



European  
Reference  
Network

for rare or low prevalence  
complex diseases



Network  
Neuromuscular  
Diseases (ERN EURO-NMD)

# 7<sup>th</sup> ERN EURO-NMD ANNUAL MEETING

## Mitochondrial diseases

21<sup>st</sup> February 2024

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Funded by  
the European Union





## Inter-ERN Mitochondrial group

Michelangelo Mancuso, group leader	Pisa	ERN Euro-NMD
Costanza Lamperti	Istituto Besta, Milano	ERN Euro-NMD
Thomas Klopstock	Fr.-Baur-Institut, Munich	ERN RND
Saskia Wortmann	Salzburg	ERN Epicare
Laura Licchetta	Bologna	ERN Epicare
Shamima Rahman	London	MetabERN
Hélène Dollfus	France	ERN-EYE
Alison Maguire	The Lily Foundation	Patient Representative
Serena Massucci	Mitocon Italia	Patient Representative
Heike Pascal, for support	Paris	ERN Euro-NMD
Manuela Lavorato, for support	Italy	ERN Euro-NMD

# Inter-ERN Aims

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- to develop guidance towards a common approach between ERNs (endorsed by EAN & PAGs, where possible)
- to know first (survey) and then to develop diagnostic pathways, treatment options through Europe
- ICD-10 codes
- the education needs:  
ERN webinars, EAN, ESHG workshops or teaching courses, training curriculum

# Closed Project I

## Inter ERN Survey: How to approach and manage primary mitochondrial diseases (PMDs) in different EU countries.


### The ERNs survey

Journal of Neurology (2024) 271:835–840  
<https://doi.org/10.1007/s00415-023-12017-1>

ORIGINAL COMMUNICATION



#### Current management of primary mitochondrial disorders in EU countries: the European Reference Networks survey

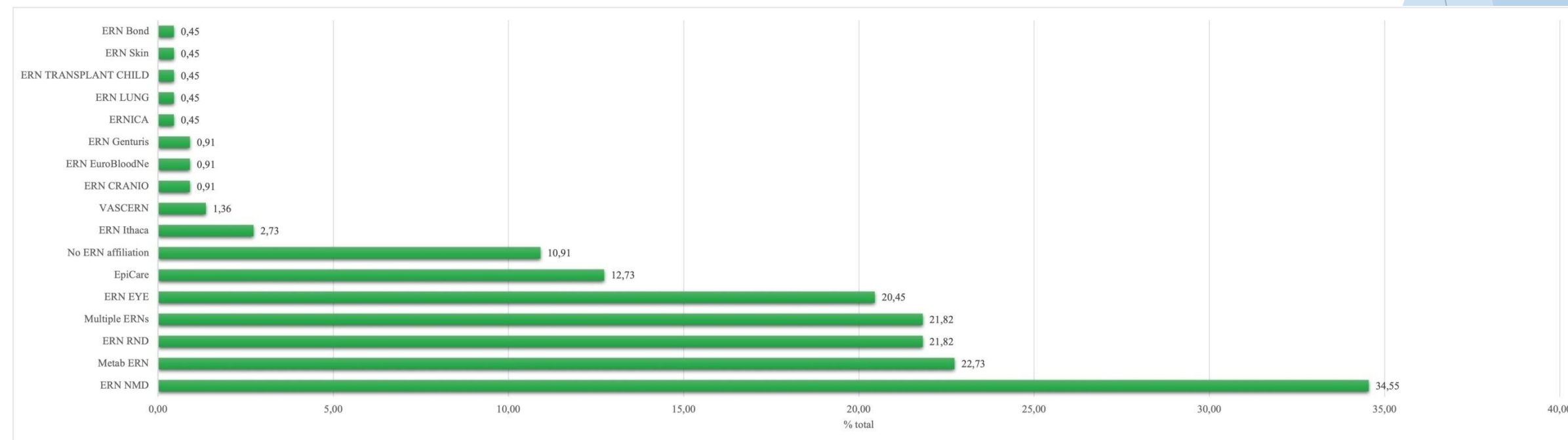
Michelangelo Mancuso<sup>1</sup>  · Piervito Lopriore<sup>1</sup> · Costanza Lamperti<sup>2</sup> · Thomas Klopstock<sup>3,4,5</sup> · Shamima Rahman<sup>6</sup> · Laura Licchetta<sup>7</sup> · Cornelia Kornblum<sup>8</sup> · Saskia B. Wortmann<sup>9</sup> · H el ene Dollfus<sup>10</sup> · Maria T. Papadopoulou<sup>11</sup> · Alexis Arzimanoglou<sup>11,12</sup> · Maurizio Scarpa<sup>13</sup> · Holm Graessner<sup>14</sup> · Teresinha Evangelista<sup>15</sup>

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N° answers

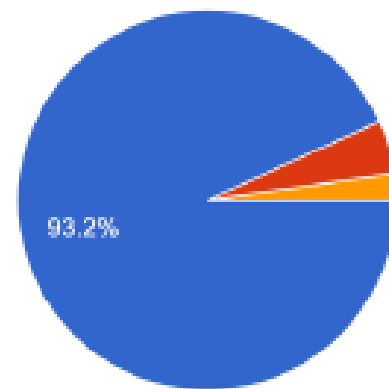


Created with mapchart.net



Do you think that PMDs have an important role in clinical practice?

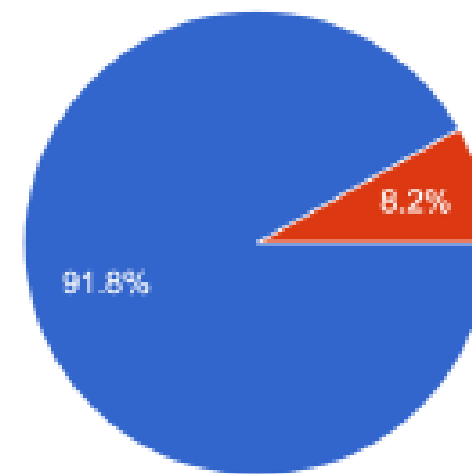
220 responses



- Yes
- No
- Do not know

Do you follow patients with PMDs

220 responses

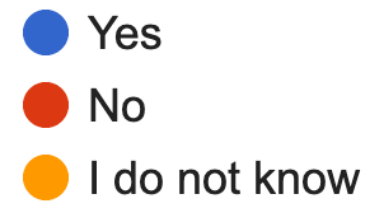
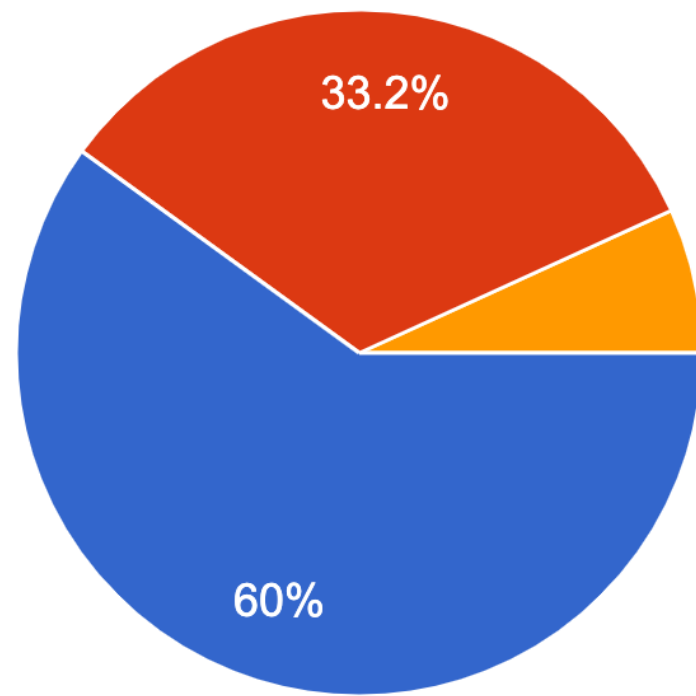


- Yes
- No



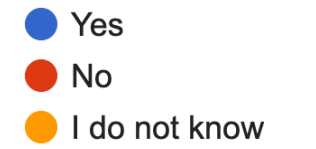
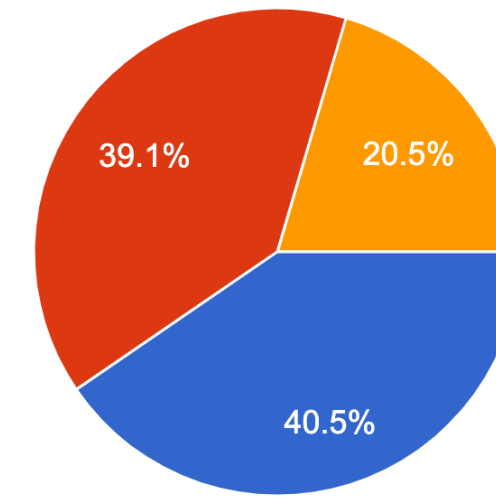
# Do you prescribe the mitochondrial cocktail to your PMD patients

220 responses



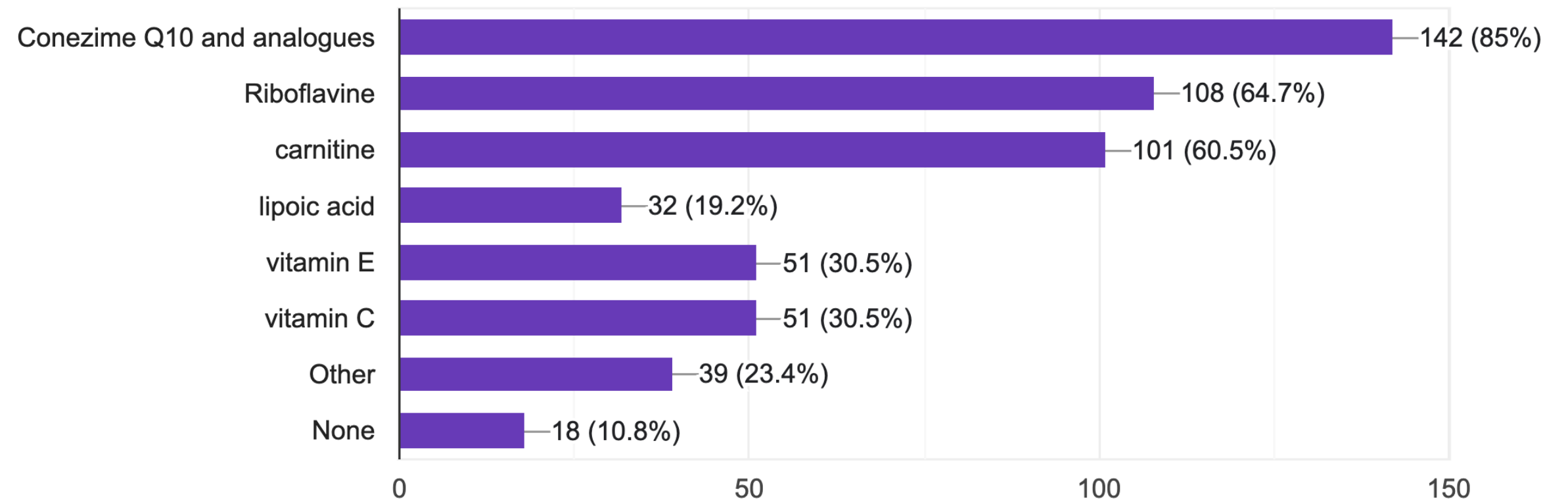
# Is the mitochondrial cocktail reimbursed by the health system of your Country?

220 responses



# if you prescribe the mitochondrial cocktail, what do you usually consider (multiple choices allowed)

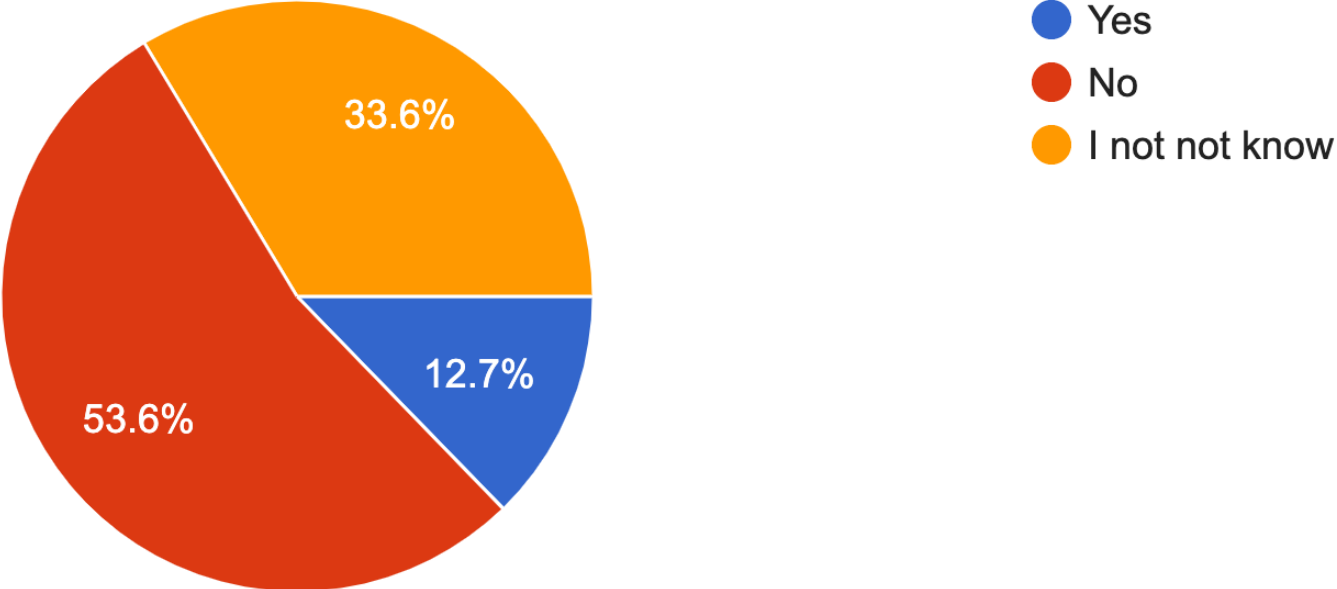
167 responses





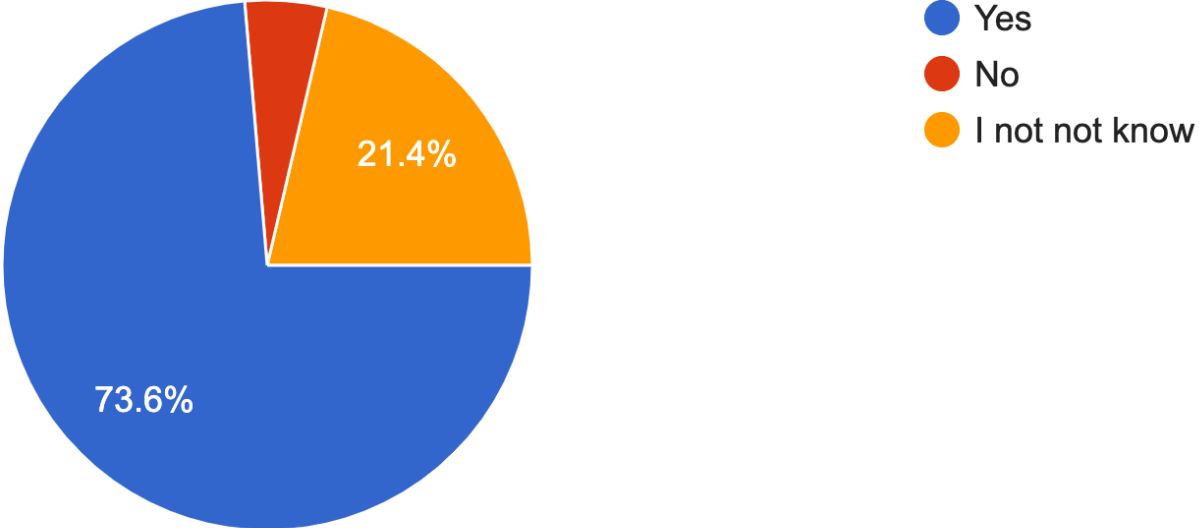
# Are you happy with the ICD-10 codes for classifying PMDs discharged from your HCP?

220 responses



# Do you think PMDs deserve specific ICD codes for reimbursement reasons or for better attention and tailored healthcare for these patients

220 responses



## **International Consensus Workshop on the Management of Seizures in Patients with Mitochondrial Diseases**

**FUNDED BY EJP RD Networking Support Scheme**

**Treatment of mitochondrial epilepsy is challenging** even for expert epileptologists. Examples of this difficulty are the risk of valproate toxicity and propofol infusion syndrome. Reason why standards of care need to be developed and disseminated.

**A panel of experts in mitochondrial medicine, representative of the five ERNs will meet in Budapest.** During this workshop, the experts will produce a consensus paper on the management of epilepsy in mitochondrial diseases. Its dissemination and a plan for updating and curating the consensus paper will also be established.



# The education needs



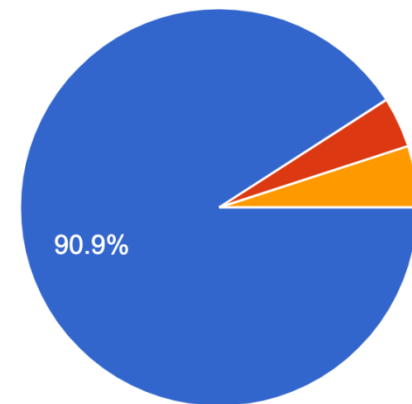
## Current management of primary mitochondrial disorders in EU countries: the European Reference Networks survey

Michelangelo Mancuso<sup>1</sup> · Pierluigi Lopriore<sup>1</sup> · Costanza Lamperti<sup>2</sup> · Thomas Klopstock<sup>3,4,5</sup> · Shamima Rahman<sup>6</sup> · Laura Licchetta<sup>7</sup> · Cornelia Kornblum<sup>8</sup> · Saskia B. Wortmann<sup>9</sup> · Hélène Dollfus<sup>10</sup> · Maria T. Papadopoulou<sup>11</sup> · Alexis Arzimanoglou<sup>11,12</sup> · Maurizio Scarpa<sup>13</sup> · Holm Graessner<sup>14</sup> · Tereziha Evangelista<sup>15</sup>

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Is there a need for education and training in PMDs?

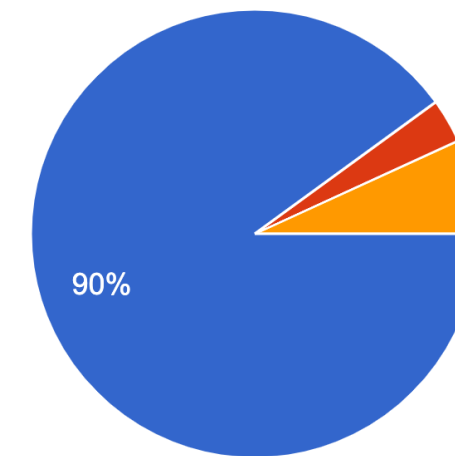
220 responses



● Yes  
● No  
● I do not know

Is there a need for education and training in the residency programmes in PMDs

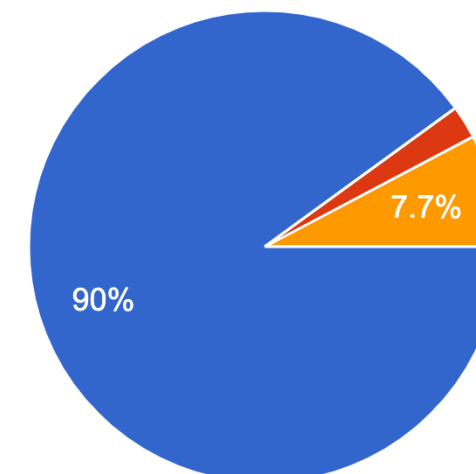
220 responses



● Yes  
● No  
● I do not know

Do you think ERNs must provide more education on PMDs through different initiatives

220 responses



● Yes  
● No  
● I do not know

# Webinars e academy

- **EURO-NMD Webinar series (February 2022: „mitochondrial month“)**
  - An introduction into mitochondrial disease in adulthood, Cornelia Kornblum **03.02.2022**
  - Primary mitochondrial myopathies, Michelangelo Mancuso **17.02.2022**
  - Targeted therapies for Leigh syndrome: Systematic review and steps towards a *Treatabolo*, Rita Horvath **24.02.2022**
- **EURO-NMD Webinar series (September 2022: „mitochondrial awareness week“)**
  - Mitochondrial Epilepsy, Michelangelo Mancuso, **01.09.2022**
  - Mitochondrial Movement Disorders, Chiara LaMorgia, **21.09.2022**
  - Mitochondrial Genetics, Costanza Lamperti, **22.09.2022**
  - Mitochondrial Optical Neuropathies, Chiara LaMorgia, **29.09.2022**



**European Reference Network**  
for rare or low prevalence complex diseases

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

# EURO-NMD Academy

Your session has t

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## Course categories

Top Level Categories

- Category: MITOCHONDRIAL DISORDERS
- Category: DYSTROPHINOPATHIES
- Category: NEUROMUSCULAR JUNCTION
- Category: INFLAMMATORY MYOPATHIES



# Group projects 2024-25

- CPMS
- Guidelines
- Educational activities
- Education (Curriculum, in collaboration with Marianne DeVisser)
- Awareness
- ICD codes
- Collaboration with EAN and European Society for Mitochondrial Research ([www.e-mit.org](http://www.e-mit.org)), and more

