

## A case of Cerebellar Ataxia with isolated vitamin E deficiency in Emergency department.

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### **ABSTRACT:**

We report the case of a 27 -year-old patient who consults for cerebellar ataxia with isolated vitamin E deficiency (AVED). It is a rare autosomal recessive condition that is caused by a mutation in the alpha tocopherol transfer protein gene [1]. Cerebellar ataxia represents an important neurological presentation in the emergency. The examination finds a cerebellar ataxia, pyramidal spasticity, and evidence of a neuropathy with absent deep tendon reflexes [2]. Diagnosis is based on physical examination, on vitamin E plasma dosage and on exclusion of known causes of malabsorption. Laboratory findings reveal a very marked deficiency of vitamin E in plasma. Neuro imaging shows an obvious cerebellar atrophy [2]. As the case we describe, the patient had clumsiness in walking. He was ataxic with impaired tandem walking. Laboratory investigations revealed, serum vitamin E of 6.89 mg/l (normal: 8.9-18.3mg/l), Computed Tomography of the head showed cerebellar vermian atrophy. High doses of vitamin E supplementation (800 mg daily) have been shown to reverse the neurological signs of AVED [3].

**Keywords :** cerebellar ataxia, vitamin E, deficiency, atrophy.

### Image in clinical medicine :



**Figure 1:** Head computed Tomography showing cerebellar vermian atrophy.

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