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Eye involvement in patients with myotonic dystrophy

Implicación ocular en pacientes con distrofia miotónica

Dear Editor:

We read with great interest the article by Gutiérrez Gutiérrez et al.[1] on the diagnosis and follow-up of patients with myotonic dystrophy.

We are sure that this comprehensive study will be extremely useful for all physicians.

We would like to contribute additional information on ophthalmological findings associated with the disease.

The authors rightly focus on the importance of detecting low intraocular pressure, and underscore the importance of ultrasound in detecting the cause of this and other pathological findings.[2—5]

However, other studies into the ophthalmological features of myotonic dystrophy report an association with Fuchs endothelial dystrophy[6,7] and eye melanoma.[8,9]

Furthermore, the authors focus on the prevalence of cataracts in these patients, and suggest the possibility of cataract removal in the event of decreased visual acuity.[10—12]

If cataract surgery is considered, a study of endothelial cells should be performed not only to gather clinical data with a view to preventing corneal endothelial decompensation, but also because the available evidence provides conflicting information on the possible association between myotonic dystrophy and Fuchs endothelial dystrophy.[13—15]

Lastly, regarding the association between myotonic dystrophy and eye melanoma, a thorough eye fundus examination should be performed in these patients to ensure early detection of this potentially fatal condition.

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Andersen-Tawil syndrome with sex-specific phenotype: usefulness of the long exercise test☆

Síndrome de Andersen-Tawil con fenotipo sexo-específico: utilidad del test de ejercicio largo

Dear Editor:

Andersen-Tawil syndrome is a clinical entity characterised by periodic paralysis, distinctive physical features, and electrocardiographic alterations. This rare disease is frequently difficult to diagnose; the long exercise test may be useful in differential diagnosis including other muscle channelopathies.[1—7] Several alterations have been described in the *KCNJ2* gene, which encodes an inwardly rectifying potassium protein of the muscle membrane. We present the case of a family with the R67W mutation of *KCNJ2*.

Our patient was a 39-year-old man who began to present episodes of generalised weakness at the age of 7-8 years; episodes lasted approximately 2 weeks, during which the patient was unable to leave his bed. Until the age of 25, episodes were frequent (presenting every month) and were exacerbated by carbohydrate and alcohol consumption, intense exercise, and rest after exercise. Such foods as bananas and potatoes did not trigger the episodes. Elevated serum potassium levels were detected at onset of the disease, leading to a diagnosis of hyperkalaemic periodic paralysis. As the disease progressed, episodes became less frequent, and some were associated with normal or even low potassium levels. The patient was referred to the neurophysiology department for evaluation due to mild generalised weakness of several months' progression. He presented elevated CPK levels at admission (1325 U/mL). We performed a neurophysiological study including the short and long exercise tests, according to the protocol described by Fournier et al.[1] The short exercise test was performed 3 times, with a 1-minute rest period between trials; results were normal. The long exercise test revealed a baseline amplitude of 10.7 mV, with a marked increase (197%) in the area of the compound muscle action potential (CMAP) after effort, followed by a progressive decrease until a minimum value of 21% was reached at 45 minutes (Fig. 1). Repetitive stimulation yielded normal results. The clinical progression, neurophysiological findings, and mild but characteristic phenotype (short stature, hypertelorism, micrognathia, and clinodactyly) observed in our patient led us to suspect Andersen-Tawil syndrome. The ECG revealed no ventricular arrhythmia; QT interval was within normal values. A genetic study detected a heterozygous mutation in the *KCNJ2* gene (nucleotide change 199C > T, resulting in the substitution of arginine for tryptophan [R67W]). A genetic study of the patient's relatives detected the mutation in his mother, but not in his sister (the only sibling). Our patient's mother underwent the long exercise test, with normal results. She presented the same dysmorphic features as her son and ventricular extrasystoles, but had no muscle symptoms.

Muscle channelopathies are rare diseases whose diagnosis is often challenging. In many cases, clinical symptoms are not sufficient to determine the type of muscle channelopathy, particularly when physicians are not familiar with this group of disorders; Andersen-Tawil syndrome is associated with mild phenotypes (some patients may even display no dysmorphic features). Potassium levels may be misleading, as in the case presented here, and genetic studies are

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