LETTER TO EDITOR (VIEWER'S CHOICE)

ATAXIA-TELANGIECTASIA, THREE CASES WITH PULMONARY INVOLVEMENT

Nooshin Baghaie, Seyed AliReza Mahdaviani, Soheila Kalilzadeh, Nazanin Parsanejad

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We describe herewith 3 children with ataxia telangiectasia (A-T) who presented to us with recurrent chest infections. The first patient was diagnosed as A-T at 4 years of age, had recurrent admissions from 14 years of age for recurrent pneumonias and was on prophylactic antibiotics and intravenous immunoglobulins (IVIG) and finally succumbed to her disease at 20 years of age. The second patient was a 13 years old girl with recurrent pulmonary infection since childhood.

She was diagnosed with A-T at 7 years old due to unsteady gait, telangiectasia in eye and skin. She has developed tubular bronchiectasis and is on prophylactic antibiotics. The third patient was diagnosed at 1 year of age with ataxia and recurrent chest infections. She is on regular ciprofloxacin prophylaxis and IVIG prophylaxis.

Ataxia telangiectasia is a rare autosomal recessive disorder characterized by unsteady gait (ataxia), telangiectasia of eye and skin and a variable immunodeficiency disorder (both cellular and humoral) and a high incidence of malignancy.[1,2] The prevalence of A-T is estimated to be 1 in 40,000 to 1,00,000.[3] Recurrent severe infections as a clinical feature of A-T is due to variable degree of immunodeficiency however some other patients present mild infection and have undetectable immunodeficiency.[4,5] Recurrent sinopulmonary infection is a common infection which occur in 50%-80% of patients. The onset of these infections may be early in life or may be presented in 10 years or more and can lead to bronchiectasis.[6] All three patients had pulmonary involvement. Bronchiectasis was seen in Computed tomography of case 2 and 3 but in case 1 ground glass pattern suggested the interstitial lung disease.

The diagnosis of A-T syndrome is established in patients over one year of age who show ataxia with a serum level of alpha fetoproteins (AFP) more than twice the upper limit of normal. Patients must also have three of the following four criteria: incoordination of the head and eyes in lateral gaze deflection; ocular or cutaneous telangiectasias before the age of five years; gait ataxia and lymphopenia, especially of CD4, and immunoglobulin deficiencies, mainly of IgA and/or IgG subclasses. Patients with less than three of these criteria

	Patient 1	**Patient 2**	**Patient 3**
Age and gender	20 years female diagnosed at 4 years of age	13 years girl diagnosed at 7 years of age	16 years girl diagnosed at 1 year of age
Age of onset of recurrent pneumonias	14 years	Since early childhood	1 year of age
Age of onset of ataxia	2 years	Not available	1 year
Age of onset Ocular telangiectasia	3 years	Not available	Not available
Born of consanguineous marriage	No	Yes	Yes
Any other family member affected	No	No	Father had lymphoma
Serum IgG (Normal = 6.58-18.37 g/L)	2.5g/L (low)	< 1.9g/l (Low)	Normal
Serum IgA (Normal = 0.71-3.60 g/L)	0.4g/L (Low)	0.4g/l nl (low)	Normal
Serum IgM (Normal = 0.40-2.63 g/L)	2.42 g/L (Normal)	14g/l nl (high)	4.07 g/l (elevated)
Alpha feto protein (Normal <30 IU/ml)	97 IU/ml (Elevated)	108 IU/ml (elevated)	130 IU/ml (elevated)
Echocardiography	Normal	-	-
CT chest	bilateral diffuse ground glass nodular infiltration with patchy alveolar infiltration	Tubular bronchiectasis and micro nodular infiltration in left lower lobe	Extensive bronchiectasis in mid lower lobes with peribronchial wall thickening and mucus impaction
Outcome	Died at 20 years due to pneumothorax, severe interstitial pneumonitis and respiratory failure	On follow up on ciprofloxacin prophylaxis	On follow up on ciprofloxacin prophylaxis and IVIG

should have the diagnosis confirmed by the finding of radiation-induced chromosomal breaks in lymphocytes and low ability of repairing them. [1,7] All our three cases were diagnosed before hospitalization in our center, based on the criteria mentioned. There is no correlation between severity of neuro-degeneration and pulmonary disease. [8-10] Respiratory complications may account for upto 50% of death in A-T patients[11]. Consequently pulmonary status is a prognostic factor. In this report, Case 1 expired due to severe interstitial pneumonitis and respiratory insufficiency.

Immunodeficiency contributes to an increased of sino-pulmonary infection. The most common humoral defects are decrease concentration or absence of IgA and IgG2. [8,10] Antibody responses to virus and bacteria antigens may be impaired too.[12] In our first and second cases low level of IgA and IgG was noted and in the third case IgA and IgG were within normal limits. Cellular immunity is also impaired in A-T cases. Impaired delayed hypersensitivity reaction is reported in 60% of patients.[12] PPD test was performed in all cases and the result was negative in all.

The use of prophylactic antibiotics or treatment with antibiotics in addition to IVIG should be considered in all of the patients with ataxia-telangiectasia.[2]

CONCLUSION

Pulmonary involvement is common in A-T and as pulmonary status is considered to be a prognostic factor in A-T patients therefore an increase in knowledge of physicians and respiratory experts involvement in early stage of disease may decrease the rate of infections and complications.

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From: Pediatric Respiratory Diseases Research Center, NRITLD, Shahid Beheshti, University of Medical Sciences,Tehran, Iran.

Address for Correspondence: Nazanin Parsanejad,M. D., Pediatric Ward, Masih Daneshvari Hospital, Darabad, Shahid Bahonar, Niavaran, Tehran, Iran. Email: p_nritld@yahoo.com

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