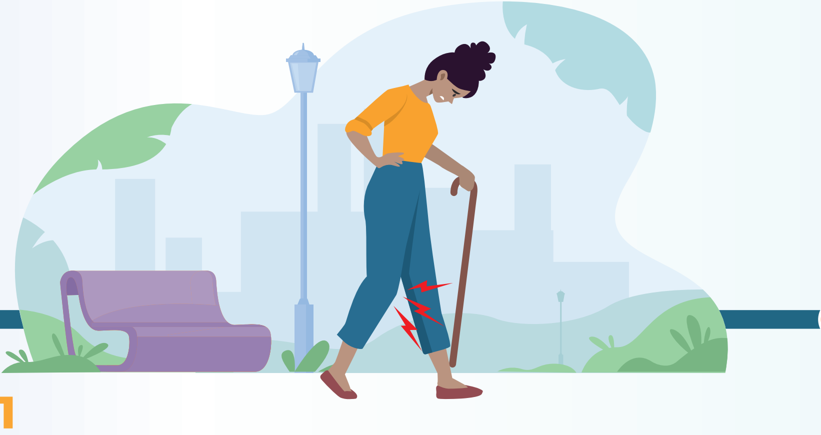
Primary Mitochondrial Myopathies *(Adults only)*
Patient Journey

European Reference Network
for rare or low prevalence complex diseases
Network
Neuromuscular
Diseases (ERN EURO-NMD)

Patient
Advisory
Board



STEP 1
EARLY SIGNS AND SYMPTOM ONSET

- Typically early, often in childhood or adolescence
- Physical activity intolerance during daily activities: exercise intolerance, walking difficulties, difficulty in sports or even light exercise
- Neurological signs (e.g., weakness, falls, clumsiness)
- Eye symptoms like ptosis and double vision (specific signs)

TAKE ACTION IF YOU:

- Notice **persistent or worsening symptoms**
  → Document symptoms (frequency, triggers)

STEP 2
DAILY LIFE IMPACT

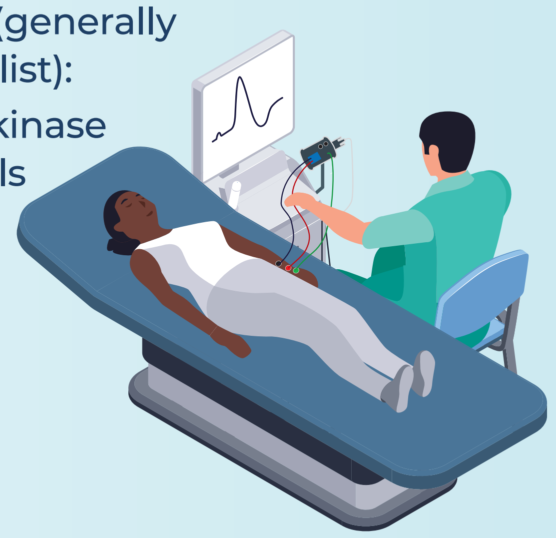
(Key Challenges)

- Fatigue and exercise intolerance
- Muscle weakness and pain
- Visual disturbances
- Employment and work adjustment, difficulties in household tasks
- Emotional and social difficulties (underreported but significant)
- Need for assistive devices and aids

STEP 3
SEEKING DIAGNOSIS

(what to do):

- **Visit a primary care physician** with a detailed list of symptoms
- **Request referral to a neuromuscular specialist** (preferably refer to a center experienced in mitochondrial disorders, e.g. seek for EURO-NMD centers, see QR Code)
- **Key tests suggested** (generally referred by the specialist):
  - Blood test, creatine kinase (CK) and lactate levels
  - Electromyography (EMG)
  - Genetic testing
  - Muscle biopsy (when needed)
  - MRI (when needed)



STEP 4
AFTER DIAGNOSIS

WHICH SPECIALISTS TO ENGAGE:

- **Neurologist** - main coordinator of care
- **Geneticist** - for diagnostic confirmation and family planning advice
- **Rehabilitation specialist** - to maintain and/or improve mobility
- **Psychologist** - to provide emotional support and help prepare for living with a chronic disease (patients often feel overwhelmed, sad or hopeless)
- **Clinical dietician** – for nutritional management (see the Nutrition Flyer in the QR code)
- **Speech therapist** - if speech/swallowing issues arise
- Other specialists responsible for managing multisystem involvement when needed (i.e. cardiologist, endocrinologist, pneumologist, gastroenterologist, nephrologist)
- **Look for referral to European reference centers and more information about the pathology** (see QR Code)

STEP 5
COMMUNICATION AND SUPPORT

- Patient and provider collaboration: open dialogue between patients, caregivers, and healthcare professionals.
- Coordinated care: A multidisciplinary team-based approach ensures comprehensive and effective management

STEP 6
TREATMENT AND DISEASE MANAGEMENT

- There is currently no definitive cure, but ongoing clinical trials and research offer hope for future treatments!
- **Seek support through mitochondrial diseases associations, specialized ERN centers** (see QR Code)
- **Seek specific mechanical supports** if needed, such as: power chairs, walkers, scooters, wheelchairs, or other supports suggested by the specialists.
- **Keep physically and mentally active**
- **Follow personalized care plan**
- **Follow specific nutrition** (see QR code for nutritional information)
- **Symptomatic treatments** to improve quality of life
- **Specialized treatment (to be determined by the clinician)**
- **Voluntary participation in clinical trials** – Patients are not obligated to take part, but it is an available option when possible (See QR Code)

STEP 7
CAREGIVER'S ROLE

HOW CAREGIVERS CAN HELP:

- Ensure special treatment and follow-up
- Monitor nutrition and medication adherence
- Emotional support and advocacy: attend appointments, join patient organizations
- Ensure social participation: help prevent isolation
- Prepare home adaptations (if needed)
- Connect with caregiver support networks
- Plan for gradual disease progression
- Assist with mobility: mechanical supports, walking aids, etc. when needed

More information here:

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