# Mitochondrial disorders: a descriptive study of a Tunisian pediatric series

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## Introduction

- Mitochondrial disorders (MD) represent a heterogeneous group of **rare** and **often misdiagnosed** metabolic disorders.
- There are **few descriptive studies** of large pediatric series both at the national and international level.
- Our objective was to describe the **demographic**, **clinical**, **paraclinical** and **evolutionary** characteristics of a Tunisian cohort followed for MD.

## Methods

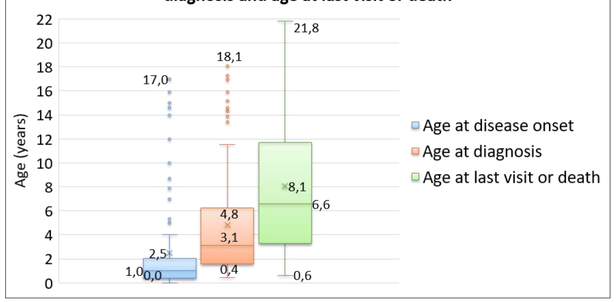
- Longitudinal **retrospective** study of patients followed in the pediatric neurology department at the National Institute Mongi Ben Hmida of Neurology in Tunis between **2004** and **2021** for MD.
- Diagnosis was made in presence of an evocative clinico-radiological presentation with biochemical evidence of energetic deficit.
- Demographic, clinical, paraclinical and evolutionary data were collected and analyzed. The different clinical phenotypes were identified.

## Results

### Demographic data: (Figure 1)

- 101 patients from 81 families (Sex-ratio: 1.1)
- Parental consanguinity: 72% of families
- Age of onset: ≤2 years in 75% of patients

**Figure 1: Distribution of patients according to age at onset, age at diagnosis and age at last visit or death**




### Clinical data: (Figure 2 and 3)

**Figure 2: Clinical manifestations**

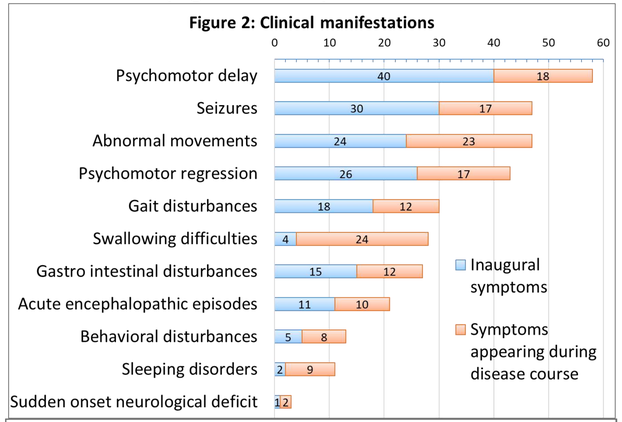
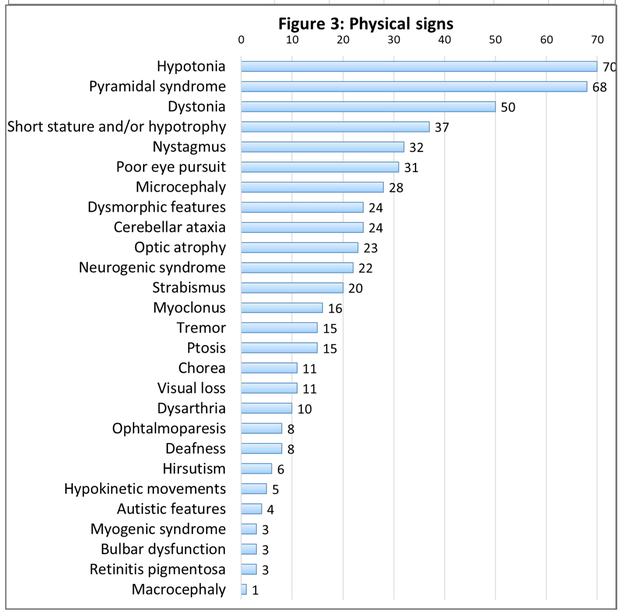



**Figure 3: Physical signs**




### Radiological findings: (Table1)

Brain magnetic resonance imaging (MRI): abnormal in 86 /97cases (88%)

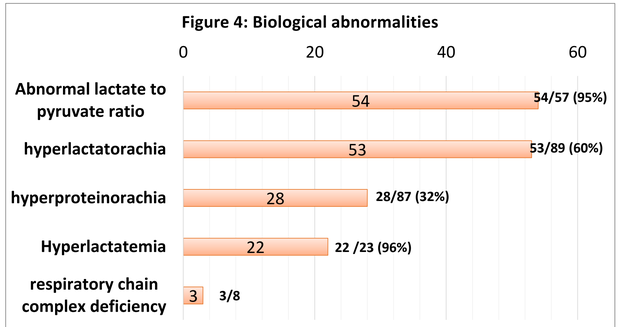
Spectroscopy: abnormal in 27/41 cases (66%)

**Table 1: Brain MRI abnormalities**

| Abnormalities | | Number of patients |
|---|---|---|
| Bilateral and symmetrical abnormal intensities | Basal ganglia | 47 |
| | Putamen | 39 |
| | Caudate nucleus | 30 |
| | Pallidum | 25 |
| | Thalamus | 17 |
| | Locus niger | 7 |
| | Subthalamic nucleus | 7 |
| | White matter | 36 |
| | Brainstem | 31 |
| | Cerebellum | 19 |
| | Corpus callosum | 8 |
| | Cortex | 6 |
| | Spinal cord | 4 |
| Atrophy | Cortical and/or subcortical atrophy | 28 |
| | Cerebellar atrophy | 12 |
| | Corpus callosum | 7 |
| Cavitations and cysts | | 11 |
| Hypoplasia or agenesis of corpus callosum | | 6 |

### Biochemical data: (Figure 4)

**Figure 4: Biological abnormalities**



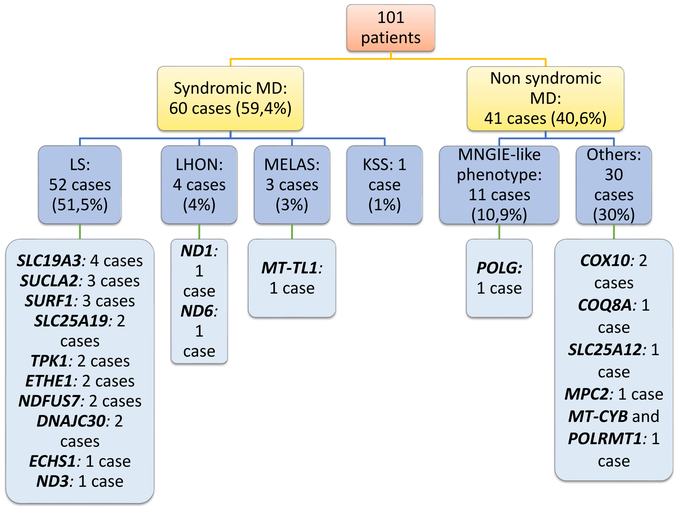

### Electrophysiological data: (Table 2)

**Table 2: Electrophysiological findings**

| Abnormalities | | Number of patients |
|---|---|---|
| Electroencephalogram (57 cases) | Background abnormalities | 36 |
| | Epileptic discharges | 29 |
| Electromyography (74 cases) | Neurogenic changes | 20 |
| | Neurogenic and myogenic changes | 7 |
| | Myogenic changes | 3 |
| Altered auditory evoked potentials (39 cases) | | 19 |
| Altered visual evoked potentials (45 cases) | | 17 |

### Phenotypes and genetic abnormalities: (Figure 5)

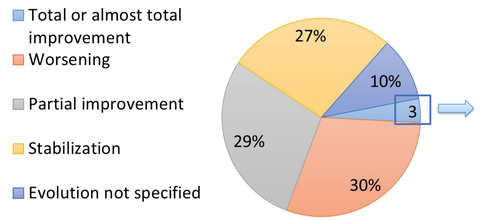
**Figure 5: Main phenotypes and genetic abnormalities**




Abbreviations: KSS: Kearns Sayre syndrome, LHON: Leber hereditary optic neuropathy, LS: Leigh syndrome, MD: mitochondrial disorders, MELAS: mitochondrial encephalopathy with lactic acidosis and stroke-like episodes, MNGIE: Mitochondrial neurogastrointestinal encephalopathy

### Clinical course: (Figure 6)

**Figure 6: Evolution in patients under energetic medications (77 patients)**




Patients followed for thiamine metabolism dysfunction syndromes and treated early with thiamine and biotine (few days after disease onset)

## Conclusions

Our study highlights the clinical and radiological heterogeneity of MD. LS remains the most frequent MD. Genetic confirmation should not delay initiation of therapy with **Thiamine and Biotin in any case with LS**, which may improve the prognosis in case of thiamine metabolism dysfunction syndrome.