A Double Dose of Triples

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Adrenal disorders and delayed puberty are a not uncommon presentation in the Endocrine Out Patient Department. However, a child presenting with both disorders simultaneously is rather uncommon and requires extensive evaluation and out-of-the box thinking to reach a valid diagnosis. Here we report a girl who presented with both primary adrenal insufficiency and delayed puberty, who on subsequent evaluation was found to have Allgrove’s syndrome and Triple X Syndrome, leading to this rare presentation.

14 year, 6 month old girl, presented to the endocrinology OP with:
- H/o hyperpigmentation of face, lips and palms since the past 8 years.
- H/O weakness of both upper and lower limbs since the past 2 years.
- H/o pain in calves noticed since 2 years, gradually associated with walking difficulty and climbing stairs.
- On follow up with neurologist and being evaluated for sensory and motor neuropathy.
- On follow up with gynecologist for delayed puberty.

Past History
- H/O occasional dysphagia and features s/o reflux esophagitis.
- No H/O seizures in past.
- No H/S/O anosmia
- 3rd degree consanguinity for 2 generations
- No H/O similar problems in family.

TRIPLE A + TRIPE X

Adrenal insufficiency
GERD and achalasia cardia
Alacrimia
Autonomic Features

Delayed Puberty
Hypergonadotropic Hypogonadism

Sensory and Motor Neuropathy

TRIPLE X SYNDROME
Incidence – 1 in 1000 females
Most common female chromosomal abnormality.
Epicantual folds, hypertelorism, upslanting palpebral fissures, clinodactyly
overlapping digits, pes planus, and pectus excavatum, hypotonia and joint hyperextensibility.
POF, unilateral kidney, AITD.

ALLGROVE’S SYNDROME – TRIPE X
Less than 100 reported cases since 1978.
Adrenal insufficiency, achalasia of the cardia, alacrima, autonomic abnormalities.
Abnormal pupillary reflexes, poor heart rate variability, and orthostatic hypotension.
Mutations of the ADRACLIN (or AAAS) gene encoding the ALADIN protein of the NPC.
No unifying pathologic hypothesis.
Progressive loss of cholinergic function.
May represent a dysfunction of melanocortin receptor signaling.

REFERENCES