

Intracranial Developmental Venous Anomaly presenting with Seizures in a patient with Wilson's Disease: A Case Report

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Developmental venous anomaly (DVA) are the most frequently cerebral malformation with an incidence of up to 2.6%. The etiology of DVAs remains uncertain¹. DVA rarely cause symptoms and are often incidental findings, and if symptomatic may rarely present with focal neurologic deficits or seizure². Wilson's disease (WD) is a genetic disorder with wide diversity of presentations; commonly by movement disorders. seizure may be the initial neurological symptom in approximately 10% of patients with WD³.

An 18-years-old male patient, positive consanguinity, with family history of hepatic diseases and unexplained deaths. Complained of gradual and progressive segmental dystonic posturing in neck and Right upper limb associated with parkinsonian features. There was past history of recurrent attacks of tonsillitis and arthritis that was misdiagnosed as Rheumatic fever and started long acting penicillin; associated with recurrent attacks of epistaxis which caused patient anaemia.

5 months later, patient presented to Kasralainy hospital, Movement disorders clinic with recent history of two attacks of generalized convulsions; associated with disease progression and development of psychiatric and behavioral symptoms, together with worsening of previous symptoms.

Slit lamp examination revealed Kayser–Fleischer rings [Figure 1]. Laboratory investigations revealed low Serum Copper, low Serum Ceruloplasmin, and normal 24hr Urinary Copper, so we performed Penicillamine challenge test which was positive. MRI Brain done and revealed Face of Giant panda + Left parietal venous malformation [Figure 2]. Electroencephalogram done but no abnormality detected. Genetic Analysis done with no mutation detected.

A diagnosis of WD was made based on Scoring system developed at the 8th International Meeting on Wilson's disease, Leipzig 2001 (patient total score =6). Patient started medical treatment D- penicillamine 250mg once/day to be increased gradually + Zinc TID, together with Antiepileptic medications, and symptomatic

treatment in the form of Quetiapine 25mg BID + L-Dopa/Carbidopa 125mg/12.5mg TID and Biperiden 2mg BID. The patient was instructed to follow a dietary pattern with avoidance of copper rich food.

During follow up visit, no seizures and no initial worsening detected with no major medications adverse effects. Patients showed minimal improvements regarding dystonia and parkinsonian features.

Seizures can complicate both WD or DVAs, but it remains difficult to attribute seizures to either disorders, and it's unknown whether seizures incidence will increase if the disorders coexist. however, the therapeutic approach of coexisting cases may not differ from the management of each disorder alone. DVAs are treated conservatively in most cases, while WD is treated with de-coppering therapy and epilepsy with appropriate antiepileptic medications¹. Urinary copper is not mandatory for diagnosis of WD and can be normal; in this case Penicillamine challenge test can help in diagnosis and further studies on adults is encouraged due to promising results. Genetic testing still remains impractical in most clinical settings and no mutation can be detected in many cases as there is over 520 WD mutations have been identified till now⁴.

In conclusion, to the best of our knowledge, our case is the second in all literature that describes seizures in a WD patient due to Intracranial developmental venous anomaly.

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Consent: Written informed consent was obtained from the patient for publication of this Case report, including any associated images. A copy of the written consent is available for review by the Editor(s) of this journal.

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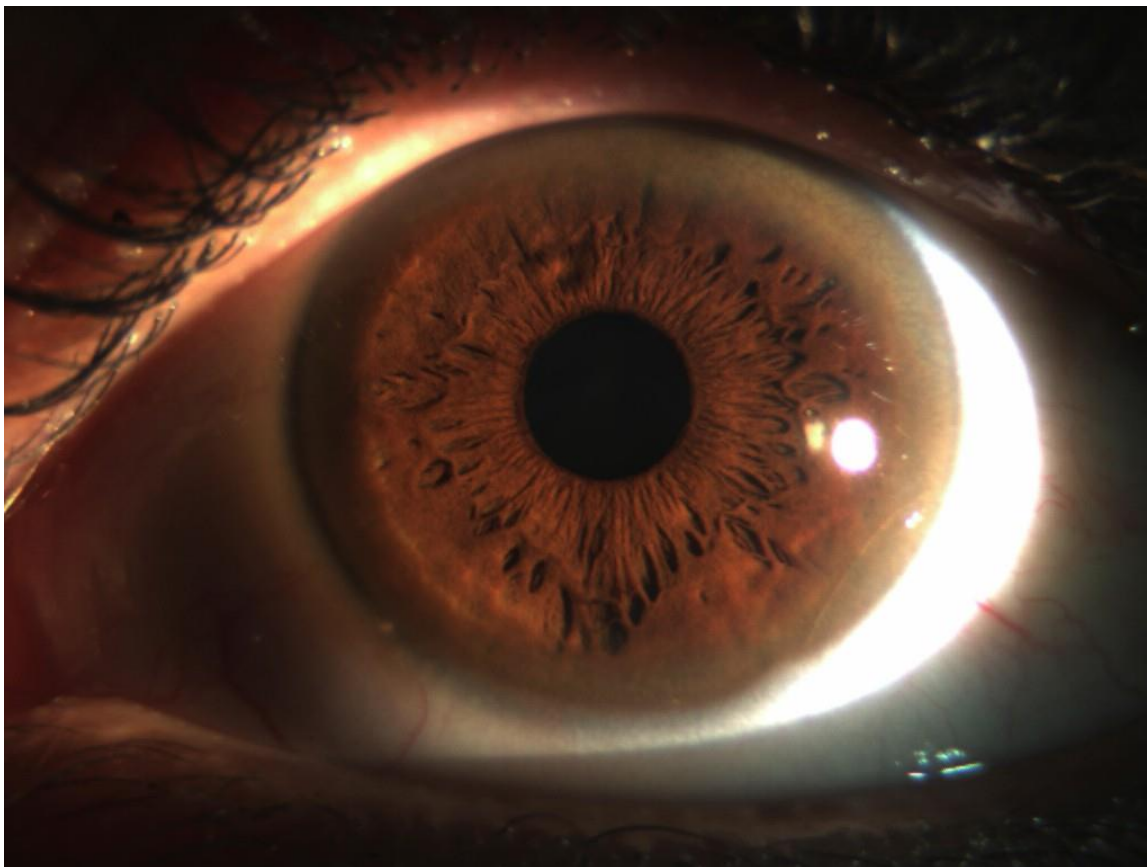


Figure 1

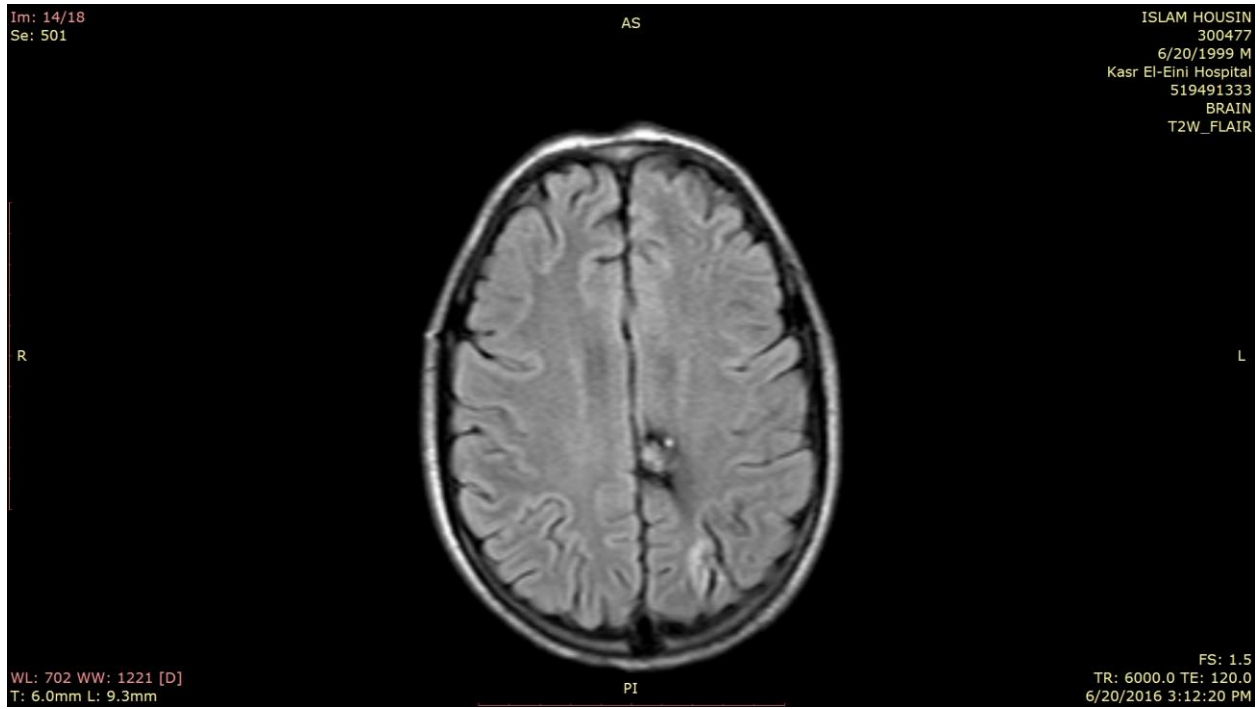


Figure 2a

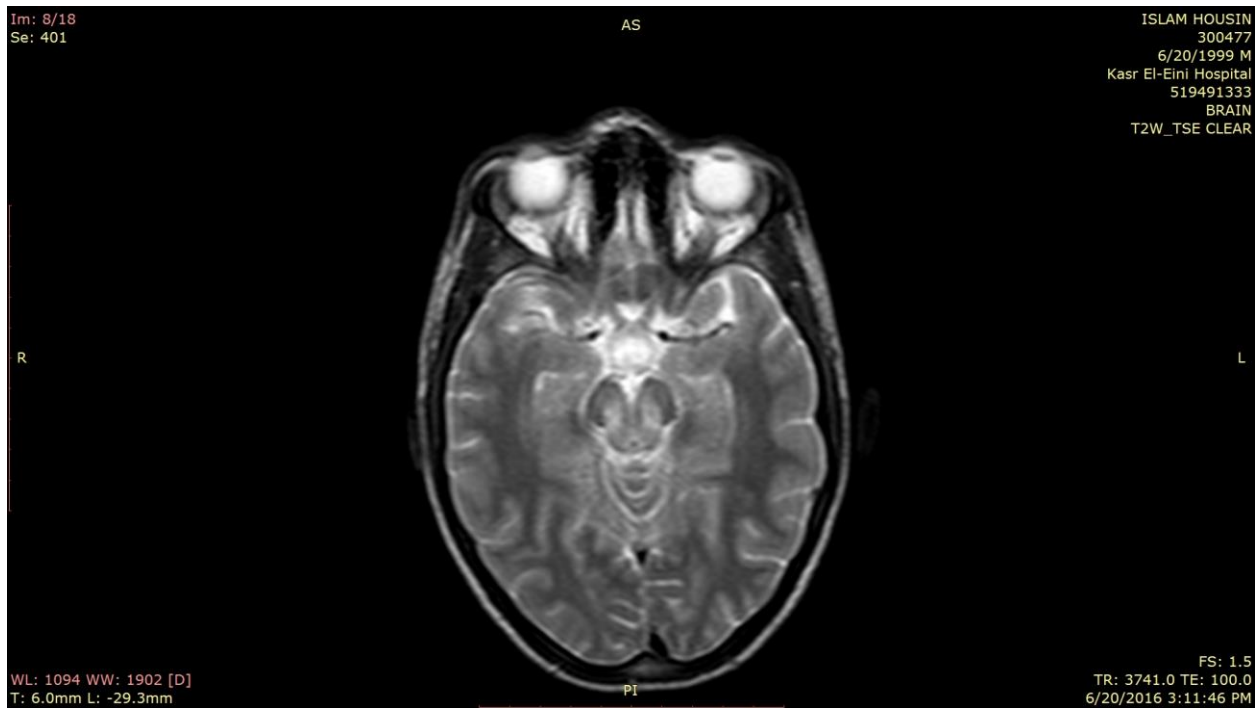


Figure 2b

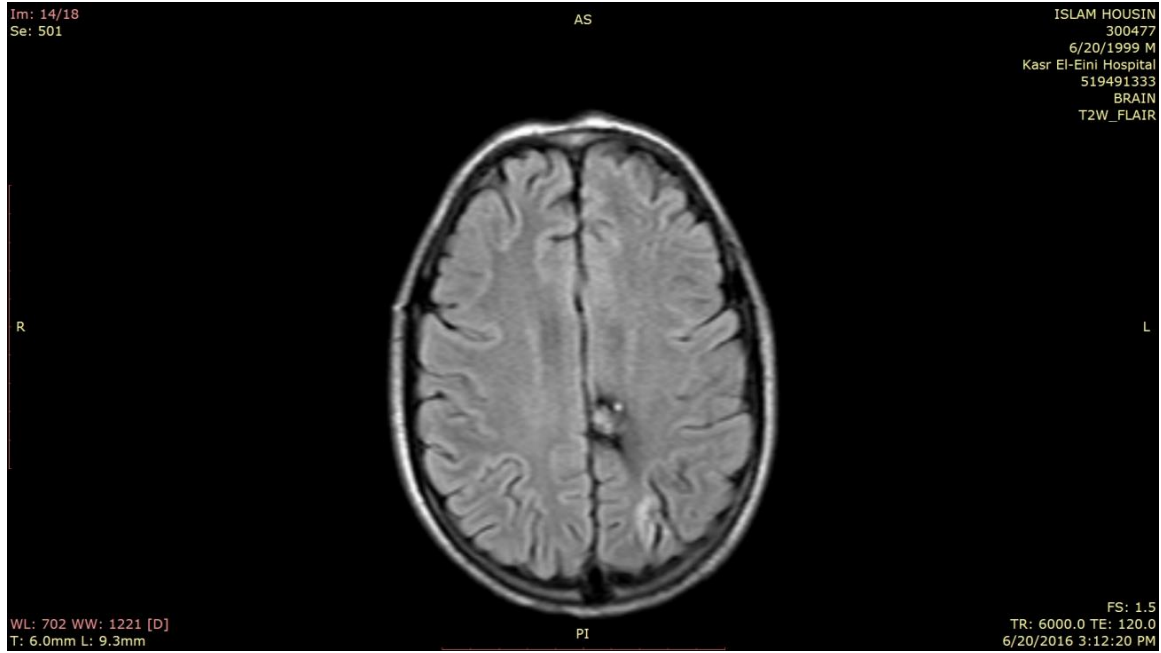


Figure 2c