Hereditary cerebellar ataxias: descriptive study of a hospital cohort

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**OBJECTIFS**

The objective is to describe the epidemiological, clinical and etiological characteristics of hereditary cerebellar ataxias in children followed in our Child neurology department.

**METHODS**

Our study is descriptive and retrospective including 46 patients (26 girls, 20 boys) with hereditary cerebellar ataxia, followed in the child neurology department of Hédi Chaker hospital over a period of 11 years (2010 to 2021). All patients underwent clinical examination, cerebral imaging and neurophysiological exams.

**RESULTS**

The main symptom for consultation was a walking disorder in 50.2%, psychomotor retardation in 45.4% and tremor in 4.4%. The mean age at onset was 5.5 years. Ataxia was associated to others signs in almost all cases (99.8%). The associated signs are oculomotor apraxia in 8.7%, Babinski's sign in 26.1%, abolished reflexes in 10.9% of cases. Peripheral neuropathy was present in 16.3% of cases. The cerebral MRI is normal in 43.5% and shows a cerebellar atrophy in 56.5%.

The phenotypic confirmation of a specific etiology was made in 19.1% (ataxia telangiectasia in 8.5% of cases, AVED in 6.4%, of friedreich's ataxia in 4.2%). The unclassified group represent 66%.

An unfavorable evolution was found in 36.74% of patients and were significantly correlated with intellectual disability (p=0.021) found in 57.40% and spasticity (p=0.036) found in 29.8% of cases. Independent walking was only acquired in 53.85% of patients. The abnormalities found in 46.15% of cases.

**CONCLUSION**

In childhood cohort, ataxia was frequently associated to other neurological or extra-neurological symptoms. Classical etiologies such as Friedreich's ataxia, ataxia telangiectasia and AVED are rare.