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Short Communication Open Access

Duchenne Muscular Dystrophy: Short Communication

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Abstract

Objective: To conduct a review about Duchenne muscular dystrophy.

Methodology: Non-systematic review of literature.

Results: Duchenne muscular dystrophy (DMD) is a disorder that occur because of mutations in the DMD gene. It is a disease that occurs in children, with central muscle weakness and muscle contractures. Dystrophin gene deletion and duplication testing is usually the first confirmatory test. When the mutation of the DMD gene is not confirmatory, muscle biopsy should be performed.

Conclusion: The treatment is supportive and the corticosteroids improve muscle strength.

Keywords: Duchenne muscular dystrophy; Cardiomyopathy; Muscles; Diagnosis; Treatment

Introduction

Alterations in the regulator gene of muscle function produce muscle weakness and loss of muscle mass. There is a diverse clinical spectrum in this group with differences in distribution and degree of motor impairment, age of onset, rate of progression, and inheritance pattern [1].

Duchenne muscular dystrophy (DMD) is a disorder that occurs because of mutations in the DMD gene, encoding the dystrophin protein. DMD is more severe, and more common, with newborn screening studies showing an incidence ranging from 1:3802 to 1:6291 live male births (rather than the 1:3500 that is commonly cited). Because the gene is X-linked, the diseases affect only boys (except in those rare cases explained by unusual genetic mechanisms such as balanced chromosomal translocations) [2].

Dystrophin binds to cytoskeletal actin via its N-terminal actin-binding domain 1 (ABD1) and to β-dystroglycan via its C-terminal domain, with the central rod domain, consisting of 24 spectrin-like repeats, in between. Alterations in the dystrophin gene cause damage to the muscle membrane, producing fibrosis and muscle replacement by fatty tissue [3].

Methodology

Non-systematic review of literature.

Clinical Manifestations

It is a disease that occurs in children, with central muscle weakness and muscle contractures; in these patients you can see the sign of Gowers in which the child has to use their hands and arms to "walk" through their own body from a squatting position. The patient loses the ability to walk when childhood ends or when adolescence begins, affecting in turn the respiratory muscles producing death in the second or third decade of life [4].

Diagnosis

In the physical examination, loss of proximal muscle strength and gait is identified, there is an alteration in hepatic function and in the levels of seric CK but dystrophin gene deletion and duplication testing is usually the first confirmatory test; testing is best done by multiplex ligation-dependent probe amplification (MLPA) or comparative genomic hybridisation array [5]. When the mutation of the DMD gene is not confirmatory, muscle biopsy should be performed [6-8].

Treatment

Glucocorticoids

The corticosteroids offer benefit to boys with DMD by stabilizing muscle strength and function, prolonging independent ambulation, and delaying the progression of scoliosis and cardiomyopathy. Daily oral prednisone (0.75 mg/kg) or deflazacort (0.9 mg/kg) is generally recommended [9].

Cardiac management

Patients with DMD have dilated cardiomyopathy and/or cardiac arrhythmias; Angiotensin-converting enzyme inhibitors or angiotensin receptor blockers should be prescribed after 10 years of age and B-adrenergic blockade is recommended when the ventricular dysfunction is observed in the echocardiogram or cardiac MRI [10].

Physical therapy

Regular physical therapy is important to maintain muscle strength and prevent contractures; In addition, experience suggests that it may help to delay the onset of scoliosis and respiratory failure [11].

Respiratory management

Baseline pulmonary function tests should begin at 5–6 years of age. Respiratory evaluations should occur annually and then biannually in non-ambulatory boys. Overnight pulse oximetry or sleep studies to detect SDB should be considered in non-ambulant boys. All boys should receive the pneumococcal and influenza vaccines [12]. Nocturnal non-invasive intermittent positive pressure ventilation (NIPPV) is indicated in patients who have signs or symptoms of hypoventilation and/or hipercapnia [13].

Conclusion

DMD is a disorder that occurs because of mutations in the DMD gene, with central muscle weakness and muscle contractures. The methods used to diagnose DMD include clinical history, physical examination, serum CK, liver enzymes, genetic testing, and perhaps muscle biopsy. The treatment is multidisciplinary with glucocorticoids being the cornerstone.

Conflicts of Interest

The authors declare that the research was conducted in the absence of any commercial or financial relationships that could be construed as a potential conflict of interest.

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