

ENMC Impact Report 2016

# Our year in highlights

EUROPEAN  
NEURO  
MUSCULAR  
CENTRE



ENMC Impact Report 2016

# Our year in highlights

EUROPEAN  
NEURO  
MUSCULAR  
CENTRE

# Contents

<b>1</b>	<b>Message from the chair of the Executive Committee</b>	<b>5</b>
<b>2</b>	<b>The mission of the ENMC</b>	<b>6</b>
<b>3</b>	<b>The impact of ENMC activities over the last two decades</b>	<b>7</b>
3.1	Number of ENMC workshops, participants and publications	7
3.2	The ENMC network	8
3.3	The gradual globalization of the ENMC	8
<b>4</b>	<b>ENMC workshops in 2016</b>	<b>9</b>
4.1	Workshop applications in 2016	9
4.2	Summary of ENMC workshops held in 2016	9
4.3	Interview with Norina Fischer, one of the Young Scientist ENMC Award winners in 2016	18
4.4	"My experience as a participant of an ENMC workshop", interview with Mrs Sarah Foye, mother of a child with titinopathy	20
<b>5</b>	<b>Role of ENMC in disseminating information</b>	<b>22</b>
<b>6</b>	<b>Resources and financial management 2016</b>	<b>23</b>
6.1	The ENMC Company Forum	23
6.2	Financial summary 2016	23
<b>7</b>	<b>Looking forward to 2017</b>	<b>25</b>
<b>8</b>	<b>Governance 2016</b>	<b>27</b>
8.1	The ENMC Executive Committee	27
8.2	The ENMC Research Committee	27
8.3	The ENMC Office	27
<b>9</b>	<b>Special thanks to all our members and supporters</b>	<b>28</b>



# 1 Message from the Chair of the Executive Committee



*Dr Raffaella Willmann, Chair  
of the Executive Committee*

*Welcome!*

I am pleased to begin this impact report by looking back at a successful 2016 and at ten workshops, selected for both the timing of the topic and for the competence of the participants. The high number of requests to hold a scientific workshop clearly demonstrates the commitment of the community to ensure progress in the field of neuromuscular conditions. In this impact report we provide details of the important issues discussed at the workshops, which highlights the current status and research priorities, for different neuromuscular conditions: in some cases, consensus guidelines of standardised care management need updating, in other cases the nomenclature or the genetics of the disease need to be first defined. One more reason to roll up our sleeves and continue the work!

I am delighted to see how this commitment is reflected by our industry partners, who support the ENMC work without receiving any particular privileges. In 2016, our Company Forum grew to five members; their financial support enables the organisation of more workshops and covers the costs of accommodation for patients and their carers to enable them to participate in the workshops.

In the past few years we have paid particular attention to the value of involving people affected by neuromuscular conditions in every workshop, which has frequently resulted in important contributions and initiatives. For the ENMC, patient empowerment is a main priority and we believe that this strategy will augment the effectiveness of the scientific and clinical research.

I want to address my special thanks to the three pillars that together drive the ENMC mission: the Research Committee and its Research Director, who review the workshop proposals to ensure they will result in high-quality outcomes; the Executive Committee, who secures the main funding, and last but not least the office personnel, who provide support both before, during and after workshops to enable the smooth operation of the ENMC, making it a modern, efficient and reliable organisation.

*Dr Raffaella Willmann,  
Chair of the Executive Committee*

## 2 The mission of the ENMC

Almost 25 years ago, a group of scientists and clinicians, together with parents of children affected by a neuromuscular condition, started the European Neuromuscular Centre (ENMC). The ultimate goals are to improve diagnosis, accelerate the search for effective treatments and improve the quality of life of people with a rare neuromuscular condition. To achieve these goals, it was, and still is, of utmost

importance that experts in this field of orphan disorders share their knowledge and experience and collaborate worldwide. The ENMC encourages and facilitates this through the organisation of small-sized, interactive workshops for multidisciplinary groups of researchers, clinicians and people 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 optimizing standards of care to improve the quality of life of people affected by neuromuscular disorders.



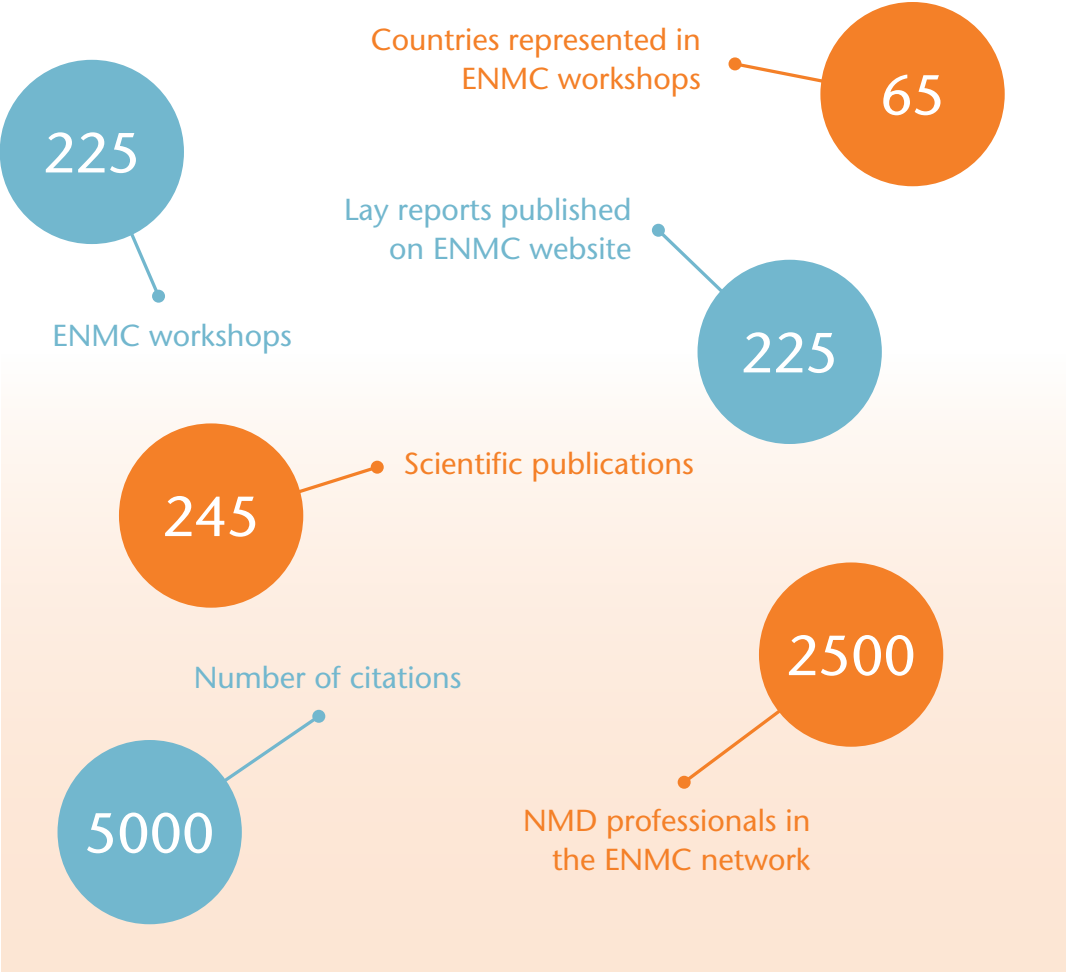
*“Connecting people”*

# 3 The impact of ENMC activities over the last two decades

## 3.1 Number of ENMC workshops, participants and publications

Since its foundation in 1992, 225 ENMC workshops have taken place. This has resulted in the publication of 225 lay reports on the ENMC website and 245 scientific publications in Neuromuscular Disorders and other related journals. We are proud to report that these publications have been cited more than 5000 times. This indicates that the outcomes of

ENMC workshops form the basis for follow-up research to improve diagnosis, treatment and care of people affected by a neuromuscular condition. In 2016, 10 workshops were held which brought together groups of 20 to 25 clinicians, researchers, patients and representatives of regulatory agencies and pharmaceutical companies.



### 3.2 The ENMC network

The ENMC has now established a network of over 2500 researchers, clinicians and health care professionals working in the field of neuromuscular research and patient care. More and more patients and their representatives join the ENMC network every year. Most members of the network have been involved in one or more ENMC workshops.

The value of promoting international collaboration at the ENMC workshops is:

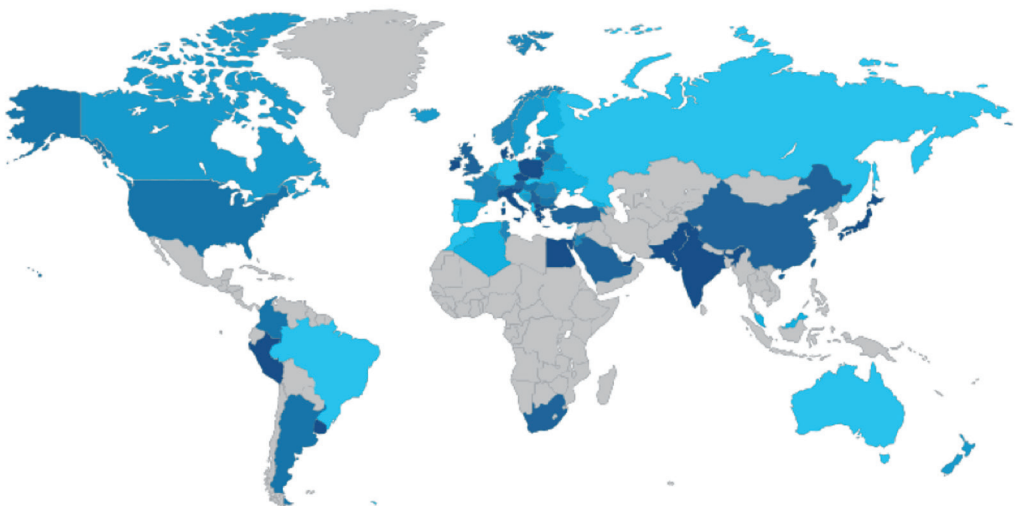
- to avoid fragmentation of research by bringing experts together and facilitating joint efforts;
- to accelerate basic research by sharing biomaterial and animal models;
- to define best practice care guidelines to improve quality of care for every individual with a neuromuscular condition in every country;
- to improve clinical trial readiness on neuromuscular disorders at an international level;
- to initiate and coordinate international clinical trials and to drive the process of bringing new drugs and treatment to the patients;
- to inform the community about the topics and outcomes of ENMC workshops through publication in scientific journals and in lay summaries.

### 3.3 The gradual globalization of the ENMC

The ENMC was originally founded as a European initiative but, due to its uniqueness, the workshops quickly raised the interest of researchers, clinicians and other health care professionals from all over the world (see map below). With large contributions from ENMC member countries and increasing attendance of experts and patient representatives from the USA and Canada over the last few years, a transatlantic network has now been firmly established.

*These 65 countries are given different shades of blue only to distinguish the country borders; the shade of blue does not indicate the number of workshop participants. Countries colored in grey were not represented at an ENMC workshop in the last 20 years.*

Over 65 countries are represented at ENMC workshops



## 4 ENMC workshops in 2016

### 4.1 Workshop applications in 2016

With the progress in neuromuscular research and the development of new drugs and therapies for neuromuscular conditions, the need to get together and collaborate increases. In 2016, a total of 11 workshop applications were submitted to the ENMC. Of these 11 applications, 8 were granted an award for an ENMC workshop to take place either in 2016 or 2017. The high number of workshops approved in 2016 is indicative of the excellent quality of the submitted applications and the need for more workshops, given the progress that is being made in the neuromuscular field.

### 4.2 Summary of ENMC workshops held in 2016

In 2016, a total of ten workshops were organised in three different venues: NH Naarden, Castle Marquette in Heemskerk and the new venue of NH Zandvoort. The workshops are listed in the table below.

### ENMC workshops in 2016

Date	Workshop No.	Workshop Title
15-17 January	216	Clinical trial readiness for FKRP related muscular dystrophies
29-31 January	217	RYR-1 related myopathies
19-21 February	218	Standards of Care in Spinal Muscular Atrophy (SMA)
29 April-1 May	219	Titinopathies, an international database of TTN mutations
27-29 May	220	The 2nd ENMC workshop on Dystroglycanopathies
10-12 June	221	Foot surgery in Charcot-Marie-Tooth disease (CMT)
1-3 July	222	Myotonic Dystrophy, a European consortium for care and therapy
16-18 September	223	AAV microdystrophin gene therapy: trial ready for DMD
14-16 October	224	Clinicopathological classification of necrotizing myopathies
18-20 November	225	A global FSHD Registry Framework

## Workshop 216 on FKRP

The first workshop in 2016 focussed on diseases with a mutation in the gene for Fukutin-related protein (FKRP), such as Limb Girdle Muscular Dystrophy type 2I (LGMD2I), Congenital Muscle Dystrophy (CMD), Muscle Eye Brain (MEB) disease and Walker Warburg Syndrome (WWS). The emergence of new therapeutic approaches like gene therapy generated the need for an expert discussion on the readiness for future clinical trials. The function of FKRP is still not biochemically proven even though it is known that FKRP at least participates in the complex process of alpha-dystroglycan glycosylation, a process which is

important for the cross-talk between the muscle cells and their environment. Later in 2016 a workshop took place which was fully dedicated to the dystroglycans and dystroglycanopathies (workshop number 220, see page 14).

Several initiatives resulted from the 216th ENMC workshop: a natural history study will be designed specially for this patient group, and a set of biochemical outcome measures will be defined, including blood and urine biomarkers, to use in future trials with potential drugs.



“To be present at an ENMC workshop as a patient makes the condition real, the patient gets a ‘face’. Spending the weekend together, having dinner together shows how disabling my condition is for patients like me”

.... Said Nynke Koelma, who joined the 216th ENMC workshop on FKRP muscular dystrophies.

## Workshop 217 on RYR-1

Ryanodine receptor 1 (RYR-1) is a calcium release channel found primarily in skeletal muscle. Since calcium plays an essential role in muscle contraction, damage to the RYR-1 protein causes muscles to weaken (RYR-1 myopathy). In humans, the ryanodine receptor 1 is encoded by the RYR-1 gene. The workshop organisers aimed to discuss the option of creating a central registry of patients with a RYR-1 myopathy and to exchange information on the correlations that exist between muscle disease

characteristics and variations in the RYR-1 gene. It was made clear that better understanding of these correlations is essential for clinicians to be able to provide families and patients with accurate information on disease diagnosis, prognosis and to help effective treatments moving forward. Also discussed were the recent advances in the ability to model the crystal-like structure of the ryanodine receptor at extremely high resolutions.



“The modelling of RYR-1 has enabled researchers to observe the working of this receptor in incredible clarity”

..... said John-Paul Cutajar, father of a son with a RYR-1 myopathy.

In turn this has significantly improved the understanding of how the different variants in the RYR-1 gene impact on the receptor function. This knowledge is essential in the quest to find new effective treatments, which can improve the function of the ryanodine receptor and alleviate disease symptoms.

This workshop was co-funded by the RYR-1 Foundation, the advocacy group for patients affected by RYR-1-related conditions.



*Dr. Michael Goldberg, MD, MPH thanked the workshop participants for their hard work and all the efforts they are pursuing to find a treatment for RYR-1 related myopathies. Michael is President and Co-Chair of Research of the RYR-1 Foundation. He is a neuroradiologist and Director of Neuroradiology at the Allegheny Health Network in the USA. He is also a patient with the condition RYR-1 myopathy and shared his experiences, concerns and expectations with the group of workshop participants.*

## Workshop 218 on SMA

The aim of the workshop organisers was to revisit the Consensus Statement of Care in spinal muscle atrophy (SMA), which was compiled eight years ago. SMA is one of the most common neuromuscular diseases and covers a spectrum of ages and impairments, ranging from infants who are unable to sit, to children and adults who can walk. It is associated with many complications related to breathing and feeding, but improvements in care have been made over the last years. The evolving understanding of the genetic basis of this disease has inspired several drug treatments that have been reviewed for approval in 2016 by the USA Food & Drug Association (FDA) and the European Medicines Agency (EMA). Eight working groups were formed to review the literature and reach consensus on the following topics:

- 1 Diagnostics and genetic counseling
- 2 Pulmonary care management
- 3 Acute care management in the hospital setting
- 4 Orthopaedics (spinal curvature, joint contractures, fractures)
- 5 Physical therapy and rehabilitation
- 6 Gastrointestinal system and nutrition
- 7 Other organ systems involved in SMA
- 8 Ethical considerations and palliative care

The outcomes of the working groups were presented and discussed at the ENMC workshop by each working group leader. Additional work was identified for each working group to be followed up after the workshop. The final recommendations of each working group were published in Neuromuscular Disorders and family-friendly versions were written and placed on advocacy group websites for easy public access.

The workshop generated enthusiastic support from the participants and achieved its initial objectives. The updated standard of care recommendations will provide a new framework for the evaluation and management of the most important aspects of SMA thereby promoting improved quality of life for these patients and reducing the burden of care for their caregivers.



*The participants of the 218th ENMC workshop on SMA*



## Workshop 219 on Titinopathies



*The participants of the 219th ENMC workshop on Titinopathies in front of Castle Marquette in Heemskerk*

The goal and purpose of this ENMC workshop was to establish an international database of Titin (TTN) gene mutations, variations and their clinical presentations, to be able to determine if particular genetic variants of the gene are the cause of disease. TTN is a giant protein, that functions as a molecular ‘spring’ responsible for the structural integrity and passive elasticity of the muscle. TTN mutations have to date been reported to be the cause of various diseases, collectively termed titinopathies, which include a range of skeletal muscle and cardiac diseases. We brought together worldwide leaders in the Titin

myopathy field, Titin family advocates, as well as representatives from other successfully developed groups. Some attendees are already collaborating together to identify disease causing genes and describe their associated clinical features.

The outcomes of the workshop included commitment of participants to work on a central TTN database, agreement on clinical questions to be included in the database and assessment of a platform for housing the database. Concrete follow up actions with responsibilities assigned to workshop participants and deadlines were agreed upon.

## Workshop 220 on Dystroglycan and Dystroglycan-related myopathies

The aim of the organisers was to bring together a multidisciplinary team of participants specialized in the field of dystroglycanopathies (DGs) and to start working on treatments for these severely disabling muscle disorders. DGs are all conditions resulting from an impaired function of a protein called dystroglycan (DG). DG along with other proteins, such as FKR (see workshop 216), maintain vital links between the inside and the outside of the cell which

serves to protect the delicate cell membrane. Perturbations in the ability of DG to interact with components, particularly on the outside of the cell, have severe implications for muscle strength.

The workshop reviewed a broad range of topics, from patient registries and clinical diagnostic approaches to the functional role of DG in muscle and brain, as well as potential new treatments for DGs. There was



*Neil Bevan and his wife Lyndsey attended the 220th ENMC workshop on dystroglycans and dystroglycanopathies, here listening to Prof Kevin Campbell who presented data on new genes and their functions related to the glycosylation process*

a presentation from a person affected by a particular type of dystroglycanopathy called LGMD2I, supported by insights from his carer and partner. The mother of a boy with LGMD2I, and a representative from Cure CMD, also gave their perspectives. These stories provided a valuable insight into the importance of finding a therapy for these patients particularly to basic scientists who have no direct contact with patients. The main achievements of the workshop

participants were: the establishment of a consortium that will work together in the future to secure funding for collaborative research projects, consensus about a new nomenclature of DGs and last but not least agreement on sharing important research reagents and tools such as antibodies, cells, therapeutic viruses, tissues and mice through existing biobanks.

### Workshop 221 on foot surgery in Charcot-Marie-Tooth disease (CMT)

One of the primary goals of this workshop was to reach agreement on some basic principles for foot surgery in CMT patients, including types of procedures, indications and outcomes as well as to identify areas for future research.

The first part of the workshop was dedicated to a review of the international contemporary practices in CMT treatment by different participants and a comprehensive review of the literature. Leading experts gave presentations on foot surgery techniques for CMT patients and discussions were held regarding all approaches. On the final day, the discussion focused on the timing of surgery, the optimum follow up protocol and the identification of the main research questions going forward.

#### The following key points were achieved:

- 1 Decision on timing and type of surgery should be taken by a multidisciplinary team, including the neurologist and the orthopaedic surgeon.
- 2 The main aim of surgery is to achieve a stable well-balanced foot.
- 3 The critical research question going forward is to standardize a follow up protocol in an international setting to allow proper comparison of surgical techniques and long term monitoring of outcomes.
- 4 A working group was formed to implement the actions identified.

## Workshop 222 on Myotonic Dystrophy - Developing a European Consortium for Care and Therapy

Myotonic dystrophy is a rare disorder characterised by enormous genetic and clinical variability. This makes clinical testing of therapeutic approaches more challenging and warrants close international collaboration between expert centres in Europe. This workshop helped to share existing, partly unpublished, natural history data, refine suitable outcome measures, identify patient populations and qualify trial sites. All of this information is essential for well-designed trials to take place in the future. Moreover, sharing existing knowledge, infrastructure and personnel will facilitate the network. It will lead to appropriate inclusion of, and communication to, patients and patient organisations, successful inter-

action with commercial as well as academic trial sponsors, and enhance the involvement of regulators and payers along the translational pathway. The “Optimistic” trial is a good example of how trials can be designed and how results can be interpreted. Findings from the “Optimistic” trial will be published during 2017 and will not only provide extensive natural history information, but also a proven framework for future trials. The community is now significantly better prepared to enter into and interpret clinical trials, and there is a high commitment to further develop along these lines (also see the website <http://optimistic-dm.eu/>).

“This workshop formed the basis of the European Myotonic Dystrophy Consortium.”



*Active discussions and exchange of information at the 222nd ENMC workshop*



## Workshop 223 on AAV Microdystrophin gene therapy for Duchenne Muscular Dystrophy (DMD)

Virus-mediated gene therapy applications to restore dystrophin expression are widely considered a promising treatment approach to directly address the underlying genetic cause of DMD. Many different viral vectors exist, each with specific properties. Adeno-associated virus (AAV) is non-pathogenic and has been well characterised in pre-clinical studies and in recent clinical gene therapy trials, for example for spinal muscular atrophy and haemophilia. There are various challenges involving AAV gene therapy, one of the main ones being that the full-sized dystrophin gene is too large to be accommodated into AAV gene

therapy vectors. For this reason, there have been a range of artificially designed microdystrophin genes, which are small enough to fit into an AAV vector, and yet retain much, if not all, of the functionality of the normal sized dystrophin. This approach was discussed in detail. The workshop participants shared and reported on the expert opinions in the field on: (i) current and planned AAV gene therapy trials, (ii) development of AAV microdystrophin vectors, (iii) production and toxicology of AAV vectors, (iv) immunogenicity of AAV vectors, and (v) DMD gene therapy clinical trial design and execution.



*The patient representatives of the 223th ENMC workshop. From left to right in the back: Dr. Jenny Versnel scientific director of MDUK and new member of the ENMC Executive Committee; Mrs. Paula Naughton from the 'Join our Boys' trust, UK; Dr. Diana Ribeiro, Director of Action Duchenne, UK; Dr. Serge Braun Scientific Director of AFM, France. In the front left, Mr. Gudjon Oskarsson, Iceland and in the front right Mr. Ward Greve, The Netherlands.*

“A lot of hard work was done by dedicated myositis experts.”

### Workshop 224 on Clinicopathological classification of immune-mediated necrotizing myopathies (IMNM)

In 2015, this consortium discussed outcome measures and clinical trial readiness in Idiopathic Inflammatory Myopathies (IIM). The timing for this 2016 workshop, focussed solely on IMNM, was well organised, given the fact that clinicians and researchers have recently discovered more about the onset of these rare subtypes of myositis. There is however an apparent lack of evidence-based guidelines for treating myositis patients. What is needed is a consensus on diagnostic criteria and preferably guidelines for therapeutic options, which can be disseminated to all specialists treating this condition.

This is very important given the fact that IMNM can exhibit a very severe course of disease. Over the two days attendants presented new data and findings from trials and research in their respective countries, which was followed by lively discussions. A lot of hard work was done by dedicated myositis experts. The group focussed on pathological and clinical criteria of IMNM, on serology (detection of auto-antibodies in blood samples), genetics (science on heredity), pathology (microscopic features observed in tissues) and also on treatment strategies.

“I was impressed how studies done by all these myositis experts in different parts of the world led to similar findings. Now it was the right time to reach agreement on subdividing the IMNM on the basis of specific auto-antibodies (anti-SRP and anti-HMGCR). Subsequently, the group agreed on clinical and pathological diagnosis criteria and proposed treatment options for each subtype. What a beautiful and promising outcome of this unique workshop!”

Ingrid de Groot, patient representative from The Netherlands.



*The participants of the 225th ENMC workshop in front of Castle Marquette in Heemskerk*

### Workshop 225 on a global FSHD registry framework

Disease registries are an essential tool for advancing research to understand a disease, improve patient care, and develop treatments. Typically, registries collect patient contact information, demographic and diagnosis data. In addition, registries may collect varying amounts of data about health and symptoms. The data will be anonymized and are accessible only to authorized individuals, to protect the privacy of patients and families who participate.

In the FSHD field, there are patient registries in at least 13 countries. This workshop aimed to create a consortium which will start collecting FSHD patient data from around the world in a single global registry or a federation of national registries that can talk to one another. In this way data from as many patients as possible will be gathered in a uniform way using

the latest standards for diagnosing and describing the condition. Traditionally, registries have not given information back to patients. Doing so takes considerable thought and work, but the consortium feels strongly that providing this information to patients and families is an obligation they owe to the patient community.

A group of registry curators worked with the FSHD Champions, an international alliance of FSHD patient advocacy organizations, to establish the key goals for this single global registry:

- 1 Accelerate research to understand and treat FSHD.
- 2 Empower patients by providing information about their condition which may help them improve their health and quality of life.

EUROPEAN  
NEURO  
MUSCULAR  
CENTRE



*"The rarity of the neuromuscular conditions makes international collaboration crucial for researchers and clinicians to share their knowledge and develop new treatments".*

[www.enmc.org](http://www.enmc.org)

### 4.3 Interview with Norina Fischer, a Young Scientist ENMC Awardee

"As I am currently working on my doctoral thesis on the topic of immune-mediated necrotizing myopathy (IMNM) in the group "Neuromuscular Diseases" of Prof. Dr. Werner Stenzel at the Charité Berlin, Germany, I was so lucky to be invited to attend this workshop as a young scientist."

"A young scientist's experience of an ENMC workshop"

Norina Fischer applied for the ENMC Young Scientists Programme (\*see page 20) and was awarded a travel grant and accommodation in the NH hotel in Zandvoort, The Netherlands.

Norina's task in the workshop was to write the lay report together with the patient representative and some of the workshop organisers. She also translated it into German. Some other workshop participants from The Netherlands, Italy, France and Sweden were asked to translate the lay report into local languages, to enable a wider distribution to patients and their families across Europe. This is one of the greatest assets of the ENMC workshops, to tell the neuromuscular community what progress is being made in genetic research and the development of medicines.

#### How to reach a consensus?

"It was immensely interesting for me to get an insight into the different scientific and clinical aspects of necrotizing myopathies and to get an overview of

the current research by experts in this field. Besides the content it was also intriguing for me to observe the procedural aspects of the consensus-finding and how the different experts and specialists contributed to the discussion and the decision-making process." The following outcomes were reached at the 224th ENMC workshop in Zandvoort, The Netherlands:

- Consensus on clinical definition of IMNM.
- Agreement on subdivision of IMNM types.
- Agreement on clinical and pathologic diagnostic criteria.
- Consensus on therapeutic guidelines.

#### A basic scientist in contact with a patient

"Besides the opportunity of meeting these experts, one of the most remarkable and memorable things of this workshop was to hear from and talk with a patient. This showed me the enormous impact this chronic disease has on one's life and it greatly reinforced my motivation to do research in this field."

"All in all I am grateful for the possibility to attend the workshop, which was an outstanding experience and very motivating for my work!"

Norina Fischer, a Young Scientist ENMC Awardee.



*\* Since the start of the Young Scientist programme in 2013, 15 people have been granted this dedicated award. This programme reserves a place in each workshop for a young scientist, clinician or health care professional working in the workshop's related field. The aim is to give young professionals the opportunity to obtain state-of-the-art scientific information and excellent networking opportunities, and to promote the concept of the ENMC amongst tomorrow's leaders.*

#### Eligibility Criteria for Young Scientists



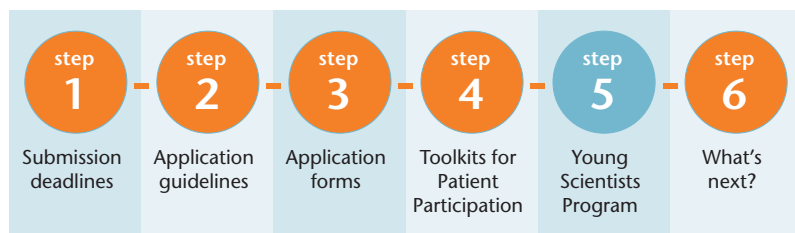
#### "Young Rising Star"

Researcher (PhD), clinician (MD), health care professional working in the field of neuromuscular disorders and younger than 35 years

#### **Are you a young rising star?**

*If you are interested, or you know the perfect candidate for this special programme, please follow the guidelines and complete the forms to apply for a Young Scientist Award (see step 5 in the application procedures at [www.enmc.org](http://www.enmc.org)).*

### Step 5: Young Scientists Program



*You can discuss what your role will be with the workshop organizers in the workshop and then work together on the Young Scientist application.*

More information about the Young Scientists Program and the application forms: see [www.enmc.org](http://www.enmc.org)

#### 4.4 Interview with Mrs Sarah Foye, mother of a child with titinopathy

"I first heard about the ENMC at a Titin dinner I organized at a strategic meeting. When I started looking into it, I was very excited about the prospect of a Titin-specific ENMC workshop. I used the ENMC website to learn about the purpose and services provided by the ENMC. I really love the ENMC 2-minute video and have shared it with family and friends."

As a patient representative Sarah had the opportunity to share with ENMC attendees her passion, fears, gratitude, and her dreams on behalf of all affected families. In life there are very few platforms that allow people to fully express themselves in an effort to

make a difference for a cause so dear to them. The ENMC workshop provided that platform for Sarah.

Sarah was one of the 18 workshop participants from many different countries with various medical and scientific backgrounds. Together with Alison Rockett Frase of the Joshua Frase Foundation, she represented the patients and their families who have to deal with titinopathy every day. They presented their experiences and emphasized the needs of patients to the clinicians and researchers in the workshop. Sarah set up her Facebook profile with the name "Team Titin" when her son was diagnosed with centronuclear myopathy, a disorder resulting from



“For me, the idea of having 20 of the world’s experts on my son’s rare muscle disease in one room for a workshop was a dream coming true!” When the application was accepted I was thrilled!”



*Sarah Foye of Team Titin on the right at the 219th ENMC workshop taking place in Castle Marquette in Heemskerk, The Netherlands. On the left is Alison Rockett Frase of the Joshua Frase Foundation. Alison is the mother of a child with titinopathy and also participated in this workshop.*

a mutation in the TTN gene. Team Titin is an online platform for families who are dealing with a titinopathy every day.



“Most people who host a party or event would agree, that it is hard for the host/hostess to enjoy themselves. Because the ENMC served as the host, it allowed the participants to be fully engaged in the process of the weekend. Having the food, housing, venue and for many their travel costs taken care of provides the support needed to have the conversation,” Sarah emphasized to ENMC staff at the meeting.

Traditionally, the lay report of an ENMC workshop is written by the scientific organizers of a workshop. Then it is published on the website to inform lay people about the outcomes of the workshop. Sarah took the initiative to write the lay report in close collaboration with Dr. Peter Hackmann and Dr. Ana Ferreiro, the two scientific organisers of this workshop. Together they compiled a report which summarized the results of the meeting both from a scientific and a lay perspective.

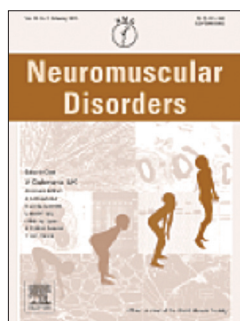
Whereas the ENMC is covering most of the workshop costs, participants coming from non-ENMC countries are not reimbursed for their travel. In this workshop, one scientist from Australia and four from the USA were able to participate with the help of the two co-sponsors, the Joshua Frase Foundation and CureCMD, acknowledged below. Sarah introduced these co-sponsors to the ENMC and helped setting up the arrangements.

“I am truly grateful to have been part of this experience. It is something that I will remember for a lifetime,”

said Sarah Foye at the 219th ENMC workshop on TTN.



## 5 Role of ENMC in disseminating information



A key role for the ENMC is to inform people within the neuromuscular community about the workshops and their outcomes. Clinicians and researchers obtain this information from the scientific publications that report content and developments from each

workshop in the journal "Neuromuscular Disorders". In 25 years, the ENMC workshop reports have been cited more than 5000 times.

In addition, the ENMC actively involves young researchers and clinicians in the experts workshops, contributing to the development of the next generation of professionals who will implement today's findings in future workshops and networks.

To meet scientists and clinicians, the ENMC team attended the World Muscle Society Congress held in Granada, Spain with a professional new booth developed this year. The ENMC had good visibility at the poster area and many young scientists and potential future workshop organisers visited the ENMC booth to discuss application procedures and share their ideas for an ENMC workshop.

21st INTERNATIONAL CONGRESS OF THE WORLD MUSCLE SOCIETY

From 4 to 8 October 2016, Granada, Spain



Another target audience of ENMC's dissemination policy is the lay public. Through publication of the lay reports on our website and messages on social media like Twitter and LinkedIn, we try to reach affected people and their families worldwide.

Follow, like and retweet us!



[www.enmc.org](http://www.enmc.org)

## 6 Resources and Financial Management in 2016

### 6.1 The ENMC Company Forum

In the last decade, the number of clinical trials for neuromuscular disorders has increased steadily. For some diseases, the first treatments are expected to become available to patients in the next few years. However, there is still much to do:

- The existing technologies for diagnosis require optimisation.
- Clinical trial readiness needs further improvement.
- International consensus of best practice care guidelines needs to be reached and implemented for all rare neuromuscular conditions.

In 2015, the ENMC started an initiative called “ENMC Company Forum”, which is a platform for pharmaceutical and biotech companies that have

neuromuscular disorders as one of their areas of interest. Members of the ENMC Company Forum support us to respond to the increasing demand for ENMC workshops and thereby facilitate collaboration of experts working in the field of neuromuscular disease worldwide. In 2016, two new members joined this forum, Sanquin and Ionis pharmaceuticals, strengthening the ENMC support.

Members of the ENMC Company Forum sponsor the general activities of the ENMC and the young scientist program. They are associated with the mission and the heritage of the ENMC. The relationship between the ENMC and companies is regulated by the established international guidelines regarding inducements and sponsorship.

If you have any questions regarding the ENMC Company Forum and/or your organisation is interested in becoming a member, please contact us at [enmc@enmc.org](mailto:enmc@enmc.org) or +31-35-5480481.

### 6.2 Financial summary 2016

Annual accounts for the year 2016 were compiled in accordance with Guideline C1 for the reporting of small sized non-profit organizations as published by the Dutch Accounting Standards Board. The financial accounts are drawn up in Euros.

In the summary table on the next page, the overall income and expenses over the year 2016 are shown in comparison with the figures for the financial year 2015. Details are given in the annual report 2016, which can be downloaded from the website.

Statement of income and expenses for the year 2016 in Euros (€)		
	2016	2015
<b>INCOME</b>		
Member contributions	210.000	210.000
Associated member contributions	5.000	5.000
Company Forum contributions	44.274	48.333
Other contributions	12.290	40.309
<b>Total income</b>	<b>271.564</b>	<b>303.642</b>
<b>EXPENSES</b>		
Personnel expenses	121.031	115.529
Rental expenses	11.126	11.346
Activity (workshop) expenses	120.361	196.272
Organisational expenses	48.617	50.778
<b>Total Expenses</b>	<b>301.135</b>	<b>373.925</b>
Operating result	-29.571	-70.283
Interest income	1.686	3.085
<b>Net result</b>	<b>-27.885</b>	<b>-67.198</b>
<b>APPROPRIATION OF RESULTS</b>		
Continuity reserve	-	200.000
Reserve for 25th Anniversary	30.000	-
Reserve for additional workshop costs	93.000	-
Other free reserves	-150.885	-267.198
	<b>-27.885</b>	<b>-67.198</b>
<b>CASH AT BANKS ON 31 DECEMBER</b>	<b>485.613</b>	<b>518.654</b>

### Opinion of the auditors

The external accountants have verified and approved the annual accounts. For a full PDF version of the annual accounts report of 2016, please visit the ENMC website.

## 7 Looking forward to 2017

Nine ENMC workshops have been scheduled for 2017 to date (see table below).

Workshops plan 2017 ([www.enmc.org](http://www.enmc.org))

Workshop no./date	Topic	Workshop leaders
Workshop no. 226 January 20-22	Towards validated and qualified biomarkers for therapy development for Duchenne Muscular Dystrophy	A. Aartsma-Rus (The Netherlands), A. Ferlini (Italy), E. McNally (USA), L. Sweeney (USA)
Workshop no. 227 February 10-11	Finalizing a plan to guarantee quality in translational research for neuromuscular diseases	R. Willmann (Switzerland), A. de Luca (Italy), M. Grounds (Australia), F. Buccella (Italy)
Workshop no. 228 March 3-5	Airway clearance techniques in Neuromuscular Disorders	M. Toussaint (Belgium), M. Chatwin (UK), J. Gonzales (France), D. Berlowitz, (Australia)
Workshop no. 229 March 17-19	Limb Girdle Muscular Dystrophies - Nomenclature and reformed classification	V. Straub (UK), B. Udd (Finland)
Workshop no. 230 February 24-26	Improving future assessment and research in IgM anti-MAG peripheral neuropathy: a consensus collaborative effort	I. Merkies (Curaçao), M. Lunn (UK), N. Notermans (The Netherlands)
Workshop no. 231 May 12-14	International Standard for CIDP Registry and biobank	F. Eftimov (The Netherlands), Y. Rajabally (UK), L. Querol (Spain)
Workshop no. 232 June 16-18	Recommendations for treatment of mitochondrial DNA maintenance disorders	R. Martí (Spain), M. Hirano (USA)
Workshop no. 233 September 15-17	Clinical trial readiness for Calpainopathies	I. Richard (France), A. Lopez de Munain (Spain), M. Krahn (France)
Workshop no. 234 December 8-10	Chaperone dysfunction in muscle disease: Therapeutic approaches	B. Udd (Finland), C. Wehl (USA), M. Hanna (UK)

A new development is that the lay reports are translated in several languages to reach more people worldwide. Furthermore, it is our goal to have at least one young scientist, but preferably two, participating in each workshop in 2017.

**Notable upcoming events:** With regard to international conferences, ENMC ambassadors will be present at the Eurordis Roundtable in Brussels, Belgium, The Dutch Neuromuscular Diseases Association congress in Eindhoven, The Netherlands and the World Muscle Society meeting in St-Malo, France.

Special announcement:  
**25 years ENMC**

**Founded on the 24th of November 1992, the ENMC celebrates its 25th anniversary in 2017.**

Proud of a successful concept and committed to its mission of facilitating scientific progress for all people with neuromuscular conditions, the ENMC plans several activities:

**1** A retrospective analysis on the impact of ENMC workshops for the neuromuscular community, both from the scientific and the patient perspective, will be performed. These results will be published in lay and scientific journals and presented at several neuromuscular meetings.

**2** A 'Call for ideas' for a special 25th anniversary workshop was launched in December 2016 to select a topic particularly focussed on the needs of the patient community. Based on these ideas, the ENMC Executive Committee and the ENMC Research Director will work out the planning of the special workshop together with the applicants and invited participants. The workshop will be held on 19 and 20 January 2018 and outcomes will be disseminated via social media and through a written workshop report.

**Title of this workshop is "The position of patients in shared decision making".**

**3** At the International Conference for Neuromuscular Disorders (ICNMD) in Vienna July 6-10, 2018, the ENMC will organize a symposium where invited speakers present the contents and results of recently held workshops. The speakers will discuss and reflect on the impact of the outcomes of their workshops and on the future outlooks for the neuromuscular community.

### **Budget implications for 2017**

Besides the usual provisions for workshops, the extra costs and resources necessary to perform the ENMC 25 year activities are also included in the budget for 2017. Both an increase in the activity expenses and personnel expenses are anticipated, which will be paid by an increase in income and out of the reserves established by the Executive Committee on 28 October, 2016.

Budget 2017	Actuals 2016	Budget 2017
<b>INCOME</b>		
Member contributions	210.000	210.000
Associated member contributions	5.000	5.000
Company Forum contributions	44.274	65.000
Other contributions	12.290	31.500
<b>Total income</b>	<b>271.564</b>	<b>311.500</b>
<b>EXPENSES</b>		
Personnel expenses	121.031	149.837
Rental expenses	11.126	11.500
Activity expenses	120.361	155.000
Organizational expenses	48.617	43.300
<b>Total operating expenses</b>	<b>301.135</b>	<b>359.637</b>
Interest income	1.686	1.600
<b>NET RESULT</b>	<b>-27.885</b>	<b>-46.537</b>
<b>Negative result will be paid out of reserves:</b>		
- Reserve for additional workshop costs		31.000
- Reserve for 25th anniversary		20.000

## 8 Governance 2016

The European Neuromuscular Centre (ENMC) was founded as a non-profit organisation under Dutch law on 24 November 1992. The foundation is supported by financial contributions of nine European patient organisations for neuromuscular disorders and other related organizations. The full members have a seat in the ENMC Executive Committee. The statutory location is in Baarn in the building of The Dutch Neuromuscular Diseases Association.

### 8.1 The ENMC Executive Committee

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

*Composition of the ENMC Executive Committee on 31 December 2016:*

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

### 8.2 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 2016:*

Prof. Dr G.P. Comi (Italy)  
Dr M. Eagle (United Kingdom)  
Prof. Dr O. Hardiman (Ireland)  
Dr D. Hilton-Jones (United Kingdom)  
Dr P. Laforêt (France)  
Prof. Dr A. Oldfors (Sweden)  
Prof. Dr G. Padberg (Research Director, the Netherlands)  
Prof. Dr M.A. Rüegg (Switzerland)  
Prof. Dr U. Schara (Germany)  
Prof. Dr B. Schoser (Germany)  
Dr W. Stenzel (Germany)

### 8.3 The ENMC Office

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

*ENMC Office staff on 31 December 2016:*

Dr A. Breukel (Managing Director)  
Mrs A. Zittersteijn (Operational Manager)  
Prof. Dr G. Padberg (Research Director)  
Mrs C. van Esch and J. Tiel-Groenestege (Workshop Assistants)

## 9 Special thanks to all our members and supporters

Thanks to the continuous support of the nine European patient organizations, the ENMC is able to facilitate and organise on average of eight workshops per year. With support from additional partner organizations, 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

FONDAZIONE



Deutsche Gesellschaft für Muskelkranke e.V. DGM



MUSKELSVINDFONDEN



fsrmm

fondation suisse de recherche sur les maladies musculaires  
fondazione svizzera per la ricerca sulle malattie muscolari  
schweiz. stiftung für die erforschung der muskelkrankheiten

### Members of the Company Forum:





Workshop-specific sponsors:



# Colophon

## Text editors

Alexandra Breukel  
Raffaella Willmann  
Ellen Sterrenburg  
Annelies Zittersteijn  
Jenny Versnel

## Lay out

Vormgevers Arnhem, The Netherlands

## Printing

Year of printing 2017, printing by DrukwerkDeal

EUROPEAN  
NEURO  
MUSCULAR  
CENTRE







European Neuromuscular Centre (ENMC)  
Lt. Gen. van Heutszlaan 6  
3743 JN Baarn  
The Netherlands

+31 35 5480481  
[enmc@enmc.org](mailto:enmc@enmc.org)  
[www.enmc.org](http://www.enmc.org)