European Reference Network
for rare or low prevalence complex diseases

Network
Neuromuscular Diseases (ERN EURO-NMD)
**Mitochondrial diseases**

- Chairs: Rita Horvath (Newcastle, UK) and Cornelia Kornblum (Bonn, Germany)
- Most centres in the ERN-NMD are adult mitochondrial disease centres, however there are a few paediatric NMD centres (Nijmegen, Barcelona, etc).
- Many paediatric mitochondrial centres are involved in the metabolic ERN (ERN-METAB)
- We discussed with the members of ERN-METAB (Enrico Bertini, Eva Morava and Shamima Rahman) to organise linked activities in mitochondrial diseases between ERN-NMD and ERN-METAB after the main structure of each ERN is established
Planned working groups in mitochondrial diseases

Chair: Rita Horvath (Newcastle, UK), Co-Chair: Cornelia Kornblum (Bonn, Germany)

Clinical diagnosis, treatments, guidelines, clinical trials – adult mitochondrial disease
Veronique Paquis (Nice, France)  
Constanza Lamperti (Milan, Italy)  
Marian Janssen (Nijmegen, Netherlands)  
Serenella Servidei (Rome, Italy)

Clinical diagnosis, treatment, guidelines, clinical trials – paediatric mitochondrial disease
Bobby McFarland (Newcastle, UK)  
Rafael Artuch (Barcelona, Spain)  
Daria Diodato (Rome, Italy)

Diagnostics
Rob Taylor (Newcastle, UK)  
Richard Rodenburg (Nijmegen, Netherlands)

affiliated:
Holger Prokisch (Munich, Germany)  
Patrick Chinnery (Cambridge, UK),  
Valerio Carelly (Bologna, Italy)
# List of named participants in mitochondrial diseases

<table>
<thead>
<tr>
<th>Country</th>
<th>Italy</th>
<th>Belgium</th>
<th>Germany</th>
<th>France</th>
<th>Spain</th>
<th>UK</th>
</tr>
</thead>
<tbody>
<tr>
<td>People</td>
<td>31</td>
<td>11</td>
<td>9</td>
<td>7</td>
<td>7</td>
<td>6</td>
</tr>
<tr>
<td>Institutes</td>
<td>11</td>
<td>3</td>
<td>5</td>
<td>3</td>
<td>2</td>
<td>1</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Country</th>
<th>Hungary</th>
<th>Poland</th>
<th>Netherlands</th>
<th>Bulgaria</th>
<th>Slovenia</th>
</tr>
</thead>
<tbody>
<tr>
<td>People</td>
<td>11</td>
<td>3</td>
<td>2</td>
<td>1</td>
<td>1</td>
</tr>
<tr>
<td>Institutes</td>
<td>2</td>
<td>1</td>
<td>2</td>
<td>1</td>
<td>1</td>
</tr>
</tbody>
</table>

**Patient Organisations**

- IMP
- AMM
- AEPM

Building bridges and breaking barriers in rare neuromuscular diseases
Mitochondrial disease: guidelines, diagnostic criteria and outcome measures


Mitochondrial disease: guidelines, diagnostic criteria and outcome measures


Next steps

- Include some institutions who did not claim mitochondrial activity, but have clear national or international leading role in mitochondrial disease (Nijmegen etc.)
- Agree in the rules of being a specialised mitochondrial centre for existing and new centres

**Discuss in the expert group:**

- how to implicate existing guidelines or create new guidelines
- how will actual patient referrals be managed
Developing novel treatments in mitochondrial disease

Gene identification/pathophysiology
- Biomarkers
- Model systems
- Delivery mechanisms
- Proof of principle studies

Trials
- Diagnosis/care standards
- Natural history
- Patient Registries
- Trial sites
- Outcome measures

Therapy delivery
- Regulatory affairs
- Ethics
- Commissioning/health economics

International Workshop on trial readiness in primary mitochondrial myopathies
Ideas for hunting down the right path to a cure
Rome, Hotel Mediterraneo
November 16-18, 2016