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RESEARCH ARTICLE

A CASE REPORT OF BRONCHIECTASIS ON PRIMARY IMMUNE DEFICIENCY TYPE ATAXIA TELANGIECTASIA

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Abstract

Bronchiectasis is defined as a permanent and irreversible increase in bronchial caliber, affecting at least two lobes of the lung. It is a frequent pathology in developing countries, but remains underestimated. There are many etiologies, including primary immune deficiency, which is considered the second most common cause of bronchiectasis in children after cystic fibrosis. We report the case of a 21-year-old patient admitted for the management of an infectious exacerbation of diffuse bronchiectasis secondary to a primary immune deficiency of the ataxia-telangiectasia type. We will cite some studies in the literature with a discussion on the main mechanism of bronchiectasis associated with ataxia-telangiectasia and its prevalence compared to other etiologies.

Introduction:-

Ataxia-telangiectasia is an autosomal recessive genetic disorder that combines a severe mixed immune deficiency, mainly in the humoral component, with progressive cerebellar ataxia.

Neurological signs associated with cerebellar ataxia include unsteady gait, eye movement abnormalities, poor limb coordination and tremor, and mucocutaneous telangiectasia. Bronchiectasis is a consequence of humoral immune deficiency, with an increased incidence of recurrent respiratory and sinus infections.

Case Presentation:

We report the observation of a 21-year-old patient with a history of recurrent respiratory infections with notion of growth retardation at the age of 7 and an as yet undocumented immune deficiency in his 5-year-old sister.

The patient presented with morning bronchorrhea with left ocular telangiectasia **[Figure 1].** The reason for which he benefited from a thoracic CT scan objectifying bronchial dilatation **[Figure 2].**

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Figure 1:- Image showing left ocular telangiectasia.

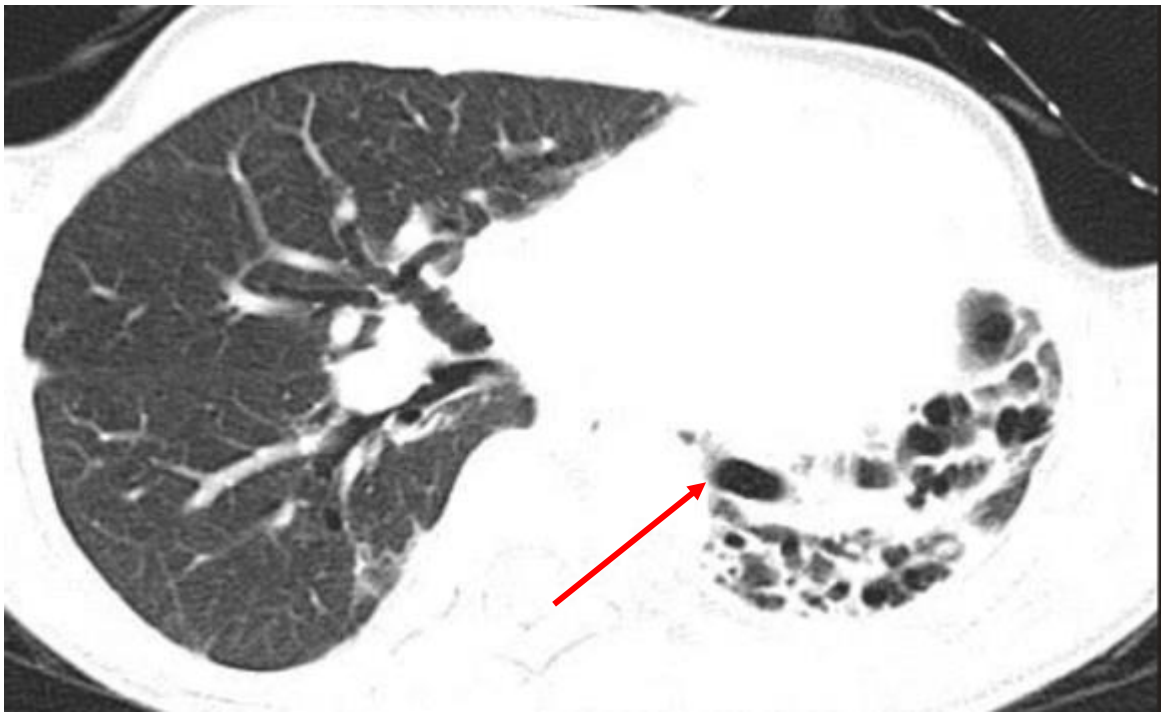

Figure 2:- Cross-section of thoracic CT scan showing a focus of bronchiectasis on a lung with sequelae.
The red arrow showing a focus of bronchiectasis on a lung with sequelae.

Two years later, the evolution was marked by the installation of a neurological symptomatology dominated by a difficult standing position, slowness in the execution of simple gestures, trembling of the upper limbs and language delay in favor of a cerebellar syndrome. However, the patient does not present with digestive or other extra-respiratory signs.

The diagnosis of ataxia-telangiectasia was retained after performing a weighted immunoglobulin assay with low IgA and IgE levels (IgG:6.01g/l , IgE< 25 IU/ml , IgA< 0.25 g/l , IgM: 2.31g/l), protein electrophoresis showing an inflammatory syndrome, HIV, hepatitis B and C viral serology coming back negative with a tumor marker assay showing a high serum level of alpha-1-fetoprotein (AFP: 56.10 ng/ml).**[See the table below]**

	Measured value	Normal values
IgG	6.01 g/l	5.4 – 18.22 g/l
IgA	< 0.25 g/l	0.63 – 6.45 g/l
IgE	< 25 UI/ml	Adult < 100UI/ml , child < 60 UI/ml
IgM	2.31 g/l	0.22 – 2.93 g/l
AFP	56.10 ng/ml	< 7 ng/ml

In addition, a bronchial fibroscopy for a bacteriological study, an immunological work-up (AAN, ANCA), aspergillus serology and a sinus CT scan were carried out, with negative results.

Discussion:-

Ataxia telangiectasia is a rare autosomal recessive disorder. It combines cerebellar ataxia, ocular and cutaneous telangiectasia and immune deficiency. It is linked to a defect in the ATM gene, located on chromosome 11q22-23, coding for a protein that plays a complex role in cell cycle control and cytoprotection [1].

Cystic fibrosis is the most common cause of diffuse bronchial dilatation in developed countries. It is the congenital disease most frequently associated with bronchiectasis. Primary immunodeficiency is a frequent cause of bronchiectasis in children [2].

The types of primary immunodeficiency most frequently implicated are variable combined immunodeficiency, ataxia telangiectasia, hyper IgE syndrome, IgA deficiency, IgM deficiency, chronic septic granulomatosis and CD8 and CD4 deficiency [3-4].

In a study of 44 cases, the etiological investigation revealed 2 cases of immune deficiency (4.54%) [5].

Thus, 4 cases were already being followed for ataxia-telangiectasia, 2 of whom developed bronchial dilatation with other associated complications, namely pulmonary tuberculosis and digestive lymphoma [1].

In another study involving 131 children, the etiology was identified in 86 cases (65.64%), with 39 cases of primary immunodeficiency (29.77%), including 8 cases (6.35%) of variable common immunodeficiency and agammaglobulinemia respectively, 3 cases (2.38%) of chronic septic granulomatosis and Ig A deficiency, 7 cases (5.55%) of hyper IgE syndrome and ataxia telangiectasia, 1 case (0.79%) of hyper IgM syndrome, CD4 deficiency and CD8 deficiency [6].

Conclusion:-

Ataxia-telangiectasia is a primary immune deficiency responsible for diffuse bronchiectasis and increased risk of recurrent sinopulmonary infections. Early diagnosis and multidisciplinary management could improve patient prognosis

References:-

[1] S. Chaouki , B.-S. Benjelloun Dakhama , K. Alaoui , L. Arqam . Ataxia-telangiectasia: Case reports and literature review. Doi:doi.org/10.1016/j.jpp.2007.12.001

[2] Sethi GR, Batra V. Bronchiectasis: Causes and management. Indian J Pediatr.2000;67:133-139.Doi: 10.1007/BF02726189

[3] Banjar HH. A reviewef 151 cases of pediatric non cystic fibrosis bronchiectasis in tertiary care center. Ann Thoracic Medicine.2007;2(1):3-8.Doi: 10.4103/1817-1737.30354

[4] Dogru D, Nik-Ain A, Kiper N, Gocmen A, Ozcelik U. Bronchiectasis: the consequence of late diagnosis in chronic respiratory symptoms. J Trop Pediatr. 2005 ;51:362–365.Doi: 10.1093/tropej/fmi036

[5] B. Oujidi, Y. Berrabah. Les étiologies des dilatations de bronches chez l'enfant : à propos de 44 cas. Doi.org/10.1016/j.rmr.2016.10.626
[6] S. Zouiter, N. Amenzoui, I. Mansouri, F. Ailal, F. Adnane, Z. Jouhadi, AA. Bousfiha.
La dilatation des bronches chez l'enfant : expérience sur une période de dix ans. Rev Mar Mal Enf2022; 51 : 16-21.http//revues.imist.ma/index.php/RMME/article/view/32588/16894.