

Delayed puberty in a girl with ataxia telangiectasia: Multiple endocrine dysfunctions

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Introduction

Introduction: Ataxia telangiectasia (AT) is a rare, genetic, primary immune deficiency disease characterized by immunodeficiency and neurological manifestations, with predisposition for infections, cancers, and autoimmune diseases.

Case Report

A 14-year-old girl presented to our clinics at Hamad General Hospital, with history of progressively increasing difficulty in walking and hand movements since age of 8 years.

She was prepubertal (Tanner 1 breast development), underweight (BMI S.D.=-2.5) with normal stature (Ht S.D. =1.1).

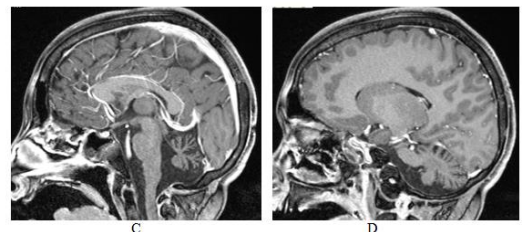
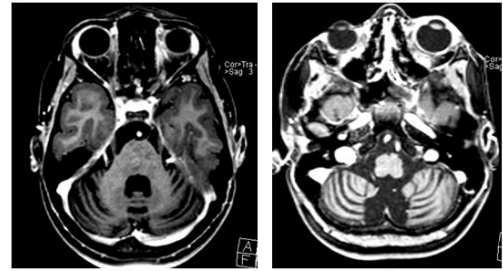
She had an expressionless face, conjunctival telangiectasia, dysarthria, diminished reflexes, dystonia, and ataxic gait.

CBC revealed lymphopenia (between 1000-1400 cells/ul) and α -fetoprotein (246 IU/ml). Liver and renal function tests were normal. Serum IgG=2940 mg/dl, IgA <6.6 mg/dl, and IgM=135 mg/dl. Serum IgG subclasses levels and anti-vaccine titers were normal.

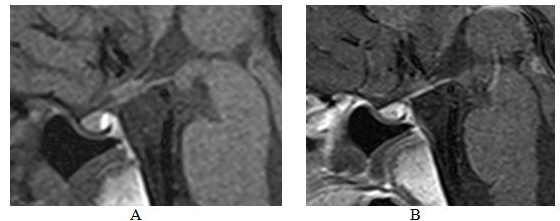
Lymphocyte subsets showed slightly low CD3, CD4, and CD19. Free thyroxine was low (7.8 pmol/l), TSH was high (>100 mIU/l), and anti-thyroid peroxidase titer high (>1000 U). Serum insulin-like growth factor1 (IGF1) and morning serum cortisol were normal.

Trans-abdominal pelvic ultrasound demonstrated small hypoplastic uterus and rudimentary ovaries.

cranial MRI showed marked cerebellar and vermis atrophy with iron deposition in the pituitary



MRI axial cuts A&B Sagittal cuts C&D demonstrating marked atrophic changes of the Vermis and cerebellar peduncles (A&C) as well as cerebellar hemispheres (B &D).



MRI T1 (precontrast A, post contrast B) demonstrating normal volume and configuration of pituitary with heterogeneous intensity due to multiple small hypointense foci which showed no enhancement after CM injection mostly due to hemosiderin deposition foci.

Discussion

Occurrence of primary hypothyroidism and hypergonadotrophic hypogonadism points out to an autoimmune aggression in cases of AT to thyroid and ovaries. Conclusion: In patients with AT endocrine manifestations may include hypothyroidism and hypogonadism and should be kept in mind when evaluating delayed puberty in this condition

Conclusion

This case illustrates the affection of endocrine system in the form of hypothyroidism and hyper-gonadotrophic hypogonadism with iron deposition in the pituitary gland .