

EUROPEAN NEURO MUSCULAR CENTRE

Bringing the voice of people with NMDs within the international NMD community.

What role for associations and charities?

Anna Ambrosini
(Fondazione Telethon Italy)
ENMC Executive Committee Chair





41st Annual General Meeting
September 8-10, 2011, Prague, CZECH REPUBLIC

EUROPEAN NEURO MUSCULAR CENTRE

Summary

- ENMC activity
- Partnership in TREAT-NMD
- Role of Associations and NMD funding agencies at national and international levels
- Future plans

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EUROPEAN NEURO MUSCULAR CENTRE

ENMC History

- First established in 1989 in Paris, France, under the auspices of the EAMDA and AFM – Y. Poortman, G. Nigro, R. Rudel + B. Baratoud
- Established as a foundation independent from EAMDA in 1992. Strategic plans - A. Emery, V. Dubovitz
- Since 1992 a Consortium financed mainly by members
- Since 2007: partner for development of Treat-NMD and role in training and communication; implementation of new core activities related to the mission

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EUROPEAN NEURO MUSCULAR CENTRE

ENMC mission

To encourage and facilitate collaborative research in Europe on neuromuscular disorders to contribute to the eradication of NMDs

The formula

- Small workshops conveying representative leaders in a field
- Aim: "to be collaborative projects, the sharing of ideas, material and patients, and hopefully joint publications" (A. Emery, 1990)
- Not a presentation of published data, but discussion of work in progress

Topics

From:	➔	To:
<ul style="list-style-type: none"> ■ Diagnostic criteria ■ Gene identification ■ Establishment of Consortia 		<ul style="list-style-type: none"> ■ Current management ■ Trial readiness

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EUROPEAN NEURO MUSCULAR CENTRE

Structure

Members



- Finland Muscular Dystrophy Association (FMDA)
- Muskelsvindfonden (MSFV)
- Deutsche Gesellschaft für Muskelkranke (DGM)
- Schweizerische Stiftung für die Erforschung der Muskelkrankheiten (SSEM)
- Österreichische Muskelforschung (OFM)
- Društvo Distrofikov Slovenije (DDS)
- Fondazione Telethon/UILDM
- Muscular Dystrophy Campaign (MDC)
- Prinses Beatrix Fonds (PBF)
- Vereniging Spierziekten Nederland (VSN)
- Association Française contre les Myopathies (AFM)
- Asociación Española contra las Enfermedades Neuromusculares (AEEM)

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EUROPEAN NEURO MUSCULAR CENTRE

Structure

Office

European Neuromuscular Centre
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3743 JN Baarn The Netherlands
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Website: www.enmc.org

■ **Location: Baarn (NL)**

Research Manager: **D. Zollinger** 

Management Assistant: **A. Zitterstein**

Workshop Assistant: **M. Rector**

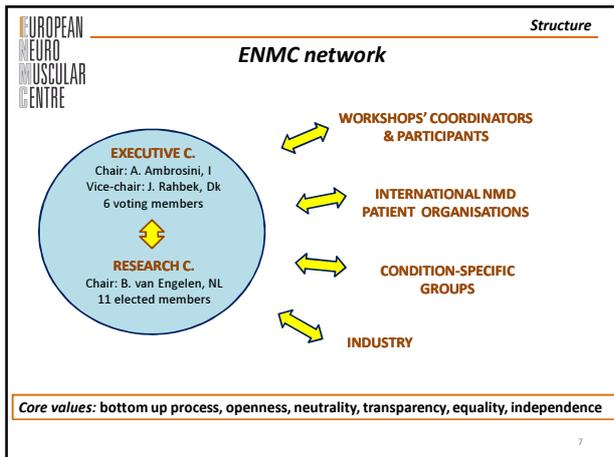


Treat-NMD Project Manager: **K. Senden**

Treat-NMD Project Coordinator: **M. Schoneveld van der Linde**

■ **ENMC is a Virtual Centre, not a facility!**

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Activities

Activities 1990-2000

Diagnostic Criteria for Neuromuscular Disorders

Neuromuscular Disorders: Clinical and Molecular Genetics

Ten years of ENMC – from a patients' initiative to a successful European research institution: the story of the European Neuromuscular Centre

Reinhild Riekke*, Giovanni Ngigi, Yshard Postman
Université Libre de Bruxelles, Belgium

Victor Dubowitz*
Imperial College School of Clinical Medicine, Hammersmith Campus, London W12 0NN, UK

Workshop report

Special Centennial Workshop – 101st ENMC International Workshop: Therapeutic Possibilities in Duchenne Muscular Dystrophy, 30th November–2nd December 2001, Naarden, The Netherlands

Victor Dubowitz*

Imperial College School of Clinical Medicine, Hammersmith Campus, London W12 0NN, UK

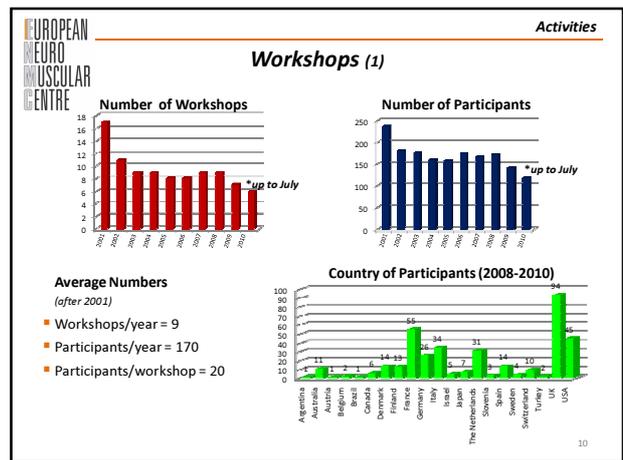
Received 2 August 2002

FORUM Clinical Medicine Vol 1 No 3 May/June 2001

European collaboration in research into rare diseases: experience of the European Neuromuscular Centre

Alan Emery and Michael Rutgers

- Activities**
- Activities 2001-2011**
- Workshops
 - Practical care workshops
 - Dissemination of information and knowledge
 - Clinical Trial Network
 - Partnership in Treat-NMD



Activities

Workshops (2)

Diseases addressed

Myopathies	17
Mitochondrial diseases	13
CMD	12
DMD	10
ALS	8
SMA	5
FSHD	1
Neuropathies /CMT	7
Emery-Dreifuss Muscular Dystrophy	7
Pompe	2
NMDs in general	12

- TREAT-NMD**
- Partnership in TREAT-NMD (1)**
- Responsibilities:**
- Training & education
 - Bringing Eastern Europe into the network
 - Expertise and infrastructure for organizing workshops
- Relied also on:**
- Existing international network of researchers (clinical and preclinical)
 - Existing network of patient organizations

EUROPEAN NEURO MUSCULAR CENTRE **TREAT-NMD**

Partnership in TREAT-NMD (2)

Training & education

- Training inside and outside the network
In loco Spring Schools (St. Petersburg and Moscow, Russia)
In loco training courses: Sofia; Budapest; Belgrade; Istanbul; Milan; Bucharest; Prague
- Development of a tool kit to support local trainers (*Standards of Care; Standards of Diagnosis; Registry development*); questionnaires
- Develop a NeuroMyology Curriculum at EU level - liaising with Scientific Society

Communication

- Bringing Eastern Europe into the network
- Develop a database with contacts, particularly from the Central Eastern Europe, newsletters dedicated to Pts' associations; surveys
- Meetings to listen to the NMD people's voice and needs and share info about Treat-NMD tools and ideas (Milan, Sept. 2010)

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EUROPEAN NEURO MUSCULAR CENTRE **TREAT-NMD training courses**

Questionnaires for professionals

Feed back from the participants on the course and on situation in their countries

- Overall rating of the course
Scientific level
Relevance of topics discussed
Structure, venue, proceedings, etc.
- Existence of national registries
- Implementation of Standards of Care
- Training needs according to professionals
- Cooperation on training courses
- Interest in translation of SoC guidelines (where not available in local language)

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EUROPEAN NEURO MUSCULAR CENTRE **TREAT-NMD training courses**

The picture highlights the contacts established in the last year with countries/academic institutions in CEE countries that have expressed interest in running or future training and meeting initiatives.

Colour indicates the number of people we are in contact with (green: 1-5 contacts; red: 6-10 contacts; yellow: 11+ contacts).

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EUROPEAN NEURO MUSCULAR CENTRE **TREAT-NMD training courses**

Suggested topics for on-site training by the professionals in CEE countries

Topics	Percentage
Diagnosis of NMD's - MD's in general, CMD, LGMD, polyneuropathies, mitochondrial diseases - Case studies - Analyses and use of muscle biopsies	50%
Standards of Care and Management - DMD and SMA basic care - Physiotherapy	23%
Therapeutic approaches - Clinical trials (ongoing and future) - Outcome measures	13%

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EUROPEAN NEURO MUSCULAR CENTRE **Survey to Pts Associations**

Survey on DMD SoC implementation sent to Pts Org in CEE countries

Questionnaire sent out to 23 Pts Org covering 18 CEE countries

1 Armenia	10	Latvia
2 Belarus	11	Lithuania
3 Bosnia-Herzegovina	12	Poland
4 Bulgaria	13	Romania
5 Croatia	14	Russia
6 Czech rep	15	Serbia
7 Estonia	16	Slovakia
8 Greece	17	Slovenia
9 Hungary	18	Turkey

Response rate:

- per Pts Org 69% (Tot responses 16)
- per Countries 72% (Tot countries 13)

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EUROPEAN NEURO MUSCULAR CENTRE **Survey to Pts Associations**

Aims of the DMD survey

- To verify awareness of SoC guidelines for DMD amongst Pts Org in CEE countries
- To verify the implementation phase of SoC according to the Pts Org view
- To have feedback on the specific training needs in SoC according to the view of Pts Org
- To verify availability and interest in developing training initiatives in their country

Target group: DMD specific Pts Org or NMD general Org in CEE countries

Overview provided of the topics covered in the SoCs for DMD and DMD family guide (incl. link to translations in other languages) in preparation of the 181th ENMC workshop on DMD SoC

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EUROPEAN NEURO MUSCULAR CENTRE Survey to Pts Associations

(A) Awareness and implementation of DMD SoC

Awareness of SoC guidelines: (no Armenia and Estonia)	83%
Document availability in the local language:	
Yes/in translation	26% / 20%
No	54%
(No: Armenia, Bosnia-H, Estonia, Greece, Lithuania, Serbia, Slovakia)	
Implementation of SoCs:	
Well implemented	13%
Partly implemented	60%
Not at all implemented	27%
(No: Bosnia-Herzegovina, Estonia, Romania, Slovakia)	
DMD registry available:	
Yes	46%
No	54%
(available in Armenia, Czech R, Romania, Serbia (local), Slovenia, Turkey)	

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Obstacles to implementation of SoCs

Reasons for partly or no implementation of SoC in CEE countries:

- Financials reasons 100%
- Lack of support within the medical community 83%
- Political reasons 50%

Other comments:

- lack of DMD specialists
- little cooperation and lobby between patient organisation
- a general passive attitude of medical staff and family, resulting from the idea of any intervention being useless

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EUROPEAN NEURO MUSCULAR CENTRE Survey to Pts Associations

(B) Training initiatives – topics of major interest for DMD care

TOPIC	N. Countries SCORE 5	N. Countries SCORE 4
Diagnosis	1	4
➔ Neuromuscular management	7	1
Pharmacological management	1	4
➔ Rehabilitation and physiotherapy	6	4
Orthopedic management including osteoporosis	3	2
➔ Respiratory management	7	1
Cardiac management and prevention	4	3
Gastrointestinal management	3	4
Psychosocial aspects	2	7
➔ Clinical trials (ongoing and future)	6	1
Outcome measures	5	3
Patients' registries	4	3

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EUROPEAN NEURO MUSCULAR CENTRE TREAT-NMD

Conclusions (1) - Major issues reported by Pts Org

1. There is still a lot of demand for training on essential problems such as respiratory care or rehabilitation/physiotherapy
2. A greater awareness on the SoCs can help to counteract the passive attitude, as reported by some Pts organisations in the survey or directly to us during the organization of training courses
3. Need for better information on registries in CEE countries. Effective use and expectations
4. Pts organisations may need help to:
 - learn to work together and with clinicians
 - get empowered and be aware of their rights (for instance to receiving funds for ventilatory support by the Natl. Health System)

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EUROPEAN NEURO MUSCULAR CENTRE TREAT-NMD

Conclusions (2)

1. Lot of interest in professionals to receiving training in several CEE countries and to contribute setting initiatives in their countries
2. Some countries have excellent one/few main centres that can act as promoter and collaborate to disseminating SoC and coordinate training initiatives. They should be involved and supported
3. Other countries still need to start a fertilization process
4. The Pts Org can be also involved. Several still need to learn to work together and to be better aware of the SoC guidelines

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EUROPEAN NEURO MUSCULAR CENTRE ENMC future plans

Future plans of the ENMC

Workshops

- Continue to support workshops
- Encourage *avant garde* groups to address NMDs types and specific problems not yet tackled in an effective way
- Promote rigorous studies on clinical problems where there is still paucity of EBM studies and guidelines (i.e. rehabilitation, practical care)

Training

- Explore possibilities for joint programs for involvement in ENMC in training activities

Patients' platform

- Identify new avenues for a more fruitful interaction between people with NMDs and clinical scientists
- Value the existence of a patient organizations' platform to play an active role in the new scenario where research on therapeutics is finally to becoming reality

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