Clinical commentary

Epileptic Disord 2017; 19 (3): 357-61

Practical clues for diagnosing WWOX encephalopathy

Oana Tarta-Arsene [1,2], Diana Barca [1,2], Dana Craiu [1,2], Catrinel Iliescu [1,2]

[1] "Prof. Dr. Alexandru Obregia" Clinical Hospital, Pediatric Neurology, Bucharest
[2] "Carol Davila" University of Medicine and Pharmacy, Pediatric Neurology Discipline, Bucharest, Romania

Received December 23, 2016; Accepted June 04, 2017

Epileptic Disorders

CASE STUDY

- We report a non-dysmorphic boy, with normal cranian perimeter, born at term from a non-consanguineously family, with:
  - epilepsy with focal as well as primary generalized seizures (infantile spasms) from the age of 4 weeks;
    - drug resistant from the age of 6 months;
  - profound global motor retardation, with hypokinetic movements, and hypotonia;
  - lack of vision or hearing, with a significant impairment of swallowing;
  - he died at the age of almost 3 years;

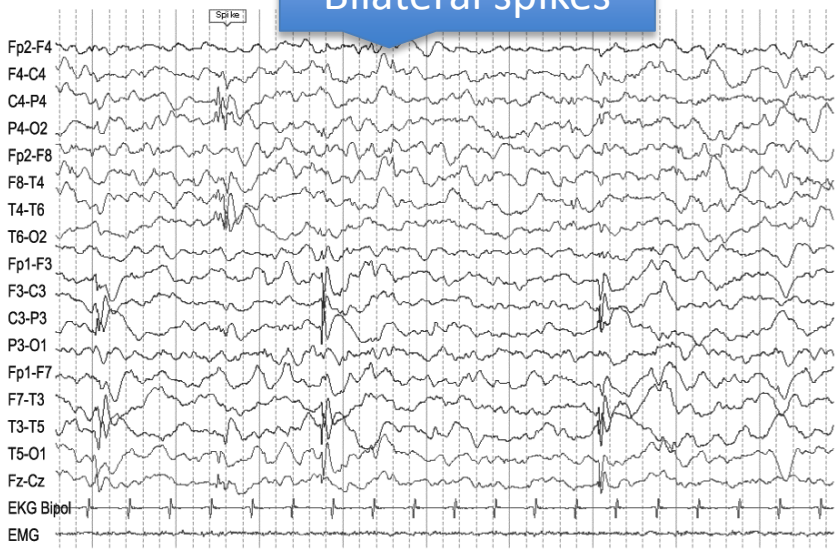
- Cerebral imaging:
  - 5 weeks: only thin corpus callosum;
  - 2.6 years: significant cortico-subcortical cerebral atrophy + flat midbrain;
    (despite the cerebral atrophy, the child had a normal cranian perimeter)

- EEG: persistent sleep hypsarrhythmia;
- WES:
  - heterozygous variant in intron 2 of the WWOX gene, c.173-1G>T

Epileptic Disorders

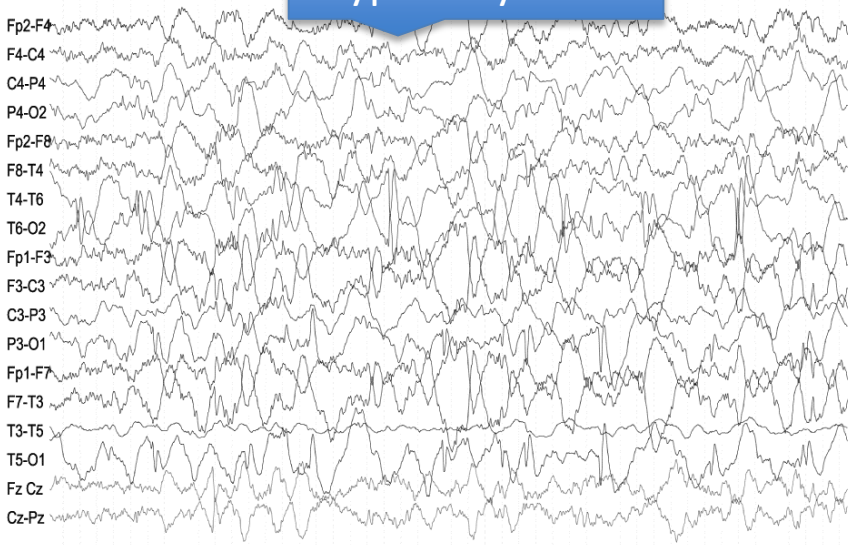
2 YEARS OLD

AWAKE EEG:
Bilateral spikes

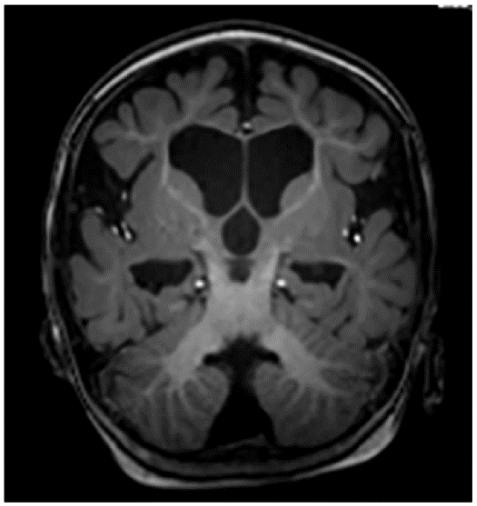


A.

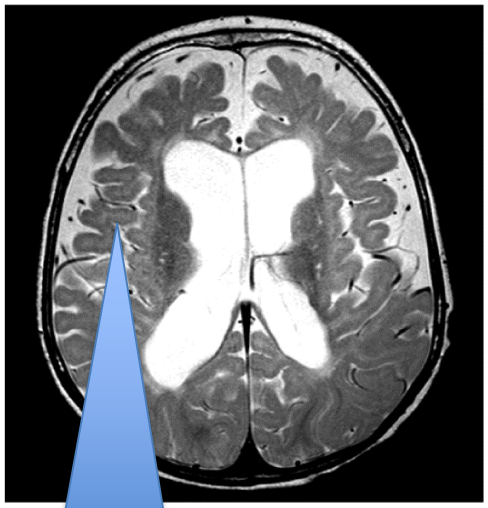
SLEEP EEG:
hypsarrhythmia



B.

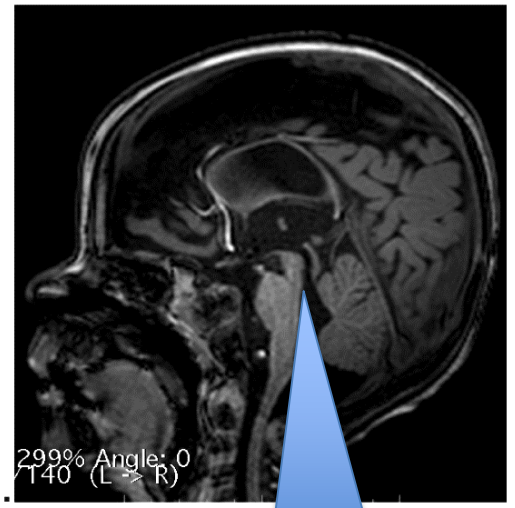


C.



D.

SUPRATENTORIAL ATROPHY



E.

FLAT MIDBRAIN

Epileptic Disorders

REFERENCES:

Ben-Salem S, Al-Shamsi AM, John A, Bassam R. Ali BR, Lihadh Al-Gazali L A Novel Whole Exon Deletion in WWOX Gene Causes Early Epilepsy, Intellectual Disability and Optic Atrophy. J Mol Neurosci (2015);56:17–23.

Mignot C, Lambert L, Pasquier L, Bienvenu T, Delahaye-Duriez A, Boris Keren B et al. WWOX-related encephalopathies: delineation of the phenotypical spectrum and emerging genotype-phenotype correlation. J Med Genet (2015); 52:61–70.

Tabarki B, AlHashem A, AlShahwan S, Alkuraya FS, Gedela S, Zuccoli G. Severe CNS involvement in WWOX mutations: Description of five new cases. Am J Med Genet (2015); Part A 9999A:1–5.