



ENMC Impact Report 2023

Our year in highlights

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1 Message from Dr Alexandre Méjat, Chair of the Executive Committee

In 2023, the ENMC continued to strengthen its position as a leading force in advancing research and collaboration in the field of neuromuscular disorders. We hosted a total of four impactful workshops, each serving as a platform for experts and stakeholders to exchange knowledge, discuss emerging trends, and foster innovative approaches to tackling challenges within our field.

Celebrating thirty years of dedication and progress, the ENMC reached a significant milestone in 2023. Three decades of unwavering commitment to excellence, collaboration, and patient-centricity have solidified our foundation and inspired us to continue doing what we are doing best, facilitating workshops for the betterment of individuals affected by neuromuscular disorders.

Furthermore, 2023 marked the first ENMC General Assembly meeting, a pivotal moment for our organisation. This gathering provided an invaluable opportunity for members to come together, reflect on past achievements, and chart a course for the future, guided by our shared vision of fostering excellence in neuromuscular research and care.

We are delighted to announce the addition of the World Muscle Society (WMS) as our new Associate Partner. This collaboration opens up exciting avenues for enhanced cooperation and shared expertise, further amplifying our collective efforts in advancing the understanding and treatment of neuromuscular disorders worldwide.

In 2023, we said goodbye to two valued colleagues. Dr Arpad von Moers had been the German representative within the Executive Committee (EC) for 14 years. We will miss his expertise, dedication and

valuable contributions during the EC meetings.

Dr Alexandra Breukel embarked on a new chapter in her career after her dedicated service as Managing Director for almost ten years. Her leadership and commitment have been instrumental in driving the ENMC forward.

To the ENMC team in Baarn, all members of the Research Committee and Executive Committee, the Associated Partners, the workshop organisers and participants and the Company Forum, I extend a sincere thank you for your unwavering support, dedication, and contributions to the ENMC. Your collaboration and commitment are the cornerstone of our success!

Dr Alexandre Méjat,
Chair of the Executive Committee
Representative of AFM Telethon, France



2 The mission of the ENMC

More than 30 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 1st ENMC General Assembly Meeting

On April 14-15, stakeholders from across the ENMC gathered in Hoofddorp, the Netherlands, for the first General Assembly Meeting (GAM). Attended by the Executive Committee, Research Committee, Associated Partners, and members of the Company Forum, this event marked an important moment in fostering collaboration and innovation within our neuromuscular community.

The primary objectives of the GAM were to facilitate networking among ENMC stakeholders, engage them in our strategic vision, and cultivate an environment conducive to the exchange of ideas and experiences. Discussions centered on the ENMC's new long-term vision and its evolving role in the future landscape of neuromuscular disorders (NMD).

One of the highlights was the exploration of themes for the upcoming Themed Workshop Call in 2024. Stakeholders from diverse backgrounds came together to prioritise topics, providing invaluable



Ingeborg Meijer, vice chair ENMC, presented the long term strategy to all participants.

insights into their respective perspectives and interests. The positive feedback we received underscores the success of the GAM. Building on this momentum, the ENMC is pleased to announce plans for the 2nd General Assembly Meeting in 2025, further solidifying our commitment to collaboration and advancement within the field of NMD research and treatment.



All participants of the 1st ENMC General Assembly Meeting.

4 The ENMC workshops in 2023

With the progress in neuromuscular research and the development of new drugs and therapies for neuromuscular conditions, the need to get together and collaborate increased again after the Covid-19 years. In 2023 twelve workshop applications were

submitted, of which one was a re-application. Eight of these applications were selected for support by the ENMC, one already took place in 2023 and seven are planned for 2024.

4.1 Summary of ENMC workshops held in 2023

Workshop no./date and format	Topic	Workshop leaders
Workshop no. 270 10-12 March 2023	Consensus for SMN2 genetic analysis in SMA patients	Prof. E. Bertini, Prof. F.D. Tiziano, Prof. E. Tizzano
Workshop no. 272 16-18 June 2023	Inclusion Body Myositis: 10 years of progress-revision of the "ENMC 2013 diagnostic criteria for IBM" and trial readiness	Prof. C. Wehl, Prof. M. de Visser, Prof. J. Schmidt
Workshop no. 271 20-22 October 2023	Third ENMC meeting on SBMA: Towards a unifying effort to fight Kennedy's Disease	Prof. M. Pennuto, Dr G. Sorarù, Prof. L. Greensmith, Dr P.F. Pradat
Workshop no. 273 27-29 October 2023	Clinico-Sero-Morphological Classification of the Antisynthetase Syndrome (ASyS) - Associated Myositis	Prof. Y. Allenbach, Prof. O. Benveniste, Dr A. Mammen, Prof. W. Stenzel

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.



Participants of the 270th ENMC International workshop on SMN2.

270th ENMC International workshop: Consensus for SMN2 genetic analysis in SMA patients

Background

In previous years, the determination of SMN2 gene copy number was mainly informative and mostly used to elaborate genotype-phenotype correlations. However, this genomic biomarker is currently one of the main determinants for therapeutic decision, especially for patients identified through newborn screening programmes. While genetic confirmation of SMA is relatively straightforward, SMN2 gene copy number assessment requires quantitative methodologies that are not easily implemented in

most laboratories. In addition, retesting DNA samples between laboratories may show surprising discordance.

Aims of the Workshop

The main aim of this workshop was the development of a common procedure to optimise the reliability of SMN2 gene copy number determination and the establishment of SMN2 gene networks for the collaboration between laboratories and clinicians.

Workshop Deliverables

The participants agreed to determine a standard operating procedure for SMN2 gene copy number determination and a specific workflow for diagnosis and management of SMA patients identified in neonatal screening as well as to engage collaborative efforts for the creation of a European network to address discrepancies among expert centres. These endeavours will be pursued in the course of 2023.



The workshop organisers of the 270th ENMC International workshop on SMN2 in SMA patients; Prof. Francesco D. Tiziano, Prof. Enrico Bertini and Prof. Eduardo Tizzano.



Prof. Maria Pennuto welcomes the participants of the 271st ENMC International workshop on Kennedy's Disease.

271st ENMC International workshop: Third ENMC meeting on spinal and bulbar muscular atrophy (SBMA): Towards a unifying effort to fight Kennedy's Disease

Background

In the recent years, significant progress has been made in understanding SBMA's pathological mechanisms and its clinical spectrum, as well as in developing tools for clinical evaluation - all vital for conducting therapeutic trials.

Aims of the Workshop

The workshop aimed to consolidate knowledge and collaborations among experts and patient representatives, align on trial designs and outcome measures, and promote participative medicine, thereby enhancing the chances of developing effective treatments for SBMA.

Workshop Deliverables

For preclinical research, several new angles of investigation and novel approaches were presented, Novel therapeutic strategies were proposed and several tools to deliver therapeutic interventions were proposed and discussed. Further studies on this matter need to be performed to find the best strategy to be undertaken. Updates from the clinical research session revealed that progress is being made in identifying biomarkers and disease outcome measures for SBMA. Discussions regarding symptom management highlighted the need for multidisciplinary management and evidence-based guidelines for SBMA. Moreover, participants acknowledged the

importance of patient-reported outcomes alongside performance assessments. The workshop attendees agreed that international protocol sharing and standard of care implementation could accelerate trial implementation. A critical issue raised by the patient representatives and patient associations was the medical community's limited awareness of SBMA, leading to diagnostic delays and inadequate management. Patient associations were agreed to be a vital support for those with SBMA as well as strong liaisons with healthcare professionals and researchers. The workshop concluded with a commitment to reinforce ongoing collaborations, emphasizing the importance of multidisciplinary approaches.



The four patients & patient representatives during the 271st ENMC International workshop on Kennedy's Disease. From left to right: Mr Kim Slowe, Mr Ed Meyertholen, Mr Marco Bertolotti and Mr Gianni Fabris.



Participants of the 272nd ENMC International workshop on Inflammatory Body Myositis (IBM).

272nd ENMC International workshop: Inclusion Body Myositis (IBM): 10 years of progress-revision of the “ENMC 2013 diagnostic criteria for IBM” and trial readiness

Background

Treatments for IBM are emerging but there is discussion on appropriate outcome measures. Therefore, the organisers proposed that it is time to revisit the diagnostic criteria for IBM that were published in 2013, to include patients needed for clinical studies and ultimately treatment. Moreover, lessons from current natural history and previous treatment trials should be shared to develop consensus guidelines on trial design and outcome measures.

Aims of the Workshop

The workshop had several aims, the first was to re-evaluate current diagnostic criteria and integrate newly developed diagnostic tools into current practice. Other aims included exploring distinguishing pathological and immunopathological features, defining clinical outcome measures and patient reported outcomes, and understanding IBM prevalence, natural history and associated co-morbidities. Lastly, the organisers aimed to review current therapeutic targets and pre-clinical data from IBM clinical research programmes and to understand the clinical care for IBM and its implications for patients.

Workshop Deliverables

A major part of the workshop was dedicated to revising diagnostic criteria. This aimed to make them as inclusive as possible for enrolment of patients into clinical trials, and, on the other hand, to avoid misdiagnosis of patients. It was emphasised repeatedly that there is an important difference between recommendations for other clinicians on how to

diagnose IBM in general healthcare settings compared to recommendations that are required for research studies. Epidemiological studies showed great variability of the frequency of IBM in different parts of the world. Recent studies showed that it is important to recognise that there may be sex- and ethnic-related differences in clinical manifestations, particularly affecting the severity of weakness of the legs and swallowing difficulty. There was consensus on using the IBM-FRS (Functional Rating Scale, a questionnaire assessing the function of arms and legs and swallowing) as an outcome measure. The participants also agreed that, in early trials, quantitative MRI as a biomarker could be used to assess whether a drug works. Finally, a research agenda was created since many presenters concluded that further research is needed for various outcome measures and biomarkers.



Prof. Merrilee Needham, Ass. Prof. Helene Alexanderson, Ingrid de Groot (patient representative), Prof. Marianne de Visser participating in the 272nd ENMC workshop on IBM. They all are collaborating in the OMERACT Myositis working group.



Participants of the 273rd ENMC International workshop on Antisynthetase Syndrome (ASyS).

273rd ENMC International workshop: Clinico-Sero-Morphological Classification of the Antisynthetase Syndrome (ASyS) – Associated Myositis

Background

Antisynthetase syndrome was not defined as a distinct entity in the 2016 ACR/EULAR classification criteria for idiopathic inflammatory myopathies (IIM). In recent years, new scientific evidence on ASyS has emerged that is not yet reflected in current classifications of myositis.

Aims of the Workshop

The aim of the workshop was to define clinical, serological, and morphological aspects of antisynthetase syndrome.

Workshop Deliverables

Unmet needs and open questions in the context of ASyS were presented and discussed, as well as the different clinical presentations, which are variable and, in many cases, linked to specific ASyS antibodies. Specific histopathological findings in the muscle biopsies of ASyS patients were highlighted. With regards to cardiac manifestations, prevalence

and optimal treatment studies are needed on larger numbers of patients to draw definite conclusions. For lung involvement, disease activity should be monitored on a regular basis to detect progression and adjust treatment. Regarding the laboratory detection of ASyS-antibodies it was stated, that depending on the method, the results can vary and especially in rare antibodies, false positive results or negative results might be problematic. As the evidence regarding specific therapies in ASyS is limited to mostly case series and expert opinions or data from myositis basket trials, more research is warranted on this topic. The group reached consensus that ASyS patients have a high risk for relapse and therefore a long-term remission should be achieved before thinking about tapering the therapy. Defining ASyS as separate disease entity within the spectrum of idiopathic inflammatory myopathies will help to improve patient care as well as future research in the context of ASyS.

4.2 Participants at ENMC workshops in 2023

The ENMC strives for a wide range of expertise and experience in its workshop participants to ensure that a broad consensus can be reached at the meetings by having all relevant stakeholders around the table.

For each workshop that took place in 2023, the numbers of different stakeholders are shown in the table below.

No	Workshop Title	Participants	Clinicians	Basic researchers	Translational researchers	Patients	Patient representatives	Early-Career researchers	Industry	Regulatory	Other*
270	Consensus for SMN2 genetic analysis in SMA patients	22	6	4	7		2	2			1
271	Third ENMC meeting on SBMA: Towards a unifying effort to fight Kennedy's Disease	29	9	7	5	1	3	2	2		
272	Inclusion Body Myositis: 10 years of progress-revision of the "ENMC 2013 diagnostic criteria for IBM" and trial readiness	29	18	3	3	2		3			
273	Clinico-Sero-Morphological Classification of the Antisynthetase Syndrome (ASyS) - Associated Myositis	22	7	3	8	1		2	1		
Average of 4 workshops (n)		25	10	4	6	1	1	2	1	0	0
%		100%	39%	17%	22%	4%	5%	9%	3%	0%	1%
Total of 4 workshops (n)		102	40	17	23	4	5	9	3	0	1

*Other: public health specialist

Clinicians, translational researchers and basic researchers formed the majority of the participants (78%), with an average of 39% clinicians, 22% translational researchers and 17% basic researchers per meeting, which reflects the predominantly clinically-orientated nature of the workshops held in 2023. Connecting basic researchers with clinicians to bridge the lab and the clinic and bring the fundamental science closer to the clinic is one of the aims of the ENMC.

Through the ENMC Patient Participation Programme we aim 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 2023, these two groups made up 9% of total participants; the patients' voice was represented, though we strive to have more representation.

On average, two Early-Career researchers attended each ENMC workshop this year, showing an improvement as compared to previous years. ENMC supports the integration of the next generation of clinicians and basic scientists in established neuromuscular networks via its Early-Career Programme and Mid-Career Mentoring Programme.

In four workshops, where it was relevant, we also had representatives from pharmaceutical companies and a public health specialist.

New numbers to be proud of in 2023



Patients and patient representatives

This year we welcomed four patients and five patient representatives (parents, patient associations, patient advocates) to ENMC workshops. They all gave a presentation, asked questions and participated in the discussions, which was very helpful for the researchers and clinicians in the workshops to learn the patients' needs and interests.



Researchers

We managed to hold four workshops in 2023, with the attendance of 17 basic researchers, 23 translational researchers and 40 clinicians, respectively 17%, 22% and 39% of the total participants. The Early-Career Programme enabled nine young researchers and clinicians to attend the ENMC workshops and promote their entry in the neuromuscular network.



Sponsors

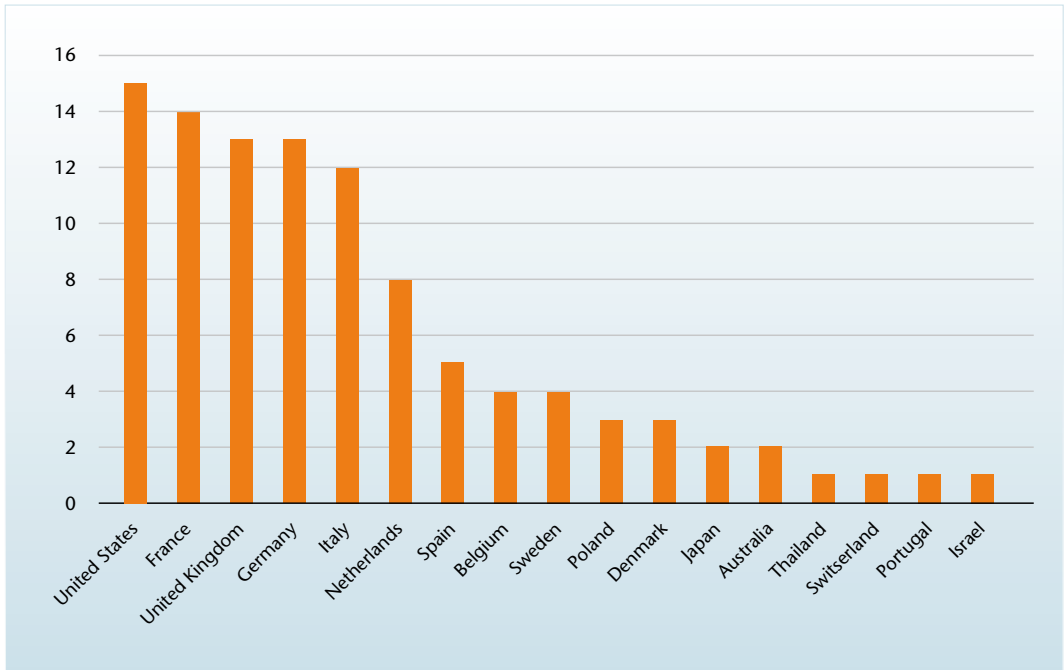
In 2023, the ENMC was sponsored by eight full partners and five associated partners. The ENMC Company Forum supported our activities through the contributions of ten pharmaceutical companies. We are very thankful for the support from all these partners and acknowledge them with gratitude. In addition to these permanent sponsors, we also would like to thank the great contributions of different organisations which co-sponsored specific workshops in 2023.

4.3 Countries represented in ENMC workshops in 2023

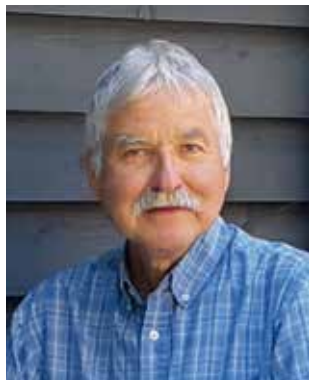
One of the key criteria for a workshop approval by the ENMC is the geographical balance of the participants. The ENMC is convinced that a wide coverage of countries in the workshops is important to make sure that broad consensus is reached at ENMC workshops. This will help to start international research 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 2023, many countries from all over the world were represented at the ENMC workshops (see table below), with a predominance of the ENMC full member countries (France, United Kingdom, Germany, Italy, The Netherlands, Denmark and Switzerland) and some non-member countries like Spain, Belgium and Sweden. Non-European countries like USA, Australia, Japan and Thailand were also represented by individual researchers and clinicians at the four workshops in 2023.

Total number of participants per country in ENMC workshops 2023



4.4 The ENMC from the perspective of a patient



ROLAND MISCHKE is 67 years old and lives near Frankfurt, Germany. He got his diagnosis, Inclusion Body Myositis (IBM), in 2010. At that time, he used to work as a process engineer. Now he works as a volunteer with the German patient organisation DGM as chairman of the Province Hessen Board. He also works with the International Myositis Society (iMyoS), International Myositis Assessment and Clinical Studies Group (IMACS) IBM Group and soon with the Myositis International & Collaborative Alliance.

How many ENMC workshops have you attended and on which topics?

I have attended three workshops:

272 (16 – 18 June 2023) Inclusion Body Myositis: 10 years of progress – revision of the “ENMC 2013 diagnostic criteria for IBM” and trial readiness

255 (9 – 11 Sep. 2022) Muscle Imaging in idiopathic inflammatory myopathies

256 (8 – 10 Oct. 2021) Myositis specific and associated autoantibodies (MSA-ab)

What motivated you to participate in these workshops?

I have learned a lot from the ENMC website regarding the workshops and I wanted to be a part of it. To learn from all the specialists where the problems in research and diagnostics of Myositis are and to make a contribution to the aim of the workshop, to bring my point of view as a patient into the discussions.

Can you describe your overall experience attending ENMC workshops?

With regard to research, diagnostic and treatment procedures we still have different standards in different countries. To be able to compare the status of investigation, we need a worldwide standardisa-

tion. Therefore, it is very important to have these international workshops to facilitate collaboration and have the patients view involved as well.

Why is it important that patients attend these workshops?

The patients are often called as the “the real experts of the disease”. Their experiences regarding quality of life on the one hand and the knowledge of the professionals on the other hand has shown, that we can learn from each other. We will all have a better understanding of myositis and how to deal with it.

What have the workshop participants learned from you?

IBM is not only, for instance, a very interesting issue under the microscope, it causes tremendous changes in someone’s life as well. The workshop participants learned something about the quality of life, needs and fears of IBM patients and they might consider this in their scientific research or clinical work in the future.

What have you learned?

It is very complicated to understand the process in IBM affected muscles. Therefore, it requires a lot of energy, time and money, to find effective treatments and global standards. All participants are very kind people and open for the patients’ view. We need to continue these workshops.

Did you collaborate with participants on projects or discussions after the workshop?

I had several discussions during the coffee or lunch breaks. Unfortunately, I didn't get the chance to collaborate on a project after the workshop.

What would you say to other patients who are thinking about joining a workshop?

The ENMC workshops are a great opportunity for us patients to have a voice, to give an input from our point of view. The scientists, researchers and clinicians in the workshops want to participate in our experiences with the disease we have.

For us as patients it is a pleasure, an honour and a responsibility as well, to take part in this great opportunity.

4.5 The ENMC from the perspective of a former Research Committee member



PROF. NICOL VOERMANS is a Professor of Muscular Diseases at Radboud University Medical Centre / Radboud University, Nijmegen, The Netherlands. As a neurologist, she dedicates herself to patients with muscle diseases, both in care and in research, with a focus on congenital myopathies and facioscapulohumeral muscular dystrophy (FSHD). She is chair of the Radboudumc Committee on Scientific Internship (COMOS), and enjoys working with the inspiring and dedicated clinical and research teams at the Radboudumc Centre of Expertise for Neuromuscular Disorders. She was a member of the ENMC Research Committee from 2017 until 2023.

How did you get involved with ENMC?

I was invited to participate in an ENMC workshop on RYR1 related myopathies in 2016.

217 (29 – 31 January 2016) RYR1-related myopathies

This was a very inspiring meeting. Being able to participate in discussion with other participants who are key opinion leaders was great. It led to a number of subsequent projects.

What is the role of ENMC in the neuromuscular field?

A great role in uniting people, including key opinion leaders and early career researchers. Enabling making step forward on specific themes.

What has been your experience of being a workshop organiser?

Inspiring and busy. It is not the weekend. It is the year of preparation, discussing various topics beforehand with the other organisers and the participants of different sessions. Performing a survey beforehand, aiming to reach consensus afterwards.

Why do you think it's so important to have a patient/patient representatives in a workshop?

To have their input, their perspective on the field. I like the example of Elizabeth Vroom in the workshop on Duchenne carriers (#263): she presented the results of a survey that she had performed. She aimed not to present her own experiences, but the voice of the international group

she represents. Sheila Hawkins did the same (#268) when she presented the results of the FSHD Europe Survey.

263 (13 – 15 May 2022) Focus on female carriers of dystrophinopathy: refining recommendations for prevention, diagnosis, surveillance, and treatment

268 (30 – 2 October 2022) Genetic diagnosis, clinical classification, outcome measures, and biomarkers in Facioscapulohumeral Muscular Dystrophy (FSHD): Relevance for clinical trials

I think that it also empowers the patient representatives. They realize that their voice is important.

What has been your experience of being a member of the ENMC Research Committee for six years?

I truly enjoy good international collaborations (“If you want to go fast, go alone. If you want to go far, go together”). Furthermore, I appreciate collaboration with patient representatives very much. Finally, I always try to think of ways in which young researchers can get involved. ENMC combines this. That has been great, both as ENMC workshop participant and as a ENMC RC member. Being part of the RC has strengthened my network and I have broadened my view on the neuromuscular research field.

What ambitions do you see for the ENMC to achieve in the near future?

- Collaborate with other organisations, such as World Muscle Society and Treat-NMD; make sure to make clear what is special for each organisation. For me, it is clear, at least for ENMC. For newcomers in the field, this is much less clear.
- Enable patient representatives to bring up topics they find important, e.g. the upcoming workshop on bone strength is a good example of a topic raised by patients representatives in the European Reference Network EURO-NMD. Marianne de Visser was the one supporting this.
- Continuously making clear that an ENMC workshop is a major step in a process of a consortium, not a single event. It is so much stronger if the collaboration continues afterwards.
- You could ask workshop organisers to report the deliverables two years after the workshop. This will increase the awareness of including ENMC in the acknowledgement of follow-up activities.

5 Creating global awareness about ENMC workshops

5.1 Publication and dissemination of workshop outcomes

Informing patients and their families about the achievements of ENMC workshops is a key priority of the ENMC. For this purpose, a workshop lay report in English is written by workshop participants and published on the ENMC website within two weeks after the workshop. Lay reports are then translated into many different languages to increase their accessibility for people worldwide. The European partner 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 (X name: _ENMC; LinkedIn: <https://www.linkedin.com/in/enmc-non-profit-a7418590/>) during the workshops and whenever lay reports are published on its website.



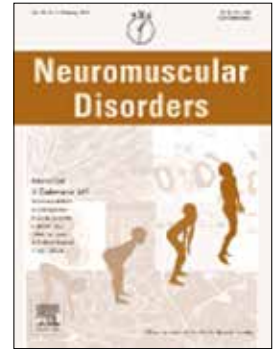
The ENMC maintains an online archive of all workshops organised since 2000, which provides access for the general public to the outcome of the workshops over the years:

<https://www.enmc.org/reports/lay-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. Therefore, it is mandatory that workshop organisers submit a full workshop report to the journal *Neuromuscular Disorders* within 6 months of the workshop.

Since 2021, Early-Career Researchers who made a significant contribution to the organisation of the workshop and the writing of the lay and full report, could become co-authors on the full report.



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

5.2 Conferences in 2023

ENMC ambassadors have attended the following national and international congresses:

- Congress of the Medical Scientific Advisory Board of the German Muscular Dystrophy Society (DGM) e.V., Essen, Germany (24 March 2023).
- German Patient Day of the DGM, Essen, Germany (25 March 2023).
- UK Neuromuscular Translational Research Conference, London, UK (29-30 March 2023).
- XXIII National Congress of Italian Association of Myology, Padua, Italy (8-10 June 2023).
- Peripheral Nerve Society Annual Meeting (PNS), Copenhagen, Denmark (17-20 June 2023).
- International Congress on Neuropathology, Berlin, Germany (13-16 September 2023).
- Dutch Patient Day (SN), Veldhoven, The Netherlands (16 September 2023).
- World Muscle Society Congress, Charleston, USA (3-7 October 2023).
- Swiss Meeting on Muscle Research, Magglingen, Switzerland (3-5 December 2023).

6 The ENMC Mid-Career Mentoring Programme

This programme has been developed for people who seek mentoring in order to acquire skills on their way toward becoming independent researchers and/or potential future leaders in the NMD field. These individuals typically are in the stage in their careers where they are developing their own research plans and have a proven track record in the neuromuscular field. They have established research teams and collaborative networks. The guidelines and the mentee and mentor forms can be found on the ENMC website:

Mid-Career Mentoring Programme



<https://www.enmc.org/mid-career-mentoring-programme/introduction/>

We asked the mentees from the first round (who started in 2021) how the ENMC mentoring programme has helped them in their careers. This is what they told us. The programme helped the mentees:

- In personal development: making decisions, setting priorities, getting prepared for associate professorship, helping to overcome shyness and anxiety, creating work/family balance.
- In career development: expanding their network, increasing visibility, working in new projects in collaboration with mentor, increasing the number of scientific articles and writing successful grant applications by making the right choices, delegating work to team in a more efficient way, members and helping them to independence.
- To better understand the landscape of the NMD field: such as biotech and pharmaceutical companies. How do they work, how do you interact and collaborate with them.

Their recommendations are:

- The programme should continue guiding mentees like them.
- Organise a meeting with mentees and mentors to foster the network.

Mentees 2023



DR SALMAN BHAI is an adult neuromuscular neurologist who is the Director of the Neuromuscular Center at the Institute for Exercise and Environmental Medicine and Assistant Professor of Neurology at the University of Texas Southwestern Medical Center in Dallas (United States).

Salman's mid-career mentors are Prof. E. Hoffman (United States), Prof. A. De Luca (Italy)



DR MARGHERITA GIANNINE is an MD PhD, rheumatologist and physiologist, working in clinical and translational research in the Physiology department of the Strasbourg University Hospital (France). She is also Associate Professor in Physiology at the University of Strasbourg (France).

Margherita's mid-career mentors are Prof. O. Benveniste (France), Dr A. Mammen (United States)



DR SASKIA HOUWEN is a paediatric rehabilitation physician in the neuromuscular centre of the Radboud University Medical Centre in Nijmegen (The Netherlands).

Saskia's mid-career mentors are Mrs S. Foye (United States), Prof. C. Gagnon (Canada)



DR PIERA SMERIGLIO is a principal investigator and group leader at the Myology Institute in Paris (France).

Piera's mid-career mentors are Prof. A. Aartsma-Rus (The Netherlands), Dr A. Beggs (United States)

7 Resources and financial management in 2023

Financial summary 2023

Annual accounts for the year 2023 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 2023 are shown in comparison with the figures for the previous financial year 2022.

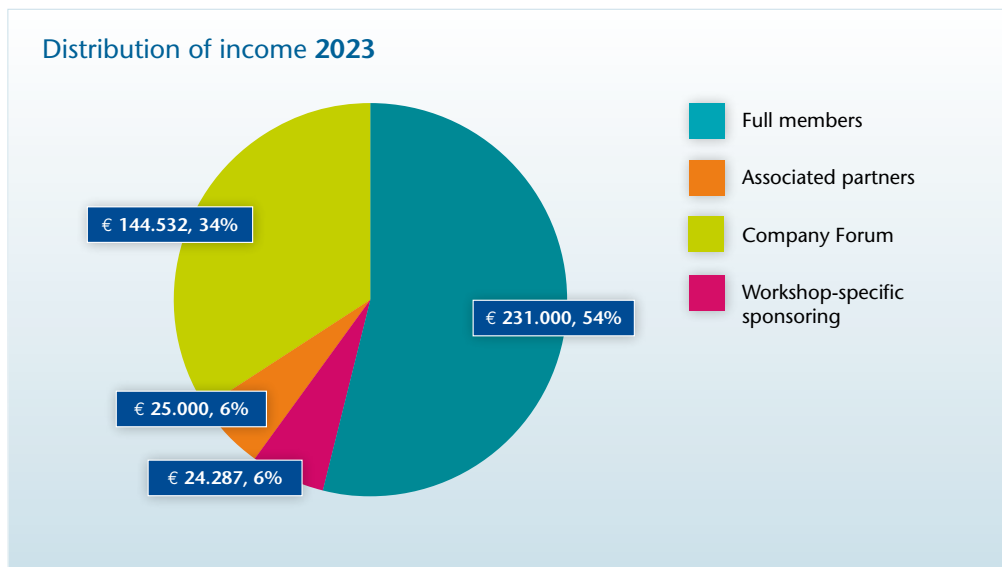
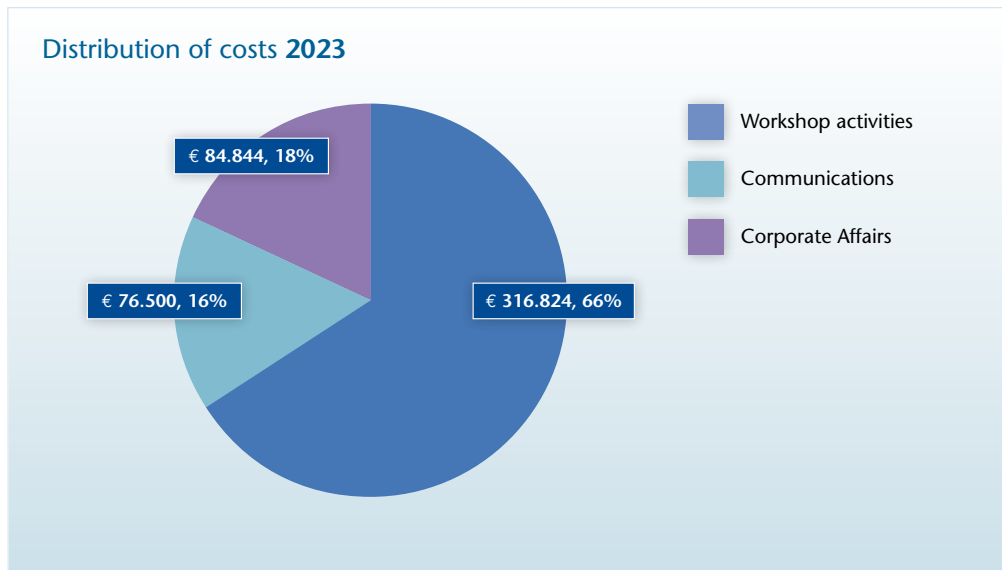
Statement of income and expenses for the year 2023 in Euros (€)		
	2023	2022
INCOME		
Full Partner contributions	231.000	231.000
Associated Partner contributions	25.000	20.000
Company Forum contributions	144.532	124.940
Other contributions	24.287	57.659
Total income	424.819	433.599
EXPENSES		
Personnel expenses	304.884	163.284
Rental expenses	11.701	11.574
Activity (workshop) expenses	89.980	113.788
Organisational expenses	71.603	60.747
Total operating expenses	478.168	349.393
Operating result	- 53.349	84.206
Interest income	2.713	- 1.602
Net result	- 50.636	82.604
APPROPRIATION OF RESULTS		
Development reserve - withdrawal*		- 20.195
Transitional reserve	- 55.722	
Other free reserves	5.086	102.799
CASH AT BANKS ON 31 DECEMBER	607.414	659.123

*Note: Withdrawal means that these costs were made in 2022 and could be taken from this provision

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

Opinion of the auditors

The independent accountants have verified and approved the annual accounts. For a full PDF version of the report of the annual accounts for 2023, please visit the ENMC website: <https://www.enmc.org/about-us/annual-report/>



8 Governance in 2023

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 European patient organisations for neuromuscular disorders and many other related organisations. The statutory location is in Baarn, The Netherlands, in the building of Spierziekten Nederland.

8.1 The ENMC Executive Committee

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

Composition of the ENMC Executive Committee on 31 December 2023

Dr K. Adcock (United Kingdom)
Dr A. Ambrosini (Italy)
Dr S. van den Berge (The Netherlands)
Mr H. Ib Jørgensen (Denmark)
Dr I. Meijer (vice-Chair, The Netherlands)
Dr A. Méjat (Chair, France)
Prof. U. Schara-Schmidt (Germany)
Dr R. Willmann (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 2023

Dr A. Buj-Bello (France)
Dr A. Ferreiro (Chair, France)
Prof. N. Goemans (Belgium)
Prof. E. Gomes (Portugal)
Prof. C. Kornblum (Germany)
Prof. C. Ottenheim (The Netherlands)
Prof. R. Quinlivan (United Kingdom)
Prof. T. Sejersen (Sweden)
Prof. G. Tasca (United Kingdom)
Prof. V. Timmerman (Belgium)
Prof. M. Weber (Switzerland)



ENMC Executive Committee meeting in November 2023



ENMC Research Committee meeting in November 2023

8.3 The ENMC Office

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

ENMC Office staff on 31 December 2023

Ms P. van Dongen (Programme Manager)
 Ms W. Hinloopen (Operational Manager)
 Ms T. van Esch (Freelance Workshop Assistant)



ENMC office staff, Wilma Hinloopen and Patricia van Dongen

8.4 Transfer of the Deutsche Gesellschaft für Muskelranke e.V. representative

After representing the Deutsche Gesellschaft für Muskelranke e.V. at the ENMC Executive Committee for 14 years (from 2009-2023), Dr Arpad von Moers passed his task to Prof. Ulrike Schara-Schmidt in July 2023. Arpad took part in many ENMC activities combining his clinical experience with the role of spokesperson for the patients' needs with the politeness and modesty that has always distinguished him. His personal touch also drew on the joy of making music, a passion that he continues to put to the service of patients nationwide, but which he passed on to the entire ENMC team, bringing joy and moments of lightness to the daily work and regular committee meetings.

Arpad had the tough responsibility of chairing the ENMC Executive Committee during the 2 years of Covid 19 pandemic (2020-2021). "What a period!" he claimed in the opening of the 2020 Impact Report. Managing tasks and relationships virtually required a great sense of belonging and soft skills, working in synergy and harmony - "Work in unison", he would say - with the research director, the Baarn team and colleagues on the research and executive committees, and inventing new ways to stay on track and maintain focus on the ENMC mission. We would like to thank Arpad's contribution and commitment for all those years to the ENMC.



Ulrike is Professor of neuropaediatrics and neuromuscular diseases. She heads the Department of Neuropaediatrics and the Centre for Neuromuscular Diseases in Children and Adolescents at the Children's Hospital of the University Hospital Essen. She is president of the German Brain Council and chair of the Ethics Committee of the Medical Faculty Duisburg-Essen and has been member of numerous national and international committees. The ENMC welcomes her to the Executive Committee and looks forward to a great collaboration.

A "Thank You" gift from Alexandra Breukel on behalf of ENMC to Dr Arpad von Moers during the EC meeting in April 2023.

8.5 Dr Alexandra Breukel says goodbye after almost 10 years working at ENMC

After nearly a decade of working with pleasure and dedication, Alexandra Breukel, the Managing Director of ENMC, said goodbye on October 1st to embark on a new chapter in her career. At the farewell reception, Alexandra expressed her gratitude to everyone for the incredible time working together to enhance the lives of individuals affected by neuromuscular conditions.

Moving forward, we are pleased to announce that Patricia van Dongen and Wilma Hinloopen will carry on the mission of ENMC.



Alexandra at her farewell reception.

9 A special thank-you to all our partners and supporters

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

ENMC full partners



Deutsche Gesellschaft für Muskelkranke e.V. DGM



ENMC associated partners



Finnish Neuromuscular Disorders Association



Members of the Company Forum



Workshop-specific sponsors in 2023



10 Looking forward to 2024 and beyond

10.1 Workshops in 2024

Three ENMC workshops took place in Q1 and at least four workshops are planned in the next quarters of 2024 (see table below).

Two review rounds for workshop applications are scheduled in 2024: one in the spring (submission deadline 1 March 2024) and one in the autumn

(submission deadline 1 September 2024).

The workshops that are selected at these review rounds will all be planned for 2024 or 2025.

For updates please visit the ENMC website:

<https://www.enmc.org/workshops/upcoming-workshops/>

Preliminary ENMC programme 2024

Workshop no. and date	Topic	Workshop leaders
Workshop no. 274 19-21 January 2024	ENMC recommendations for optimizing bone strength in neuromuscular disorders	Prof L. Ward, Prof. A. Moretti, Dr D. Weber, Prof. N. Voermans
Workshop no. 275 9-11 February 2024	Seronegative MG: an update paradigm for diagnosis and management	Prof. A. Evoli, Dr L. Maggi, Dr J. Palace, Prof. J. Verschuuren
Workshop no. 276 15-17 March 2024	ENMC recommendations on optimal diagnostic pathway and management strategy for patients with exertional rhabdomyolysis worldwide	Prof. P. Laforêt, Prof. J. Vissing, Prof. N. Voermans, Dr S. Bhai
Workshop no. 277 21-23 June 2024	Congenital myopathies: revising and revisiting nomenclature and diagnostic guidelines	Dr J. Dowling, Dr C. Bönnemann, Dr E. Oates, Dr A. Ferreiro
Workshop no. 278 20-21 September 2024	European standards for harmonization of myasthenia gravis (MG) registries and emerging digital solutions	Prof. S. Sacconi, Dr R. Mantegazza, Dr E. Cortés-Vicente, Prof. A. Meisel
Workshop no. 279 1-3 November 2024	Classification, clinical care, outcome measures and biomarkers in childhood onset FSHD: towards standardising clinical care and ensuring clinical trial readiness	Dr C. Erasmus, Prof. K. Mathews, K. de Valle, Prof. T. Willis
Workshop no. 280 22-24 November 2024	Diagnostic criteria and outcome measures in primary mitochondrial myopathies	Prof. M. Mancuso, Prof. C. Kornblum

10.2 International conferences in 2024

ENMC ambassadors will attend the following international congresses.

- 4th Scientific International Congress on SMA, Ghent, Belgium (14-16 March 2024).
- 8th International Congress Myology, Paris, France (22-25 April 2024).
- Peripheral Nerve Society Annual Meeting (PNS), Montreal, Canada (22-24 June 2024).
- European Academy of Neurology (EAN), Helsinki, Finland (29 June – 2 July 2024).
- World Muscle Society Congress, Prague, Czech Republic (8-12 October 2024).
- ICNMD 2024, Perth, Australia (25-29 October 2024).

10.3 Budget for 2024

This table presents the budget forecast for 2024 as of 31 December 2023.

Budget 2024 in Euros (€)	Actuals 2023	Budget 2024
INCOME		
Full Partner contributions	231.000	231.000
Associated Partner contributions	25.000	25.000
Company Forum contributions	144.532	145.000
Other contributions	24.287	30.000
Total income	424.819	431.000
EXPENSES		
Personnel expenses	304.884	222.188
Rental expenses	11.701	12.000
Activity expenses	89.980	180.000
Organisational expenses	71.603	66.384
Total operating expenses	478.168	480.572
Operational result	- 53.349	- 49.572
Interest income	2.713	7.000
NET RESULT	- 50.636	- 42.572

Colophon

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ENMC attended the German Patient Day (DGM) in Essen with a booth. From left to right, Julia Roll (DGM), Patricia van Dongen (ENMC), Roland Mischke (DGM) and Alexandra Breukel (ENMC).



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