Abstract:
In our opd we come across many cases of RP but syndromic phenotypes associated with retinitis pigmentosa are not so frequent particularly alstrom syndrome. Alstrom syndrome is a rare autosomal recessive multiorgan disorder. Involvement ranges from ocular, aural, endocrinial, hepatorenal, gastrointestinal systems, among many others. We hereby present one such case who came to us for blindness certificate.

Vision was perception of light in both eyes. IOP was 17.3 mm of hg.

Keyword:
RETINITIS PIGMENTOSA, ALSTROM SYNDROME, PIGMENTARY RETINOPTHY, DEAFNESS, OBESITY, DIABETES, NORMAL INTELLIGENCE.

A 13 years old boy was brought to us with complaints of loss of vision in both eyes from past 4 years. Increased sensitivity to light and also decreased hearing from childhood. Child was born of second degree consangineous marriage, full term normal delivery with birth weight of 2.8 kg, with history of delayed developmental milestones. Other siblings were normal. On general examination he was obese with BMI of 32.6 and pale. and had acanthosis nigricans. Ocular examination of anterior segment with slit lamp was normal except both pupils were reacting sluggishly to light.

Both eyes showed waxy pale disc of normal size and shape, with marked attenuation of blood vessels and diffuse mottling of peripheral retina. An obese child with loss of vision in nearly first decade with pigmentary retinopathy without frank bony spicule pigmentation made us to think about syndromic phenotypes of RP like laurence moon syndrome and bardet biedl(1). (2) Detailed systemic examination was done and we noticed patient had splenomegaly. He had normal intelligence. CVS and respiratory systems were normal, there was no polydactyly or syndactyly, external genitalia was normal. We investigated him further.
Blood reports showed Hb 6.4gm/dl (anemia)

FBS 147mg/dl PPBS 228mg/dl (diabetes)

Lipid and thyroid profiles were normal. Renal and liver function tests were normal.

ERG showed extinguished responses.

AUDIOPGRAM showed sensorineural deafness.

USG ABDOMEN showed cirrhosis of liver with splenorenal collaterals. and LIVER BIOPSY CONFIRMED CIRRHOSIS. UPPER GI ENDOSCOPY showed grade 3 esophageal varices.

OUR PATIENT HAD,

1) Pigmentary retinopathy.
2) Obesity
3) Sensorineural deafness.
4) Diabetes with acanthosis nigricans.
5) Cirrhosis of liver.
6) Normal intelligence and no polydactyly.

OUR CASE FULFILLS 1 MAJOR, 3 MINOR CRITERIA AND 2 VARIABLE SUPPORTIVE EVIDENCES, THUS QUALIFYING FOR DIAGNOSIS OF ALSTROM SYNDROME.

DISCUSSION.

Was first reported in 1959 by ALSTROM AND HALLGREN. Alstrom syndrome is rare AR disorder with prevalence less than 1:1,00,000. Mutation of ALMS1 gene located on chr 2p13 is resposible. Exact function of ALMS1 is not known. Baldness hyperurecemia scoliosis hyperostiosis frontalis interna are other features. The primary cause of mortality is cardiomyopathy in young and in older age group renal failure.

To diagnose alstrom syndrome in 3-14 years age group we need presence of 2 major or 1 major + 3 minor criterias.(3)

MAJOR CRITERIA.
1) VISION PATHOLOGY (decreased vision, photophobia, cone dystrophy by erg) 2) ALMS1 mutation in one allele and or family h/o alstrom syndrome.

MINOR CRITERIA.
1) Obesity and insulin resistance and or diabetes.
2) H/O dilated cardiomyopathy.
3) Hearing loss.
4) Hepatic dysfunction.
5) Renal failure.
6) Advanced bone age.

VARIABLE SUPPORTIVE EVIDENCES.
Normal digits
Delayed developmental milestones
Hyperlipidemia
Scoliosis
Hypertension
Recurrent UTI.

As currently it has only 266 reported cases in medical literature, and over 501 known cases in 47 countries across the world.(4) 

BIBLIOGRAPHY.
1) American journal of ophthalmology october 1986
2) Ophthalmology july 1998
3) Orphanet journal of rare genetic diseases 2007
4) IJA vol 54 mar-april 2010