Juvenile absence epilepsy is a type of idiopathic generalized epilepsy recognized by the International League Against Epilepsy.\textsuperscript{1} Onset is from age 8 to age 16 years, with a peak occurrence at 10 to 12 years of age. The frequency of absence seizures in juvenile absence epilepsy is lower than that in childhood absence epilepsy. A higher frequency of generalized tonic-clonic seizures is seen, compared with childhood absence epilepsy, and there is an increased probability of epilepsy continuing into the adult years.\textsuperscript{2}

The precise incidence and prevalence of juvenile absence epilepsy are not known. Wolf and Inoue report that, typically, patients with juvenile absence epilepsy are physically and neurodevelopmentally normal. They also notes that 11\% of patients with the disorder report a family history of epilepsy.\textsuperscript{3}

**CLINICAL PRESENTATION**

Absence seizures are predominant. The impairment of consciousness in juvenile absence epilepsy is moderate and not generally as severe as in childhood absence epilepsy. The level of retained consciousness may vary significantly from seizure to seizure in the same patients. Absence attacks have been termed spatiolepetic, meaning “scarce.” Unlike childhood absence epilepsy they may occur once a day or in a cluster in the hour after awakening. The duration of absence seizures is probably similar to that in childhood absence epilepsy at 4 to 20 seconds. The classic clinical feature is “simple absence” with staring and altered alertness (sometimes “complex absence” with blinking or head nodding).\textsuperscript{2} A large majority of patients will have experienced at least one generalized tonic-clonic seizure, and this may lead to the diagnosis. Myoclonic seizures associated with juvenile absence epilepsy occur in a minority of cases.\textsuperscript{4} Seizures are typically triggered by hyperventilation or sleep deprivation. A photoparoxysmal response is described in a minority of children. However, some experts view the presence of a photoparoxysmal response as excluding a diagnosis of juvenile absence epilepsy.

Tovia and associates\textsuperscript{5} studied 17 patients with juvenile absence epilepsy retrospectively and found that the outcome of patients with the disorder is less favorable than in children with childhood absence epilepsy and that the presence of generalized tonic-clonic seizures is a predictor for poorer outcome, that is, fewer such patients were seizure free when treated.

**EEG FINDINGS**

Interictal background activity is usual normal in juvenile absence epilepsy.\textsuperscript{4} Posterior bilateral delta activity has been described as an interictal phenomenon in children with absence epilepsy more commonly in childhood absence epilepsy and less commonly in juvenile absence epilepsy.\textsuperscript{6} Absence seizures demonstrate generalized “surface negative spikes” seen predominately in the frontocentral head regions, followed by a surface negative slow wave discharge with frequency often faster than 3 Hz (3.5–4.0 Hz).\textsuperscript{6} Overall, the ictal discharges are similar to those in childhood absence epilepsy, although polyspikes on the slow wave are common.

**MRI FINDINGS**

Betting and colleagues\textsuperscript{7} report, in a study of 124 patients with idiopathic generalized epilepsy, that 24\% of patients had nonspecific MRI abnormalities. Woermann and coworkers\textsuperscript{8} note that microdysgenesis in gray and white matter has been found in cases of idiopathic generalized epilepsy; their group performed MRI studies of patients with idiopathic generalized epilepsy and found that 4 of 10 patients with juvenile absence epilepsy demonstrated significant abnormalities of the regional distribution of cerebral gray and subcortical matter.

**GENETICS**

Heron and colleagues\textsuperscript{9} report that in a study of 100 patients with a generalized epilepsy syndrome, including juvenile absence epilepsy, variants in the T-type calcium channel gene CACNA1H, which alters channel properties, are present. In a study of 55 families with idiopathic generalized epilepsy, no mutation of sodium, calcium, or \(\gamma\)-aminobenzoic acid (GABA) channels was found. In this study, phenotypic concordance...
within families of juvenile absence epilepsy probands was 10%. In a genetic survey of families with the four most common subtypes of idiopathic generalized epilepsy (e.g., childhood absence epilepsy, juvenile absence epilepsy, juvenile myoclonic epilepsy, and epilepsy with grand mal seizures on awakening), three different heterozygous mutations in the chloride-channel gene CLCN2 in three unrelated families were found. Durner and coworkers report strong evidence for a genetic locus common to most idiopathic generalized epilepsies on chromosome 18. Sander and associates report that allelic variants of the kainate-selective GluR5 receptor gene (GRIK1) on chromosome 21q22.1 are a factor in the pathogenesis of juvenile absence epilepsy–related phenotypes.

**TREATMENT AND PROGNOSIS**

Wolf and Inoue report that 82% of patients with juvenile absence epilepsy will be seizure free when treated with ethosuximide or valproic acid. Newer-generation agents have shown efficacy, although usually in noncontrolled open-label studies. The seizures of juvenile absence epilepsy may also be worsened by certain antiepileptic drugs.

**CASE 4-1: Juvenile Absence Epilepsy**

**Patient History:** A 12-year-old boy with diagnoses of attention-deficit disorder and hyperactivity was noted to have “staring episodes” beginning over 1 year before this evaluation. About 6 months earlier, the staring episodes became accompanied by some eye blinking and mouth twitching.

**Seizure Risk Factors:** He had a fraternal twin without clinical evidence of epilepsy. He was born at 34 weeks’ gestational age. A family history of epilepsy was reported in a paternal aunt and paternal cousin.

**Medications:**
- Methylphenidate extended-release tablets
- Clonidine

**Video-EEG:** Normal physiologic rhythms were seen. Approximately 3-Hz spike and wave discharges lasting 4 to 6 seconds were associated with behavioral arrest. Two- to 3-Hz spikes were seen associated with some slow waves (Figs. 4-1 to 4-3).

**Outcome:** The patient was discharged on lamotrigine.

**Figure 4-1** EEG: Longitudinal bipolar montage.
CASE 4-1: Juvenile Absence Epilepsy—Cont’d

Figure 4-2 • EEG: Referential montage to ears.

Figure 4-3 • EEG: Transverse montage.
References


