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Epileptic Encephalopathies: an update

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Disclosures

- Advisory board member, consultant or lecturer in agreement with my Institution
  - Amzell, Arvelle, Biomarin, Eisai, GW Pharmaceuticals, Neuraxpharma, Shire, Takeda, UCB Pharma, and Zogenix
- Received authorship royalties
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- I fully assume my collaborations with Pharma, they are always totally transparent, and represent provided work based on expertise
- I personally assume the opinions expressed in this presentation

Learning Objectives

- Share up to date knowledge on definitions of neuro-developmental and epileptic encephalopathies
- Describe the main aetiological categories involved
- Identify and describe most common seizure semiology observed
- Share up to date knowledge on best diagnostic clinical practices and investigations
- Share up to date knowledge on best treatment strategies

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Evolution of the concept of “epileptic encephalopathy”

“**Epileptic encephalopathy**” = where the epileptic activity itself contributes to severe cognitive and behavioural impairments above and beyond what might be expected from the underlying pathology alone (e.g., cortical malformation). Global or selective impairments can worsen over time. These impairments can be seen along a spectrum of severity and across all epilepsies, and can occur at any age.

“**Developmental encephalopathy**” :

- can pre-exist: anoxic-ischaemic encephalopathy prior to epileptic spasms (followed by further stagnation or further regression) ;
- Appear on a background of normal development : Dravet syndrome at the age of 1-2 years of age while EEG may still be normal;
- Epilepsy may settle down relatively early in the child’s history, but the developmental consequences may remain profound (KCNQ2 or STXBP1 encephalopathy)

Evolution of the concept of “epileptic encephalopathy”

Epileptic encephalopathy and inter-ictal EEG

- In an epileptic encephalopathy, it is assumed that the abundant epileptiform activity interferes with development resulting in cognitive slowing and often regression, and sometimes is associated with psychiatric and behavioural consequences.
- The epileptiform activity can cause regression in an individual with normal development or pre-existing developmental delay, who then shows developmental plateauing or regression.
- **A key component of the concept is that amelioration of the epileptiform activity may have the potential to improve the developmental consequences of the disorder**.

Predefine your treatment goals

Good examples of "Epileptic Encephalopathy":

- Regression in a normal child, with isolated epileptic spasms, hypsarrythmia and negative screening (including MRI; Metabolic; Genetic)
- Regression in a normal child, with isolated epileptic spasms, and a focal dysplasia;
- Regression in a child with Continuous Spike-Waves during Slow Sleep (ESES)
- Regression in a normal child with epilepsy related to a DNET; to Sturge-Weber; etc.
- Lennox-Gastaut syndrome **depending on aetiology**

Good examples of "Epileptic **AND** Neurodevelopmental Encephalopathy":

- Regression in a child with a history of anoxic-ischaemic encephalopathy (acquired/fixed) that develops electro-clinical characteristics of West syndrome
- Regression in a child with a Progressive Myoclonic Epilepsy
- Dravet syndrome; CDKL5 encephalopathy; KCNQ2 encephalopathy; SYNGAP1; Other...
- Lennox-Gastaut syndrome **depending on aetiology**