

# Juvenile absence epilepsy

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## Abstract

*Juvenile absence epilepsy (JAE) is one of the age-related idiopathic generalized epilepsies (IGE) that accounts for approximately 10% of all epilepsies. Age at onset is usually near or around puberty, between 10 and 17 years of age, in young people with normal neurologic examination. Its clinical features are characterized by sporadic (non-pyknoleptic) occurrence of absence seizures frequently associated with generalized tonic-clonic seizures (GTCS) predominantly on awakening. Interictal and ictal EEG shows generalized spike and wave discharges with normal background activity. Hereditary factors play a major role in the etiology of JAE. Sodium Valproate (Depakine®) is the first choice anti-epileptic medication, controlling both absences and tonic-clonic seizures.*

## Key words

Juvenile absence epilepsy (JAE), idiopathic generalized epilepsies (IGE), generalized tonic-clonic seizures (GTCS)

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## Disease name and synonyms

Juvenile absence epilepsy (JAE)

## Definition

According to the Revised International Classification of Epilepsies and Epileptic syndromes (Commission 1989), Juvenile absence epilepsy (JAE) is an age-related idiopathic generalized epilepsy, occurring around puberty.

## Differential diagnosis

There is some overlap between JAE, [juvenile myoclonic epilepsy](#) and epilepsy with grand mal seizures on awakening. JAE is distinguishable from childhood absence epilepsy (CAE) by age of onset in adolescence and by the frequency of absences.

## Etiology

JAE is an idiopathic epilepsy and approximately 11% of patients report a familial history of epilepsy (Wolf 1992). The exact cause of this disorder remains unknown.

### Clinical description

Absences of JAE are the same as in CAE but they occur around puberty (peak at 10-12 years) and the seizure frequency is lower than in CAE, with absences occurring mostly sporadically. The majority of patients also have generalized tonic-clonic seizures (GTCS), whose manifestations precede that of the absences. Most frequently, GTCS belong to the awaking type.

### Diagnostic methods

JAE diagnosis is based on clinical signs and EEG recordings on awake and during sleep. EEG background activity is normal. Interictal and ictal EEG is characterized by focal generalized spike-wave discharge occasionally prominent in the frontal region. Spike and wave frequency is faster (3.5-4 Hz) than in typical CAE (3 Hz). Duration of absences is longer than in CAE. Polyspikes can occur on discharge onset. EEG paroxysms are precipitated by hyperventilation, sleep deprivation.

### Epidemiology

The incidence and prevalence of JAE in the general population are not known.

### Genetics

Familial and twin studies suggest that genetic factors play a major role in the etiology of JAE. An autosomal dominant gene seems to be involved. Sander et al (1997) studied the relation of JAE to glutamate receptor polymorphism. Evidence was found that allelic variants of [the kainite-selective glutamate receptor gene] GRIK1 (in 21q22) contribute as a major genetic determinant to the pathogenesis of JAE-related phenotypes. A family with JAE was found to be linked to chromosome 3q26 (Sander et al, 2000). More recently, Haug et al (2003) identified in families with IGE localized to 3q26 a heterozygous mutation in the chloride channel-2 gene *CLCN2*.

### Management and treatment

Response to therapy is often good. Sodium Valproate (Depakine®) is the first choice anti-epileptic medication, controlling both absences and tonic-clonic seizures. Lamictal (Lamotrigine®), in combination with Depakine, can be effective. If the strategies fail, ethosuximide (Zarontin®) or acetazolamide (Diamox®) can be tried in combination with Depakine®.

### Unresolved questions

JAE, JME and generalized tonic-clonic seizures on awakening share several clinical features, thus giving rise to questions of phenotypic overlap suggesting underlying possible neurobiological and genetic, relationships

(Reutens & Berkovic, 1995). Photosensitivity is a frequent electroencephalographic finding in IGE (Wolf & Goosses 1986). In this study, 13 % of patients with absence-epilepsies were photosensitive with a marked difference between CAE (7.5 %) and JAE (18 %). This difference was not reduplicate by Waltz et al (1990), who found an overall rate of photosensitivity of 18 % in both absence epilepsies. Nevertheless photosensitivity occurrence in JAE must be taken into account for treatment strategy.

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