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Clinical Profile of Muscular Dystrophies in Burkina Faso

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Abstract

To describe clinical profile of muscular dystrophies in Burkina Faso, we retrospectively reviewed 17 patients who had clinical phenotypes of Muscular Dystrophy between 2004 and 2004 in the neurology Department of Yalgado Ouedraogo Teaching Hospital in Ouagadougou.

There were 16 male patients (94.1%) and one (5.8%) female patients. The mean age of patients was 19.82 years (6-39 years).The disease onset was characterized by motor retardation (58, 8%). The age at onset was 10 years in 52.9% of patients. The disease duration was 9.41 years. At first visit, the muscle weakness was progressive in all patients. The most neurological finding was weakness (94.1%), Gower's sign (88.5%) and proximal amyotrophy in 55.8%. The mean Serum creatine kinase level was 3016.64 U/l (range 344 - 8896 U/l).

Physiotherapy (100%), corticosteroid (41.1%) and orthopedic management (23.5 %) were the most treatment. During the study period, the most common complications were loss of walk (23.5%), cardiac deterioration (29.4%) and orthopedic deterioration (41.1%). The mortality rate was 5.8%.

The present study revealed that muscular dystrophy is not an uncommon disease in Burkina Faso. We have concluded that if clinical diagnosis is possible nevertheless some difficulties exist to do muscles biopsy analysis.

Keywords: Muscular Dystrophies, Clinical Features, Burkina Faso

Introduction

Muscular dystrophies are characterized by primary damage to muscle tissue outside of any lesion of the nervous tissue. The term muscular dystrophy encompasses a range of disorders including Duchenne, Becker, congenital, myotonic, Emery-Dreifuss, facioscapulohumeral, oculopharyngeal, and limb-girdle muscular dystrophies [1]. *Muscular dys*trophies of Duchenne (DMD) and Becker (DMB) were the common. Each disorder varies in severity, age of onset, pattern of inheritance, and affected muscle groups and other organs [2]. Crude prevalence per 100,000 for all muscular dystrophies across the included studies ranged between 3.8 in Japan to 26.8 in Egypt [3,4]. Prognosis varies across the muscular dystrophies with some patients experiencing mild, though usually progressive symptoms, while others experience severe disability and early mortality [1]. Muscular dystrophies are ubiquitous pathologies that affect all continents but particularly affect certain populations in Maghreb [5,6], Asia [7], America and in Scandinavian countries [8,9]. There are relatively few investigations in Africans. The most studies were done in South Africa. There are no more studies regarding the prevalence of muscular dystrophy in Burkina Faso. The objective of our study is to give the experience of Burkina Faso in the management of Muscular Dystrophies.

Patients and Methods

Area of Study

Burkina Faso is a country of West Africa which occupies an area of 274,300 K msq for a population of 16,248,558 according to the 2006 Burkina population census. This study was performed at the Neurology Department of Yalgado Ouedraogo Teaching

Hospital January 2004 to December 2014 (10years), a tertiary national referral center for neurological disorders.

Participants

Seventeen patients with clinical diagnosis of myopathies (presence of proximal weakness, elevation of serum creatine kinase and the history of disease) were including retrospectively in the study. All patients included in this study were examined by neurologists. The history included onset of illness, distribution of weakness, atrophy, hypertrophy, course of the disease and family history. Muscle involvement was evaluated clinically using MRC grading scale.

Routine blood investigations and CK level were done in all. Muscle biopsy, muscle imaging immunohistochemistry and genetic diagnosis were done in certain patients. Molecular and genetic diagnosis was carried out in a reference laboratory in France. Patients with acute polymyotitis were non- in the study

Data Collection and Analysis

The analysis concerned Sociodemographic features of the study population (age, sex, residence, profession), clinical features (past medical history of epilepsy or not. We collected and analyzed epidemiological, clinical data using the Sphinx 5.0 software. Incomplete records were not included in the study. The confidentiality of the data was preserved by the anonymity of the fact sheets.

Ethical Considerations

We preserved the anonymity of all patients included in the study.

Results

Out of 17 patients, 16 (94.2 %) were men and one (5.8 %) woman. The mean age of patients was 19.82 years (6-39 years). The population adult older than 15 years was 14 (82.3%) patients. Fifteen patients were coming from urban regions and 5 from rural areas. Table 1 describes sociodemographic features of 17 patients with Muscular dystrophies. The mean age at disease onset was 6.7 years (ranges 3 years-25 years). Muscular dystrophy age of onset was between 5-10 years in 9 (52.9%) patients followed by 0-5 years in 4 (23.5%) patients. Only 11.7% (n=2) of the patients had age of onset at 25-30 years of age. Overall, 11.7% (n=8) of the patients had consanguineous parents. Eight (47%) patients had family history of similar cases. Disease onset was characterized by motor retardation in 10 (58, 8%) patients, difficulties in walking in 8 (47%) patients and difficulties in running in 7 (41.1%) patients. The mean age of patients at the first visit was 10.94 years (Ranges 5-26 years). The patients presented at 5-10 years of age in 5(29.4%) patients, between 10-15 years in 7 (41.1%) patients, between 15-20 years in one (5.8%) patient and between 25-30 years in 4 (23.5%) patients. The interval from the age of onset to the time the patient first presented (disease duration) was 9.41 years. The Table 2 describes the interval from the age at onset to age at first visit. At first visit, muscle weakness started proximally and had selective symmetrical distribution in the upper and lower limbs in 16 (94.1%) patients. One (5.8 %) patients had lost the walk before first visit (patient 1).

Demographic features	Number (%)
Mean age	19.82 years
Age (years)	
5-10	2 (11.7%)
11-15	3(17.6%)
16-20	7(41.1%)
>20	5 (29.4%)
Male gender	16 (94.1%)
Pediatric patients	12 (70.5%)
Urban residence (%)	15 (88.2%)
Consanguineous family	2 (11.7%)
Familial case	8 (47%)

Table 1: Sociodemographic features of 17 patients with muscular dystrophies

There was no systematic psychometric tests were done for the patients, but informal assessment revealed normal language function. There was not evident symptomatic cardiac involvement. The Table 3 describes the clinical data of muscular dystrophy patients at the first visit. The most clinical phenotype was Duchenne muscular dystrophy (52.9%) followed by Becker muscular dystrophy (41.1%) and Limb girdle muscular dystrophy (5.8%). According to biochemical features, serum creatine kinase level (CK) was elevated in the whole patients. The mean CK level was 3016.64 U/l (range 344 - 8896 U/l), normal value for our laboratory < 150 U/l. Nine patients (52.9%) patients had done genetic testing and immunohistochemical analysis. The diagnostic

Clinical features	Number =17
Motor deficit, n (%)	16 (91.1)
Gower's sign, n (%)	15(88.2)
Proximal amyotrophy, n (%)	10(58.8)
Distal amyotrophy, n (%)	9 (52.9)
Scapula alata, n (%)	7 (41.1)
Calf hypertrophy, n (%)	5 (29.4)
Cachexia, n (%)	3 (17.6)
Cognitive impairment, n (%)	1 (5.8)
Cardiac deterioration, n (%)	1 (5.8)
Orthopedic deterioration, n (%)	7 (41.1)
Loss of walk, n (%)	1 (5.8)

Table 2: clinical features of Muscular dystrophy patients at the first visit

of muscular dystrophies was confirmed in 5 patients (55.5%). Immunostaining showed evidence of pattern for dystrophin in 3 cases, dysferlin in one case and congenital myopathy in another one case. Genetic testing had found duplication of exons 3 and 4 in Dystrophin gene one case, deletion of exons 48 to 52 in two cases and absence of duplication and deletion in 2 cases. Deletions and duplications were reported in the Dystrophin gene. The confirmed diagnosis was DMD in 2 cases, BMD in one case, dysferlinopathie (one case) and congenital dystrophy in another one case. During study period, patients had benefited of three pluridisciplinary consultations with neurologist, pediatric pneumologist and cardiologist. The number of pluridisciplinary consultation was 2.52 per patient and 14 patients had done more than one consultation. During this follow up, 8 (47%) patients had deterioration and 2 (11.1%) had stopped their follow-up without medical advice. In totality, loss of the ability to walk was observed in 4 (23.5%) patients, respectively before the follow up (patient 1) and during the follow up (patient 4, 5 and 6). The mean age when last walked was 14.25 years. Patients had loss the walk respectively at the age of 24 years (patient 1), 14 years (patient 4), 10 years (patient 5) and 9 years (patient 6). Cardiovascular deterioration occurred in 5 (29.4%) at the age of 18 years (patient 1,4,6,7 and 11). The cardiovascular abnormalities was represented by decline in left ventricular (LV) systolic function and left ventricular hypertrophy respectively in 2 patients. Ventricular arrhythmias were seen on electrocardiogram in only one patient. %). Orthopedic deterioration was observed in 7 (41.1%) patients. Scoliosis, hyperlordosis, equinovarus and kyphosis were seen respectively in 2 patients. Death was observed in an 18-year-old boy who had cardiac and respiratory deterioration (patient 4). The Table 3 describes the deterioration in patients with muscular dystrophies. According to treatment, all the patients with muscular dystrophies had received physiotherapy. Corticosteroid was prescribed in 7 (41.1%) patients and angiotensin-converting enzyme inhibitors (ACE) in one (5.8%) patient Table 4. All this treatment was only used in patients with DMD and BMD. The Table 5 summarizes the demographic and clinical features of patients with muscular dystrophies.

Interval onset and first visit (years)	Population study (N=17)
0-5	15
5-10	0
10 -15	1
15-20	0
21-25	1

Table 3: Interval between onset and first visit (year)

Deterioration	Number (%)
Loss of walk	4(23.5%)
Arrhythmia	2 (11.7%)
Cardiomyopathy	2 (11.7%)
Cardiac insufficiency	1 (5.8%)
Scoliosis	2(11.7%)
Lordosis	2(11.7%)
Foot varus	2(11.7%)
Kyphosis	2(11.7%)
Death	1 (5.8%)

Table 4: Clinical outcome of patients with muscular dystrophies (N=17)

Patient N°	Age at onset (years)	Age at first visit (years)	Age at inclusion (years)	Sex	Family cases	Consanguinity	Clinical features	CK level	Genetic testing	Molecular diagnosis	Complication
1	11	26	34	M	None	No	BMD	1526	Duplication of exons 3 and 4	BMD	Loss of walk Cardiopathy scoliosis
2	5	27	39	M	None	Yes	BMD	1765	N/A	N/A	None
3	13	17	25	M	None	No	BMD	725	N/A	N/A	None
4	6	9	18	M	1	No	DMD	987	Duplication of exons 3 and 4	DMD	Loss of walk, cardiopathy, respiratory , scoliosis, death
5	5	8	15	M	None	No	DMD	2141	N/A	N/A	Loss of walk, kyphosis
6	3	7	15	M	None	No	DMD	1815	No deletion or duplication of exon	CMD	Loss of walk, cardiopathy kyphosis
7	7	10	15	M	None	No	DMD	3345	Deletion of exons 48 to 52	DMD	Cardiopathy lordosis
8	8	10	16	M	3	Yes	DMD	1925	N/A	N/A	Foot varus
9	3	6	10	M	3	No	DMD	3754	N/A	N/A	None
10	9	11	18	M	None	No	BMD	4825	N/A	N/A	Lordosis
11	21	25	29	M	1	No	BMD	8896	No deletion or duplication of exon	LMGD 2B	Cardiopathy foot varus
12	8	10	12	M	3	No	DMD	4634	N/A	N/A	None
13	9	11	18	M	4	No	DMD	834	N/A	N/A	None
14	5	5	6	F	None	No	DMD	2565	N/A	N/A	None
15	9	12	18	M	None	No	BMD	7475	N/A	N/A	None
16	8	12	20	M	2	No	BMD	1975	N/A	N/A	None
17	25	26	27	M	None	No	LGMD	2096	N/A	N/A	None
Abbreviations : M : male, F : female, DMD: Duchenne muscular dystrophy, BMD: Becker muscular disease, CMG : Congenital muscular dystrophy, LGMG : Limb girdle muscular dystrophy , CK: Serum creatine kinase, N/A : no available											

Table 5 : Demographic and clinical features of patients with muscular dystrophies

Discussion

This study demonstrate that muscular dystrophies was present in African black, as reported in 17 patients in Burkina Faso. The few number of patients included in the study is in consistence with others studies carried in West Africa [6,7]. The number of cases is probably underestimated in our case. The majority of patients were coming from urban areas (88%) in which there are health workers (neurologists, pediatrician) able to care neuromuscular diseases. Patients who were coming from urban areas had been referred by a local association of myopathy which explains that they are specialized health care access. In rural areas, there are specially a lack of knowledge of disease by health workers and ignorance of patients. Knowledge could be disseminated towards caretakers, physicians (neurologists, pediatricians, general patricians) and paramedical staff by continuous medical training, advanced training course in the neurology department. Indeed, build a local/national network of specialist and patient registry is an opportunity for us. The diagnosis based on CPK level is not available in rural zone. Currently there are 3 specialists on muscular dystrophies (composed of 2 neurologists and one anatomopathologist) in Burkina Faso for a population of 16 millions of persons. There is a weekly consultation for muscular dystrophy in the neurology department. For next years, it is necessary to have more consulations in neurology department in the main hospital of Ouagadougou and to make screening campaigns in rural areas. The mean age at disease onset was 6.7 years (ranges 3 years-25 years), in line with Dey *et al.* and Swaminathan *et al.* who observed respectively an average age at onset at 3.9 years and 3.1 years [9,10]. In contrast, Manjunath M *et al.* had found that age at onset mean was 45.3 years [11]. The majority of our patients (52.9%) had disease onset at the age between 5-10 years while Viswajyothi in India observed that 50% of patients had disease onset between the ages of 0-5 years [12]. Thus, muscular dystrophy is seen in early childhood. The majority of our population studied were male (94.2 %), in line with the study of Viswajyothi P *et al.* in India which report that 90 % of this study population was men [12]. This study showed that the diagnosis of muscular dystrophy was tardy at the age 10.94 years. The majority of patients presented at the age between 10-15 years (41.1%) patients, in line with Viswajyothi P in India who observed the same result. The first symptoms at onset are not recognizing by families as muscular features and are unknown by most of the health agents. Consanguinity was observed in 12 percent of our patients. In X-linked diseases consanguineous marriage was detected in 28.1% of patients with DMD [13]. Consanguinity is widely practiced in countries of Asia and Africa especially in societies where Islam prevails [14]. Forty seven percent of patients had family history of similar cases. People with a family history of muscular dystrophy are at a higher risk of developing the disease or passing it on to their children. The most clinical phenotypes were Duchenne muscular dystrophy (52.9%), Becker muscular dystrophy (41.1%)

and Limb girdle muscular dystrophy (5.8%). In terms of the prevalence of dystrophy types, our results are consistent with the results of Khadijeh *et al.* in Iran and Paul in the United States which showed the higher prevalence of Duchenne dystrophy than Becker dystrophy in the United States [15,16]. Some different methods can be used to diagnose the various types of muscular dystrophy, such as a Serum creatine kinase (CPK) test, an electromyography (EMG) test, a nerve conduction velocity (NCV) test, a genetic test, and a muscle biopsy. In our study, electromyography is not available for muscular dystrophies. Muscle biopsy was done in only one patient because it was not practical in Burkina Faso. Serum CK level was assessed and elevated in all our patients. This investigation is only available in two towns of Burkina Faso, Ouagadougou, the capital city and Bobo Dioulasso. An epidemiological study by El-Tallawy et al. on muscular dystrophies in Egypt found that more than 80% of subjects had an elevated CPK (> 225 IU/l) [4], which is consistent with our results, which showed elevated CPK in about 88.8% of patients and constitute with clinical findings the key of the diagnosis [17]. In our study, the diagnosis of Muscular dystrophy was confirmed in about 42% of cases by molecular genetic testing. According to Alao *et al.* [18], the diagnosis of muscular dystrophy was confirmed by molecular genetic testing in Benin, Niger and Guinea respectively in 50%, 28% and 0% of blood samples. In our study, deletions and duplications have been reported in the dystrophin gene in tree patients with Duchenne and Becker muscular dystrophy. Only one patient had deletion in dysferlin gene. Dystrophin gene was reported in African patients with Duchenne in Ghana [19], in Rwanda and South Africa [6,20,21]. In our study, the confirmed diagnosis was DMD in 2 cases, BMD in one case, dysferlinopathie (one case) and congenital dystrophy in another one case. In opposition with the study of Alao, we did not found any case of Gamma sarcoglycanopathy, spinal amyotrophic or Becker Myotony. To improve diagnosis and care in our context, it is we must facilitate the availability of a genetic and molecular screening device, access to paraclinical assessments (biopsies, muscle imaging) and promote medical center equipment (EMG, needed for biopsies, etc.). To date, no effective treatment is available to patients with muscular dystrophy. In our study, corticosteroids and ACE and were used for symptomatic treatment respectively 41.1% and 5.6 in patients with DMD and BMD. Corticosteroids are useful to improve muscle strength and function in the short-term (six months to two years), especially in DMD. In addition, ACE inhibitors were useful in the management of cardiomyopathy in patients with Duchenne or Becker muscular dystrophy [22]. If we considered the number of patients with clinical features of DMD or BMD, corticosteroids and ACE were less used. This situation could be explained by high cost of these treatments. Physiotherapy was used in all patients but was limited by orthopedic complications (41.1%). The short duration of this treatment was observed in patients who had financial constraints. To facilitate access to specific treatments, we should provide g corticosteroid treatment, electric wheelchairs and inclusion of patients in clinical studies. The mortality rate (5.8%) can be explained by the severity of complications, including cardiac deteriorations [23].

Limits of the Study

This was a retrospective case-note review. Some data were missing from clinical notes, in particular information about consanguinity. This may reflect difficulty in asking families about this issue in a clinic setting, or poor documentation, but it did restrict analysis of this variable. Even though this was a full population study, the specific conditions being studied are rare and numbers in individual diagnostic groups were small, which limited the power to undertake more detailed analysis. As it is a prevalence study, it was only possible to assess association rather than causation.

Conclusion

The present study revealed that Muscular Dystrophy is not an uncommon disease in Burkina Faso. We have concluded that if clinical diagnosis is possible nevertheless some difficulties exist to do muscles biopsy analysis. Physiotherapy and prevention of cardiac deterioration are available in our context.

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