Abstract:
A 24 years old lady presented with dysphagia and regurgitation since her childhood, inadequate weight gain and delayed milestones. History of consanguineous marriage and history of surgery for dysphagia to her paternal aunt were present. At 3 years of her age, barium swallow and thyroid function tests were reported to be normal while ophthalmological consultation revealed she had refractory error and dry eye for which corrective glasses and artificial tears were prescribed. Psychiatric consultation for delayed milestones revealed she had mild mental retardation. At 14yrs of age upper GI scopy was reported to be normal. At 19 yrs of age, barium swallow showed features suggestive of achalasia and she has been treated medically. At 24yrs of age, barium swallow showed sigmoid esophagus with tight LES. She presented to our institution only at this juncture and evaluation revealed her to have 4A or Allgroves syndrome. After complete evaluation, she underwent long cardiomyotomy and she is fine on regular follow up. This case is presented for its rarity and need for high index of suspicion. 

Keyword : Allgrove syndrome, achalasia, addisonian disease, alacrimia, autonomic dysfunction, cardiomyotomy

AAA syndrome is a rare autosomal recessive congenital disorder with Achalasia, Addisionianism and Alacrimia

It occurs due to a mutation in the AAAS gene, coding a protein known as Aladin. It was described by Jeremy Allgrove and his colleagues in 1978. The above 3 A's along with Autonomic dysfunction such as abnormal sweating, orthostatic hypotension, disturbances of heart rate, pupillary abnormalities and abnormal reaction to intradermal histamine has made it as a 4'A' syndrome. In most of these cases there was no family history. It is a progressive disorder and takes several years to develop a full blown clinical picture. Alacrimia is usually
the earliest symptom. Sometimes, hypoglycemia may be the earliest sign. Adrenal insufficiency is due to ACTH resistance. Patient may have mild intellectual impairment. Adrenal insufficiency may not be present initially. Adrenal insufficiency is assessed using ACTH stimulation test, Fasting blood sugar, Serum sodium and serum potassium levels. It may appear years after diagnosis. They commonly experience muscle weakness, movement problems and nerve abnormalities (peripheral neuropathy). Some patients develop optic nerve degeneration. Many of the neurological symptoms may worsen with time. The pathogenesis in achalasia is fibrosis of intermuscular plane (auerbach plexus) and lack of NO synthase.

Our patient is a 24 years old lady, who presented with dysphagia and regurgitation since her childhood. She had inadequate weight gain and delayed milestones. There was a history of consanguineous marriage and history of surgery for dysphagia to her paternal aunt.

**AT 3 YEARS:**
At 3 years of her age, she had upper GI symptoms and barium swallow and thyroid function tests were reported to be normal. She had an ophthalmological consultation which revealed refractory error and dry eyes, for which corrective glasses and artificial tears were prescribed. Psychiatric consultation for delayed milestones revealed she had mild mental retardation. At 14yrs and 19 years of age, upper GI scopy was done at another institution and was reported to be normal but Barium swallow has shown features suggestive of achalasia cardia for which she was on medications.
At 24yrs of age, she had severe dysphagia and barium swallow showed sigmoid esophagus with abrupt smooth narrowing at GE junction and stasis of barium in esophagus. She presented to our institution only at this juncture. Clinical examination revealed right hemiatrophy of the face. Further evaluation revealed her to have “4A” or Allgrove’s syndrome. Her serum ACTH was 34.89pg/ml, serum cortisol was 17.73 mcg/dl, ACTH stimulated cortisol was 32.67mcg/dl. Upper GI endoscopy revealed a hugely dilated esophagus with plenty of food residue, absent peristalsis in the body and tight LES. Manometry revealed 100% simultaneous contractions and sphincter response to swallows were not appreciated with LES pressure of 40 mm Hg. After complete physiological and endocrine evaluation, she underwent long cardiomyotomy

**LONG CARDIOMYOTOMY**

She had good symptom relief with weight gain and on regular follow up for the past 13 months. This case is presented for its rarity, need for high index of suspicion and the need for a regular long term follow-up.