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A Comprehensive Overview of a Rare Case of Type 2 Autoimmune Polyglandular Syndrome

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Abstract:-

- ***Introduction:***

Polyglandular deficiency syndromes reflects a wide spectrum of disorders. Autoimmune polyglandular syndrome (APS) is a rare condition generally divided into two categories APS -1 and APS -2.

- ***Case Report:***

We report a case of APS-2 in a 28 years old male with marfanoid habitus, presented with significant weight loss, fever and hemoptysis. upon examination, he had exophthalmos, pallor, marfanoid habitus with systolic murmur in all cardiac areas and coarse crepitation in left hemithorax. Laboratory evaluation revealed left upper lobe pneumonia secondary to Acinetobacter. Further evaluation revealed severe hyperthyroidism, anti TPO, TRAB antibody was elevated, megaloblastic anemia with atrophic gastritis and positive 21 alpha hydroxylase.

- ***Diagnosis:***

The above findings were consistent with the diagnosis of APS type 2 (Graves' Disease, Adrenal Insufficiency, Pernicious Anaemia, Vitiligo). The presence of two or more endocrine deficiencies defines APS-2 which may include graves' disease, type 1 Dm, primary adrenal insufficiency, hypogonadism and features like pernicious anaemia, vitiligo and alopecia.

- ***Conclusion:***

Circulating antibodies may precede development of clinical disease by many years but would allow clinician to follow the patient and identify the disease onset at the earliest.

Keywords:- *APS, APS-2, Autoimmune Polyglandular Syndrome, Polyglandular Deficiency Syndrome*.

I. INTRODUCTION

Autoimmune polyglandular syndromes (APS) comprise a diverse group of rare diseases characterized by autoimmune attacks targeting multiple endocrine glands, though non-endocrine organs may also be involved. These syndromes represent a broad spectrum of disorders with varying clinical presentations and complexities. These disorders are classified into two main categories: APS type 1 (APS-1) and APS type 2 (APS-2). Autoimmune Poly glandular Syndrome Type 1 (APS-1) is an autosomal recessive disorder caused by mutations in the AIRE gene. In contrast, Autoimmune Polyglandular Syndrome Type 2 (APS-2) is an autosomal dominant disorder involving multiple genes [1].

APS-2 is more common than APS-1, occurs more often in females, has onset in adulthood. Presence of two or more of the following endocrine deficiencies in the same patient defines the diagnosis of APS-2: primary adrenal insufficiency, graves' disease, type 1 diabetes mellitus and primary hypogonadism. Associated with auto immune conditions such as myasthenia gravis, vitiligo, alopecia, serositis and pernicious anemia [2].

II. CASE REPORT

We present the case report of a 28 years old male who is a known case of Marfan syndrome with features suggestive of APS-2. The patient came to us with a chief complaints of significant weight loss (about 24 Kgs in 3 months), fever for two weeks (episodic, low grade) and hemoptysis (one episode of blood-streaked sputum). There is no significant family history. He had no relevant past history.

- *Clinical Findings:*

Upon examination patient was conscious, oriented, afebrile. Pallor present, pedal oedema present, no lymphadenopathy, on detailed head to toe examination he had exophthalmos, high arched palate, positive wrist thumb sign, vitiligo present in trunk. His Blood pressure was 110/70 mmhg; pulse rate was 106 / min, Respiratory rate was 18/ min with Spo2: 98% in room air.

- Cardio vascular system examination: S1, S2 present, short systolic murmur heard in all cardiac areas.
- Respiratory system examination: coarse crepitation present over left hemi thorax.
- Per abdomen examination: soft, bowel sounds present with no organomegaly
- Central nervous system examination: no focal neurological deficit.

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III. TIMELINE OF EVENTS

The patient was clinically apparently normal three months back, when he slowly developed the above-mentioned symptoms and presented to the OPD. Thorough clinical examination was done and the patient had features suggestive of graves' disease, anemia and lower respiratory tract infection.

➢ *Diagnostic Assessment:*

(Table 1) On evaluation TsH was found to be very low suggestive of hyperthyroidism with decreased hemoglobin level. Hence, peripheral blood smear was sent and it showed dimorphic anemia with thrombocytopenia. Antibodies such thyroglobulin antibody, anti TPO, TRAB antibody were profoundly elevated suggesting the diagnosis of graves' disease. Bone marrow cytology showed erythroid hyperplasia with megaloblastic maturation. Upper Gastro-intestinal endoscopy (figure:1) was done in view of anemia and it showed atrophic gastritis suggestive of pernicious anemia. To look for other auto immune condition 21 alpha hydroxylase antibodies were sent and it came positive suggestive of Addison's disease. Echo cardiography was done and it showed anterior mitral leaflet prolapse. Sputum AFB and CBNAAT came negative but culture showed Acinetobacter species sensitive to meropenem. CT chest showed left upper lobe and lingular consolidation. HbA1C and fasting C-peptide were normal ruling out type 1 diabetes.

➢ *Therapeutic Interventions:*

Patient was adequately treated with injection meropenem, tab carbimazole, tab propranolol and injection vit b12. Patient recovered symptomatically after 2 weeks of treatment, and hence discharged with anti-thyroid drugs and antibiotics. Genetic counselling was given to the patient and to the family members.

➢ *Follow Up:*

Patient was followed up after 1 month and patient's consolidation was resolved and he was referred to higher centre for hormonal replacement therapy

Table 1 Diagnostic Assessment

Investigation	Value
WBC	4200 Cells / cu mm
Hb	6.5 g/ dl
Platelet	37000 plt / micro L
Total bilirubin	4.1 mg/dl
Direct bilirubin	0.3 mg/dl
Hba1c	5.6%
C-peptide	0.8 ng/ml
Sputum AFB	Negative
CBNAAT	Negative
Sputum culture	Acinetobacter species Sensitive to meropenem
Ct chest	Left Upper lobe and lingular consolidation
TsH	<0.005 micro IU/L
Free T3	>20pg/ml
Free T4	10.52 ng/ml
Thyroglobulin antibody	462ng/ml
Anti TPO	232.7 IU/ml
TRAB antibody	>40 IU/L
21 alpha hydroxylase antibodies	Positive

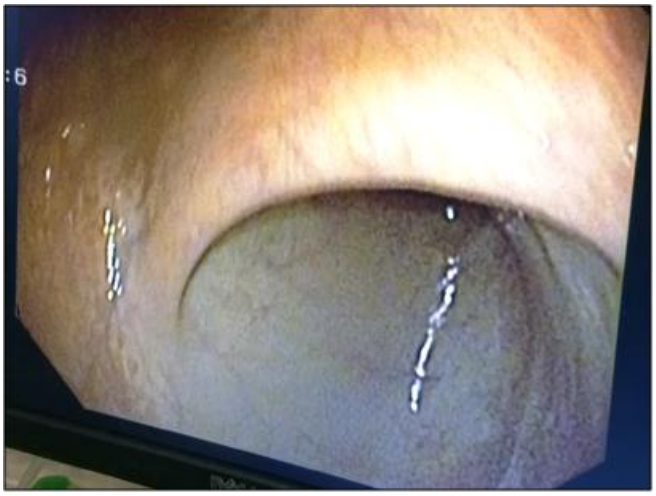

Fig 1 Upper GI Scopy Showing Atropic Gastritis.

IV. DISCUSSION

APS-2 was described earlier as a disease where Addison's disease occurred with chronic lymphocytic thyroiditis concurrently [3]. Its association with type 1 diabetes mellitus was later observed and the entity was named APS type 2. This condition was previously known as adrenal insufficiency with one or more of the following disorders in the same patient: Graves' disease or autoimmune hypothyroidism, Type 1 diabetes mellitus, Primary hypogonadism, Myasthenia gravis, Coeliac disease. Other conditions such as pernicious anaemia, vitiligo, alopecia and serositis occur at a greater frequency among these patients, but they are not a part of the diagnostic criteria.[4]

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Clinically, the frequent presentation of disorders associated with APS-2 is Addison's disease with Hashimoto's thyroiditis (56%), and the least frequent is the triad of Addison's disease, Graves' disease, and type 1 diabetes mellitus (3%) [5]. Akhila Arya PV et al reported a case of APS-2 which presented with Addison's diseases, autoimmune thyroiditis, and vitiligo [5]. Additionally, Ikram Abdullah et al reported type 1 diabetes mellitus along with other 2 disorders mentioned before [6].

V. CONCLUSION

Different ways of presentation of disorders were observed in the syndrome it is worth noticing the appearance of autoantibodies precedes the occurrence of clinical disease. It can be useful in early identification of the disease and treatment.

➢ *Additional information:*

- Consent: Informed consent was obtained from the study participant for publication.
- Conflicts of interest: Nil
- Funding: Nil

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