■ Original article

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Epileptic Disorders

A characteristic occipital epileptiform EEG pattern in ADCK3-related mitochondrial disease

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ADCK3-related disease

- Rare mitochondrial disorder associated with abnormality of coenzyme Q_{10} metabolism
- Ataxia and epilepsy are common
- The phenotype overlaps with other mitochondrial diseases
- CoQ_{10} supplementation may be beneficial
- Early diagnosis i crucial

AIM

We noted a remarkable epileptiform pattern in ADCK3-related disease and wished to assess the evolution of EEG characteristics over time

METHOD

- All EEG recordings of 4 known patients were reviewed.
- A total of 96 recordings over 15-32 years were studied, mean 24 per patient (range 17-28):
  - 50 digital recordings, including four long-term
  - 46 selected paper segments.
- EEG graphoelements were classified according to the standardized computer-based organized reporting of EEG (SCORE)
- The evolution of EEG features was assessed.

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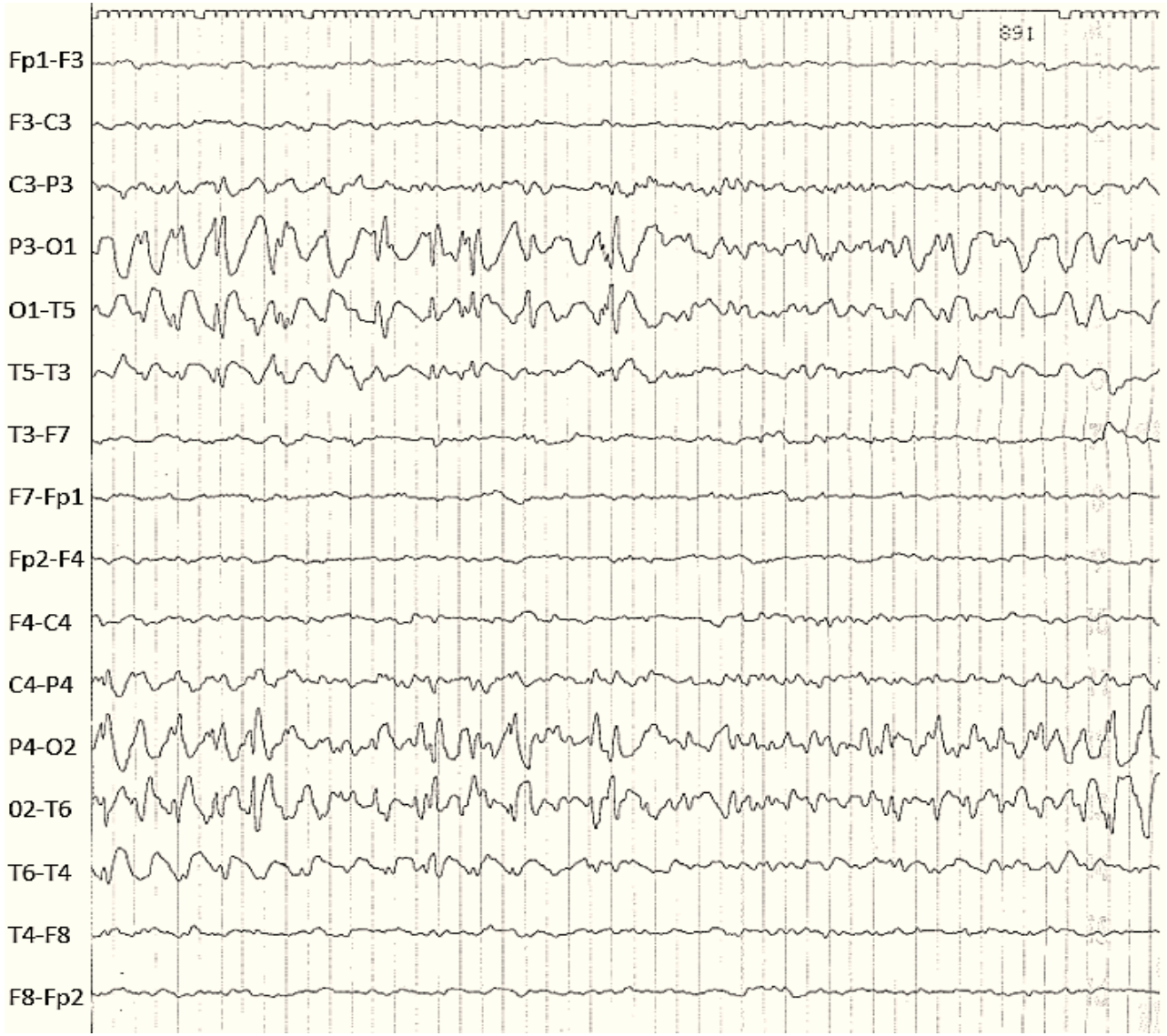
RESULTS

EEG findings

- Three patients showed prominent bilateral asynchronous and synchronous epileptiform discharges in occipital and posterior-temporal regions
- The pattern occurred continuously, nearly continuously or in prolonged runs.
- The findings remained stable over many years.

Patient 1

(paper segment)



Montage:
Longitudinal bipolar

First recording at age 10: Continuous bilateral synchronous and asynchronous epileptiform activity with discrete slowing in post-temporal and occipital regions

Patient 1

Montage:
average reference

At age 22: The principal findings of continuous bilateral synchronous and asynchronous epileptiform activity with occipital predominance persisted.

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Patient 1

Montage:
average reference

At age 38: EEG pattern unchanged

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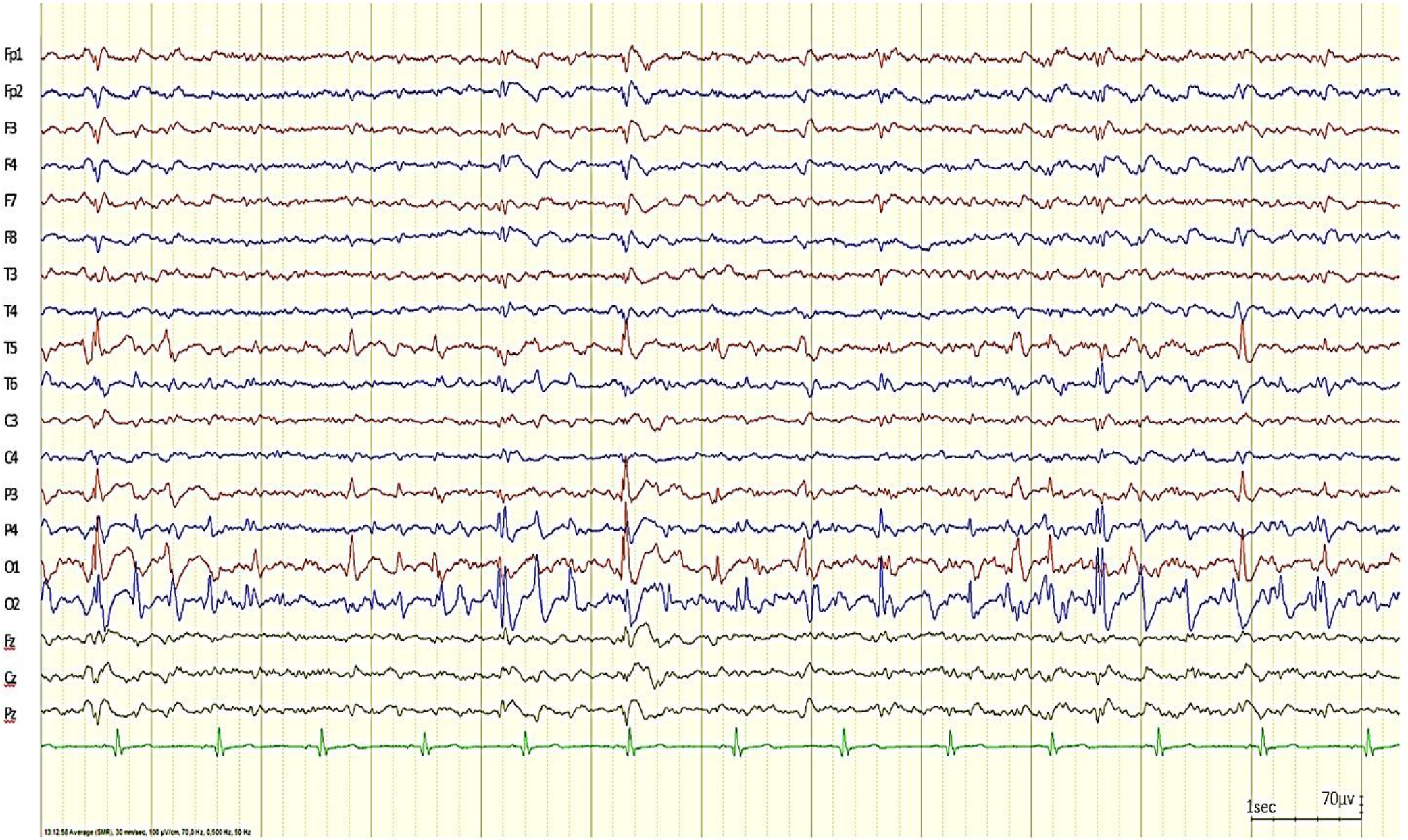
Patient 2

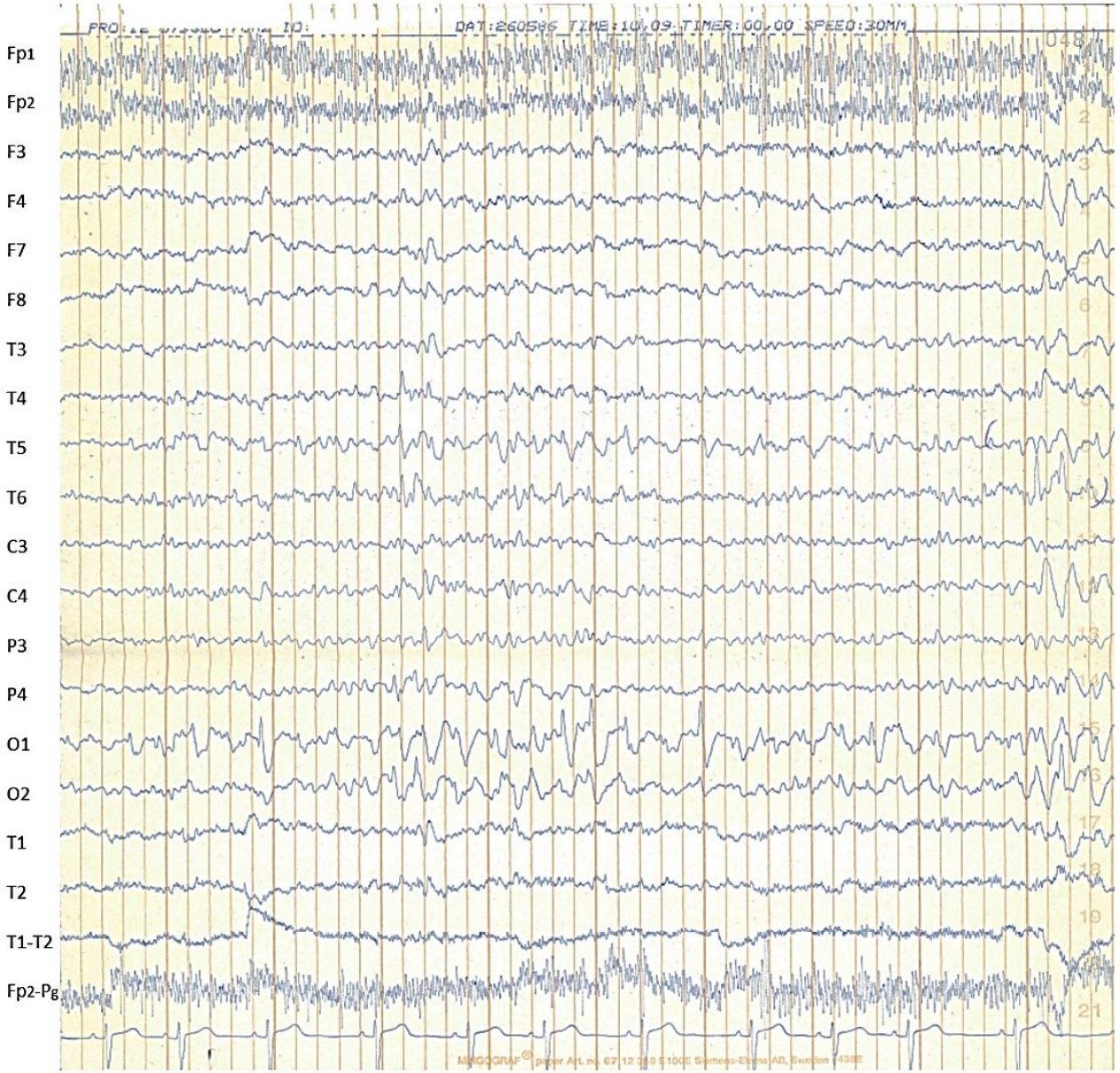
Montage:
average reference



EEG at age 18. Continuous/nearly continuous bilateral epileptiform discharges over posterior head regions, mainly the occipital and posterior temporal areas.

Patient 3

(paper segment)

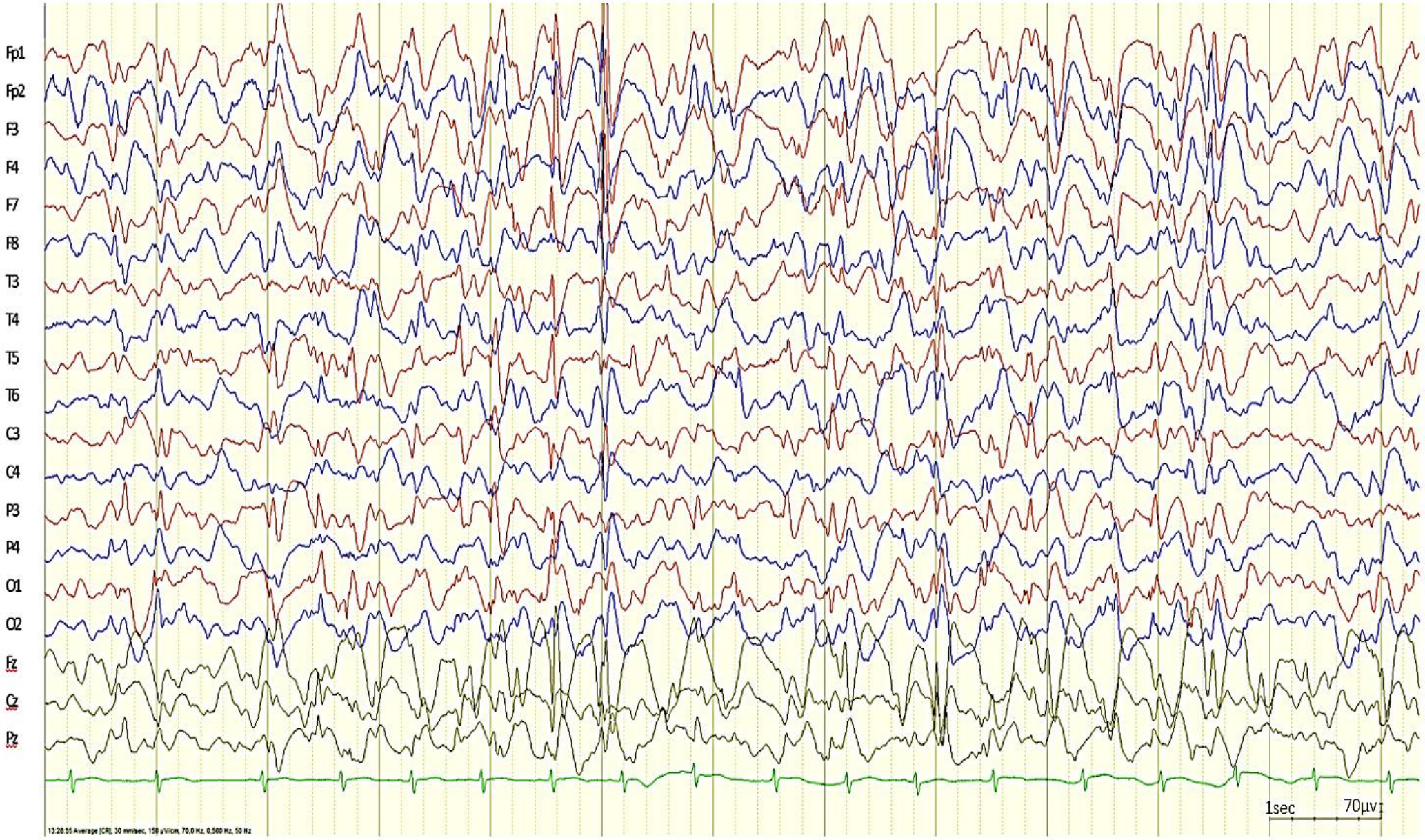


Montage:
average reference

EEG at 10 years. Nearly continuous or prolonged runs of bilateral asynchronous and synchronous epileptiform discharges in occipital and posterior-temporal leads

Patient 4

Montage:
average reference



Interictal EEG at age 7: Combined multifocal and bilateral synchronous and asynchronous epileptiform discharges with spike focus in the posterior region mostly left side.

CONCLUSION

- Continuous/nearly continuous bi-occipital spike-waves may serve as a biomarker for this potentially treatable condition.

- This EEG pattern might help to differentiate ADCK3- related disease from the more common POLG-related disease, which is usually characterized by lateralized or focal slowing with more sporadic epileptiform elements of similar topography.

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