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# Rare Cause of Generalised Pigmentation

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## Introduction

Primary adrenal insufficiency is a disorder of adrenal cortex characterized by deficiency of glucocorticoid and mineralocorticoid. It has various causes which includes infective, autoimmune, neoplastic, congenital, iatrogenic and others. Autoimmune etiology is the commonest cause of adrenal insufficiency in developed country where as in developing country it is the infection which is the leading cause. Prompt diagnosis is necessary in this case as delay in initiation of treatment might cause fatal outcome. This is a cause of primary adrenal insufficiency in a young boy due to rare genetic cause.

## Clinical Presentation

9 year old boy presented with insidious onset, gradually progressive anorexia, weight loss, occasional vomiting, progressive blackening of whole body for last 2 years. No history of past childhood tuberculosis or any chronic systemic illness. No history of convulsion, unconsciousness, hypoglycemic symptoms and delayed milestones in childhood.

## Examination

Height-128.2cm (-0.9SDS), weight -19.2 kg(-2.4SDS) BP-86/64(<3rd percentile). Hyperpigmentation present over knuckles, lips and buccal mucosa.

***Figure 1:*** Pigmentation over lips. ***Figure 2:*** Pigmentation over knuckles.

## Investigation

| | |
|---|---|
| Cortisol | 1.89ug/dl |
| ACTH | 1400pg/ml |
| Sodium | 132mg/dl |
| Potassium | 3.47mg/dl |
| CECT Adrenal Protocol | Hypoplastic left adrenal<br>Absent Right adrenal |
| Anti TPO | Negative |
| Chest X Ray | Normal |
| Hb | 11.2gm/dl |
| Whole Exome Sequencing | Homozygous c.43 C>A in exon 1 in AAAS gene.<br>(Autosomal recessive) |
| Schirmer test | Positive (1mm) |
| Barium swallow | Normal study |
| Esophageal Manometry | Normal LES pressure |

## Management

The child was diagnosed with Allgrove syndrome. He was managed with tab Hydrocortisone 15mg/$m^2$ in two divided doses along with eye lubricants. Gradually his apetite and skin color improved.

## Discussion

AS is a rare disorder with a prevalence of 10 cases per 100 000 people. Glucocorticoid secretion affects up to 85% of patients, most of whom are in their first or, less frequently, the second decade of life. It is the leading cause of death due to severe hypoglycemia and can manifest as a variety of symptoms such as recurrent vomiting, hyperpigmentation of skin and mucous membranes, or developmental delays. One of the important peculiarity in allgrove syndrome is preservation of mineralocorticoid axis. Alacrima is the most common early presenting sign occurring at birth or within the first year of life, but its significance is often overlooked [1]. Achalasia usually developed in second decade of life. A neurological syndrome including central, peripheral, and autonomic nervous system impairment is often associated with Allgrove syndrome; neurological manifestations appear at a later age when compared with other manifestations. Distal sensorimotor polyneuropathy is a common manifestation. Other features that have been reported in association with Allgrove syndrome include microcephaly, short stature, dysmorphic features (long narrow face, long philtrum, down-turned mouth, thin upper lip, and lack of eyelashes), palmar and plantar hyperkeratosis, osteoporosis, and long QT syndrome. The molecular basis for this rare autosomal recessive disorder syndrome is the mutated *AAAS* gene, located on chromosome 12q13, that codes for ALADIN protein. Most of the many reported mutations produce a truncated protein [2]. The exact role of ALADIN protein in the nuclear pore is not known. Its probable functions are structural scaffolding, protein, and/or RNA trafficking between cytoplasm and nucleus, redox homeostasis, and steroidogenesis. Different *AAAS* mutations lead to nonfunctional ALADIN protein and its mislocalization in the cytoplasm, hence no longer available at the nuclear pore. This protein is highly expressed in the brain, gastrointestinal tract, and adrenal cortex, the major sites of disease expression [3]. Early recognition of adrenal insufficiency is very much important because delay in initiation of steroid may lead to recurrent hypoglycemia, neurological impairment and fatal consequences. Similar case was reported from india where child with allgrove syndrome presented with alacrimia and addisonian crisis without achalasia [4]. All children presenting with Alacrima in conjunction to Addisons disease should be suspected of Allgrove syndrome and periodically evaluated for achalasia and neurological defects as the onset can be variable [4].

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