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Diagnosis of Cushing's Disease in an Adolescent female.

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Abstract-
Cushing's syndrome or hypercortisolism refers to the excess secretion of glucocorticoids from any sources, whereas Cushing's disease (CD) refers to hypercortisolism due to pituitary adenoma causing excess secretion of adrenocorticotropic hormone (ACTH). It is rare in paediatric population and confirming diagnosis of Cushing's Disease can become quite challenging in some cases. Hypercortisolism is associated with high morbidity and mortality, largely due to cardiovascular diseases. Here we present a case of an adolescent female patient who presented with a two- year history of excessive sweating, joint pain, headaches, hirsutism, acne, weight gain and muscle cramps.

Key words: -
Cushing's syndrome, Cushing's disease, hypercortisolism, pituitary adenoma, pseudohypercortisolism.[1]

Introduction-

Cushing's disease (CD) is a condition characterized by hypercortisolism due to excessive secretion of adrenocorticotrophic hormone (ACTH) by a pituitary adenoma. Though uncommon in children and adolescence, prevalence of Cushing's disease is around 70% with a marked female predominance, from 3:1 to 10:1 [(1)]. Diagnosing Cushing's disease, especially in young patients, can be challenging due to the difficulty in confirming and differentiating it from other types of hypercortisolism, namely ectopic ACTH or CRH (corticotropin releasing

hormone) production [2].The disease significantly increases the risk of mortality and morbidity, particularly due to cardiovascular complications, therefore early diagnosis and timely management is of utmost importance [3,6].

Case presentation-

A 17- year-old female with a body mass index (BMI) of 30.5 kg/m², presented with a two- year history of excessive sweating, daily headaches, hirsutism, acne, weight gain, and muscle cramps. Two years prior, she was seen by a paediatric endocrinologist who diagnosed hyperinsulinemia and vitamin- D deficiency. However, the patient did not start the prescribed medications. Laboratory investigations including dehydroandrosterone sulphate (DHEAS), 17-hydroxyprogesteorne (17-OHP) and total testosterone did not reveal any abnormalities. She attained menarche at the age of 12, regular since then with 26- 29-day cycles. Has an unhealthy diet, mild physical activity. She does not smoke or drink alcohol. On physical examination, 5mm wide and 2- 3 cm long purple striae were seen on the front wall of abdomen and hirsutism was scored at 15 by Ferriman- Gallwey scale. Instrumental investigations revealed steatotic liver disease.

Initial laboratory investigations revealed normal glucose tolerance with elevated C- peptide levels and HDL-cholesterol deficiency. Low dose dexamethasone suppression test (LDDST) revealed no suppression of cortisol of 6.2 mcg/dL. Repeated LDDST again showed hypercortisolism (4.6 mcg/dL). Adrenocorticotropic hormone (ACTH) was measured to differentiate between ACTH- dependent and ACTH- independent hypercortisolism. Results showed slightly elevated ACTH levels at 70ng/L (N= 7- 63), which prompted the performance of pituitary MRI. The MRI revealed a Rathke cleft cyst, 1.5mm in diameter, and a pituitary microadenoma, 2.1*1.6*1.7 mm in size. Considering patient's young age, low clinical suspicion on Cushing's syndrome (due to absence of proximal myopathy and easy bruisability), inability to perform inferior petrosal sinus sampling (IPSS) or CHR/ dexamethasone test, we did not proceed with transsphenoidal surgery (TSS) to remove the pituitary adenoma. It was decided to initially manage metabolic alterations, namely obesity and hyperinsulinemia with a combination of metformin tablets daily and semaglutide injections once weekly and a follow-up in 3-6 months' time. Patient came on a follow up visit another year later. Apparently, she had stopped taking both medications in a couple of months. LDDST now revealed suppressed cortisol of 0.74 mcg/dL with similar findings on pituitary MRT (a 1.5 mm Rathke cleft cyst and a 2.1*1.6*1.7 mm pituitary adenoma). It was assumed that the initial investigations a year ago were more suggestive of pseudohypercortisolism due to obesity and steatotic liver disease. At this point,

the patient was again commenced on semaglutide injections once weekly with titrating doses and was recommended to repeat LDDST in 6 months' time.

Conclusion-

The presence of Cushing's disease in adolescent patients may pose challenges in diagnosis as metabolic alterations may mimic Cushing's syndrome. Our clinical case demonstrates that LDDST may also show falsely elevated results that has led us to unnecessary further investigations. Therefore, careful clinical and laboratory evaluation is needed in order to avoid false diagnoses and unnecessary treatment.

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