ATAXIA-TELANGIECTASIA IN CAMEROONIAN CHILDREN

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ABSTRACT

Background: Ataxia-Telangiectasia (A-T) is a rare genetic disease characterized by progressive ataxia and multisystem involvement, which requires early diagnosis and multidisciplinary management. **Methods:** We report a series of seven patients with A-T. This is a retrospective study conducted from 2016 to 2020, in patients with a history of progressive cerebellar ataxia and ocular telangiectasia on physical examination as well as elevated serum alpha-fetoprotein. Data were collected from the medical records of the patients. **Results:** There were three boys and four girls. The mean age at onset of symptoms was 36.4 months (range 12 to 72 months) and the mean age at diagnosis was 84 months (range 60 to 144 months). Family history of A-T was noted in 3 patients. Ataxia was the first sign of the disease and the reason for consultation in all patients. Cerebellar ataxia and ocular telangiectasia were seen in all patients. Malnutrition was present in 6 (85.7%) patients, dysarthria was observed in 3 (42.8%) patients, intention tremors in 2 (28.6%) patients, dystonia and epilepsy in 1 (14.3%) patient each. Recurrent infections with repeated hospitalizations were present in 1 (14.3%) patient. Elevated serum alphafetoprotein level was seen in all patients with a mean level of 311.39 ng/ml (range from 113 ng/ml to 911.6 ng/ml). Four (57%) patients underwent brain imaging which showed cerebellar atrophy in three of them. Genetic testing for A-T was not done in any patient due to unavailability. All these patients were still alive at the submission of this abstract. A motor deficiency worsening was observed in 2 patients, who currently use a wheelchair since the age of 6 and 7 years old, respectively. **Conclusion:** In children developing gait disorders with progressive cerebellar ataxia, physicians should look for ocular telangiectasia. In our setting, the diagnostic confirmation is based on elevated serum alpha-fetoprotein level.

Keywords: ataxia, telangiectasia, children, Cameroon

Conflict of interest: none