CASE REPORT

Vomiting, Weakness, and Skin Discoloration in a 10Year Old Child

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CASE PRESENTATION

A 10 year old Pakistanian boy presented to my OPD pediatric clinic with complaints of protracted vomiting and weakness , for the last few days and that his weakness had progressively worsened to the point that he was not able to bear weight on the day of presentation. The emesis was no bilious and none bloody and was associated with vague, diffuse abdominal pain. His mother reported no fever, diarrhea, travel or exposures. No medication had been given for his symptoms.

Medical history

The patient's mother described him as an otherwise healthy boy, with normal birth history and no medical problems in the past. The mother reported that the patient's visual acuity started deterioration before one year for that reason she consult ophthalmologist and prescribed glasses for his visual impairment.

The ophthalmologist showed pigmentary retinopathy (retinitis pigmentosa) and retinal flat with hyper pigmented geographic macular lesion, lens clear, squint with refractory error showed in his both eyes. And diagnosed as (Non accommodative Esotropic)

Physical Examination:

Vital signs: Afebril, heart rates from 94- to 113 beats per minute-

-Blood pressure,from 91/48- to 80/40 mmHg,-respiratory rates 20-30 breathe per minute

-oxygen saturation% =100%.-weight 23.7 kg BMI 23

General: Thin, weak, and dry tongue, alert and awake but listless

Head, eyes, ears, nose, and throat, normocephalic;Squint and pupils equal& reactive at 3 mm, mucus membranes dry and dark Neck: supple with no adenopathy; no signs of meningismus.

Zulekha Hospital /Dubai UAE.

Cardiovascular: tachycardia, regular rhythm, no murmurs, rubs, or gallops.

Lungs: lungs clear to auscultation bilaterally

Abdomen: soft & flat with diffuse tenderness no rebound or guarding, no organomegaly, masses, or hernias

Extremities: thin but with nail dystrophic changes and bluish discoloration no joints swelling.

Skin: tanned skin, no rash, bruises, purpura, or petechie

Capillary refill delayed at> 3 sec.

Neurological examination: Glasgow coma scale score=15, generalized weakness but with normal muscle strength, tone, sensation and coordination

Management

-the child was admitted in the hospital -Intravenous access was obtained and fluid resuscitation was started with normal saline boluses 20ml/kg .and initial laboratory workup included- Venous blood gas analysis, complete blood count CBC, c-reactive protein-CRP,-ESR,-Peripheral smear,-metabolic paneland tissue transglutaminase, glygated haemoglobin (HBA1c),-Hormonal assays: 1-thyriod Function tests 2-parathyroid hormone 3- 9 am cortisol 4-ACTH 5-Aldosteron 6- rennin 7- 17 hydroxyprogesteron

Laboratory and radiographic studies

Venous blood gas pH 7.33 PCO2 40mmHg -CBC, WBC : 12 000 Cells/ mm3

Hb, 16/dl, Heamatocrit 48%, Platelets 401000 cell/ml,differential counts (59% neutrphils, 32% lymphocyte, 8% monocytes, 0% eosinophils).coagulation normal.

Hormonal assays: thyroid function test and 17 hydroxyprogesteron 0.06 ng/mL within normal and parathyroid hormone 5ng/ml (10-65ng/L) and 9 am cortisol 1.5ug/dl (4.3-22.4 ug/dl), aldosteron <10 ng/L (40-310 ng/L) were low and ACTH 19.5pg/ml (<10pg/ml), and plasma renin 4.9 ng/mL/hr (1.31-3.95) were high.

Metabolic panel- Na=131 mEq/L K= 5.4mEq/L Chloride=93 mEq/L Bicarbonate=22meq/L, Blood urea nitrogen = 35 mg/dl, Creatinine= 0.8mldl Calcium10mg/dl, Ferritin = 94 ng/L, Iron = 77mic/dl, Total iron binding capacity and HBA1c 5% and tissue transglutaminase antibodies were normal

After the patient was hydrated, he became more alert and active. Electrocardiography showed normal sinus rhythm. Chest X-RAY to evaluate the size of heart was normal. Abdominal films (i.e. kidneys, ureter, and bladder and left lateral decubitus) were also obtained to assess for sign of obstruction. Abdominal X-ray showed paucity of gas in the abdomen, therefore, abdominal computed tomography was performed. The scan could not depict the adrenal glands, which confirmed the suspicion of adrenal insufficiency and crisis. Upon, further discussion with parents, we confirmed that no family members had darker skin pigmentation

The presenceof dystrophic changes of nails and hypoparathyroidsm with hypoadrenalism also known as autoimmune polyendocrinopathy with cutanous ectodermal dystrophic (APECED)

Case summary

This patient's presentation can be concisely summarized to contribute to the diagnosis of Addison disease with adrenal crisis: weakness, vomiting, abdominal pain, abnormal skin and mucus membrane coloring, hypotension, tachycardia, Hyperkalemia, hyponatremia and hypochloremia.

Patient Follow –up:

The patient continues to consult me and is taking daily hydrocortisone and Fludrocortison.He is feeling better and his electrolytes have returned to normal on repeated laboratory work. Addison disease (i.e. primary adrenal insufficiency) was diagnosed after confirmatory laboratory testing. Although adrenal insufficiency is a relatively rare paediatric condition, the constellation of clinical signs (ie, weakness, vomiting, hypotension and dehydration) must lend to its consideration.(1)

DISCUSSION:

Addison disease is a disorder of the adrenal glands that results in decreased production of steroids. It was first described by British physician Thomas in 1855. The incidence of Addison disease is 40-60 cases per million and the disease usually presents in adults who are 30-50 years old[(2)]. Mortality is directly associated with an addisonian crisis. The causes of this disease are broadly identified as primary or secondary. Primary disease reflects impairment of the adrenal glands. Example includes autoimmune (70%) congenital adrenal hyperplasia, adenoma, and tuberculosis infection and subsequent damage. Secondary causes occur with impairment of the hypothalamic-pituitary axis. Examples include malignancy, Sheehan syndrome, and tuberculosis.[(2)] Adrenal insufficiency occurs when 90% of the cortex has been destroyed adrenocorticotropic hormone (ACTH) and rennin levels are elevated in primary adrenal insufficiency. Addison disease and its clinical manifestations occur because of impaired cortisol and aldosterone secretion. Secretion of cortisol, a stress hormone , results in blood pressure elevation. Aldosterone causes increased reabsorption of sodium and chloride, as well as secretion of potassium. Hyponatremia can occur with urinary sodium loss, whereas a lack of cortisol results in hyperkalemia. Progression of this disease may be insidious and nonspecific.[(2)]

Common symptoms of acute crisis include:

Abdominal pain,Vomiting,Weight loss, Weakness or fatigue,Myalgias or arthrlgias,Decreased body hair,Dehydration,Diarrhea or constipation, Dizziness or syncope, Low blood pressureHypoglycemia and Cravings for salt

Hyper pigmentation is common in patients with Addison disease and involves dark patches of skin, especially in the skin folds black freckles on the forehead and face. And discoloration around areas such as the nipples, lips, and rectum.

Cutaneous pigmentation is a well-known manifestation of Addison disease. However, oral mucous membrane hyperpimentation is less common and is virtually pathognomonic [(3,4)].The diagnosis of adrenal insufficiency begins by demonstrating low cortisol secretion, typically in the morning while fasting .An ACTH stimulation test (ie, cosyntropin stimulation test) confirms the diagnosis with greater sensitivity in patients with primary disease (specificity, 95% sensitivity 97%) [(5,6)].The result of these 2 tests determine primary vs. secondary causes. Low cortisol levels and elevated ACTH level suggest primary disease in which case adrenal antibodies should be measured. If patient is negative for antibodies, causes such as tuberculosis adrenoleukodystrophy. Adrenoleukodystrophy or congenital adrenal hypoplasia should be examined. Mineralocorticold activity should also be evaluated but deficiency

can be assumed if patients with normal kidney function have hyperkalemia and hyponatremia. Plasma rennin activity and direct renin concentration will be elevated in these patients[7] . Treatment of primary adrenal insufficiency is with 15-25 mg hydrocortisone every 8 hours and fludrocortisones 0.05-0.2 mg/d.[8]

CONCLUSION:

In children, adrenal insufficiency results from numerous causes and should be a concern in any patient with vomiting and hypotension. Primary adrenal insufficiency and addisonian crisis are less common in children. However they carry significant morbidity if not urgently addressed with supportive measures and treatment with glucocorticoids and mineralocorticoids .our case is an example of a patient with addisonian crisis presenting to a paediatric clinic without any previous diagnosis of disease, highlighting the need for clinical suspicion and timely management in the paediatric patient

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