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Oral Presentation

Approach to Syndromic Epilepsy

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Abstract

Epilepsy can be observed during the course of many inborn errors of metabolism (IEMs) usually as part of a large clinical spectrum. However, several IEMs may manifest with inaugural epileptic seizures. Inborn errors of metabolism (IEMs) represent poorly known causes of epilepsy in adulthood. Although rare, these are important to recognize for several reasons:

1- some IEMs respond to specific treatments

2- some antiepileptic drugs interfering with metabolic pathways may worsen the clinical condition

3- and specific genetic counselling can be provided.

When should one suspect an IEM in an epileptic patient?

In a patient with epilepsy, it should be stressed that several clinical, radiological or electrophysiological features suggest an IEM:

1-The epileptic syndrome does not match with any classical epilepsy syndrome i.e. atypical electro-clinical presentation, atypical response to antiepileptic drugs or mixture of generalized and partial epileptic manifestations (for example association of myoclonus and partial seizures in a given patient;

2-Progressive myoclonic epilepsy;

3- Association with other neurological impairments (cerebellar, pyramidal, etc.) or with unexplained mental retardation, or other organ disorders (eyes, muscles spleen, etc);

4-Familial history suggestive of a genetic disease;

5-Seizures related to the times of eating, fasting, protein-rich meal;

6-Inefficacy or worsening with classical antiepileptic drugs;

7-Unexplained status epilepticus;

8-Abnormalities on proton magnetic resonance spectroscopy: for instance, creatine deficiency or increased in lactate;

9-EEG showing slowing of the background activity or photo-paroxysmal responses during the photic intermittent stimulation at low frequencies (1–6 H).

To recognize the type of IEM, clinical history needs to be analyzed considering the following points;

(a) Age at onset

(b) clinical presentations

(c) Pattern of inheritance

(d) Key clinical symptoms and signs with special focus on sites of neuraxis and extra-neural involvement

(e) Course of the disease and

(f) Severity of impairment.

Keywords: Epilepsy, Impairment, Patient, Metabolic.

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