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ALLGROVE SYNDROME: A CASE REPORT FROM PINK CITY

General Surgery

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ABSTRACT

Allgrove syndrome is a Described by **Allgrove** et al in 1978, Allgrove syndrome consists of triad: ACHLASIA CARDIA,ADRENAL INSUFFICIENCY,ALACRIMIA. Allgrove syndrome is also known as Triple-A-Syndrome.Rare **autosomal recessive** disorder. Very few cases of this syndrome have been reported in literature.Usually cases present with symptoms in 1st decade of life. Other features that have been reported in association with Allgrove syndrome include microcephaly, short stature, dysmorphic features , palmar and plantar hyperkeratosis, osteoporosis, and long QT syndrome.

KEYWORDS

INTRODUCTION:

Allgrove and colleagues in 1978 first described this syndrome in two pairs of siblings (aged 4–6 years)[1]. Triple A or Allgrove syndrome is charecterized by three main conditions:Achlasia,Alacrimia(defective tear formation)& addisons disease[2].The combination of these three manifestations was known as Allgrove syndrome or Triple A syndrome.The cases usually present during the first decade of life with dysphagia or severe hypoglycaemia or hypotensive attack related to adrenocortical insufficiency. The molecular basis of this rare autosomal recessive disorder is the mutated achalasia, adrenocortical insufficiency, alacrimia syndrome (*AAAS*) gene, located on chromosome 12q13, that codes for ALacrima Achalasia aDrenal Insufficiency Neurologic disorder (ALADIN) protein[3].One such patient reported to us with this rare syndrome.

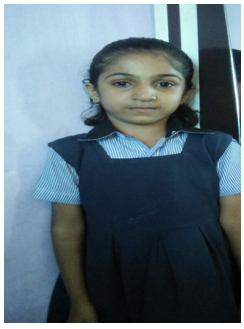
CASE REPORT:

A 10 year –old girl was referred to Gastro surgery department at MGMCH with

Chief complaints- Difficulty in swallowing which was more for liquids and it was associated with vomiting after ingestion of food. She was a born full-term baby, from a non-consanguineous marriage.

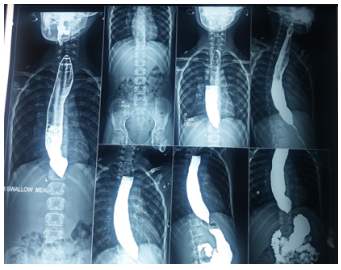
Medical history Patient reviled a history of generalized weakness, asthenia ,fatigue , anorexia 2 years back for which patient was taking Tab.Prednisolone 5mg daily as advised by phycician.

Examination revealed a lean build patient ,with nasal twang to voice. Her blood pressure was 100/60 mmhg with no other obvious clinical finding.



Baseline investigations Reviled normal CBC, sr. creatinine & normal electrolytes. **Basal serum Cortisol was low**(0.36mic/dl) & **Plasma ACTH** was **elevated** (1000pico/ml).A diagnosis of primary adrenal insufficiency was made.

Barium swallow revealed persistent narrowing in distal region of oesophagus making diagnosis of **Achlasia.**



On basis of history of absence of tears patient was referred to opthalmologist where Schirmer test was suggestive of Alacrimia.

On the basis of above findings & investigations a diagnosis of Allgrove syndrome was made.

Patient underwent **Hellers myotomy** for achlasia cardia and patient was discharged post-operatively on Tab .Prednisolone and carboxy methyl cellulose drops for alacrimia.

DISCUSSION:

The description of Allgrove syndrome is limited to case reports and thus prevalence is unknown. 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[4].

The etiology of achalasia esophageal motility is unclear. It is charecterized by degeneration of enteric nervous system that innervates the esophageal muscles & lower esophageal sphincter.studies have found no gender distribution[5].

Previous genetic analysis of eight families with Allgrove's syndrome demonstrated linkage to markers on 12q13[3,6].

The earliest feature of syndrome is alacrimia. Adrenal insufficiency and achlasia are usually diagnosed during **first decade** of life.Achlasia cardia is seen in **75% cases** of allgrove syndrome.

Adrenal insufficiency in Allgrove syndrome is a unique form of adrenal insufficiency with preservation of mineralocorticoid production.

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[7].

Our patient presented with symptoms Similar to cases reported with adreno-cortical insufficiency & dysphagia during 1st decade of life. But alcrimia was not noticed by her parents which was diagnosed on her visit to hospital for dysphagia.

Allgrove syndrome is a multisystem disorder which usually present during 1st decade of life with any cardinal triad of disease.

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