Clinical commentary

Epileptic Disord 2019; 21 (1): 117-21

Berardinelli-Seip syndrome and progressive myoclonus epilepsy

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Received August 28, 2018; Accepted December 01, 2018

Epileptic Disorders

Celia's encephalopathy: distinguishing features in relation to "classic" lipodystrophy type 2

- Early-onset myoclonic seizures
- Photosensitivity on EEG
- Neurodegenerative encephalopathy
  - Ataxia
  - Action myoclonus
  - Progressive cognitive impairment
  - Cortical myoclonus
- Progressive cerebral atrophy
- Exitus between 8 and 12 years of age

Epileptic Disorders

Key Points

- 3-year-old boy with Progressive Myoclonus Epilepsy and Lipodystrophy type 2
- Moderate cognitive impairment, hyperactivity, progressive ataxia
- Slow and disorganized EEG background activity with trains of atypical high-voltage multifocal spike-wave complexes
- Normal brain MRI scan
- Clinical characteristics
  - Myoclonus, atypical absence seizures, myoclonic absence seizures, drop attacks
  - Progressive degeneration of EEG background activity
  - Progression towards refractory status epilepticus
  - Progressive movement and gait disorder

- Lipodystrophy type 2 may be considered in rapidly progressive myoclonus epilepsies associated with metabolic disorders.
- Application of a vagal nerve stimulator might be a valid palliative therapy

Epileptic Disorders