

Rett syndrome due to mutation in the *MECP2* gene and electroencephalographic findings

Síndrome de Rett devido a mutação no gene MECP2 e achados eletroencefalográficos

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Arq. Neuro-Psiquiatr. 2024;82(8):s00441787801.

A 6-year-old girl with *MECP2*-positive Rett syndrome presented for investigation of seizures. The electroencephalogram (EEG) showed markedly disorganized cerebral activity, epileptiform paroxysms and a quasiperiodic pattern, followed by attenuation after paroxysmal activity (► **Figures 1** and **2**). In *MECP2*-positive patients, there is a progression of EEG findings in stages. First, seizures are not a prominent feature, and the EEG may be normal. Second, focal spikes may be observed, especially in the centrotemporal regions. Third, there is an accentuation of epileptiform activity, abnormalities during sleep, and the presence of bilaterally synchronous discharges of pseudoperiodic delta activity and generalized rhythmic spike discharges. Fourth, there are theta waves in the central and frontal regions, multifocal epileptiform activity in the waking state, and generalized slow spike and wave activity during sleep.^{1,2}

Authors' Contributions

MRC: conceptualization and design of the work, data acquisition, analysis, and interpretation, and writing of

the manuscript; TTC: data acquisition and writing of the manuscript; PSO, PVFC: analysis and interpretation of data, review of the manuscript; PELC: analysis and interpretation of data, and writing and review of the manuscript. All authors have approved the final version of the manuscript and agreed to be responsible for all aspects of the work.

Conflict of Interest

The authors have no conflict of interest to declare.

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received
February 3, 2024
accepted
April 6, 2024

DOI <https://doi.org/10.1055/s-0044-1787801>.
ISSN 0004-282X.

Editor-in-Chief: Hélio A. G. Teive
Associate Editor: Luciano de Paola.

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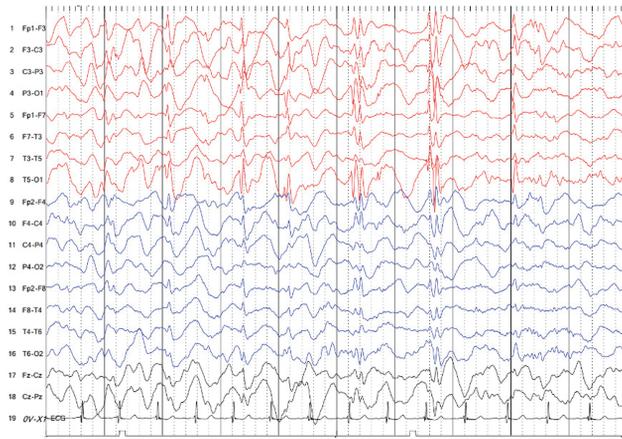


Figure 1 Bipolar longitudinal. Source: Image collection of the Clinical Neurophysiology Sector of Hospital Universitário de Brasília. (Sensitivity: 10 μ V; High filter: 70 Hz; Low filter: 1 Hz).

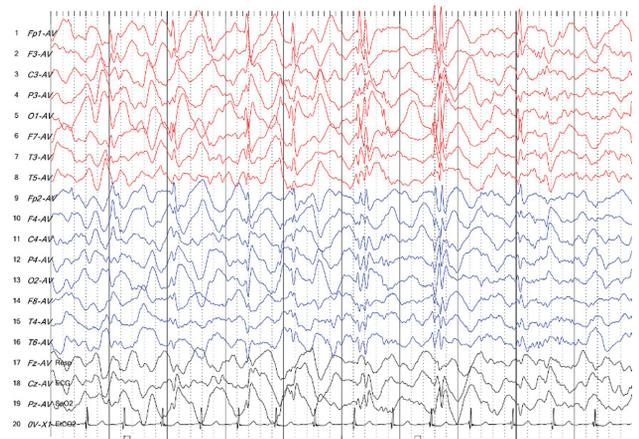


Figure 2 Average referential. Source: Image collection of the Clinical Neurophysiology Sector of Hospital Universitário de Brasília. (Sensitivity: 10 μ V; High filter: 70 Hz; Low filter: 1 Hz).

Appendix A

Quiz

1. What is the main gene involved in typical cases of Rett syndrome?

- a) *CDKL5*
- b) *FOXP1*
- c) *MECP2*
- d) *SCNA1*

answer: c

2. In a patient with Rett syndrome due to a mutation in the *MECP2* gene, in how many stages does the progression of electroencephalographic findings occur?

- a) 3
- b) 4
- c) 5
- d) 6

answer: b

3. A patient with Rett syndrome due to a mutation in the *MECP2* gene who presents a pattern of pseudoperiodic discharges on the electroencephalogram can be classified into which stage?

- a) 3
- b) 4
- c) 5
- d) 6

answer: a