A case of Alstrom's syndrome

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Abstract:
We report here, a case of a 14 year old boy who was admitted to our hospital with features of cirrhosis liver and reduced visual acuity. Further investigations revealed him to be case of Alstroms syndrome, a very rare genetic disorder. He was treated symptomatically and was discharged in reasonably good health.

Keyword: Alstrom syndrome, Cirrhosis liver, childhood blindness.

Alstrom syndrome patient

Introduction:
Alstrom’s syndrome is a very rare autosomal recessive genetic disorder caused by mutation of the ALMS1 gene. Only 266 cases have been reported in medical literature till date.

Clinical presentation:
A 14 year old boy was brought to our hospital with history of a single bout of hema
temesis. His parents gave a history of two similar episodes during the past two years. They also revealed that the boy had difficulty in hearing and vision, which first came to their attention when he was 5 years old. During his first hospital admission 2 years back, he was found out to be diabetic and was started on insulin injection. He was born out of a second degree consanguineous marriage and there were no peri
tum complications. His developmental mile stones were delayed and scholastic performance was poor. His father, mother and sister are apparently normal. On examination, he was obese, with a BMI of 32.6. Pallor was noted. There was no cyanosis, clubbing, lymphadenopathy or oedema. Pulse rate was 86/min regular and BP was 100/70 mm Hg in supine position. He had Acanthosis nigricans. No digital or bony abnormalities were noted.
Acanthosis nigricans
His abdomen was distended. Spleen was palpable 4 cms below the left costal margin. Liver was not palpable. Shifting dullness was present and Traube’s space was dull on percussion. His external genitalia was pre pubertal and the testis measured 3 ml, with an orchidometer. His cardiovascular and respiratory systems were normal. His intelligence was subnormal for his age, and visual acuity was limited to perception of light bilaterally.

Normal central foveal thickness = 182 ± 23 m Central foveal thickness in this case = 88 m
Audiogram – Bilateral Sensory Neural Hearing Loss.

These investigations revealed that we were dealing with a 14 year old boy, who was obese and had physical and biochemical evidence of insulin resistance. He also had bilateral severe sensory neural hearing loss and grossly impaired vision due to retinal atrophy. And he is now admitted with complications of cirrhosis liver. The two close differential diagnoses that would
prop up in this setting would be Alstrom syndrome and Bardet Biedel syndrome. The absence of polydactyly, presence of retinal atrophy and cirrhosis liver at a such a young age helped us narrow down the diagnosis in favour of Alstrom syndrome.

| HEMOGLOBIN | 6.4 GMS/FL |
| TOTAL COUNT | 3800 CELLS/CMM |
| DIFFERENTIAL COUNT | P58 L37 E2 M3 |
| RBC | 3.1 MILLION/CMM |
| PLATELETS | 1.5 LAKHS/ CMM |
| ESR | 45 MM/HR |
| BLEEDING TIME | 2 MIN |
| CLOTTING TIME | 4 MIN 30 SEC |
| PROTHROMBIN TIME | 21 SEC |
| INR | 1.5 |
| APTT | 30 SEC |
| PERIPHERAL SMEAR | DIMORPHIC ANEMIA |

| SERUM TSH | 2.19 miU/DL (0.27 - 4.2 miU/DL) |
| LEUTINIZING HORMONE | 0.08 miU/ML |
| FOLLICLE STIMULATING HORMONE | 0.08 miU/ML |

**Urine Examination**
- Albumin — Nil
- Sugar — Nil
- Deposits — 2-3 pus cells
- Acetone — Negative

**ECG** — Heart rate — 120/ min. sinus tachycardia. Normal axis. No ST/T changes.

**Echocardiography** — Normal study

**Bone age** — 14 – 18 yrs.

**USG Abdomen** — Cirrhosis with portal hypertension.
- Moderate ascites present
- Spleno renal collaterals present
- Other intra-abdominal organs normal.

**USG Testis**
- left — 2.1 x 1.2 x 0.8 cm volume = 2.016 mL
- right — 2.2 x 1.4 x 0.8 cm volume = 2.464 mL

**CT Abdomen**
- Early cirrhosis liver
- portal hypertension
- extensive collaterals present.

**Upper GI endoscopy**
- Three columns of grade III Oesophageal varices present.
- Fundal varices present.
DISCUSSION:
DIAGNOSTIC CRITERIA FOR ALSTROM’S SYNDROME At 3–14 years of age: 2 major criteria or 1 major and 3 minor criteria. Major criteria: 1) ALMS1 mutation in 1 allele and/or family history of Alstom Syndrome, 2) Vision pathology (nystagmus, photophobia, diminished acuity), cone dystrophy by ERG. Minor Criteria: Obesity / insulin resistance / Type 2 Diabetes 2) History of dilated cardiomyopathy with congestive heart failure 3) Hearing loss 4) Hepatic dysfunction 5) Renal failure 6) Advanced bone age ALMS 1 gene analysis was not available in our state. Our case fulfils 1 major and 3 minor criteria, thus qualifying for the diagnosis of ALSTROM’S SYNDROME.

References:
1 Alström syndrome: insights into the pathogenesis of metabolic disorders. Girard D, Petrovsky N.

