA disease”

Genetic counselling is uniquely difficult in the group of muscle diseases caused by shortage of energy known as “mitochondrial diseases”, in which the mitochondrial DNA (mtDNA) is inherited exclusively from the mother. In mitochondrial diseases, patients may harbour both normal and damaged mtDNA, so babies may inherit both normal and damaged mtDNA from their mothers.

A multi-disciplinary group of 29 participants from Australia, Austria, Belgium, Brazil, France, Germany, the Netherlands, Spain, the United Kingdom and the United States attended this ENMC workshop to discuss current perspectives and knowledge in reproductive options in patients with mtDNA-related mitochondrial disease.

Options

There are several different genetic options aiming to prevent the birth of a child affected by severe disease by reducing or preventing transmission of damaged mtDNA:

- to offer a healthy egg cell donated by an unaffected woman;
- pre-implantation genetic diagnosis (PGD);
- mitochondrial replacement therapy (MRT).

The participants aimed to develop guidelines for the new reproductive options that are becoming available for families with maternally inherited mtDNA disease.

They discussed the scientific, social and ethical background leading to the introduction of MRT. Also counselling protocols and clinical guidelines of who can be offered MRT were reviewed. Patient representatives were enthusiastic about MRT and felt that the potential benefits out-weigh the risks of these new options.

The following key deliverables were achieved:

- 1 consensus on referral of patients for MRT;
- 2 counselling guidelines for units carrying out MRT.

Next steps

The clinical guidelines will be reported at international meetings including EUROMIT 2020. A follow up ENMC meeting reporting on implementation of these guidelines in different countries and the short- and long term outcomes of the procedures is planned. This information will be important for countries discussing legalisation of these approaches. The need for continuing research was emphasised and participants have made plans to collaborate in future projects.

Prof. Joanna Poulton

“Even though it is very early days, the possibility of mitochondrial replacement therapy augments the reproductive options for families with maternally inherited mtDNA disease. We anticipate that the guidelines we developed will need revision once there are sufficient births to inform about the safety, efficacy and potential of MRT. Affected families are enthusiastic.”



During the workshop weekend the dinner is the moment where conversations are continued and new collaborations arise. The organisers of this workshop are Joanna Poulton (second from left), Julie Steffann (third from left) and Joerg Burgstaller (fifth from left).

The full scientific report of this workshop can be found in *Neuromuscular Disorders*: **September 2019** Volume 29, Issue 9, Pages 725–733.

244th ENMC international workshop: “Newborn screening in Spinal Muscular Atrophy”

Researchers, clinicians and patient representatives from Belgium, Finland, France, Germany, Italy, the Netherlands, Poland, Russia, Spain, Switzerland, the United Kingdom and the United States discussed newborn screening (NBS) for Spinal Muscular Atrophy (SMA). The aim of this workshop was to synthesise the currently available information on

several NBS aspects and to identify the gap in the current knowledge of patient identification and communication of diagnosis and treatment following NBS. During the weekend scientific presentations are interspersed with interactive discussions about the pros and cons of the topic. Both the presentations and the group discussions are important to show



In this SMA workshop the organisers were Prof. Laurent Servais (Belgium) and Dr Eduardo Tizzano (Spain); the Young Scientist Dr Tamara Dangouloff helped with writing the lay and full report.

unpublished data, to share centre and country experiences, to identify the gaps in knowledge and plan future collaborations.

Spinal Muscular Atrophy is a genetic disorder with autosomal recessive transmission, characterised by alpha motor neuron degeneration within the spinal cord. SMA is classified into three main types: type I (most severe form), type II and type III. SMN1 is the responsible gene in all SMA types and more than 90% of SMA cases show absence of both copies of the gene. Two treatments are currently approved for SMA; Nusinersen (Spinraza®) from Biogen (approved by FDA and EMA in 2017) and AVX-101 (Zolgensma®) from Avexis/Novartis (approved by the FDA in 2019). A third one is in clinical trial, called RG7916/RO7034067 (Risdiplam®) from a collaboration between Roche, PTC and the SMA foundation.

All data converge to show that the earlier the treatment is given, the more efficient it will be. In this view, pre-symptomatic treatment is much more

efficient than post-symptomatic treatment. Various SMA NBS programs in progress or in preparation were presented at the ENMC workshop.

What was discussed

Patients' associations put forward the idea of issuing recommendations and guidelines for countries that would like to develop NBS. Representatives of pharmaceutical companies presented their data. The medical-economic aspect of NBS and pre-symptomatic treatment was discussed, as well as an ethical vision of NBS.

Working groups have been set up to provide the necessary information and evidence to decide on the issues concerning treatment and more specifically the time required to handle according to the number of SMN copies.

The full scientific report of this workshop can be found in *Neuromuscular Disorders*: **January 2020** Volume 30, Issue 1, Pages 93–103.



Participants of the 244th ENMC workshop on “Newborn screening in Spinal Muscular Atrophy”.



Participants of the 246th ENMC workshop on “Protein Aggregate Myopathies (PAM)”.

246th ENMC international workshop: “International Workshop on Protein Aggregate Myopathies”

Protein aggregate myopathies (PAM) are a group of neuromuscular disorders marked and defined by the detrimental accumulation of proteins in striated muscle fibers based on defects in a wide variety of genes. The disease manifestations vary from childhood to late adulthood. Non-skeletal muscle disease manifestations are typically found in distinct PAM sub forms. To date, no effective treatments exist for this group of diseases.

Promoting multidisciplinary contact

Twenty participants, including one patient representative, from Australia, Austria, Belgium, Finland, France, Germany, Japan, Spain, Sweden and the United States attended this ENMC workshop. The aim of the workshop was to promote multidisciplinary contact and discussion between clinical and basic researchers working on PAM-related issues. The experts exchanged knowledge and data: they discussed recent discoveries of disease entities, the morphological spectrum of protein aggregation and new pathophysiological and pharmacological treatment aspects derived from respective animal and cell models.

Despite technical advances in genetic testing approaches and the fact that many genes and mutations have already been characterised for PAM, the discussion highlighted that many genetically unsolved cases still remain. The debate on the guidelines for diagnosis and classification demonstrated that this group of myopathies currently defies clear definition and thus classification. Ongoing preclinical evaluation of pharmacological studies in different PAM animal models shall help to pave the way to new treatment options.

Achieved goals of this workshop:

- the improved comprehension of disease mechanisms in various PAM sub forms;
- the establishment of new scientific collaborations;
- agreement to promote efforts for more treatment oriented preclinical research;
- agreement to enlarge the national German patient registry to an international scope.

The full scientific report of this workshop has been submitted to Neuromuscular Disorders.



Dr Montse Olivé

"This was the third workshop on PAM I have attended, and for the first time, Rolf Schröder and I were the organisers. It was a pleasure to meet very good colleagues and friends again with whom we have collaborated for many years, and to welcome new participants on board. As usual, discussions took place in a very friendly atmosphere."

Dr Montse Olivé from the Institute of Neuropathology at the Bellvitge University Hospital in Barcelona.

250th ENMC international workshop: "Clinical trial readiness in nemaline myopathy"

Nemaline myopathy (NM) is a rare pediatric neuromuscular disorder, characterised by genetic and clinical variability and is most commonly caused by mutations. Currently, there is no treatment.

Clinical testing of therapeutic approaches for NM can be challenging, as there are several known genetic causes and some of the underlying genetic mechanisms are still being determined. To this end, the goal of this ENMC workshop was to discuss trial design and develop a well-planned and meaningful Natural History Study (NHS) approach. The design of the NHS should be aimed towards key clinical features typical of NM, which can be reproducibly obtained across multiple sites to maximise good use of the patients' time and, importantly, encourage broad patient participation.

The limited availability of natural history data through unified, validated outcome measures that can be reliably used to make decisions in clinical trials, is one of the most significant barriers to drug discovery and development in neuromuscular disease. This workshop, with participants from Brazil, Finland, France, Germany, Italy, the Netherlands, Spain, Sweden, the United Kingdom and the United States, was dedicated to address this key gap.



Steph Colquhoun from UK, affected by nemaline myopathy herself and representative for other patients, discussing the right outcome measures for patients at the 250th ENMC workshop on "Clinical trial readiness in nemaline myopathy".

Future

The consortium is now significantly better prepared to initiate the development of natural history studies across continents, making the nemaline community 'ready' for upcoming clinical trials.

The USA and Canada will have a plan in place for a collaborative natural history study data collection in NM, utilizing the Muscular Dystrophy Association (MDA) Network. European countries and Brazil will work together to develop a similar framework for their NHS.

The full scientific report of this workshop has been submitted to Neuromuscular Disorders.

From left to right: Steph Colquhoun (UK), Gustavo Dziewczapolski (Scientific Director of CureCMD and A Foundation Building Strength, USA) and Charles Park (father of a daughter with nemaline myopathy and founder of the MAP nemaline foundation in the UK).



Steph Colquhoun

"It was very important to me as patient and person working within the health sector that the experience of the service users is valued and taken into account."



Participants of the 247th ENMC workshop on “Muscle Magnetic Imaging in NMD”.

247th ENMC international workshop: “Muscle Magnetic Imaging: Implementing muscle MRI as a diagnostic tool for rare genetic myopathy cohorts”

Twenty participants, including one patient representative, from Canada, Denmark, Finland, France, Italy, the Netherlands, Poland, Spain, the United Kingdom and the United States, attended this ENMC workshop. MRI assists with the diagnosis of genetic myopathies by identifying the pattern and extent of muscle tissue replacement by fat, signs of muscle inflammation, and wasting of single muscles or muscle groups. Muscle MRI can identify relevant muscles for biopsy that could not be easily identified clinically and subsequently can decrease the risk of a ‘negative’ or futile biopsy.

Quantitative muscle MRI can also be applied to assess the progression of muscle diseases longitudinally in a non-invasive fashion, which is also relevant to assess the efficacy and safety of treatments in clinical trials.

The aim of this ENMC workshop was to assemble the required expertise to establish standardised muscle MRI protocols, to identify key imaging findings for different disorders, to discuss an special platform to share images and finally to develop an education plan to convey these findings to the wider medical community.

Participants in the workshop reviewed the new MYO-MRI online imaging platform “MYO-SHARE” and the coordination of image sharing with the European Reference Network for neuromuscular diseases (EURO-NMD). They improved their comprehension of distinct and overlapping imaging findings in various genetic muscle diseases.

Major workshop outcomes

- Development of recommendations for ‘best practice’ standardised protocols of acquiring muscle MRI scans.
- Establishment of the initial framework to begin cohort development to utilise a central imaging repository, MYO-SHARE.
- Expanding a strategic educational plan to increase awareness and knowledge in the radiological/medical community by developing a publicly available online muscle imaging atlas.

The full scientific report of this workshop has been submitted to Neuromuscular Disorders.

248th ENMC international workshop: “Myotonic dystrophies, molecular approaches for clinical purposes. Framing a European molecular research network”

Myotonic dystrophy (DM) is a rare disorder, characterised by huge genetic and clinical variability.

Close co-working is required to ensure optimal sharing of existing materials, models, protocols, partly unpublished data, refinement of suitable and harmonised trial outcomes etc. Additional clinical cohort descriptions have been uncovered and two subtypes that are more precisely discriminated were associated with the respectively DM1 and DM2 mutations.

Goals

The participants of this ENMC workshop consisting of academic scientists, patient representatives and clinical DM experts from Belgium, France, Germany, Italy, the Netherlands, Poland, Spain, Sweden, Switzerland and the United Kingdom, identified two major aims:

- 1 The basic science of DMs: What do we already know and what do we urgently need to understand further?
- 2 Therapy development for DMs: where do we stand?

The still limited knowledge of the disease mechanism is one of the most significant barriers to drug discovery and development.

The participants shared their expertise in research on a range of molecular mechanisms and therapeutic avenues, reviewed existing animal models and their role in therapy development as well as the understanding of the DM mutation and function. They tried to identify the key unknowns and defined missing tools. Furthermore, participants explored the barriers to new therapy identification and established an action plan fostering novel drug discovery in the future.

The consensus from the workshop was the establishment of a European Consortium that would share the

diversity of expertise and material, methods, and patient data to inform therapy paradigms through a standardised and harmonised data set.

Next step

The group will apply for an EU-funded Innovative Training Network to ensure exchange and integration of early career scientists.

The full scientific report of this workshop is in press (Neuromuscular Disorders).



The two Young Scientists, selected for this workshop, were Mr Kees Okkersen (left) and Mrs Lise Ripken (right). They both are working as PhD students at the Radboud University, Nijmegen, The Netherlands.

Ahead of this ENMC workshop, two Young Scientists applied for the “ENMC Young Scientist Programme”. This programme enables early career researchers, clinicians and health care professionals to participate in an ENMC workshop related to their field of expertise, with the aim of giving them the opportunity to obtain state-of-the-art scientific information and networking opportunities, promoting the concept of ENMC and making the field attractive for early career professionals (see also www.enmc.org).

249th ENMC international workshop: “The role of brain dystrophin in muscular dystrophy: Implications for clinical care and translational research”

The topic of brain comorbidities in Duchenne (DMD) and Becker muscular dystrophies (BMD) brought together 24 experts in neuromuscular research, neuropsychology and neuropsychiatry. Also patient representatives from respectively Action Duchenne, AFM-Telethon and the Duchenne Parent Project attended the meeting. The participants represented the following countries: Denmark, France, Germany, Italy, Japan, the Netherlands, Switzerland, the United Kingdom and the United States.

Background and discussion

Cognitive, behavioral, and psychiatric symptoms may affect up to approximately 50% of individuals with DMD and recent evidence indicates a high prevalence of brain comorbidities in patients with BMD as well. This finding is related to the deficiency of multiple dystrophin variants during brain development in different brain locations. These central nervous system (CNS) symptoms have important clinical implications for the affected individuals.

Several topics were discussed during the workshop and included experimental and clinical aspects relevant to dystrophin in the brain such as:

- the genetic regulation of the various forms of dystrophin in the brain and their distribution in different brain regions;

- similarities and differences between the human and mouse models;
- the effect of restoring different forms of dystrophin after birth using different genetic therapies.

The experts discussed the frequency and severity of brain related symptoms, what brain imaging can tell about the features of dystrophin deficiency and also examined recommended standards of care and treatment options.

Representatives of patient advocacy groups articulated how common and significant the cognitive symptoms are in affected individuals.

Award

Participants will further work together and address brain dystrophin in patient registries, experimental models and clinical studies, supported by the recently Horizon 2020 awarded BIND project (**B**rain **I**nvolvement **I**n **D**ystrophinopathies).



Published with courtesy of World Duchenne Organization



Organisers of the 249th ENMC workshop on “The role of brain dystrophin in muscular dystrophy: Implications for clinical care and translational research” with from left to right: Dr Hermien Kan (the Netherlands), Dr Jos Hendriksen (the Netherlands), Prof. Francesco Muntoni (UK) and Ass. Prof. Mathula Thangarajh (USA).

The full scientific report of this workshop has been submitted to Neuromuscular Disorders.

251th ENMC international workshop: “Polyglucosan storage myopathies”

Polyglucosan storage myopathies are a group of glycogen storage diseases with accumulation of polysaccharides that are less branched than normal glycogen. These polysaccharides form inclusion bodies in the muscle fibers that are the pathological hallmarks of the diseases, which comprise a group of fewer than ten different genetically characterised entities. Although the diseases have the muscle polyglucosan storage in common, some of them also affect other tissues which in some cases causes the main symptoms.

Multidisciplinary forum

Twenty-one participants including one patient and three industry representatives from Canada, Denmark, France, Israel, Spain, Sweden, the United Kingdom and the United States, attended this workshop focusing on polyglucosan storage myopathies. The aim of this workshop was to create a multidisciplinary forum for discussion between clinical and basic researchers working on polyglucosan storage-related issues.

Among other things, they focused on composition and biochemical characteristics of polyglucosan, molecular mechanisms that lead to accumulation of polyglucosan and genotype-phenotype correlation in patients with glycogenin-1 deficiency.

The attending experts discussed recent discoveries of new and established disease entities, their genetic background and pathophysiological mechanism. This was followed by discussions concerning pharmacological or gene treatment options derived from current knowledge of disease mechanisms. The promising results from preclinical treatment studies in several animal models were presented; the models all aimed to reduce the amount of polyglucosan storage to cure the disease or prevent its progression.



Organisers of the 251st ENMC workshop on "Polyglucosan myopathies".

From left to right: Prof. Pascal Laforêt (France), Prof. John Vissing (Denmark), Prof. Anders Oldfors (Sweden) and Dr Edoardo Malfatti (France).

Major benefits of this workshop include exchange of knowledge amongst participating experts, the establishment of new scientific collaborations leading to improved diagnostic possibilities and intensified efforts to promote preclinical and clinical treatment investigations.

The full scientific report of this workshop has been submitted to Neuromuscular Disorders.

3.2 Participants at ENMC workshops in 2019

The ENMC strives for diversity in its workshop participants, to ensure that consensus can be reached at the meetings by having all relevant decision-makers around the table.

For each workshop that took place in 2019, the number of different stakeholders are shown in the table on the right page.

Each workshop had on average 38% clinicians and 41% basic researchers, showing that these two groups were equally distributed and formed the majority of the participants (79%). With ENMC workshops we primarily aim to connect basic researchers with clinicians to overcome the bridge between the lab and the clinic. When a workshop is aimed at clinical deliverables, one can see the number of clinicians increase (for example workshop nr 242 and 247). When a workshop has a more molecular or pathological nature, you see the number of basic scientists increase (for example workshop nr 240 and 248). It should be noted that here "clinicians" are defined as research professionals with a medical degree (MD) and that "basic researchers" are defined as scientists holding a PhD, but not a MD.

Both experts may be involved in translational projects, to bring the fundamental science closer to the clinic and vice versa.

On average 5% patients and 6% patient representatives attended the ENMC workshops in 2019, which shows that we reached our goal to have the patient's voice well-represented (11%) this year. By means of the ENMC Patient Participation Programme we try to ensure that at least 10% of the participants of each workshop are persons affected by a neuromuscular condition, parents or advocates of these patients and/or representatives from a disease-specific patient or funding organisation.

In addition, we always had either 1 or 2 Young Scientists attending an ENMC workshop this year, because one of our main aims is to increase the entrance of early career researchers in the established neuromuscular network via the ENMC Young Scientist Programme.

In workshops where it was relevant (n=4 workshops), we also had representatives from pharmaceutical companies.

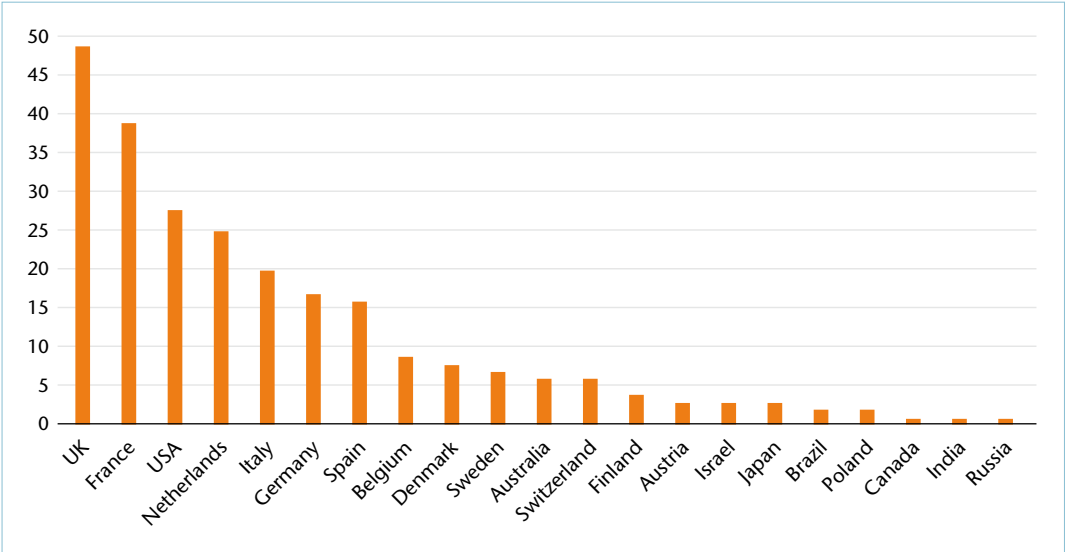
The number of workshop participants (%) and the different stakeholders attending each workshop in 2019

No	Workshop Title	Total number of participants	Clinicians	Basic re-researchers	Patients	Patient representatives	Pharma representatives	Young Scientists
240	The involvement of skeletal muscle stem cells in the pathology of muscular dystrophies	26	5 (19%)	17 (65%)	1 (4%)	1 (4%)	0 (0%)	2 (8%)
241	Towards a European unifying lab for Kennedy's disease	24	7 (29%)	10 (42%)	2 (8%)	1 (4%)	2 (8%)	2 (8%)
242	Diagnosis and management of Juvenile Myasthenia Gravis	19	15 (79%)	1 (5%)	0 (0%)	2 (11%)	0 (0%)	1 (5%)
243	Developing guidelines for management of reproductive options for maternally inherited mtDNA disease	28	6 (21%)	16 (57%)	3 (11%)	1 (4%)	0 (0%)	2 (7%)
244	Newborn screening in Spinal Muscular Atrophy	18	3 (17%)	7 (39%)	1 (6%)	3 (17%)	3 (17%)	1 (6%)
246	Protein Aggregate Myopathies (PAM)	21	8 (38%)	11 (52%)	1 (5%)	0 (0%)	0 (0%)	1 (5%)
247	Muscle Magnetic Imaging: Implementing muscle MRI as a diagnostic tool for rare genetic myopathy cohorts	19	14 (74%)	3 (16%)	1 (5%)	0 (0%)	0 (0%)	1 (5%)
248	Myotonic dystrophies: molecular approaches for clinical purposes. Framing a European molecular research network	26	3 (12%)	17 (65%)	1 (4%)	2 (8%)	1 (4%)	2 (8%)
249	The role of brain dystrophin in muscular dystrophy: Implications for clinical care and translational research	25	11 (44%)	9 (36%)	0 (0%)	3 (12%)	0 (0%)	2 (8%)
250	Clinical trial readiness in nemaline myopathy caused by mutations in the nebulin and actin (ACTA1) genes	22	12 (55%)	6 (27%)	1 (5%)	2 (9%)	0 (0%)	1 (5%)
251	Polyglucosan storage myopathies	22	7 (32%)	10 (45%)	1 (5%)	0 (0%)	2 (9%)	2 (9%)
Mean of 11 workshops		23	8,3 (38%)	9,7 (41%)	1,1 (5%)	1,4 (6%)	0,7 (3%)	1,5 (7%)
Total of 11 workshops		250	91	107	12	15	8	17

3.3 Countries represented in ENMC workshops in 2019

One of the key criteria for workshop approval by the ENMC is the geographical balance of the participants. The ENMC considers that a wide coverage of countries in the workshops is important, to make sure that consensus is reached at ENMC workshops. This will help to start international research collaborations, ensure the standardisation of healthcare provision for people affected by a neuromuscular condition, and improve the quality of diagnosis and treatment for patients worldwide. In 2019, many countries from all over the world were represented at the ENMC workshops (see diagram below).

Total number of participants per country in ENMC workshops 2019



The countries/states with the highest number of workshop participants were from Europe and USA. The academic and clinical centres which were represented most frequently in the 2019 workshops were the Dubowitz Neuromuscular Centre (UK), Newcastle University (UK), University of Oxford (UK), Radboud University Medical Center Nijmegen (Netherlands), Leiden University Medical Centre (Netherlands), National Institute of Health (USA), Institut de Myologie (France), Centre for Research in Myology (France), Hospital Universitari Vall d’Hebron (Spain), Friedrich-Baur-Institute Munich (Germany), Charité Univerzitatzmedizin (Germany), University of Milan (Italy), University of Padova (Italy) etc.

3.4 The ENMC from the perspective of patient advocate Mrs Ria Broekgaarden

For more than 25 years Mrs Ria Broekgaarden has worked at the Dutch patient organisation for neuromuscular disorders. She was chair of the ENMC Executive Committee for several years and has been very active as board member of SMA Europe, FSHD Europe and the International Pompe Association (IPA). In all these years, Ria has paved the way to provide medicines for patients with Pompe disease and SMA and contributed to the establishment of FSHD Europe and IPA. Because Ria had a very large international network, she connected the right people with one other and thereby facilitated cross-interactions like no other. In March 2020 she retired.

Patient participation in ENMC workshops

Mrs Broekgaarden regularly suggested that the ENMC should invite patients to the workshops, allowing the voice of the patient to be heard by scientists and clinicians. “You need the best lay people around the table to represent the patients”, she used to say. “A good representative should not only have proficient knowledge and overview of the specific neuromuscular disorder, but should also be able to represent the patients’ voice of the entire patient community. Experience with meetings at the international (or national) level is an added value for participating in the ENMC workshops.”

What is your view of the emerging landscape for neuromuscular disorders?

“In the coming 10 years, many new medicines will become available. Precision medicine should identify which medicine is the best for which patient, at what moment it should be given and in what combination with other treatments. With biomarkers in place, it now becomes possible to predict whether a new medicine will work for a particular patient. The ENMC can play a crucial role in bringing people together to work on the implementation of this precise medicine concept and hence the assurance that the new medicines come to the right patient.”

Patient journey

Ria states that it is important to prepare patients, their families, clinicians and hospitals for the journey and the challenges regarding prices and affordability of medicines. “Newly developed and approved medicines are not readily available to everyone, because reimbursement by the health insurance is not guaranteed.”

What role can ENMC play in this new landscape?

“To me, ENMC is the adhesive medium which binds together all researchers, clinicians, patient representatives and nowadays also the new stakeholders in the neuromuscular landscape, like pharmaceutical companies, payers and insurances. ENMC is a well-established and sustainable organisation.”



Ria Broekgaarden:

“The strenght of the ENMC is that it is an independent organisation, able to offer a safe, constructive and open platform.”

3.5 The ENMC from the perspective of a neurologist/scientist: Prof. Benedikt Schose



The 248th ENMC workshop on “Myotonic dystrophies, molecular approaches for clinical purposes: Framing a European molecular research network” was kicked off by workshop organiser Prof. Benedikt Schoser from the *Friedrich-Baur-Institute* in Munich, Germany.

Prof. Schoser welcomed the workshop participants and started by asking the spontaneous question: ***“Who of the participants has never visited an ENMC workshop before?”***

Half (50%) of the participants in this workshop raised their hands; this was their first ENMC workshop.

“Attracting new faces and brains to neuromuscular research, the golden issue of ENMC, is a key element for creating new ideas and exploring innovative research areas” explains Prof. Schoser to the question why he considers it important that new people participate in workshops. “It adheres and binds people to the topics we want to cover.”



50%

Benedikt Schoser has attended ten ENMC workshops in the last 17 years and has been organiser and chair of five of them. Besides that, Benedikt was also a member of the ENMC Research Committee for four years. The Research Committee assesses all received applications on their scientific quality, clinical relevance, sense of urgency and list of participants bi-annually. It provides recommendations to the Executive Committee about the applications and their readiness for a workshop. So Benedikt Schoser knows the ‘heart of the ENMC’.



He likes the universal perspective of the ENMC workshops given by patients, doctors and scientists and adds “that we need to include more health care persons.”

Prof. Schoser feels that it is his obligation as workshop chairman to motivate and energise the participants. “Overcoming shyness is a job for the chair – we need frank and controversial discussions”, he emphasises.

To ensure engagement by all workshop participants, Prof. Schoser likes the concept of small-sized workshops, that discussions are open and preliminary results are shared, and that everyone attending has to give a presentation and/or chair a session.

To the question what great ambitions he sees for the ENMC to achieve for the near future he answers: “Share your voice in the global neuromuscular community and at the same time keep providing a home base for European researchers and clinicians.”

4 New numbers to be proud of achieved in 2019



Patients and patient representatives

This year we welcomed 12 persons affected by a neuromuscular condition and 15 patient representatives (parents, patient associations, patient advocates) to our workshops. They all gave a presentation, asked questions and intermingled in the discussions, which was very helpful for the researchers and clinicians in the workshops to learn the patients' real needs and interests.



Researchers

We held a record high number of 11 workshops in 2019 with the attendance of 107 basic researchers and 91 clinicians, respectively 43% and 36% of the total participants. We are very proud of our Young Scientist Programme which made it possible for 17 early career researchers to attend the ENMC workshops and pave their way to enter into the neuromuscular network.



Sponsors

In 2019, the ENMC was sponsored by seven full members and two associated members. Seven patient associations/foundations co-sponsored condition-specific workshops and 11 pharmaceutical companies supported us either by a membership of the ENMC Company Forum or by workshop-specific funding. We are very thankful for the support from all these parties and acknowledge them with gratitude.

5 Creating global awareness about ENMC workshops

5.1 Publication and dissemination of workshop outcomes

The workshop results are reported and disseminated to the patient and scientific community, respectively.

Patients and families

Informing patients and their families about the achievements of ENMC workshops is a key priority of the ENMC. This is done by the workshop lay report, which is written by workshop participants and published on the ENMC website within two weeks after the workshop. Nowadays, all lay reports are translated into many different languages other than English, which increases the accessibility of these reports for people worldwide. The nine European patient member organisations and other co-sponsors help to disseminate these translated lay reports via their local patient networks and the ENMC creates awareness on social media (Twittername: _ENMC; LinkedIn: ENMC group) during the workshops and whenever lay reports are published on its website.



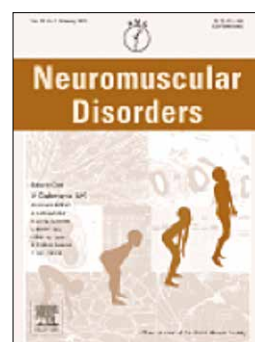
The ENMC maintains an online archive of all workshops organised since 2000, which provides access for the general public to the lay reports produced after each ENMC workshop.

See:

<https://www.enmc.org/publications/workshop-reports/>

Research community

Researchers, clinicians and healthcare providers who are active in the research field of rare neuromuscular disorders need to be able to read about the scientific results of ENMC workshops in the literature. Therefore, it is mandatory that workshop organisers submit a full workshop report to *Neuromuscular Disorders* within 6 months after the workshop.



Citation score
124%

ENMC-workshop derived publications are cited 24% more than average (100%). In bibliometric terms this means that they have a "high impact" in the research field.

5.2 International conferences in 2019

ENMC representatives attended the following local and international conferences:

- International Congress on Myology in March 2019, Bordeaux, France;
- Neuromuscular Translational Research Conference in April 2019, Newcastle upon Tyne, UK;
- Peripheral Nerve Society Annual Meeting in June 2019, Genoa, Italy;
- International Congress of the World Muscle Society in October 2019, Copenhagen, Denmark;
- TREAT-NMD Conference in December 2019, Leiden, The Netherlands.



From left to right:
Dr Davide Pareyson, Dr Anna Ambrosini,
Prof. Mary Reilly and Prof. Mike Shy on the 2019
Peripheral Nerve Society Annual Meeting which
took place in Genoa, Italy.

Anna Ambrosini is one of the ENMC Executive
Committee members and represents the Telethon
Italy Foundation, which has been a member of
the ENMC since its early days. Anna has been
ambassador for the ENMC at international
congresses many times.



Dr Alexandra Breukel, Managing Director of the
ENMC at the Myology Conference in Bordeaux,
France.



Prof. John Vissing and Dr Konni Kass from the
Rigshospitalet welcoming all attendees of the
World Muscle Society meeting in Copenhagen in
Denmark, with the breathtaking song

'You've got a friend ...'

6 Resources and financial management in 2019

Financial summary 2019

Annual accounts for the year 2019 were compiled in accordance with Guideline C1 for the reporting of small-sized non-profit organisations as published by the Dutch Accounting Standards Board. The financial accounts are drawn up in Euros.

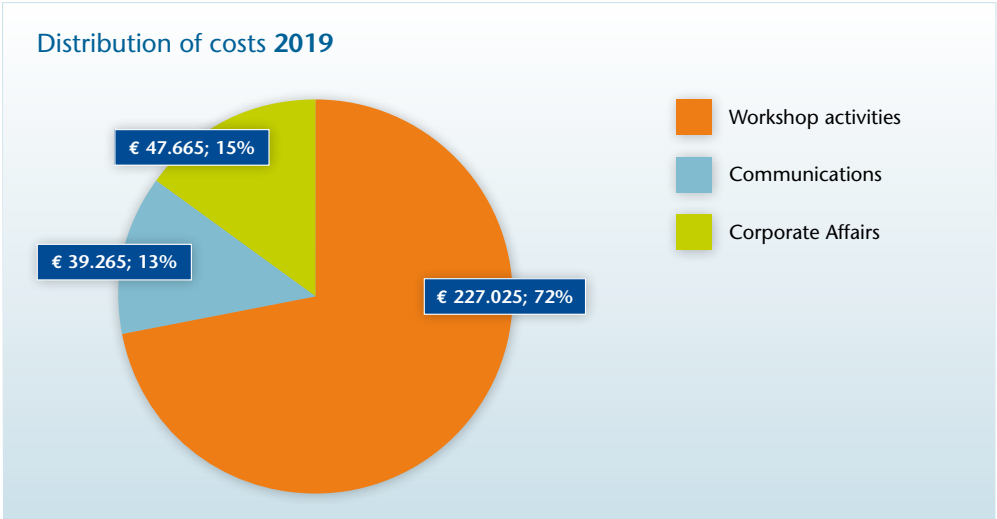
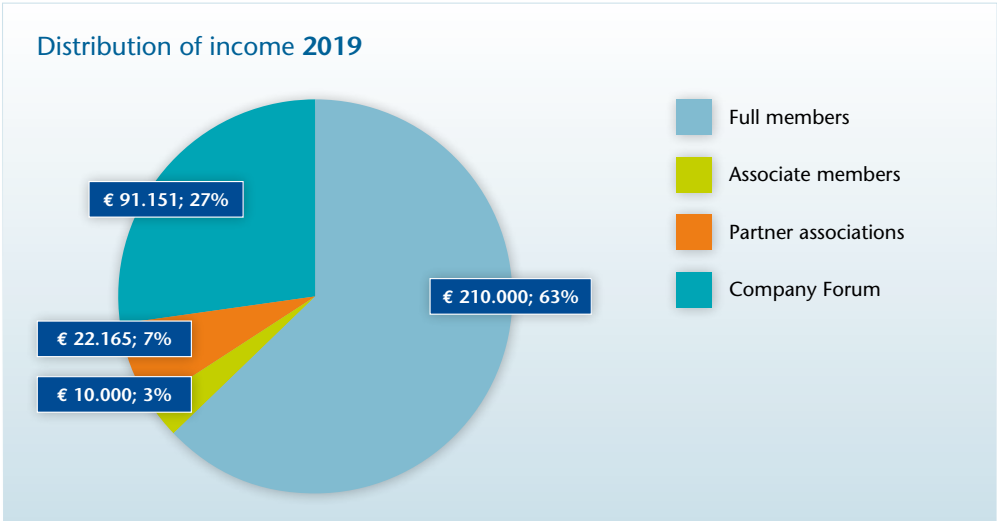
In the summary table below, the overall income and expenses over the year 2019 are shown in comparison with the figures for the financial year 2018.

Statement of income and expenses for the year 2019 in comparison with 2018 in Euros (€)		
	2019	2018
INCOME		
Member contributions	210.000	210.000
Associated member contributions	10.000	5.000
Company Forum contributions	91.151	77.425
Other contributions	22.165	3.724
Total income	333.316	296.149
EXPENSES		
Personnel expenses	144.090	149.436
Rental expenses	11.421	11.300
Activity (workshop) expenses	115.233	157.814
Organisational expenses	44.365	43.248
Total operating expenses	315.109	355.798
Operating result	18.207	– 59.649
Interest income	117	193
Net result	18.324	– 59.456
APPROPRIATION OF RESULTS		
Reserve for 25th Anniversary – withdrawal*	– 3.131	– 14.848
Reserve for 25th Anniversary – release*	– 4.012	
Reserve for additional workshop costs	– 29.845	– 48.000
Other free reserves	55.312	3.392
CASH AT BANKS ON 31 DECEMBER	477.725	507.136

*Note: Withdrawal means that these costs were made in 2019 and could be taken from this provision. Release means that the amount was left-over and could be deducted from this provision and transferred to Other free reserves.

Outside the ENMC administration, five patient organisations (Lily Foundation, Mito Foundation, Muscular Dystrophy UK, Patient Association for Mitochondrial Pathologies and Parent Project Muscular Dystrophy) supported the flights of participants from Spain, the UK and the USA directly with a total amount of € 5.700.

The distribution of income from the different ENMC supporters and of costs over the key accounts: workshop activities, corporate affairs and communications, is provided in the two diagrams below.



Opinion of the auditors

The independent accountants have verified and approved the annual accounts. For a full PDF version of the annual accounts report of 2019, please visit the ENMC website.

7 Governance in 2019

The European Neuromuscular Centre was founded as a non-profit organisation on 24 November 1992 under Dutch law. The foundation is supported by financial contributions of nine European patient organisations for neuromuscular disorders and many other related organisations. The statutory location is in Baarn, in The Netherlands, in the building of the Dutch Neuromuscular Diseases Patient Association.

7.1 The ENMC Executive Committee

The ENMC is governed by an Executive Committee consisting of representatives of ENMC member organisations.

Composition of the ENMC Executive Committee on 31 December 2019

Dr K. Adcock (United Kingdom)
Dr A. Ambrosini (Italy)
Dr I. Meijer (The Netherlands)
Dr A. Méjat (France)
Dr A. von Moers (Vice Chair, Germany)
Dr J. Rahbek (Denmark)
Dr E. Sterrenburg (Chair, The Netherlands)
Dr R. Willmann (Switzerland)

7.2 The ENMC Research Committee

The ENMC Research Committee is responsible for reviewing the scientific content and quality of the workshop applications and advises the Executive Committee on awarding the grants for ENMC workshops.

Composition of the ENMC Research Committee on 31 December 2019

Prof. Dr G.P. Comi (Italy)
Dr A. Ferreira (Chair, The Netherlands)
Prof. N. Goemans (Belgium)
Prof. Dr H. Jungbluth (United Kingdom)
Prof. Dr P. Laforêt (France)
Dr M. Olivé (Spain)
Prof. Dr M.A. Rüegg (Switzerland)
Prof. Dr T. Sejersen (Sweden)
Prof. Dr W. Stenzel (Germany)
Prof. Dr V. Timmerman (Belgium)
Dr N. Voermans (The Netherlands)

7.3 The ENMC Office

The office takes care of the daily business of the ENMC.

ENMC Office staff on 31 December 2019

Dr A. Breukel (Managing Director)
Ms A. Zittersteijn (Operational Manager)
Dr A. Ferreira (Research Director)
Ms M. Edens (Management Assistant)
Mrs C. van Santen (Workshop Assistant)

8 A special thank-you to all our members and supporters

It is thanks to the continuous support of the ten European patient organisations that the ENMC is able to facilitate and organise, on average, eight workshops per year. With support from additional partner organisations, such as condition-specific associations and members of the ENMC Company Forum, we are also able to invite participants from non-ENMC countries and facilitate the attendance of Young Scientists and patient representatives.

ENMC full and associated members



Finnish Neuromuscular Disorders Association



Deutsche Gesellschaft für Muskelkranke e.V. DGM



Members of the Company Forum



Workshop-specific sponsors



9 Looking ahead to 2020

Due to Covid-19, some of the nine approved workshops had to be postponed to later dates in 2020 and the beginning of 2021 (see table below). We acknowledge that the workshop programme may be interrupted and delayed because of Covid-19. Two review rounds for workshop applications are scheduled: one in the spring and one in the autumn. The workshops that are selected at these review rounds will take place in 2020 and 2021.

Preliminary ENMC programme 2020 (www.enmc.org)

Workshop no./date	Topic	Workshop leaders
Workshop no. 252 6–8 March 2020	Developing best practice guidelines for management of mouthpiece ventilation in neuromuscular disorders	Dr M. Toussaint, Dr M. Chatwin, Prof. J. Gonzales and Prof. M. Gonclaves
Workshop no. 257 4–6 September 2020	The 3rd ENMC workshop on Dystroglycan and the Dystroglycanopathies	Prof. S. Winder, Prof. V. Straub and Prof. K. Campbell
Workshop no. 258 16–18 October 2020	Genetic epidemiology and clinical trial readiness in Encephalomyopathy of Leigh Syndrome	Prof. E. Bertini, Prof. S. Rahman, Dr M. Schiff and Dr B. Cohen
Workshop no. 253 Postponed to: 30 October–1 November 2020	Skeletal muscle laminopathies – natural history and clinical trial readiness	Dr G. Bonne, Dr L. Maggi, Prof. S. Quijano-Roy and Prof. C. Bönnemann
Workshop no. 254 Postponed to: 23–25 October 2020	Formation of a European network to initiate a European data collection and development of treatment guidelines for adult SMA patients	Prof. P. Laforêt, Dr E. Pegoraro, Dr L. van der Pol and Prof. M. Walter
Workshop no. 259 11–13 December 2020	Anaesthetic management in neuromuscular disorders	Prof. H. Jungbluth, Dr N. Voermans and Dr M. Snoeck
Workshop no. 255 Postponed to: 15–17 January 2021	Muscle imaging in idiopathic inflammatory myopathies	Prof. M. de Visser, Prof. J. Vencovský and Prof. P. G. Carlier
Workshop no. 256 Postponed to: 5–7 February 2021	Myositis specific and associated autoantibodies	Dr Y. Allenbach, Prof. O. Benveniste, Dr J. Damoiseaux and Dr A. Mammen



Welcome to the new Chair of the Executive Committee Dr Arpad von Moers

Every two years, the chair of the ENMC Executive Committee rotates. As of 1 January 2020, Dr Ellen Sterrenburg (on the right) stepped down and her successor Dr Arpad von Moers (on the left) took over the responsibilities as ENMC chair.

Dr von Moers is pediatric neurologist. He graduated at the Freie Universität Berlin (Germany) in 1984 and completed a residency in General Pediatrics at the Berlin University Medical Centre. He worked as Senior Pediatric Neurologist at the Charité Universitätsmedizin Berlin and at the Göttingen University Medical Centre.

Dr von Moers serves as medical director of the DRK Kliniken Berlin, Westend in Germany and is head of the Department of Pediatric and Adolescent Medicine since 2005. He is co-director of the nationwide Child Abuse and Neglect helpline for medical professionals and he is head of the Berliner Transitions Program (BTP), by now a nationwide applicable transition platform.

Since 2009, Dr von Moers is a member of the ENMC Executive Committee as representative of the “Deutsche Gesellschaft für Muskelkranke (DGM)”. “It is a pleasure to be part of the stimulating and enriching work of the ENMC. The international workshops provided essential contributions to clinical and basic research and to standards of care in numerous neuromuscular diseases. Nowadays ENMC is a powerful representative of patients in the neuromuscular community, which has been demon-

strated by increased patient participation in the workshops and by the special workshop in 2018 dedicated to “The role of the neuromuscular patient in Shared Decision Making.”

Beside the disease related workshops, which are the core competence of the ENMC, Dr von Moers would be pleased to broaden the spectrum of activities to some disease-wide topics such as transition of patients from child to adulthood or pricing and availability of advanced therapy medicinal products. In his limited free time, Dr von Moers plays the tenorsax in a band called “Echte Ärzte” (Real Doctors) and travels by bike all over Europe. Besides this he is also a great fan of the Berlin Philharmonic Orchestra.

Welcome to a new member of the Executive Committee Dr Simone van den Berge

With Dr Ellen Sterrenburg leaving the ENMC as of 1 January 2020, her colleague Dr Simone van den Berge becomes the new representative of ENMC member Prinses Beatrix Spierfonds, The Netherlands.



"I am senior research coordinator at the Prinses Beatrix Spierfonds, the Dutch neuromuscular foundation. After my PhD in neuroscience, I started to work for this foundation in 2011. I am coordinating different grant rounds and a talent programme." In addition, Dr van den Berge is actively involved in patient participation and in lay communication of research subjects and results.

As a member of the ENMC Executive Committee, Dr van den Berge is thrilled to contribute to the wonderful international network of researchers and other stakeholders that has been built. Things in the neuromuscular field are changing rapidly and with multiple new therapies in the pipeline, collaboration is more important than ever.

International conferences in 2020

ENMC representatives will run a booth, are invited to speak and to participate in the scientific programme of the following local and international conferences:

- Flanders Charcot-Marie-Tooth Disease (CMT) patient day in April 2020 (cancelled), Antwerp, Belgium;
- International Congress on Neuromuscular Diseases (ICNMD) in July 2020 (changed into a shortened online meeting, programme will take place face-to-face in 2021), Valencia, Spain;
- International Congress of the World Muscle Society in September-October 2020 (changed into a shortened online meeting), Halifax, Canada.

Budget for 2020

This table presents the budget forecast for 2020 established on 31 December 2019.

Budget 2020 in Euros (€)	Actuals 2019	Budget 2020
INCOME		
Member contributions	210.000	231.000
Associated member contributions	10.000	15.000
Company Forum contributions	91.151	90.000
Other contributions	22.165	10.000
Total income	333.316	346.000
EXPENSES		
Personnel expenses	144.090	146.000
Rental expenses	11.421	11.500
Activity expenses	115.233	140.000
Organizational expenses	44.365	47.300
Total operating expenses	315.109	344.800
Interest income	117	0
NET RESULT	18.324	1.200

Colophon

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