A most distinctive reflex idiopathic generalized epilepsy (IGE) syndrome with well-defined clinico-EEG manifestations.

Prevalence

~3% of adults with epileptic disorders and 13% among IGEs with absences. Age at onset Range is 2 to 14 years; peak at 6 to 8 years. Sex 2-fold female preponderance. Neurological and mental state Normal.

Etiology

Genetically determined.

Clinical manifestations

The triad of manifestations are (1) eyelid myoclonia with and without absences; (2) eye closure-induced seizures and EEG paroxysms; and (3) photosensitivity.

Eyelid myoclonia, the hallmark of this syndrome, consists of marked jerking of the eyelids and often with jerky upward deviation of the eyeballs and the head (eyelid myoclonia without absences). This may be associated with or followed by mild impairment of consciousness (eyelid myoclonia with absences). The seizures are brief (3 to 6 sec) and occur mainly after eye closure and consistently many times per day. All patients are photosensitive.

Generalized tonic-clonic seizures (GTCS), induced by lights or spontaneous, are probably inevitable in the long term and are particularly provoked by precipitating factors (sleep deprivation, alcohol) and inappropriate AED modifications. Typically, GTCS are sparse and avoidable. Myoclonic jerks of the limbs may occur but are infrequent and random. Eyelid myoclonic status epilepticus (1/5 of patients) consists of repetitive and discontinuous episodes of eyelid myoclonia with mild absence. Seizure-precipitating factors Eye closure and photosensitivity (flickering or uninterrupted light). Photosensitivity declines in adulthood. Self-induction is rare.

EEG Video-EEG is the most important procedure. It shows frequent and brief (2 to 3 sec) high-amplitude 3 to 6 Hz generalized spike and wave discharge of mainly polyspikes, which generally occur after eye closure. These are frequently associated with eyelid myoclonia. Photoparoxysmal responses occur in all untreated young patients.

Differential diagnosis

Eyelid myoclonia is often misdiagnosed as facial tics, although if seen once, will never be forgotten or confused with other conditions. It should not unquestionably be considered as a deliberate attempt to self-induction.

Prognosis

Lifelong disorder. Eyelid myoclonia persists often without apparent absences and even without demonstrable photosensitivity. It is highly resistant to treatment and occurs many times per day.

Management options

Avoidance of precipitating factors. Valproate, clonazepam, ethosuximide, and levetiracetam are treatment options.