Workshop 2
Getting involved in Research
How can patient organizations trigger an EU-funded rare disease project?

“The role of patient groups in an EU-project”

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Organization: ENMC (European NeuroMuscular Centre)
EAMDA (European Alliance of neuroMuscular Disorders Associations)
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Neuromuscular Disorders characterised by:

- Degeneration of muscles/(motor)neurons
  → leading to severe paralysis & death
- Heterogeneous group of diseases (±600 (sub)types)
- In majority inherited (75%)
- Rare disorders
- No cures available, treating symptoms and slowing down progression
Some examples of NMD:

- Duchenne Muscular Dystrophy
- Limb Girdle Dystrophy
- Facio Scapulo Humeral Dystrophy (FSH Dystrophy)
- Myasthenia Gravis
- Myotonic Dystrophy (Dystrophia Myotonica)
- Guillain Barré Syndrome
- Spinal Muscular Atrophies (SMA types I/II/III/IV)
- Amyotrophic Lateral Sclerosis (ALS/Lou Gehrig’s disease/Motor Neuron Disease)
- Pompe’s Disease (glycogen storage disease type 2)
- Charcot Mary Tooth (CMT)
TREAT NMD

Translational Research in Europe
Assessment & Treatment of
Neuromuscular Diseases

Network of Excellence

FP6, Life Sciences, Genomics and Biotechnology for Health
Getting involved: Being Aware

- Being aware of the drug development process

- Being convinced that it is possible to have influence on the drug development process
Getting involved: Being Aware

- Being aware of the drug development process

- Being convinced that it is possible for patient organizations to have influence on the drug development process
Getting involved: Being There

- Being organised on national level: AFM, VSN, DGM, ....

- Being organised on the European patient level: EAMDA

- Being organised on the scientific level: ENMC

- Being represented on the political, regulatory, scientific and industrial level: EURORDIS
European Alliance of neuroMuscular Disorders Associations (EAMDA)

- Established 1971 London
- Objectives
  - Help for people suffering from NMD
  - Improve well being (physical/mental)
  - Support research into causes, treatment and cure
- 21 Members throughout Europe
  - Heterogeneity of members
Getting involved: Being There

- Being organised on national level: AFM, VSN, DGM, ....
- Being organised on the European patient level: EAMDA
- Being organised on the scientific level: ENMC
- Being represented on the political, regulatory, scientific and industrial level: EURORDIS
European Neuro Muscular Centre (ENMC)

- First established in 1989 in Paris, France
- Independent in 1992
- Secretariat based in Baarn, The Netherlands

Aim:
- To usefully and efficiently contribute to the eradication of neuromuscular diseases
- To improve efficiency in European neuromuscular research
- To facilitate and support research communication between European (and international) researchers and clinicians
Topics of Workshops (1990 - 2006)
Total of 145 Workshops

- Spinal Muscular Atrophy 10
- Duchenne Muscular Dystrophy 8
- Congenital Muscular Dystrophy 8
- Charcot-Marie-Tooth 7
- Emery-Dreifuss Muscular Dystrophy 7
- Myotubular Myopathy 6
- Facioscapulohumeral Dystrophy 5
Results of ENMC 1990-2006

- Collaborative Workshops: 145
- Consortia: 18
- Participants: 2123
- Publications: 225
Getting involved: Being There

- Being organised on national level: AFM, VSN, DGM, ....

- Being organised on the European patient level: EAMDA

- Being organised on the scientific level: ENMC

- Being represented on the political, regulatory, scientific and industrial level: EURORDIS
The call

- This network of excellence will aim at sharing expertise between basic and clinical academics and industrial partners in order to develop technological and methodological tools with a view to accelerate the elaboration of new therapies for rare neuromuscular diseases. Important tools include animal models, databases, biobanks, well defined patient cohorts, methods for efficacy assessment.
Taking an international perspective in NMD research

- Time to build on networking for translational research building on:
  - Respected work of ENMC workshops and consortia
    - Basic research
    - Standards of care
    - Clinical trials
  - Increasing resources of national networks
  - Networks of patient organisations
21 partners, including national networks, research and clinical organisations, charities and companies.

10 million Euro budget over 5 years

Aim is to concentrate on integration activities
What are the barriers to trials in NMD?

- NMD encompasses a wide number of diseases
- Most types of NMD are individually rare
- Full molecular diagnosis of NMD is still not always widely available/ fully applied
- Standards of care are not uniformly applied
- Assessment tools are lacking
- Trials to date are few
- No “trial culture” amongst patients or doctors
- Fragmentation of both research groups and patient groups
Aims and objectives of the NoE

- To bring together the EU researchers, clinicians, patients groups and industry working in NMD
- To address the fragmentation currently hindering the progress of promising therapies
- To develop a co-ordination centre linking centres of excellence for translational research in NMD
- For delivery of innovative treatments for NMD
- To educate clinicians and researchers in these processes
- To extend the network to Eastern Europe
ENMC main involvement in work packages: (leading participant)

- **WP 01.2** Extend TNCC towards Eastern Europe
- **WP 11.1** International collaboration with research groups
- **WP 11.2** Cross linking with European Consortia (to avoid duplication NoEs, IPs, STREPs)
- **WP 12.1** Establish a mobility plan for scientists and key staff
- **WP 12.2** Develop internal and external training
- **WP 14.1** Ethical aspects and science communication
- **WP 14.2** Dissemination of accessible information and integration op patient groups
Deliverables WP 14.2 for the next 12 months

- Overview of all European patient groups/support groups

- Overview of European expert centers (experts)

- Organize patient conferences in conjunction with EAMDA annual general meetings (Warsaw 2007 and Bulgaria 2008)
Challenges WP 14.2

- Managing patient expectations
  - Informing patients on the drug development process
  - Issues like compassionate use, inclusion criteria, placebo controlled trials etc.
  - Balancing and judging the interests of both scientists, industry and patients
Additional Challenges:

- Overcoming language barriers needs special attention
  (mainly addressing non-scientists!)

- What about non EU countries in Europe

- How to enable (empower & support) patient organisations to build and maintain national infrastructures for communication
Is it possible for patient organizations to play a role in EU research projects?

- Being aware
- Being there
- Having access to resources (AFM, ACIES, ENMC)
- Having dedicated scientists

**but there still is a need for:**

- Recognition for potential roles patient org. on national level
- Need for training and education
- Structural funding of patient organisations
Thank you, for your attention!