A rare presentation of triple a syndrome with prominent neurological and hematological Features: A case report

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A rare presentation of triple a syndrome with prominent neurological and hematological Features: A case report

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Abstract

Triple A (AAA) syndrome was first described by Jeremy Allgrove in 1978 as uncommon. We reported a 30-year-old woman who presented in our clinic with symptoms of Adrenal Failure, Achalasiacardia and alacramia with a rare hematological manifestation rarely seen.

Introduction

Triple A syndrome is a rare, autosomal recessive disease characterized by clinical triad of adrenal failure, achalasiacardia and alacrima. The syndrome is associated with variable and progressive neurological impairment involving the central, peripheral and autonomic nervous systems [1]. Mutation in the AAAS gene on chromosome 12q13 which encodes a protein known as ALADIN (alacrima, achalasiacardia, adrenal insufficiency and neurological disorder) has been described as a causative agent [2]. We reported a rare case of a 30-year-old woman who presented with Triple A syndrome with hematological manifestations.

Case Report(s)

A 30-year-old lady presented to our hospital with a one month history of prolonged coughing and progressive breathlessness. Her past medical history included a worsening of her dysphagia for the last three years. She was born to non-consanguineous parents. It was noted that she had cried without tears since birth. She had a significant past medical history of thalassemia intermedia that was diagnosed two years ago. Her family history was significant in that her brother also suffered from achalasia cardia and alacrima. Physical examination revealed a blood pressure of 80/60 mm Hg and hyper-pigmentation of oral mucosa and knuckles. Neurological evaluation revealed absent gag and palatal reflex with thenar and hypothenar muscles atrophy. Ophthalmic features included decreased tear production confirmed by Schirmer's test. Blood investigations revealed a hemoglobin of 12.5 g/dL (normal: 11-16), a total leukocyte count of 9300/uL (4,000-11,000). Peripheral smear showed microcytosis, target cells, Burr cells, and a doubled population of RBCs. Liver and kidney function tests were normal. Chest x-ray and CECT thorax revealed consolidation of right upper lobe and enlarged mediastinal lymph nodes suggestive of tuberculosis with dilated esophagus in thoracic segment up to the gastroesophageal junction. Upper gastrointestinal endoscopy was performed and revealed esophageal candidiasis with a dilated esophagus and there was some retained residual food with a tight Lower Esophageal Sphincter (LES) suggestive of achalasiacardia. She was started on antitubercular treatment and intravenous steroids followed by oral steroids. Endoscopic dilatation of LES was done. Blood pressure improved to 120/80 mm Hg. She was advised to frequently apply artificial tears. Her symptoms improved and she was discharged after twenty days. At present, the patient is clinically better.

Discussion

Recent medical literature reports Triple A syndrome as a rare disorder which manifests itself within the first decade of life with alacrima, other symptoms include glucorticoid deficiency and achalasia. A few case reports that have been documented so far with triple A syndrome include Brooks and Kleta [2] who in 2004 reported a 12 year old boy with classic systemic features of Triple A syndrome with several prominent ophthalmic features including accommodating spasm, dry eye and superficial punctate keratopathy. They had performed a DNA sequencing that revealed a mutations in the AAAS gene on chromosome 12q13. Handschug et al [3] mapped the syndrome to a 6cM interval on chromosome 12q13 and refined the critical region to 0 cm between KRT8 and D12S1651. Nakamura and Yoshida et al [4] described another case of a 60 year old Japanese man with genetically confirmed adult or late onset Triple A syndrome. Neurological manifestations of the disease included motor neuron disease like presentations, motor-sensory or autonomic neuropathy, optic atrophy, cerebellar ataxia, Parkinsonism and mild dementia. In
one Indian published case report [5] a 22-year-old male presented with erectile dysfunction, loss of spontaneous morning erections for six months and nocturnal diarrhea and with recurrent postural dizziness for three months.

Our patient had presented with alacrima, achalasia cardia and neurological features. Hematological features included thalassemia intermedia and dimorphic population of RBCs. There is no case noted so far that has reported Triple A syndrome with such hematological findings. This patient is being reported because of rarity of Triple A syndrome with hematological manifestation. Autonomic dysfunction at the level of lacrimal glands explains the alacrima seen in this condition. Careful assessment for alacrima following molecular genetic analysis of AAAS had been considered in the patients who have also showed neurological manifestation as described in the limited literature available.

References


