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Case Report

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Duchenne muscular dystrophy in Niger: a family history

Abstract

Duchenne muscular dystrophy is an inherited disease characterized by progressive muscle degeneration and usually affects boys. The authors reported at the neurology department of the national hospital in Niamey, the case of a family whose boys presented proximal motor deficits in all four limbs, and whose mothers were carriers of cardiomyopathy. The first generation consisted of seven boys and four girls, among which, three boys died of walking disability and breathing disorders, and two of unknown cause. Also, the three girls were carriers of cardiomyopathy and the other died of unknown cause. In the second generation, three boys had died (unknown cause), two were alive and aged 10 and 14 years with walking disability whose balance sheets were abnormal, including CPK (creatinine phosphokinase) and myoglobin. The genetic test showed an out-of-phase duplication of exons 8 to 18 in the latter. Duchenne muscular dystrophy is a rare disease. It is important to think about it when there is a family history of limb-girdle deficit in boys, and to systematically search for cardiac disorders in mothers.

Keywords: duchenne muscular dystrophy, familial, niger, tabouret's sig, gowers's sign

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Abbreviations: CPK, creatinine phosphokinase; DMD, duchenne muscular dystrophy

Introduction

Neuromuscular diseases are conditions normally present in all ethnic communities and all latitudes.[1,2] Duchenne muscular dystrophy (DMD) is the most common of dystrophinopathies.[3] In 2010, the prevalence of DMD was estimated in the United States to be 1.4 per 10,000 male sexes.[4] However, there are very few reports of DMD in Africa, especially in the sub-Saharan region, except in South Africa and the Maghreb countries where they are published.[3] Neuromuscular diseases in West Africa are dominated by Duchenne muscular dystrophy.[5,6] The objective of this work was to present the clinical characteristics of a DMD family in Niger.

Clinical cases

Family history

This was a sibling group of 11 born, including 7 boys and 4 girls (Figure 1). The first generation consisted of seven boys and four girls. Three boys died within a pattern of walking disability and respiratory disorder. Three of the four girls were carrier of cardiomyopathy, and the other died of unknown causes. In the second generation, three boys had died (unknown cause), and two were alive and aged 10 and 14 years (observation 1 and 2). So, we have a family of eleven children from the same father and mother without consanguinity.

Observation 1

This was a 14-years-old boy who had a school interruption two years ago (III 13, Figure 1). He was the second born to a sibling of four, including two boys and two girls which were apparently healthy. There was a notion of cardiomyopathy in his mother and the death of his elder son in the same picture. The onset of the symptomatology would date back to childhood, at the age of 8 years, marked by frequent falls and difficulties in standing. The clinical examination revealed deficiencies of the shoulder girdle (difficulty in dressing, symmetrical detachment of the scapula) and pelvic (Tabouret's sign *and* Gowers's sign were positive). There was also hypertrophy of the calves, abolished idiomuscular reflexes, spine deformity, waddling gait with asymmetric equinus and retraction of the Achilles tendons. In addition, there was no intellectual disorder but anxiety was noted on the child's face. On paraclinical examination, CPK was 15150 IU/l (N equals 25-195 IU/l), LDH was 948 IU/l (N=230-430 IU/l) and myoglobinemia was higher than 500 ng/ml (N lower than 80ng/ml). ECG, chest X-ray and cardiac ultrasound were normal. The diagnosis of Duchenne muscular dystrophy was confirmed by genetic test which showed an out-of-phase duplication of exons 8 to 18. The patient was put on corticosteroid therapy, physical therapy and Captopril with a slight improvement of motor function.

Observation 2

Older child aged 10 years old (III 18, Figure 1), male, attending school and second born of a sibling group of four, including two boys and two apparently healthy girls. There was also a notion of cardiomyopathy in his mother with a follow-up in cardiology. The onset of the symptomatology would date back to childhood at the age of 7 years marked by frequent falls and difficulties in standing and running. The clinical examination revealed a waddling gait, a moderate equinus and a detachment of the scapula. There was also a pelvic girdle deficit with positive Gower sign, calf hypertrophy, abolition of idiomuscular reflexes and moderate retraction of the Achilles tendon. There was no intellectual deficit or cardiorespiratory disorder. On paraclinical examination, CPK was 8944 IU/l, LDH 560 IU/l,

myoglobinemia was 489 ng/ml. Chest X-ray, ECG, echocardiogram did not reveal any abnormality. The diagnosis of DMD was confirmed by genetics tests which found an out-of-phase duplication of exon 8 to 18 of the short arm of the X chromosome. The child was put on corticotherapy and physiotherapy with psychological support. The clinical evolution was marked by an improvement.

Discussion

On the clinical aspects, we can see that the dates of onset of the disease are variable. This clinical heterogeneity was mentioned by[7] who found an age of onset varying between 3 and 6 years. The signs of onset are globally an inability to run, to walk and to climb stairs. Then, the Gowers sign (difficulty in getting up from the floor) is observed in a chronological order. This clearly demonstrates pelvic involvement long before that of the shoulder girdle. The same chronology was described in the study by.[8] Pseudo-muscular hypertrophy, especially in the calves was found in all our patients. This is classically suggestive of dystrophinopathy, but it can also be found in many other muscle diseases.[3] Psychological impairments were noted in all our patients, such as anxiety and school dropout for the oldest. These elements are usually absent in limb-girdle myopathies, but are sometimes found in Duchenne myopathy. The age of onset of these disorders depends on the precocity of the signs of onset.[3,4,9] Have found mental retardation in children with Duchenne muscular dystrophy with a significant decrease in the intellectual coefficient.[10] have found cognitive impairment, but concluded that this depends on the site of mutation.[4,11] have also mentioned that cognitive impairment would be mainly correlated to the type of dystrophin affected, in particular DP140, which is the dystrophin of the brain. The ECG, the cardiac ultrasound and the chest x-ray, performed as part of the evolutionary assessment in search of cardiac complications did not show any abnormality. It should be noted, however, that cardiac and pulmonary complications develop secondarily after the age of 20 years.[12] Therefore, regular follow-up of affected individuals will be required to detect the occurrence of slowly progressive cardiomyopathy.[7] CPK elevation is found in all our patients. A very high CPK level (more than 10 times normal) in a child or a young adult with a limb-girdle muscular deficit of insidious onset and progressive evolution, strongly suggests a progressive muscular dystrophy.[8] Duplications and deletions represent about 70-80% of the mutations while rearrangements and punctual mutations represent 20-30% of the genetic mutations observed in Duchenne muscular dystrophy,[8,13] which is the case for our patients. All our patients were put on corticosteroids with an immediate effect on the regression of the signs, especially in the two patients who were put on ACE inhibitors (captopril) in addition to the corticosteroids with an improvement of the muscle strength. Corticosteroid therapy prolongs the age of onset of walking loss and improves motor function.[10,12] All our patients underwent functional rehabilitation, which improved the deformity of the limbs and the hypertonia related to tendon retraction. Multi-weekly physiotherapy sessions improve muscle trophicity and fight against the retraction of all the joints linked to muscle fibrosis.[7] Elle rallongerait de ce fait la phase ambulatoire de la maladie. It would therefore lengthen the ambulatory phase of the disease. The 10-year-old child presents a retraction of the Achilles tendon and the 14-year-old has in addition a retraction of the knee tendon.

Some difficulties must be noted, in particular the impossibility of carrying out complementary assessments in the context of neuromuscular diseases which currently require sophisticated immunohistochemistry and molecular biology techniques. There were also difficulties in finding the financial resources to carry out some examinations available in Niger, in particular the muscle scanner, EMG and immunohistochemistry.

Otherwise, in the absence of therapeutic intervention, children lose their ability to walk by the age of 10 to 12 years.[4,7]

Conclusion

This work highlights the fact that in Niger, complementary examinations for the diagnosis of myopathies are limited to routine examinations. This may explain the lack of knowledge, and especially the underestimation of these diseases in our country. There is a need for genetic counselling to prevent the disease. Finally, this study opens perspectives, notably the research and follow-up of other cases in this family.

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None.

Conflicts of interest

The authors declare that they have no conflicts of interest.

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