

MEETING ABSTRACT

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Ataxia-telangiectasia: immunologic profile and clinical outcome

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Background

Evaluate the immunologic profile and clinical outcome of Ataxia-Telangiectasia (AT) patients followed on a immunology service of a health unity in Brasília.

Methods

This is a retrospective study and the informations were retrieved from the patients medical records.

Results

In our service, a hundred and thirty patients with the diagnosis of a primary immunodeficiency are followed-up and from this sample we have four patients with AT. Accessing clinical history and the laboratory findings becomes clear that a large variability occurs concerning their immune system.

All subjects presented recurrent infections, especially sinopulmonary. In laboratory evaluation it was shown immunoglobulin A (IgA) deficiency, lymphopenia due to low lymphocyte T count and selective antibody deficiency with normal immunoglobulins levels. Some of these patients also require human immunoglobulin replacement and two of them evolved with lymphoid malignancy (Hodgkin and non-Hodgkin lymphoma).

Conclusions

This study demonstrates that clinical aspects and level of immunodeficiency has a large variation and that both cellular and humoral immunity might be affected. This presentation is the same one found in databases worldwide.

A multidisciplinary approach allows adequate control of infectious episodes and related comorbidities with a positive impact on their quality of life.

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