Wolf-Hirschhorn syndrome and Dup15q – review of the literature and case report

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- Wolf-Hirschhorn Syndrome (WHS) is caused by the subtelomeric deletions of chromosome 4p and it is characterized by: growth delay, intellectual disability, seizures, and distinctive craniofacial features.

- Dup 15q syndrome is caused by duplications of the Prader-Willi/Angelman critical region and is characterized by: hypotonia neurodevelopmental delay, variable intellectual disability (ID), autism spectrum disorders (ASD), and epilepsy.

OBJECTIVES

The objective of this case presentation was to describe the clinical manifestation, EEG pattern and share our experience with seizures in a patient with WHS and Dup15q and review the current data on this subject.

MATERIALS & METHODS

- We present a case of a 2 year old boy with recurrent febrile seizures. His personal history revealed two genetic syndromes, Wolf-Hirschhorn (WHS) syndrome and Dup15q syndrome (Dup15q) (Microdeletion 15q13.2q13.3 both of which can cause seizures in early childhood.
- Patient evaluation:
  1. medical history
  2. neurological exam
  3. blood work-up
  4. videoEEG
  5. MRI

•A literature review was performed using MEDLINE data base.

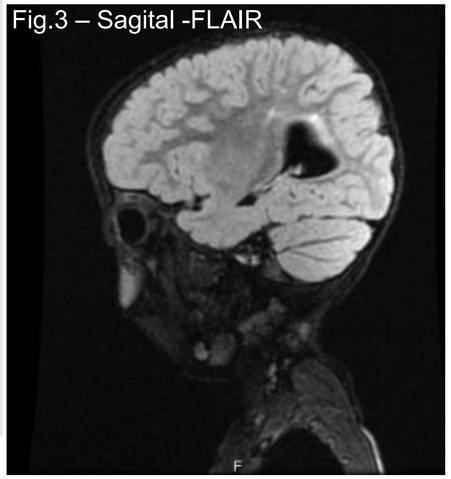

Fig.3 – Sagital -FLAIR

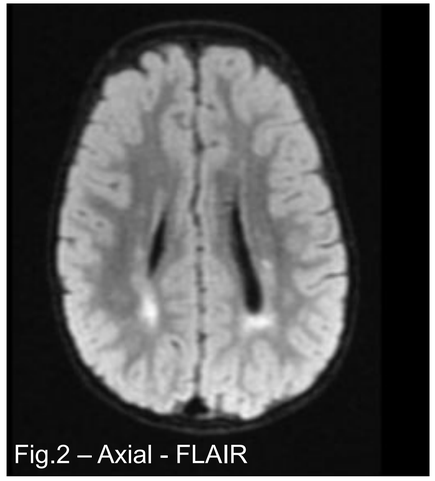

Fig.2 – Axial - FLAIR

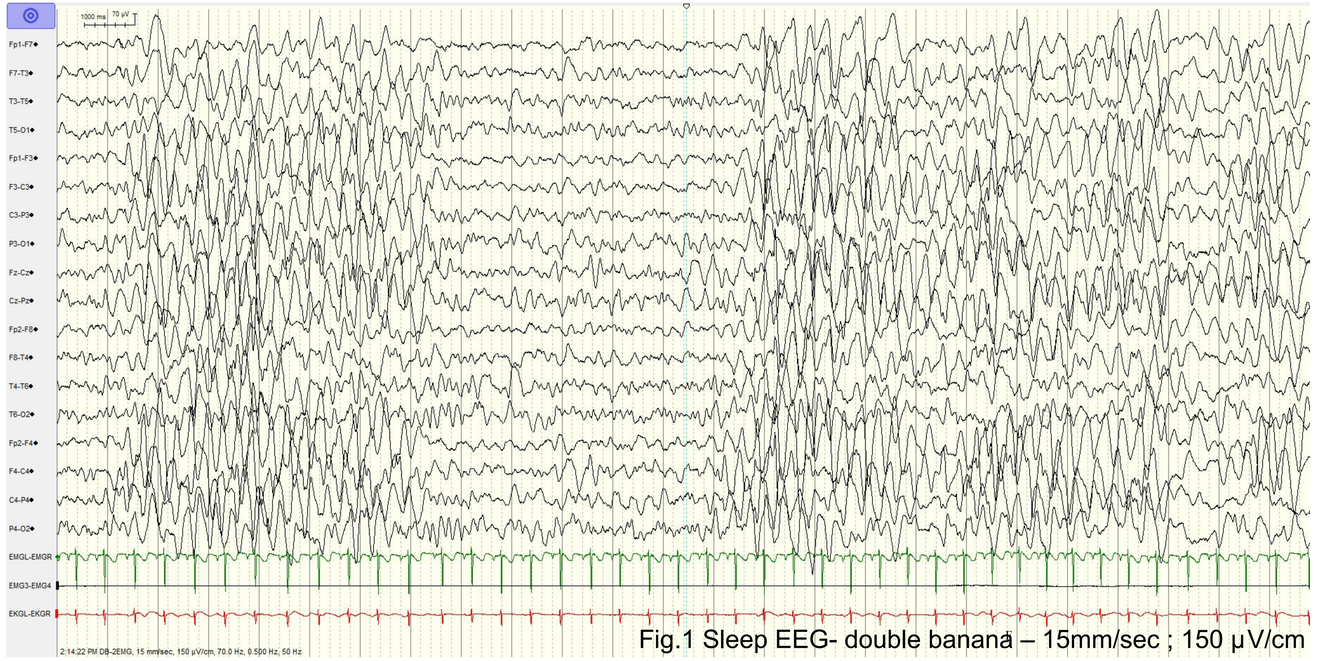

Fig.1 Sleep EEG- double banana – 15mm/sec ; 150 µV/cm

RESULTS

- Neurodevelopmental delay – Patient age – 2 years ; development age of 7 months
- The EEG showed intermittent bursts of generalized high amplitude slow waves, activated by sleep, characteristic for WHS. (Fig.1)
- The EEG trace had similar pattern to those described in the literature as characteristic for WHS: long bursts of high-amplitude, sharp element spike/wave complexes at 2-3.5Hz; frequent high-amplitude spikes-polyspikes/wave complexes at 4 to 6Hz, over the posterior regions.
- MRI demonstrated ventricular asymmetry, periventricular T2- weighted hyperintensity which may be secondary hypoxic-ischemic injury. (Fi.g.2,3)
- Antiseizure treatment with topiramate was initiated, with a good response.

CONCLUSIONS

The presented clinical case is worthy of notice since the observed genetic syndromes can share multiple features, including seizures, with different prognosis. According to the literature seizures in WHS have a good prognosis while in Dup15q syndrome are difficult to control.

The EEG pattern and treatment response might suggest WHS as the etiology of seizures and a good outcome of the patient.

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