

Diaper changing-induced reflex seizures in CDKL5-related epilepsy

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CDKL5-related epilepsy

REFLEX SEIZURES Diaper-changing reflex seizures



Ictal Polygraphic recording during diaper changing

During EEG recordings, we documented focal seizures induced by diaper changing: the events clinically started with staring, related to EEG paroxysmal low-voltage rhythmic theta activity in the left centro-parietal and vertex regions (arrows), followed by generalized delta activity and a series of spasms characterized by high-voltage slow complexes related to EMG periodic contractions of the four limbs (arrowheads).

Upper panel and lower panel are consecutive.

R-delt= Right deltoideus; R-quad= Right quadriceps femoris; L-delt= Left deltoideus; L-quad= Left quadriceps femoris.

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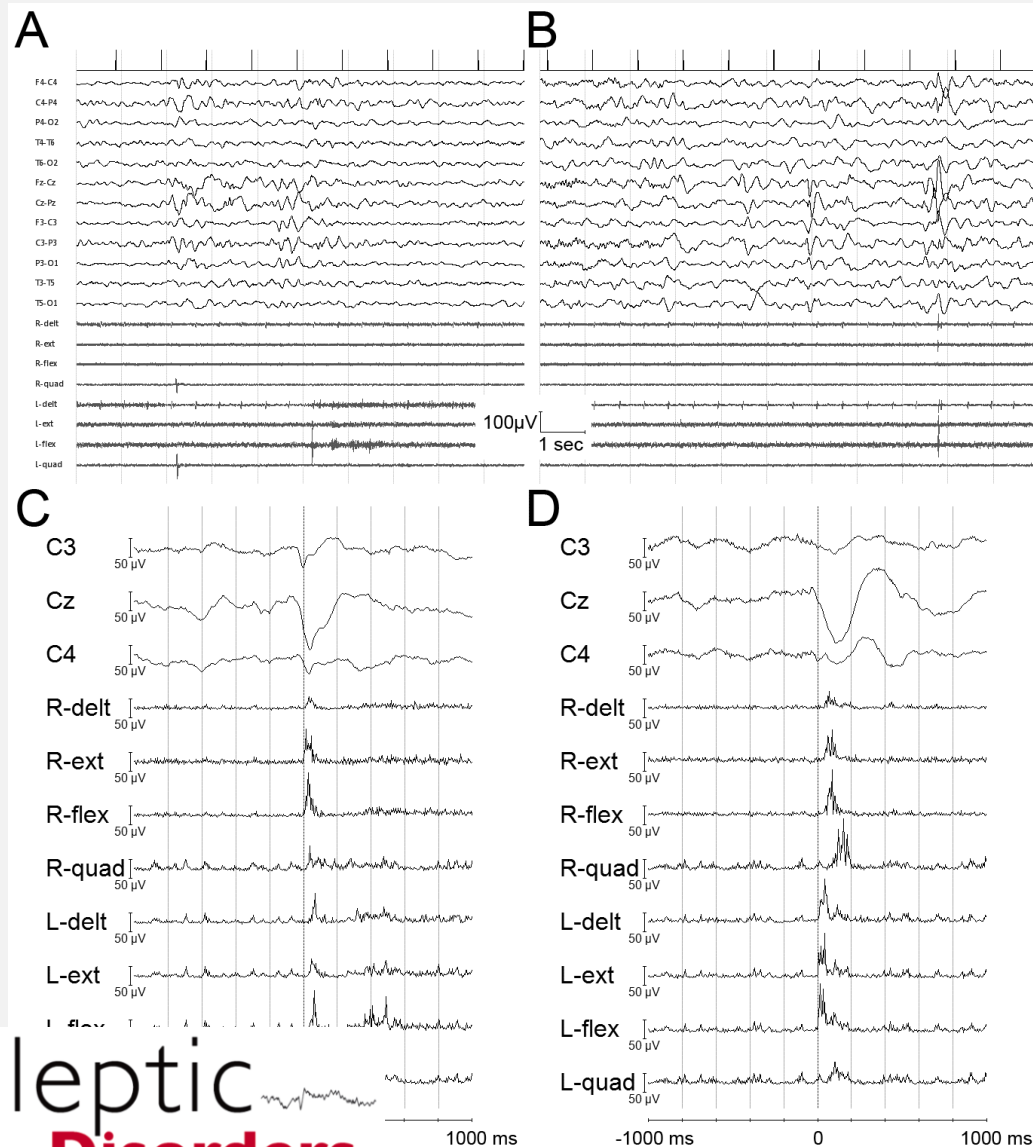
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CORTICAL MYOCLONUS



EEG during sleep: sporadic isolated spikes in the vertex region and both right and left centro-parietal regions, often related to single myoclonic jerks of the lower limbs (panel A) or upper limbs (panel B), asynchronous on both sides.

Back-averaging of myoclonic events: evidence of co-contraction of agonist muscles, followed by a contralateral myoclonic event (probably due to a propagation of the discharge); the myoclonic event is preceded by a positive transient discharge, maximal on the central vertex and central region in the hemisphere contralateral to the earliest myoclonic potential (panel C, D).

EMG abbreviations: R-delt= Right deltoideus; R-ext= Right extensor carpi; R-flex= Right flexor carpi; R-quad= Right quadriceps femoris; L-delt= Left deltoideus; L-ext= Left extensor carpi; L-flex= Left flexor carpi; L-quad= Left quadriceps femoris.

CDKL5-related epilepsy

Early-onset drug-resistant epilepsy

Cognitive impairment with absent language

Hand stereotypies

Deceleration of head growth

- Focal seizures as first manifestation of disease (< 5 months of age)
- Infantile spasms
- Myoclonic encephalopathy



Early features suggesting
DIFFUSE CORTICAL HYPEREXCITABILITY

REFLEX SEIZURES
Diaper changing-
induced reflex seizures

**CORTICAL
MYOCLONUS**

as documented in our patient