369- Alpha-fœtoproteine prognosis value in monitoring patients with ataxia-telangiectasia

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INTRODUCTION

Serum alpha-fetoprotein (AFP) is used as a biomarker for the diagnosis of some hereditary diseases such as ataxia-telangiectasia (AT), that is an autosomal recessive neurodegenerative disease with early onset at childhood. The prognosis value as well as the individual and the interindividual variation of AFP levels over time has been few studied [1] .

OBJECTIVES

Though the present study, we aimed to look for correlations between AFP kinetics and the patients outcome and consequently to determine the interest of monitoring this marker

MATIERIALS AND METHODS

We reported a series of 16 patients followed in child Neurology department for AT, having benefited from a regular serum dosage of AFP in Immunology Laboratory using electrochemiluminescence technique on the Cobas e411 automaton (Roche®). We evaluated the AFP level and its kinetics.

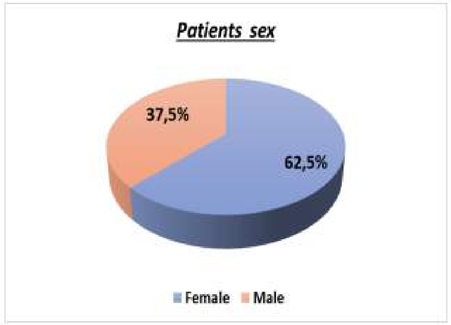
RESULTS

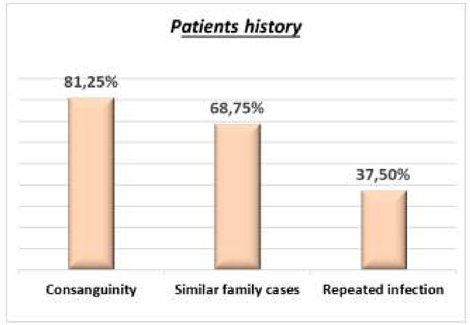
1- Demographic and anamnestic features:

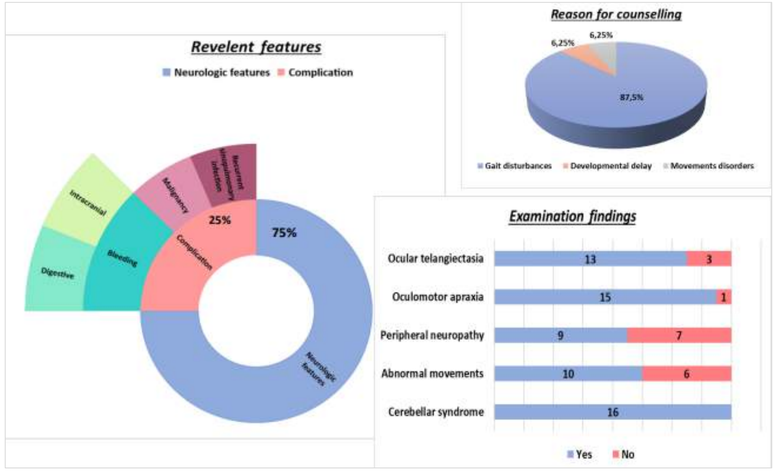
The study included 10 girls and 6 boys. The mean ages at onset symptoms and at diagnosis were 39 (10-96) and 67.5 months (10 - 144), respectively.

2- Clinical features and outcome:

Two patients had inaugural bleeding at onset, one patient presented initially for hematological malignancy and another developed lymphoma during follow-up.

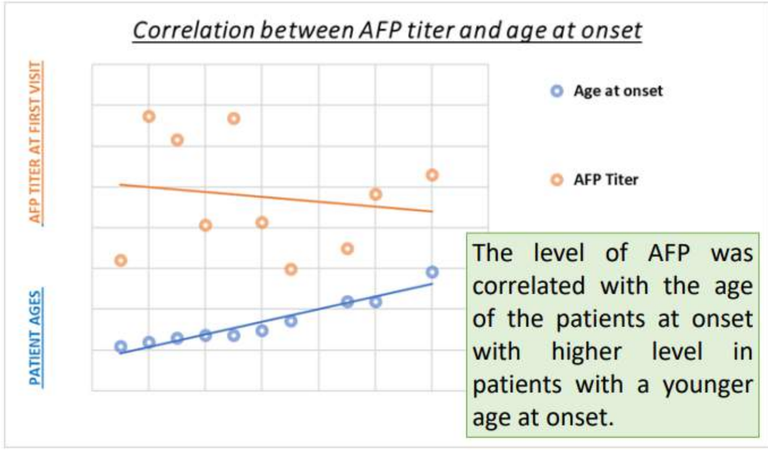




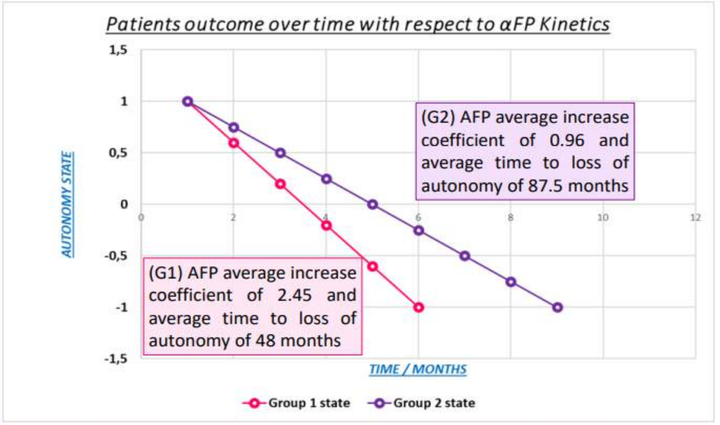


3- Paraclinical findings:

Immune deficiency was noted in 7 patients of whom 5 had Ig A deficiency and only 4 associated IgG deficiency. AFP level _ recorded for 12 out of 16 patients_ ranged from 99.5 to 656.3 ng/ml. In patients who had regular monitoring over a period of up to 8 years; AFP kinetics in relation to time showed pattern of a positive straight curve with a guiding coefficient between 0.76 and 3.



The level of AFP was correlated with the age of the patients at onset with higher level in patients with a younger age at onset.



Faster growth in AFP levels was associated with a worse functional prognosis. Thus, the study of the kinetics of AFP made it possible to identify two groups (G) of patients: G1 and G2.

CONCLUSIONS

Ataxias represent a challenging group of disorders due to significant clinical overlap [2]. The utility of AFP for the investigation of ataxia syndromes is evident as it association with AT is well recognized [2,3]. Previous report demonstrated common increase of AFP level with age during monitoring Serum AFP level. Similarly, high level of AFP were constantly predictive of malignancies [1-3]. In the current study, we also proved that faster growth in AFP levels seemed to be associated with worse functional prognosis. Previous report, however, did not confirm the utility of AFP as a biomarker for disease progression [2]. Further studies with large population are necessary to confirm these data.

REFERENCES

1-**Alpha fetoprotein is increasing with age in ataxia-telangiectasia.** A Stray-Pedersen **et al. Eur. J. Pediatr. Neurol. 2007**

2- Progressive Ataxia with Elevated Alpha-Fetoprotein: Diagnostic Issues and Review of the Literature. Martin Paucar **et al.** Tremor Other Hyperkinet Mov (N Y). **2019**

3-Diagnosis and Management of Ataxia-Telangiectasia in Resource-Limited Settings. Journal of the International Child Neurology Association.2022