

# Limb-Girdle Muscular Dystrophy (LGMD) in Burkina Faso: About a Case of Dysferlinopathy LMGD 2B and Review of the Literature

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## Abstract

**Objective:** To describe clinical features of the first diagnosed case of dysferlinopathy in Burkina Faso. **Clinical observation:** This is a 33-year-old Burkinabe married patient, born from a non-consanguineous polygamous family of 37 children. Disease onset was when the patient was 24 years; he experienced weakness of the 2 lower limbs with gait disorders, balance and painful muscle cramps. There was apparent muscle weakness at a level of 3/5 in the proximal muscles on neurological examination; no weakness of the distal muscles, pathological reflex, or cranial findings. CK level was increased to 24,414 U/L (0-248). Echocardiography was found left ventricular hypertrophy. Muscle MRI of the shoulders, pelvis and limbs founded diffuse amyotrophy. Immuno-histo-chemistry in Italy revealed sarcoglycannopathy. Western blot from blood samples and genetics were performed in France and revealed dysferlinopathy. Since 2009, he was treated with corticosteroid the CK levels temporarily decreased to 7518 U/L in February 2014. Currently, at 6 years post disease onset, the patient is independent for daily life activities but needs a cane for walking long distances. **Conclusion:** Dysferlinopathies are rare in Black African. In Burkina Faso, we observed one case and 4 non diagnosed cases in the same family..

**Keywords:** Dysferlinopathies; Limb girdle muscular dystrophy; Burkina Faso

## Introduction

Limb girdle muscular dystrophies are a heterogeneous group of inherited muscle diseases characterized by wasting and weakness of skeletal muscle. It's affect the muscles of the scapular and pelvic belts. The prevalence of LGMD is estimated to be between 14,500 and 123,000. [1] Dysferlinopathy is an autosomal recessive-limb girdle muscular dystrophy (AR-LGMD) caused due to the defect in gene encoding dysferlin, a sarcolemmal protein. Dysferlinopathies are rarely described in an African black. We reported the first clinical case of dysferlinopathy with genetically confirmed diagnosis in native Burkinabe patient.

## Clinical Case Presentation

This is a 33-year-old man born from a non-consanguineous polygamous family of 37 children. He had got married and has a child of 2 years old. His disease began when the patient was 24 years; he experienced weakness of the 2 lower limbs with gait disorders, balance and painful muscle cramps. There were 4 others similar cases in his family. At the time of his first medical visit, at the age of 25, his blood pressure was 130/ 80 mmHg and his temperature was 36.5°C. There was

apparent muscle weakness at a level of 3/5 in the proximal muscles on neurological examination; no weakness of the distal muscles, pathological reflex, or cranial findings. His intellectual status was normal. The Respiratory, cardiac, and abdominal examinations were normal. There was significant increase of CK level to 24,414 U/L (0-248), lactate dehydrogenase to 1566 U/L (0-248), aspartate aminotransferase level to 69 U/L (0-35) and alanine aminotransferase level to 44 U/L (0-35). The others investigations (erythrocyte sedimentation, C-reactive protein, hemogram, creatine, electrolytes, HIV test, hepatitis B serology, thyroid-stimulating hormone) were normal. Echocardiography was found left ventricular hypertrophy. Muscle MRI of the shoulders, pelvis and limbs founded diffuse amyotrophy. Muscle biopsy analysis in Burkina revealed advanced myopathy but

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immuno-histo-chemistry in Italy revealed sarcoglycanopathy. Western blot from blood samples and genetics were performed in France and revealed dysferlinopathy. Since 2009, he has been treated with dexamethasone at a dose of 30 mg of prednisone deflazort per day and Delazort after January 2013. Following this treatment, the CK levels temporarily decreased to 15 933 U/L in March 2013 and 7518 U/L in February 2018.

Physiotherapy sessions were performed two times per week. Currently, at 6 years post disease onset, the patient is independent for his daily life activities (eating, have a bath, sanitary); he needs a cane for walking long distances. The walk perimeter is reduced. Echocardiography found the same features than the initial investigation. After that, we prescribed ICE to prevent cardiac complication.

## Discussion

We reported the first clinical case of dysferlinopathy type LMGD 2B in Burkina Faso. Publications about dysferlinopathy in black African or in African Americans are rare. An atypical dysferlinopathy was discovered in London in a West African boy of 10 years old.<sup>[2]</sup> In USA, only 5% of patients with dysferlinopathies were African Americans.<sup>[3]</sup> In the present case, clinical features (age of onset, pelvic involvement, absence of calf hypertrophy, rarity of cardiac involvement) and CK levels were according to the literature.<sup>[4]</sup> CK level is 10 to 150 times higher than normal (24,414 units / l). Family history of weakness is suggesting to genetics disease. In the present case, there are difference between immunohistochemistry of muscle biopsy (sarcoglycanopathy) and Western Blot results. Western Blot is more specific than immunohistochemistry. Immunohistochemistry, western blot and genetics were not available in our context. Muscular MRI was performed in our context but there are financial limits and radiologists have not experience about this investigation. According to literature,

Delazort is not an effective therapy for dysferlinopathies but in our case, we observed important decrease of CK levels. Physiotherapy is a palliative treatment, in the absence of treatment able to curing patients with dysferlinopathy.<sup>[5]</sup>

## Conclusion

Dysferlinopathies are not so rare in Burkina Faso. We observed 4 non diagnosed cases in the same family. We need some trainings and laboratory investigations (immunohistochemistry, Western blot) to improve the management of neuromuscular diseases in the West Africa Region.

## Conflict of Interest

I declare there is no conflict of interest

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