EPILEPSY – PATHOPHYSIOLOGY, CLINICAL MANIFESTATIONS, AND DIAGNOSTIC APPROACHES

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Abstract: Epilepsy is a chronic neurological disorder characterized by a persistent predisposition to generate epileptic seizures and by the neurobiological, cognitive, psychological, and social consequences of this condition. It affects approximately 50 million people worldwide, making it one of the most common serious neurological diseases. This article reviews the pathophysiological mechanisms underlying epilepsy, its clinical classification, and current diagnostic strategies.

Keywords: epilepsy, seizures, epileptogenesis, EEG, MRI, focal seizures, generalized seizures, SUDEP.

Introduction

Epilepsy is not a single disease but a spectrum of disorders with diverse etiologies and clinical presentations. The International League Against Epilepsy (ILAE) classifies seizures into focal, generalized, and unknown-onset types. Despite significant therapeutic advances, epilepsy remains associated with stigma, reduced quality of life, and increased mortality.

Neurobiological Mechanisms

The fundamental mechanism of seizures is an imbalance between excitatory and inhibitory neuronal activity. Excessive glutamatergic excitation and reduced gamma-aminobutyric acid (GABA)-mediated inhibition lead to neuronal hyperexcitability and hypersynchrony. Ionic channel dysfunctions, including sodium, calcium, and potassium channelopathies, contribute to genetic epilepsies.

Alterations in synaptic plasticity, neuroinflammation, oxidative stress, and blood–brain barrier dysfunction further contribute to epileptogenesis. Structural brain abnormalities such as cortical dysplasia, hippocampal sclerosis, tumors, and vascular malformations are frequent substrates of focal epilepsy.

Clinical Manifestations

Clinical manifestations vary widely depending on seizure type and epileptogenic zone. Focal seizures may present with motor, sensory, autonomic, or psychic symptoms, while generalized seizures include absence, myoclonic, tonic, clonic, and tonic–clonic seizures. Postictal symptoms such as confusion, headache, and transient neurological deficits are common.

Children and elderly patients may exhibit atypical presentations, often complicating early diagnosis.

Diagnostic Approaches

Electroencephalography (EEG) remains the cornerstone of epilepsy diagnosis, allowing detection of interictal epileptiform discharges and seizure patterns. Video-EEG monitoring is essential for epilepsy classification and presurgical evaluation.

Neuroimaging, particularly high-resolution magnetic resonance imaging (MRI), is used to identify structural causes. Functional imaging techniques such as positron emission tomography (PET) and single-photon emission computed tomography (SPECT) assist in localizing the epileptogenic zone in refractory cases.

Differential Diagnosis

Conditions that can mimic epileptic seizures include syncope, psychogenic nonepileptic seizures, transient ischemic attacks, and sleep disorders. Accurate diagnosis is essential to avoid inappropriate treatment.

Complications and Comorbidities

Epilepsy is associated with cognitive impairment, depression, anxiety disorders, and increased risk of sudden unexpected death in epilepsy (SUDEP). Social limitations and decreased educational and occupational opportunities are common among patients.

Conclusion

Epilepsy is a complex neurological disorder requiring a comprehensive and individualized diagnostic approach. Advances in neuroscience continue to refine understanding of epileptogenesis and improve diagnostic accuracy.

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