Linear Growth and Endocrine Function in Patients with Ataxia Telangiectasia; A Cohort of 13 patients in Qatar

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Introduction

Ataxia telangiectasia (AT) is a rare, genetic, primary immune deficiency disease characterized by immunodeficiency and neurological manifestations, with increased tendency to infection, malignancy, and autoimmune diseases. Both growth delay and endocrine abnormalities are occasionally reported in these patients.

Patients and Methods

We studied growth parameters (height (Ht) , weight, body mass index (BMI) and measured the HtSDS) of 13 patients (age 7.7 +/- 3.5 years) with AT in relation to their mid-parental HtSDS. We measured their biochemical (serum calcium (Ca), phosphorus (PO4), Alkaline phosphatase (ALP) , alanine transferase (ALT), ferritin, creatinine and albumin, endocrine (Free thyroxine (FT4), thyrotropin (TSH) , insulin-like growth factor-I (IGF-I) , 8 AM cortisol) and immune functions (IgG, IgM and IgA antibodies).

Results

Abnormalities of growth and IGF-I in AT

	HtSDS	BMI	MPHtSDS	IGF-I
		Kg/m2		ug/L
Mean	-1.4	15.1	-1.3	149
SDS	1.2	2.4	1.1	110
No=	13	13	13	8
Abnormal	31%	38%	31%	38%

Thirty one % of patients with AT had short stature (HtSDS <-2). However, their MPHtSDS denoted that their short stature is familial as 4/13 had MPHtSDS <-2.

Results

Abnormalities of thyroid and liver functions

	FT4	TSH	Anti TPO	Albumin	ALT
	pg/ml	mIU/L	U/ml	g/L	U/L
Mean	15.2	7.3	88.7	39.2	42.2
SDS	3.8	13.6	131	7.2	24.2
No=	13	13	13	13	13
Abnormal	15%	31%	15%	15%	38%

Interpretation and conclusion

1.They had low BMI and 2 of them had low serum albumin and IGF-I denoting malnutrition.
2.Low IGF-I (IGF-I SDS < -1.5) can be explained in part by under- nutrition or disturbed GH secretion.
3.Elevated serum ALT and ferritin in some patients suggest immune-related inflammation in the liver.
4.Thirty % of patients had high TSH, two of them had low FT4 diagnosing clinical (15%) and sub-clinical (15%) hypothyroidism. Anti-peroxidase antibodies were high in 2/13 denoting immune-related thyroid aggression.
5. 8/13 had vitamin D deficiency (<20ng/ml)
6. One adolescent girl (14.5 yr) had hyper-gonadotropic hypogonadism (low estradiol and high FSH)
7. None of the growth parameters were correlated with the immunoglobulin (IgG, IgM or IgA) levels.

Patients with AT had high prevalence of growth delay and endocrine dysfunction in the form of low IGF-I, clinical and subclinical hypothyroidism and hypogonadism.
Early diagnosis and management of these endocrinopathies is important or improving the prognosis of the disease.

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