



# Childhood Myopathy: Service Experience Neurology at Ignace Deen National Hospital and Simbaya Neurological Institute

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## Abstract

Childhood myopathy (CM) is rarely reported in sub-Saharan Africa. The objective of this study was to describe the clinical, paraclinical and therapeutic aspects of children with myopathies and to assess their quality of life as well as that of their parents. This is a mixed study with a retrospective part of 3 years and a prospective cross-sectional period of one month of children aged 2 to 17 years. Our variables were clinical, paraclinical and therapeutic. The quality of life of children and parents was assessed by the Euroqol and PAR-QOL scales respectively. Statistical analysis was performed using R studio software. The study involved 17 cases of EDM, *i.e.* a frequency of 1.4%; the mean age was  $8 \pm 4.2$  years. The M/F sex ratio was 1.8 with a male predominance of 64.7%. Amyotrophy was 58.8% of cases, 8 gait disorders 47.2%, calf hypertrophy 35.2%, breathing difficulties 29.4%, intestinal disorders 17.6%, oculomotor disorders 11.7%, cardiac disorders 5.8%. CPKs were increased in 13 patients; 88.2% had high LDH. The electroneuromyograms performed in our patients showed myogenic tracings. The evaluation of the quality of life by the PAR QOL scale concluded has an average impact on the general quality of life with an average score of 2.87. MDEs are underestimated in our context. Patients consult at an advanced stage, thus favoring late treatment. This conjunction of facts considerably alters their quality of life.

## Subject Areas

Pediatrics

## Keywords

MDE, Electroneuromyogram, BY QOL, HNID

## 1. Introduction

Neuromuscular diseases are ubiquitous diseases and therefore present in all ethnic communities and at all latitudes [1].

Progressive forms appearing in children include Duchenne muscular dystrophy (DMD) which is an X-linked recessive disease affecting between 1/3600 and 1/6000 male live births. Spinal muscular atrophy (SMA) is an autosomal recessive disease with a spectrum of disease severity, and a number of other neuromuscular conditions [2].

Childhood muscular dystrophy (MDE) is rarely reported in Africa, particularly in its sub-Saharan region [3]. Rare studies published in South Africa (1994) showed a prevalence of 1/100,000 for Duchenne muscular dystrophy and 1/755,000 for Becker muscular dystrophy [4].

However, a study carried out by the French Society of Myology on myopathies in West Africa showed an overall molecular confirmation rate of 45.3% [5].

In Guinea, to our knowledge, no studies on these conditions in children have been found.

The symptomatology of MDE is dominated by the classic myogenic syndrome characterized by: muscle weakness, myalgia and cramps, proximal and bilateral motor deficit, amyotrophy or more rarely hypertrophy, hypotonia, hyporeflexia, tendon retractions [6].

The most widely used diagnostic tools in patients suspected of myopathy are: serum creatine phosphokinase (CPK) assay, electroneuromyogram (ENMG), muscle biopsy, magnetic resonance imaging (MRI) and blood. Molecular genetics has DNA extraction [5].

In the absence of curative treatment, the current management of these chronic pathologies is essentially palliative. However, improving the medical follow-up of these patients can increase their life expectancy. Myopathic patients are highly dependent on those around them. No study has focused on measuring the health-related quality of life of caregivers who regularly care for these people [7].

The objective of this study was to describe the clinical, paraclinical and therapeutic aspects of children with myopathies and to assess the quality of life of these children and their parents.

## 2. Material and Methods

We conducted a mixed study with a retrospective part of 3 years (from January 1, 2017 to December 31, 2019) and a prospective cross-sectional period of one month (October 2020). The study involved all children aged 2 to 17 suffering from muscular dystrophy and followed within the framework of a consultation at the Neurological Institute of Simbayah and at the Neurology department of the National Ignace Deen Hospital in Conakry. We proceeded to the identification of the medical files of the patients followed for myopathy. These patients were summoned and re-examined by a neurologist and a neurology intern concomitantly to verify the diagnosis. The diagnosis of myopathy was made by a

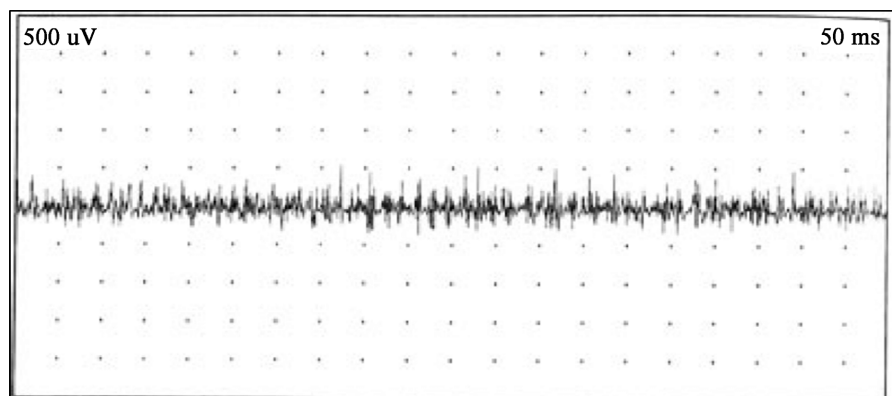
neurologist certified in pediatric neurology. All children underwent a neuro-pediatric examination. The diagnostic criteria were essentially clinical based on: a duration of the disease greater than 06 months, an onset beyond 01 years and proximal and distal muscle weakness; often associated with an elevation of CPK and a myogenic tracing on ENMG in accordance with the recommendations of the International Committee for Congenital Myopathies (ICMC) [8] (**Iconography 1** and **Iconography 2**).

### 2.1. Exclusion Criteria

Presence of one or more of the following: 1) human immunodeficiency virus



**Iconography 1.** Bilateral calf hypertrophy + bilateral Scapula allata.



**Iconography 2.** ENMG trace of an 8-year-old child with gait disorder, calf hypertrophy evolving since birth showing microvolt waves.

(HIV) infection, 2) obstructive hydrocephalus, 3) history of malignancy, 4) evidence of developmental regression, 5) diagnosis of a known genetic syndrome, 6) primary neuromuscular disorder or 7) major extracerebral birth defects. HIV serology was determined by chart review. All children enrolled in the study were HIV negative.

## 2.2. Data Gathering

We interviewed the mother of each child with a questionnaire including the following variables:

- socio-demographic data (age, sex, origin, level of education of parents).
- clinic (reasons for consultation as well as the circumstances of discovery, the severity of motor function disorders was graded according to the Gross Motor Function Classification System (GMFCS) [9]. The type of myopathy distributed according to the international classification of Walton in 1981 [8].
- At the paraclinical level, all the patients benefited from a dosage of muscle enzymes (CPK, LDH) and an additional biological assessment according to the clinical orientation. ENMG was systematic.
- The quality of life of children and caregivers was assessed using the Euroqol EQ and PAR-QOL scales [10] [11]. The Euroqol scale includes five items representing five dimensions: mobility, personal autonomy, daily activities, pain and discomfort, anxiety and depression. Each item has five response levels. The answers to these five questions describe the state of health as five consecutive numbers [10].

## 2.3. Interpretation of PAR-QOL Scores

The scores are transformed into a standardized score by dividing them by the total number of items that compose them. Thus the scores can vary from 1 (no impact at all) to a value of 5 (very impact).

To facilitate the interpretation of the results and to classify the scores by level of quality of life, the scores were grouped in the Grimm-Astruc study [11].

- Good quality of life: (score less than 2)
- Average quality of life: (score between 2 and 3.5)
- Low quality of life (score between 3.6 and 5)

## 2.4. Statistical Analyses

Statistical analysis was performed using Rstudio software version 1.1456. Descriptive statistics were performed for all variables.

## 2.5. Ethics

The study was approved and authorization was obtained from the ethics committee of the University Hospital of Conakry. Informed consent was obtained from the parents of each patient.

### 3. Results

Out of 1195 patients seen in consultation, 17 children presented with a clinical and electrophysiological myogenic syndrome, *i.e.* a frequency of 1.4%. The average age was  $8 \pm 4.2$  years with extremes of 2 and 17 years. The age group of 6 to 10 years was the most represented with 41.1%. There was a male predominance (64.7%) with a sex ratio M/F = 1.8. An urban origin was found in 64.7%. The average consultation time for our patients was 18.8 months. All patients had motor deficit and axial hypotonia, 10 atrophy (58.8%, 8 gait disturbances (47.2%), 6 hypertrophy (35.2%) (**Table 1**). Motor deficit according to the GMFCS score 10

**Table 1.** Distribution of patients according to socio-demographic and clinical characteristics.

Sociodemographic and clinical characteristics	Efficient	Frequency%
<b>years of age</b>		
2 - 5	6	35.2
6 - 10	7	41.1
11 years and over	4	23.5
Average: $8 \pm 4.2$ years		
<b>Sex</b>		
Boy	11	64.7
Daughter	6	35.3
Sex ratio: 1.8		
<b>Origin</b>		
Urban	11	64.7
Rural	6	35.3
<b>Reasons for consultation</b>		
Limb motor deficit	17	100
Axial hypotonia	17	100
Amyotrophy	10	58.8
Gait disorders	8	47.2
Calf hypertrophy	6	35.2
Eye disorders	2	11.7
Cardiac disorders	1	5.8
Intestinal disorders	3	17.6
Breathing difficulties	5	29.4
<b>Consultation period</b>		
$\leq 12$ months	5	29.4
$> 12$ months	12	70.5
Average: 18.8 months		

patients were at stage V; 5 at stage IV and 2 at stage III with an average of 4.4 (Table 2). The CPK level was increased in 13 patients, *i.e.* 76.4% (Mean: 790.5  $\pm$  680.5); 15 (88.2%) patients had elevated LDH levels (mean: 859  $\pm$  667.5). The 17 children had a clinical myogenic syndrome and were confirmed electrophysiologically by the demonstration of a rich and microvolt tracing. All our patients received physiotherapy and 10 (58.8%) corticosteroid therapy combined with vitamin therapy and one (5.88%) received an ACE inhibitor. Descriptive analysis of responses through the PAR QOL scale concluded that myopathy had a medium impact on overall quality of life with a mean score of 2.87 (Table 3) and (Figure 1).

#### 4. Discussion

Our study made it possible to highlight a hospital frequency of CM at 1.4% out of a workforce of 1195 patients. To our knowledge, this is the first study carried out in our context on CM. These pathologies are well described in developed countries with a well-codified international classification [12]. In Africa, very few studies have been carried out on ME given the difficulties of etiological diagnosis and management, the last clinical study dating back 35 years [3]. The limited access to ENMG and the high cost of CPK as well as the absence of a laboratory for the analysis of neuromuscular biopsies explain this scarcity of data. A male predominance was reported in our study which can be justified by the importance that the African community gives to the male sex. The average age of onset is 8.6  $\pm$  4.2 years. A multicenter study conducted in North Africa and Saudi

**Table 2.** Distribution of patients according to the GMFCS.

GMFCS	Number (n = 17)	Frequency%
Stage I	-	-
Stage II	-	-
Stage III	2	11.7
Stage IV	5	29.4
Stage V Average: 4.4	10	58.8

**Table 3.** Distribution of patients according to quality of life (Euroqol scale).

Euroqol	no problem not/%	Minor problems not/%	Moderate not/%	Strict not/%	Unable not/%	Student not/%
Mobility	-	-	5 (21.4)	3 (17.6)	2 (11.7)	7 (21)
Autonomy	1 (5.8)	6 (35.2)	5 (21.4)	3 (17.6)	2 (11.7)	-
Current activities	1 (5.8)	6 (35.2)	4 (23.5)	4 (23.5)	2 (11.7)	-
Pain	-	6 (35.2)	4 (23.5)	5 (29.4)	2 (11.7)	-
Anxiety	1 (5.8)	6 (35.2)	4 (23.5)	3 (17.6)	3 (17.6)	-



**Table 4.** Types of myopathies (clinical-biological classification).

	TYPES	Duchenne not (%)	belt-limbs not (%)	Becker not (%)	Facioscapulohumeral not (%)	Spinal atrophy (%)
<b>Kindly</b>	Man	3 (27.3)	4 (36.3)	3 (27.3)	1 (9.1)	-
	Feminine	-	2 (33.3)	1 (16.7)	-	3 (50)
<b>Clinical signs</b>	Motor deficit	3 (17.6)	6 (35.4)	4 (23.5)	1 (5.9)	3 (17.6)
	Axial hypotonia	2 (11.8)	-	6 (35.4)	3 (17.6)	6 (35.4)
	Amyotrophy	1 (5.9)	5 (29.4)	4 (23.5)	5 (29.4)	2 (11.8)
	Gowers signs	3 (23)	6 (46.2)	2 (15.4)	-	2 (15.4)
	Scapula Alata	3 (25)	6 (50)	2 (16.7)	1 (8.3)	-
	Calf hypertrophy	3 (75)	-	-	1 (25)	-
	<b>PCK</b>	Positive	3 (23.1)	4 (30.7)	2 (15.4)	1 (7.7)
	Negative	-	2 (50)	2 (50)	-	-
<b>LDH</b>	Positive	2 (13.4)	6 (40)	3 (20)	1 (6.6)	3 (20)
	Negative	1 (50)	-	1 (50)	-	-
<b>ENMG</b>	Myogenic trace	3 (17.6)	6 (35.4)	4 (23.6)	1 (5.8)	3 (17.6)
	Nope	-	-	-	-	-

The difficulties of the etiological diagnosis encountered in our context lead us to base the management on the monitoring of symptoms and on the attempt to slow down their evolution and their consequences [15].

Therefore, the choice relies quite heavily on the clinician's experience in this area. Corticosteroids remain, to date, the first-line treatment for several reasons: their entry into action is one of the fastest among the powerful immunomodulators; they are the best studied therapeutic agents and whose benefits are clearly documented. The rule is initial high-dose corticosteroid therapy (prednisone 1 mg/kg/day). Higher doses showed no benefit, mostly increasing side effects. In our context, quite classically, a good initial response to corticosteroids is observed, but at the same time, corticosteroid-dependent therapy of lesions and relapses affects the long-term prognosis [16].

Beyond corticosteroid therapy, other complementary treatments are necessary given the complexity of neuro-orthopedic, cardiac or respiratory damage.

Currently, there does not seem to be any formal scientific proof of the efficacy of a treatment, whether from a functional or orthopedic point of view [17]. However, technical aids and human resources to preserve physical functions (walking and standing) and quality of life must be put in place as soon as possible in order to make life easier for patients by giving them more autonomy [18] and at the same time ease the burden placed on those around them.

In addition to an analytical rehabilitation focused on the osteoarticular structures to preserve the flexibility of the rib cage (overall flexibility exercises, passive segmental mobilizations, muscle stretching) and on the respiratory muscles



to maintain their function (long-term strengthening of the inspiratory muscles, breathing exercises, incentive spirometry), rehabilitation is also based on the early use of mechanical ventilation. In order to maintain the intrinsic qualities of the lung and promote thoraco-pulmonary expansion, these mechanical ventilation supports are largely responsible for increasing the life expectancy of patients with myopathy over the past 20 years [19].

The negative impact of myopathies on the psychological state of patients and their families is not only related to the diagnosis, but also to the fluctuation of signs and symptoms according to the phases [20].

Perceived health measures established by the Euroqol tool are possible within a sample of people with myopathy. In our study, 35.2% of children had mild problems performing routine activities as well as other activities such as bathing and dressing. The parents' quality of life was moderately emotionally and adaptively degraded with an overall score of 2.87. Boyer *et al.* reported that 30% of parents of myopathic children expressed psychological harm and a clinically significant decrease in quality of life [7].

The ethical and psychological training of health professionals who must support families and patients with myopathies in order to avoid depression seems to be a matter of common sense [8].

## 5. Conclusion

In our context, the clinical symptomatology of CM was dominated by the complete occurrence of a myogenic syndrome. Beyond the diagnostic problem, the late management of this chronic disabling pathology has a negative impact not only on the quality of life of the child by reducing his autonomy, but also on that of the parents emotionally and adaptively. The establishment of infrastructures allowing the realization of muscular biopsies and their consequent analysis could be an asset for the etiological diagnosis in order to direct a good care.

## Conflicts of Interest

The authors declare no conflicts of interest.

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