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CHILDHOOD-ONSET IDIOPATHIC GENERALIZED EPILEPSIES: PATHOGENESIS, CLINICAL SPECTRUM, DIAGNOSIS, AND MODERN THERAPEUTIC APPROACHES

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Abstract

Childhood-onset idiopathic generalized epilepsies (IGEs) represent a heterogeneous group of genetically determined epileptic syndromes characterized by generalized seizure types and the absence of structural brain abnormalities. These disorders include a spectrum of syndromes such as benign myoclonic epilepsy of infancy, childhood absence epilepsy, epilepsy with myoclonic-astatic seizures, and epilepsy with myoclonic absences. This review provides a comprehensive analysis of their pathophysiology, clinical manifestations, electroencephalographic features, differential diagnosis, and treatment strategies. Although many forms exhibit favorable outcomes, certain syndromes demonstrate pharmacoresistance and neurocognitive impairment, emphasizing the importance of early diagnosis and individualized management.

Keywords: Idiopathic generalized epilepsy, childhood epilepsy, absence seizures, myoclonus, EEG, treatment

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Introduction

Idiopathic generalized epilepsies constitute a significant proportion of pediatric epilepsies, accounting for approximately 16–30% of cases. These syndromes typically manifest during childhood or adolescence and are characterized by generalized seizure types, including absence, myoclonic, and generalized tonic-clonic seizures.

The defining feature of IGEs is the absence of identifiable structural brain pathology on neuroimaging. Instead, these disorders are believed to arise from functional disturbances in neuronal networks, primarily involving thalamocortical circuits. Genetic predisposition plays a central role, with increasing evidence supporting involvement of ion channel dysfunction (channelopathies), particularly affecting voltage-gated sodium, calcium channels, and GABAergic inhibitory systems.

Pathophysiology and Genetic Basis

The pathogenesis of IGEs is closely linked to abnormalities in neuronal excitability and synchronization. The following mechanisms are considered central:

Dysfunction of voltage-gated ion channels (Na^+, Ca^{2+} channels)

Alterations in GABAergic inhibitory transmission

Abnormal thalamocortical oscillatory activity

Genetic heterogeneity with both monogenic and polygenic inheritance patterns

Mutations in genes such as SCN1A, GABRA1, and CACNA1H have been implicated in various IGE syndromes. Notably, a single genetic mutation may produce different clinical phenotypes, highlighting the complexity of genotype–phenotype correlations.

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Electroencephalographic Features

EEG is a cornerstone in the diagnosis of IGEs. Characteristic findings include: Generalized, bilaterally synchronous spike-wave or polyspike-wave discharges

Frequencies typically:

~3 Hz in absence epilepsy
Faster (>3 Hz) in myoclonic syndromes
Preservation of background rhythms
Possible photoparoxysmal response
Hyperventilation is a strong provocation method, especially in childhood absence epilepsy, while photic stimulation is particularly relevant in photosensitive forms.

Clinical Spectrum of Childhood-Onset IGEs

1. Benign Myoclonic Epilepsy of Infancy

This rare syndrome presents within the first two years of life. It is characterized by brief, generalized myoclonic jerks involving the neck, shoulders, and upper limbs.
Development: usually normal
EEG: generalized polyspike-wave discharges
Prognosis: favorable, remission by 5–6 years
However, some patients may later develop juvenile myoclonic epilepsy, suggesting a continuum of disease.

2. Childhood Absence Epilepsy (CAE)

One of the most common pediatric epilepsy syndromes.
Onset: 4–10 years (peak 5–7 years)
Clinical features:
Frequent brief absences (seconds)
Sudden interruption of activity

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Impaired consciousness

EEG: classic 3 Hz spike-wave pattern

Seizures may occur dozens to hundreds of times per day. Prognosis is generally excellent, though some patients may develop generalized tonic-clonic seizures later.

3. Epilepsy with Myoclonic-Astatic Seizures (Doose Syndrome)

A complex syndrome with multiple seizure types:

Myoclonic seizures

Atonic (drop attacks)

Absence seizures

Generalized tonic-clonic seizures

Onset: 1–5 years

EEG: generalized spike/polyspike-wave activity

Clinical course is variable:

~50% maintain normal cognition

Others develop intellectual disability

Pharmacoresistance is common.

4. Epilepsy with Myoclonic Absences

A rare and often severe epilepsy syndrome:

Prolonged absences (10–30 seconds)

Prominent rhythmic myoclonic activity

EEG: 3 Hz spike-wave discharges

This form is frequently associated with:

Cognitive impairment

Poor response to treatment

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5. Eyelid Myoclonia with Absences (Jeavons Syndrome)

A photosensitive epilepsy syndrome characterized by:

Eyelid jerking

Upward eye deviation

Brief absences

Distinct features:

Strong photic sensitivity

Self-induction behavior

Chronic course with persistence into adulthood

Differential Diagnosis

Differentiating IGEs from other epileptic and non-epileptic conditions is essential:

Focal epilepsies with secondary generalization

Structural epilepsies (tumors, malformations)

Non-epileptic paroxysmal events (syncope, tics)

Metabolic or genetic encephalopathies

MRI is typically normal in IGEs and helps exclude structural causes.

Treatment Strategies

First-line therapy

Valproate – broad-spectrum, most effective across seizure types

Alternative and adjunctive medications

Ethosuximide (absence seizures), Lamotrigine, Levetiracetam, Topiramate, Benzodiazepines

Drug considerations

Ethosuximide is highly effective for absences but not other seizure types

Lamotrigine is preferred in females of reproductive age

Levetiracetam is effective for myoclonic seizures

Contraindicated drugs

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Certain antiepileptic drugs may worsen IGEs:
Carbamazepine, Oxcarbazepine, Phenytoin, Gabapentin
Prognosis
Prognosis varies significantly depending on syndrome:

Syndrome	Prognosis
Childhood absence epilepsy	Excellent
Benign myoclonic epilepsy	Good
Doose syndrome	Variable
Myoclonic absence epilepsy	Often poor

Early diagnosis and appropriate therapy significantly improve outcomes.
Monitoring and Long-Term Management

- Regular EEG monitoring (1–2 times per year)
- Video-EEG in uncertain cases
- Careful withdrawal of therapy after remission

Treatment duration:

- 2–4 years remission → possible withdrawal (CAE, benign forms)
- ≥4–5 years → cautious withdrawal in chronic forms

Conclusion

Childhood-onset idiopathic generalized epilepsies represent a diverse and complex group of disorders with shared neurophysiological mechanisms but distinct clinical phenotypes. Advances in genetics and neurophysiology have significantly improved understanding of these syndromes. Nevertheless, challenges remain in managing pharmacoresistant forms and preventing cognitive impairment. A personalized approach based on syndrome classification, EEG findings, and clinical course is essential for optimal outcomes.

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