Ataxia telangiectasia case report from tertiary-care hospital of North Karnataka

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Abstract

Ataxia-telangiectasia (AT) is a primary immunodeficiency disease with multisystem disorder characterized by progressive neurologic impairment, variable immunodeficiency, impaired organ maturation, oculo-cutaneous telangiectasia, and a predisposition to malignancy. It is a variable immunodeficiency involving both cellular and humoral responses and a predisposition to cancer. In 1995, a large gene was identified on chromosome 11q22-q23, known as AT Mutant(ATM) gene and the lack of its gene product, the ATM protein, is responsible for the clinical features of AT. Here we present a case of ataxia telangiectasia in a 16 year old female who presented with progressive ataxia.

Keywords: Ataxia telangiectasia, humoral immunity, immunodeficiency.

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