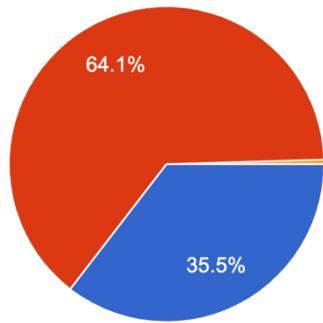


ERNs Survey on Primary mitochondrial diseases: Results

GENDER

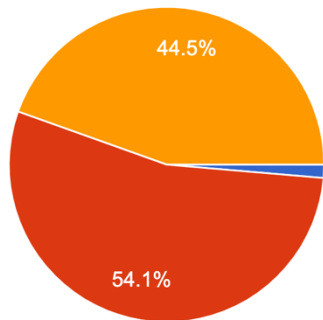
220 responses



- MALE
- FEMALE
- Prefer not to say

AGE RANGE (YEARS)

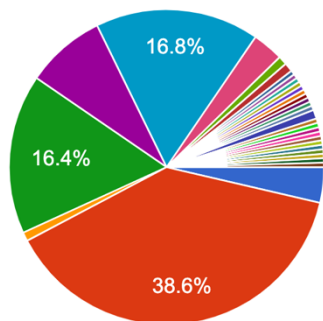
220 responses



- UNDER 30
- 30-50
- MORE THAN 50

Current professional situation:

220 responses

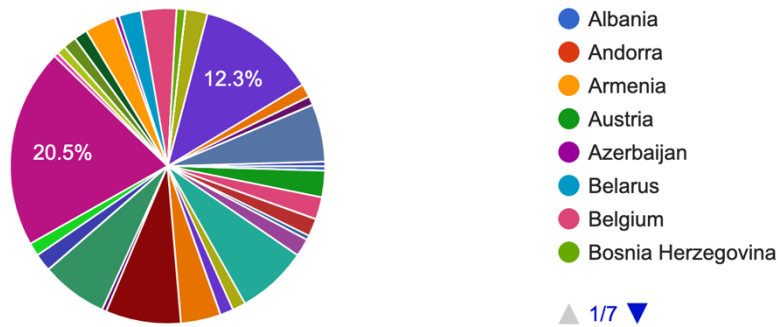


- NEUROLOGY RESIDENCY PROGRAM
 - NEUROLOGIST AFFILIATED TO HOSPITAL
 - NEUROLOGIST IN PRIVATE PRACTICE
 - NEUROPEDIATRICIAN
 - CLINICAL GENETIST
 - OTHER MD SPECIALIST
 - ePAG representative
 - PM
- ▲ 1/4 ▼

ERNS Survey on Primary mitochondrial diseases: Results

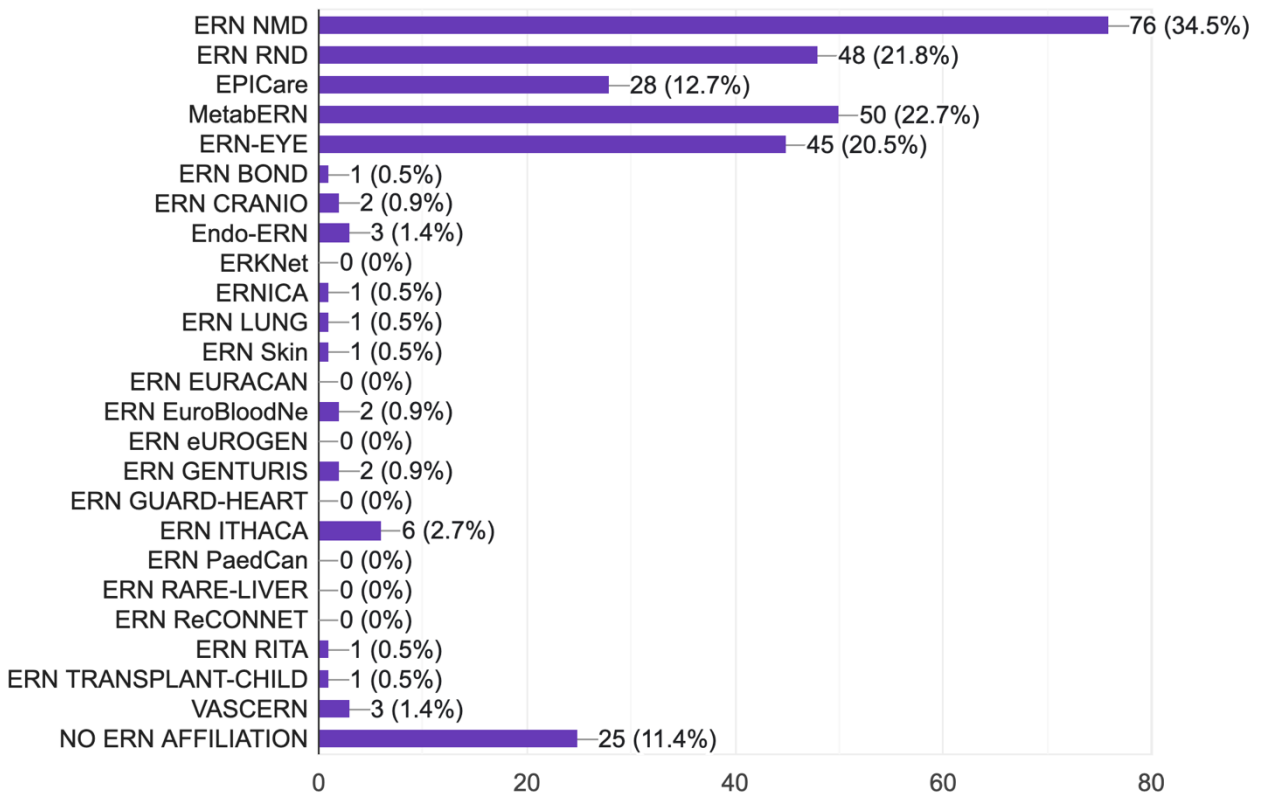
COUNTRY WHERE YOU PRACTICE (COUNTRY WHERE YOU LIVE IF YOU ARE A PAG member)

220 responses



ERN(S) YOU ARE AFFILIATED (multiple choices allowed)

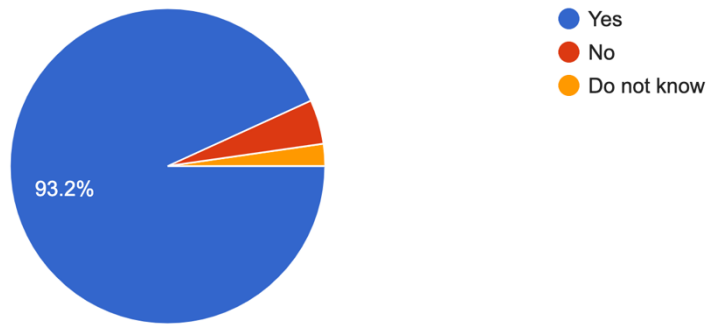
220 responses



ERNs Survey on Primary mitochondrial diseases: Results

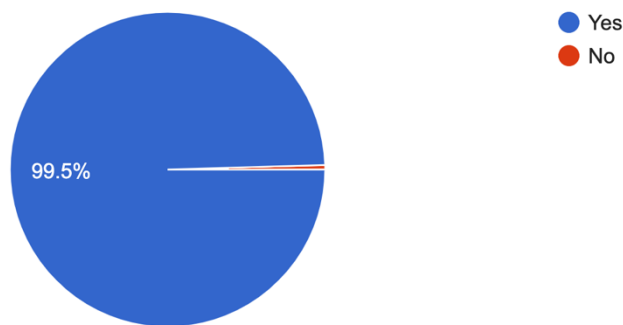
Do you think that PMDs have an important role in clinical practice?

220 responses



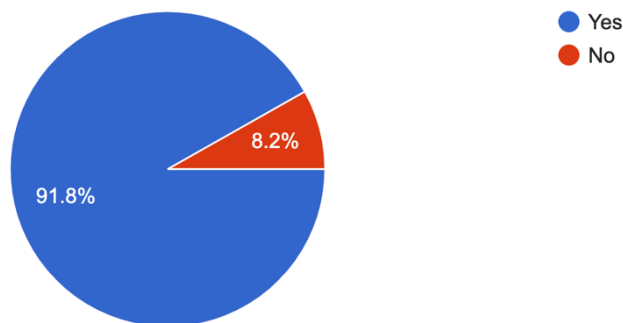
Are you aware of PMDs

220 responses



Do you follow patients with PMDs

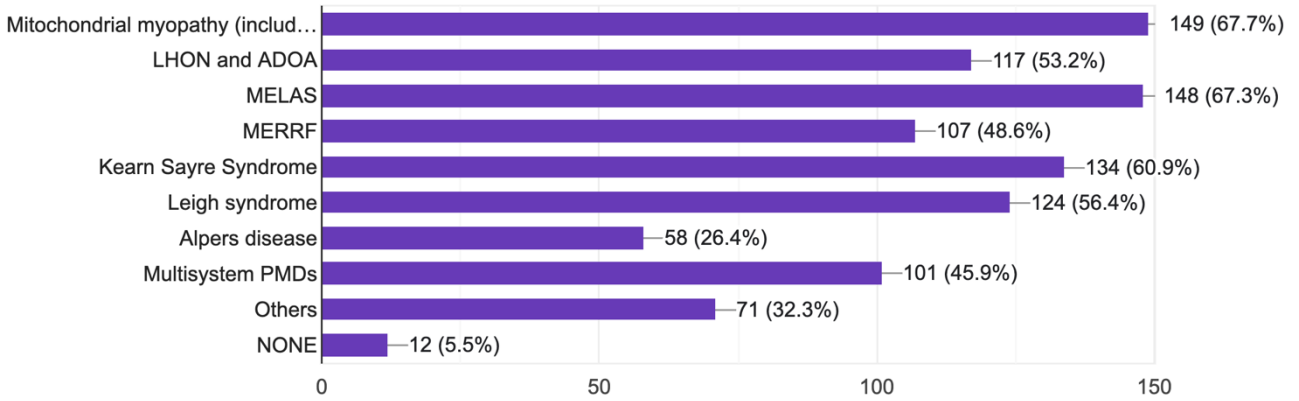
220 responses



ERNs Survey on Primary mitochondrial diseases: Results

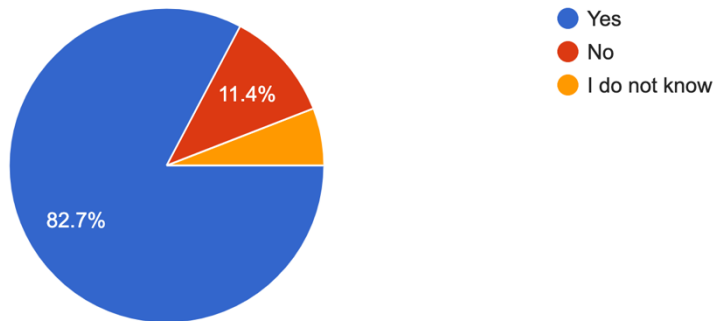
If you follow patients with PMDs, which one of the listed groups are you familiar with (multiple choices allowed)

220 responses



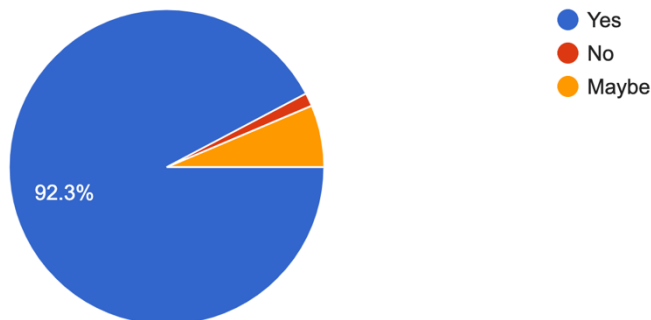
Do you think that a family history, including matrilinear inheritance, is usually collected?

220 responses



Do you think that family history is an important finding in the diagnostic flowchart?

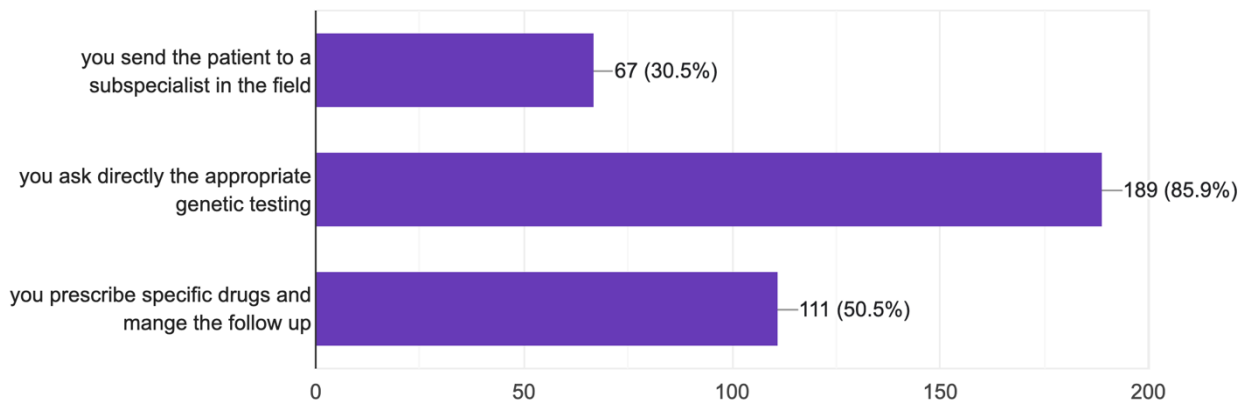
220 responses



ERNs Survey on Primary mitochondrial diseases: Results

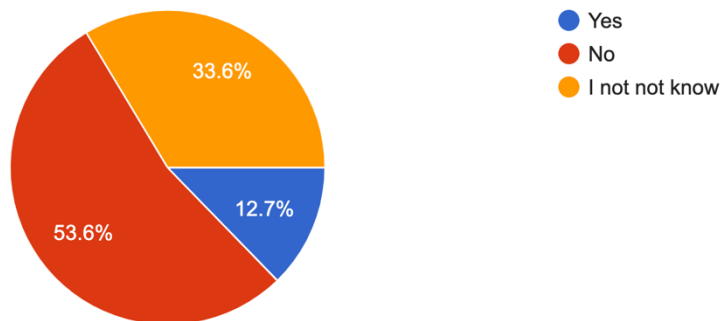
When do you have a patient with a possible PMD, what is your attitude (multiple choices allowed):

220 responses



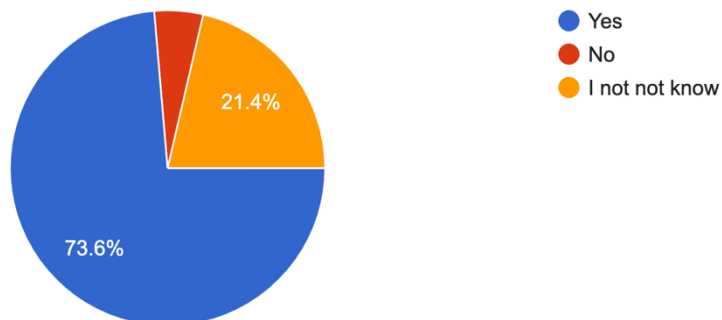
Are you happy with the ICD-10 codes for classifying PMDs discharged from your HCP?

220 responses



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

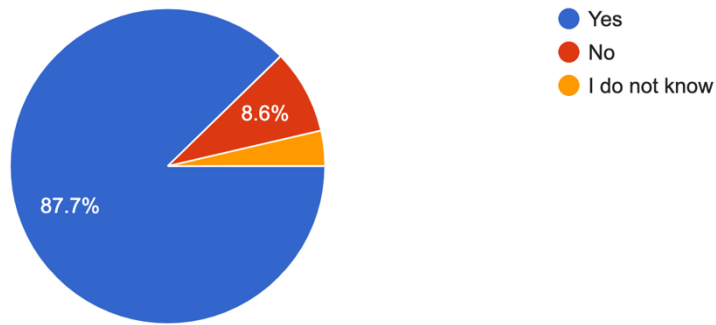
220 responses



ERNs Survey on Primary mitochondrial diseases: Results

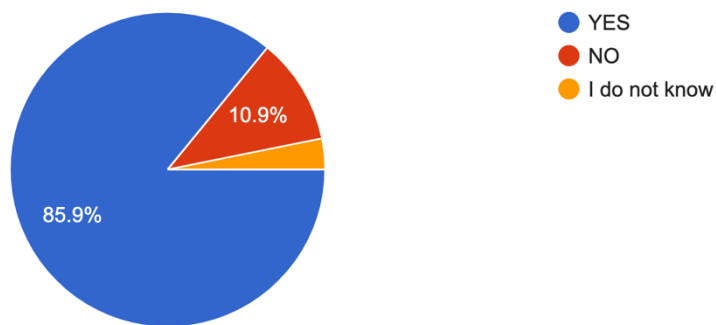
Muscle biopsy available at your HCP

220 responses



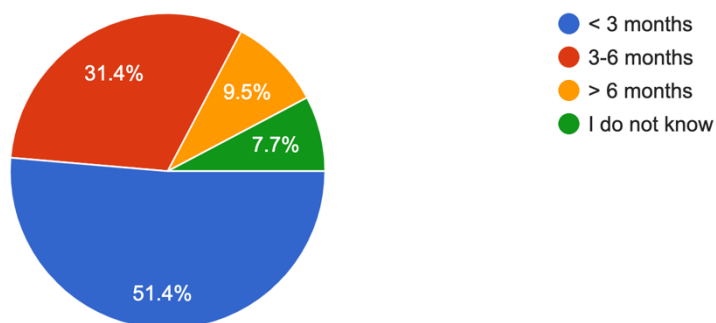
Single gene analysis available at your HCP

220 responses



Single Gene analysis RESPONSE waiting time

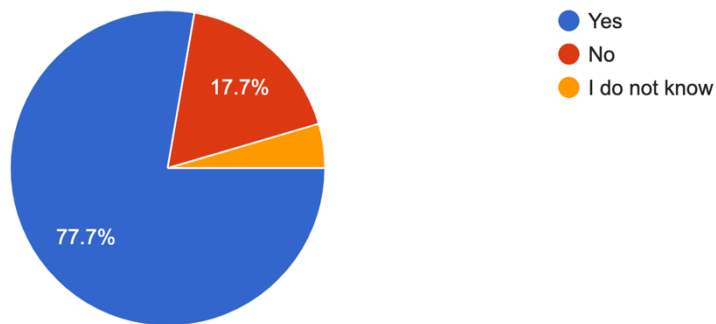
220 responses



ERNs Survey on Primary mitochondrial diseases: Results

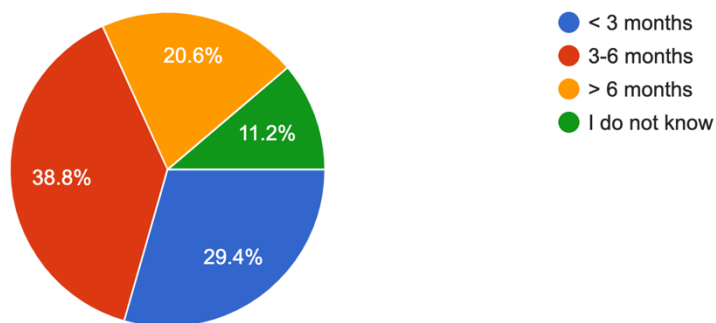
NGS Panels for PMDs available at your HCP

220 responses



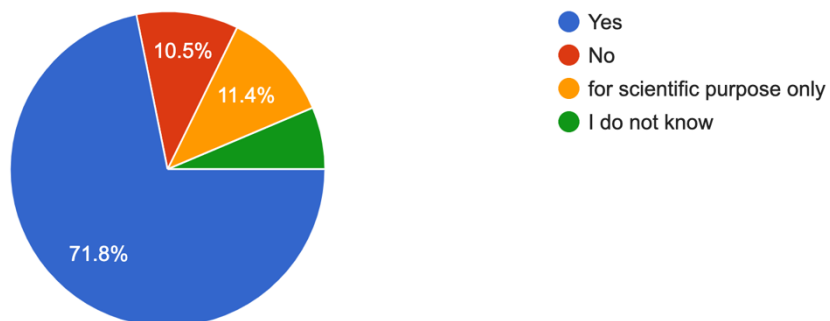
NGS Panels for PMDs RESPONSE waiting time

214 responses



Exome available at your HCP

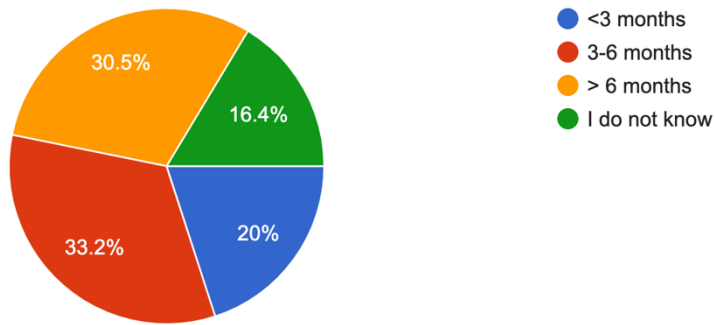
220 responses



ERNs Survey on Primary mitochondrial diseases: Results

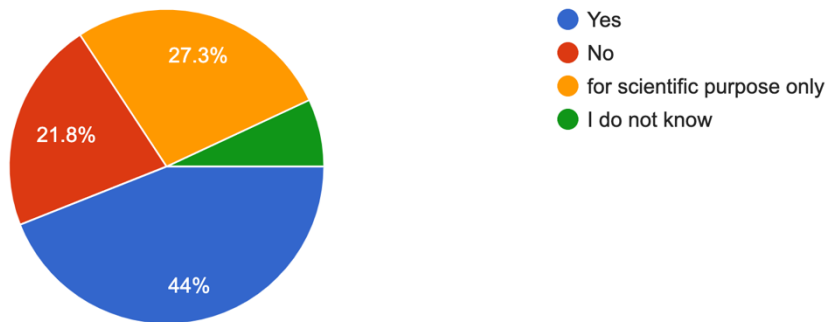
Exome for PMDs RESPONSE waiting time

220 responses



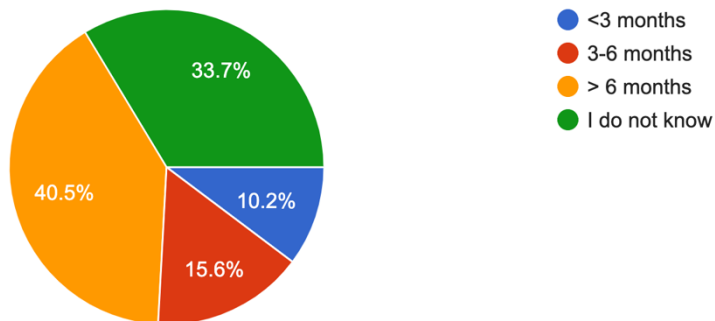
Whole Genome (WGS) available at your HCP

216 responses



WGS for PMDs RESPONSE waiting time

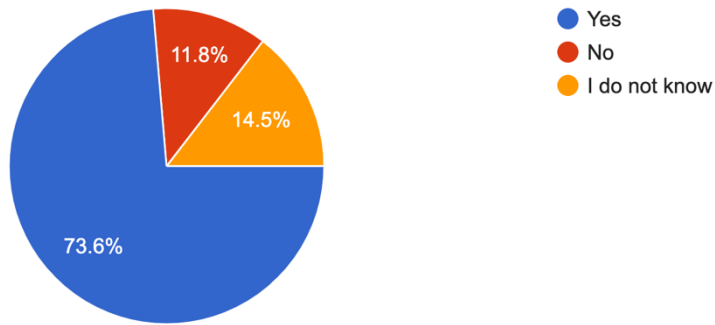
205 responses



ERNs Survey on Primary mitochondrial diseases: Results

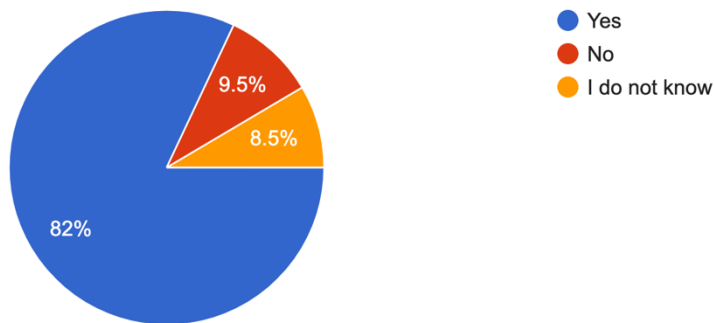
IS THE PRESYMPTOMATIC DIAGNOSIS SERVICE AVAILABLE IN YOUR COUNTRY?

220 responses



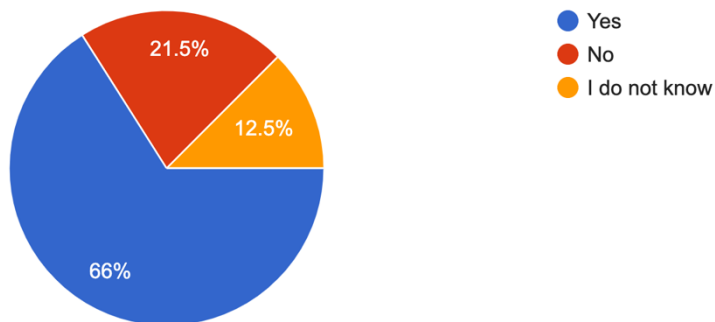
IS THE PRESYMPTOMATIC DIAGNOSIS PRECEDED BY A MEDICAL GENETIC COUNSELING CONSULTATION?

200 responses



IS THE SPECIALIST WITH EXPERTISE IN PMDs ALLOWED TO PRESCRIBE GENETIC TESTING IN PRESYMPTOMATIC SUBJECTS?

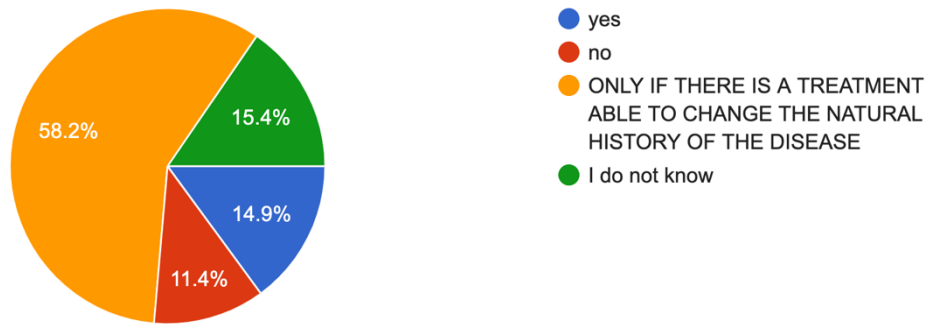
200 responses



ERNs Survey on Primary mitochondrial diseases: Results

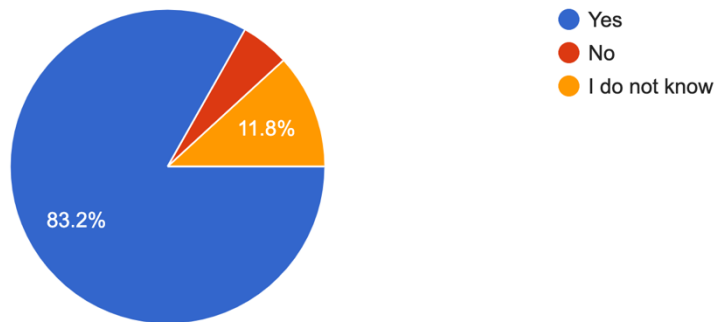
IS THE PRESYMPTOMATIC DIAGNOSIS SCREENING PERFORMED IN MINORS (<18-YRS)?

201 responses



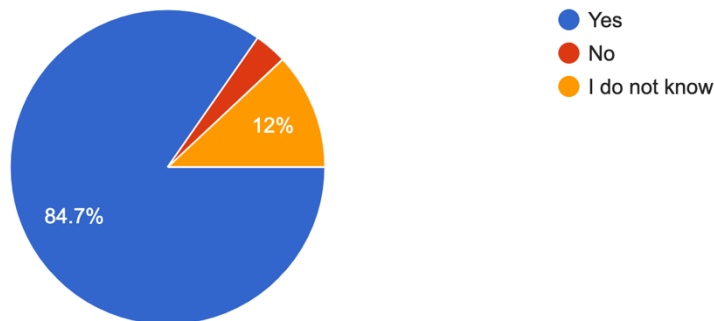
IS THE PRENATAL DIAGNOSIS SERVICE AVAILABLE IN YOUR COUNTRY?

220 responses



IS THE PRENATAL DIAGNOSIS PRECEDED BY A MEDICAL GENETIC COUNSELING CONSULTATION?

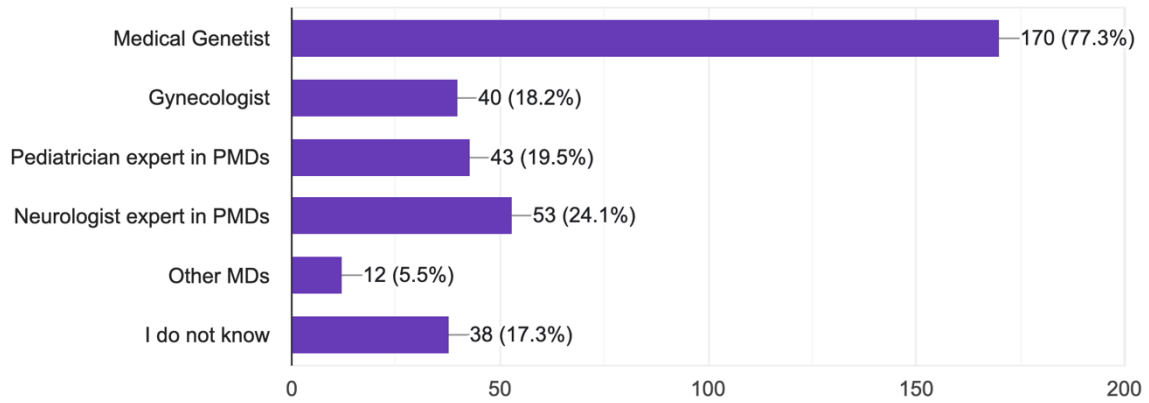
209 responses



ERNs Survey on Primary mitochondrial diseases: Results

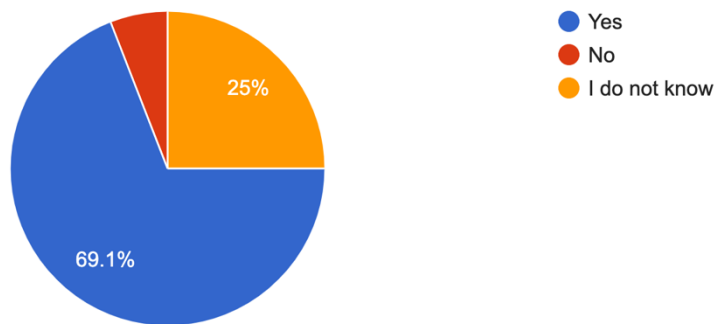
Which specialist is allowed to prescribe genetics test for prenatal diagnosis in your country (multiple choices allowed)

220 responses



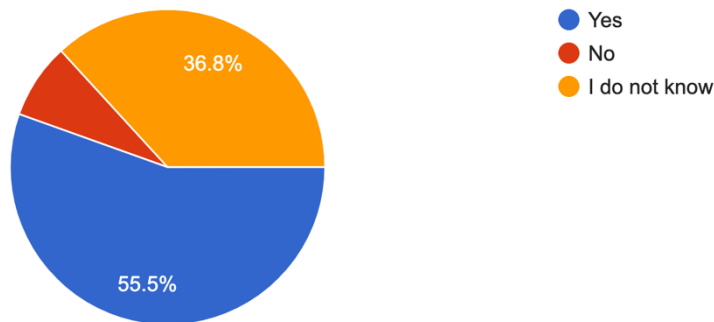
IS PGD AVAILABLE IN YOUR COUNTRY?

220 responses



Is there a law of the management of PGD in your country?

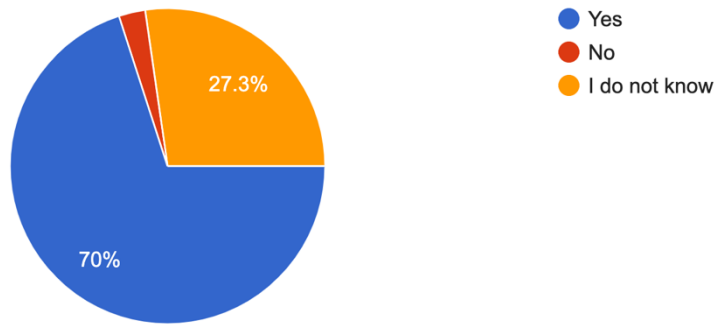
220 responses



ERNs Survey on Primary mitochondrial diseases: Results

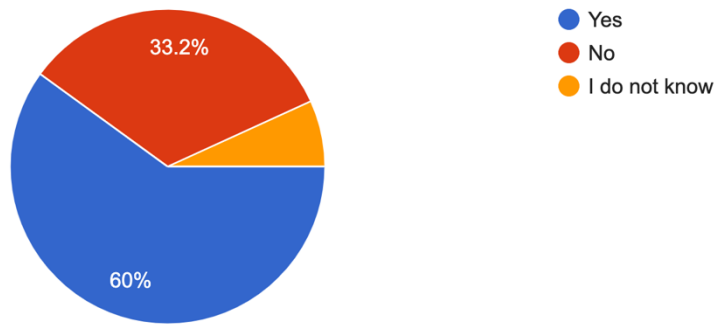
IS PGD PRECEDED BY A MEDICAL GENETIC COUNSELING CONSULTATION?

220 responses



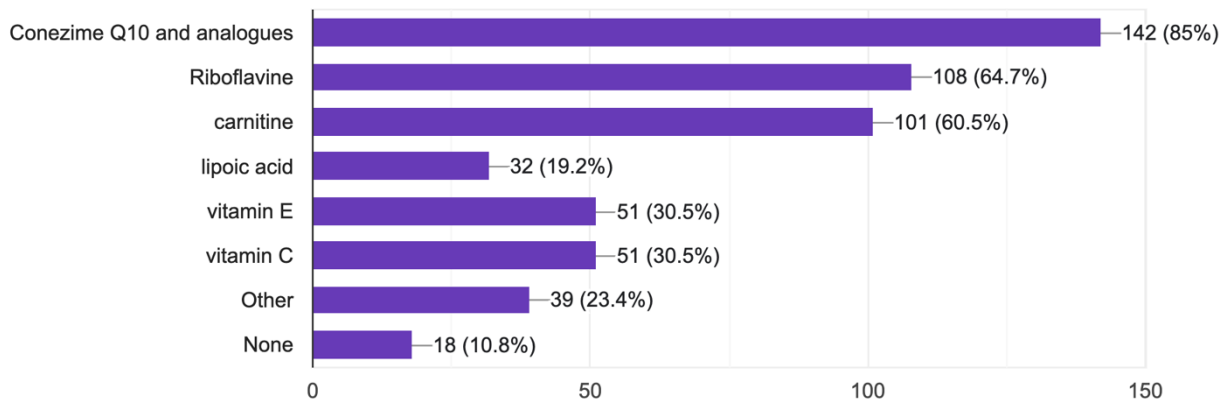
Do you prescribe the mitochondrial cocktail to your PMD patients

220 responses



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

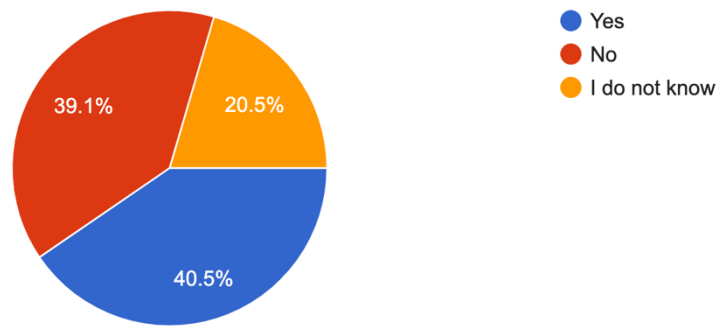
167 responses



ERNs Survey on Primary mitochondrial diseases: Results

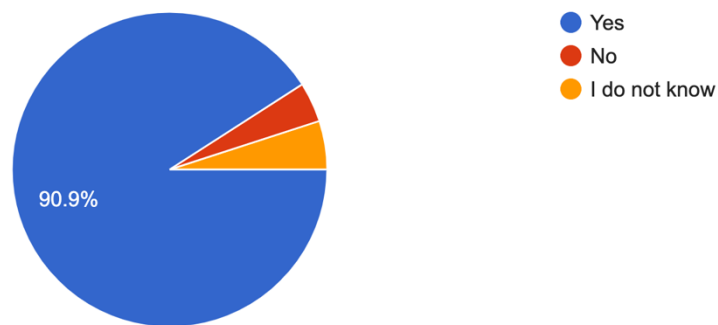
Is the mitochondrial cocktail reimbursed by the health system of your Country?

220 responses



Is there a need for education and training in PMDs?

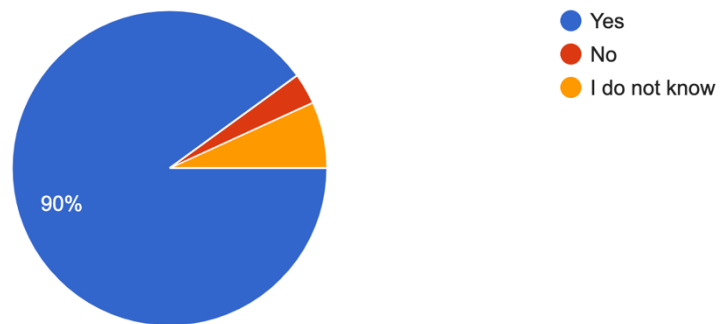
220 responses



ERNs Survey on Primary mitochondrial diseases: Results

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

220 responses



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

220 responses

