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RESEARCH ARTICLE

A CASE REPORT ON ATYPICAL PRESENTATION OF CONGENITAL ADRENAL HYPERPLASIA

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Abstract

We report a case of 52-day old infant presented to casualty with complaint of failure to gain weight,10-day history of regurgitation of feeds, poor acceptance of feed and lethargy from one day, not passed urine in last 8hours. On examination baby had tachycardia with signs of dehydration, blood gas shows metabolic acidosis with serum sodium-124meq/l, serum potassium- 6.2meq/l. A probable diagnosis of failure to thrive with severe sepsis, inborn errors of metabolism (IEM), and CAH was considered and baby was treated with intravenous fluids, antibiotics, calcium gluconate, salbutamol nebulisation and hydrocortisone. Following the above interventions, baby's hydration status and ECG changes improved. The diagnosis of CAH was considered as 17- hydroxyprogesterone levels were elevated. Genetic analysis was not done due to patient's economic constraints and baby was discharged with oral Hydrocortisone, Fludrocortisone and table salt (Nacl). Currently baby is thriving well with adequate weight gain.

Introduction:-

Congenital adrenal hyperplasia (CAH) refers to a group of disorders that arise from defective steroidogenesis. CAH due to 21-hydroxylase deficiency is a potentially life-threatening endocrine disorder if not diagnosed and treated timely [1].The prevalence of CAH in India is reported 1 in 5762 babies as per newborn screening data [2].

The disorder has variable phenotypic expressions ranging from overt symptomatic disease with signs of acute adrenal insufficiency and virilization at birth in female infants [salt- wasting (SW) CAH], to only virilization in female babies and precocious puberty in boys without features of adrenal insufficiency [simple-virilizing (SV) CAH], to non-classical CAH which may remain asymptomatic or present during adolescence with features of hyper-androgenism.[3]

Newborn screening of all babies for CAH will help improve infant mortality rate and quality of life by assisting in early diagnosis and well-timed initiation of steroid therapy.[4]

We report an unusual case of CAH presented without any hypoglycaemia, hyperpigmentation, or any features of disorders of sexual development (DSD), but with features of septic shock.

Case Study

A 52-day old male infant presented to our emergency department with history of poor feeding, decreased activity one day post vaccination, not passing urine for the last 8hours.

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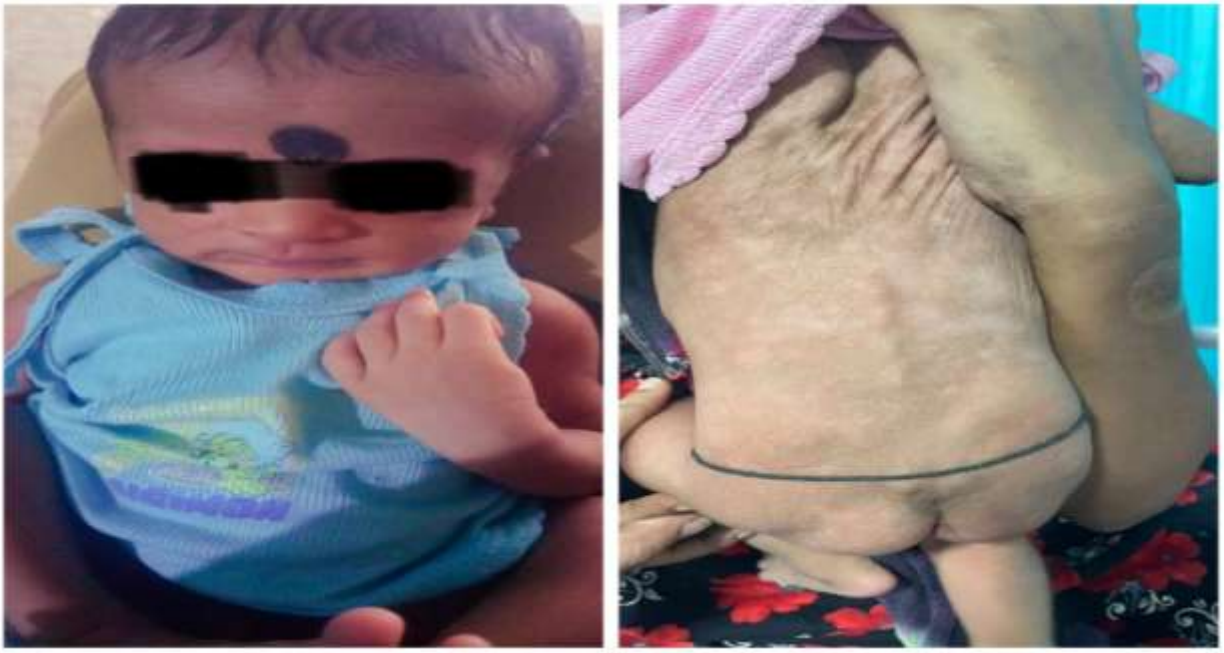
Baby was first born child out of non-consanguineous marriage with no significant antenatal history, delivered at 38weeks of gestation through elective LSCS with birth weight- 3.3kg and immediate postnatal period being uneventful. At one month of age mother noticed baby has not gained weight inspite of exclusive breastfeeding but did not seek any medical attention. From past 10 days baby had regurgitation of feeds, poor feeding, decreased activity for one day post vaccination and no urination for past 8 hours.

On examination baby was sick looking, dull with sunken eyes, dry, lusterless loose folds of skin with multiple hypopigmented patches present over the trunk, anterior fontanelle was at level admitting tip of 2 fingers. External genitalia w normal. He was noted to have poor weight gain with weight at admission being 3.4kg (100gm weight gain in 52days), Heart rate – 152 breaths per min, Respiratory rate – 52 cycles pr minute, maintaining saturation (98%) on room air, blood pressure – 82/44mm of hg over right upper arm, capillary refill time – 3 seconds. Systemic examination was noted to be normal.

A probable diagnosis of sepsis with dehydration, inborn errors of metabolism (IEM), gastro esophageal reflux disease (GERD) were considered and investigated in these lines. Blood gas showed pH 7.38 (735- 7.45),



HCO3 17.1 (22-28), PCO2 23 (40-60), BE 9.8, lactate – 1.2, Na+ - 124meq/l, k+ - 6.2meq/l, sr. Creatinine- 1.5mg/dl, glucose – 82mg/dl.

Baby was treated in lines of sepsis with dehydration with intravenous fluids and antibiotics (piperacillin tazobactam, clindamycin) and continued on exclusive breast feeds. Tandem mass spectrometry (TMS), Gas chromatography mass spectrometry (GCMS) were done in view of IEM (urea cycle and fatty oxidation defects) which turned out to be normal. Inspite of adequate hydration and treatment with higher antibiotics baby continued to have signs of dehydration and electrolyte disturbances (hyponatremia and hyperkalemia) along with ECG changes showing tall peaked "T" waves with good urine output, for correction of hyperkalemia stat dose of calcium gluconate and salbutamol nebulization were given. A probable diagnosis of CAH and pseudo hypoaldosteronism (type-4 renal tubular acidosis) was considered and evaluated.

Image Of Baby At Presentation

Investigations

Serum cortisol – 2.2 ug/dl,

•17- OH Progesterone. - > 30 ng/ml,

•Serum aldosterone – 70.90 ng/dl,

•Direct plasma renin - > 500 microIU/ml.

Baby was treated with injection Hydrocortisone, table salt and k- bind after which signs of improvement were seen in terms of activity, hydration status, ECG changes and electrolyte imbalance. Regular monitoring of the ECG, electrolytes, and blood pressure were done in hospital and were gradually improved to satisfactory levels. A probable diagnosis of CAH (salt wasting type secondary to 21hydroxylase deficiency) was considered. Elevation of serum aldosterone in our case was probably considered as aldosterone resistance due to sepsis. Baby was discharged on oral hydrocortisone (15mg/m2/day in 3 divided doses), oral fludrocortisone (200mcg/day), table salt (1gm/day). An instruction was given to the parents to double the dose of oral hydrocortisone in intercurrent illnesses (fever, cough, vomiting, diarrhea). They were also given an emergency chart which can be assessed and managed immediately by the hospital team whenever present to the hospital.

Image Of Baby After Treatment

Conclusions:-

Congenital Adrenal Hyperplasia (CAH) is autosomal recessive disorder caused by defect in steroidogenesis with reported incidence of 1:10,000- 1:20,000 births worldwide. CAH has varied spectrum of presentation ranging from acute adrenal crisis and virilization in newborns to subtle menstrual irregularities in the adolescent population [4].

Most important variant is the salt-wasting crisis presenting as a medical emergency. Females with classic 21-hydroxylase deficiency are exposed to excess androgens prenatally and are born with virilised external genitalia. Males have no genital ambiguity to alert physicians before the onset of dehydration and shock; hence the diagnosis is particularly critical in them. If CAH is not diagnosed and treated early, neonates are susceptible to sudden death at the early age of life due to arrhythmias [5].

Hallmark features in order to suspect CAH:

Symptoms of failure to gain weight or progressive weight loss inspite of regular feeding without any gastrointestinal disturbances, vomiting, lethargy, pigmentation, clitoromegaly (in females) and signs of dehydration, hypotension, hypovolemia, hypoglycaemia, hyponatremia, hyperkalaemia.

Learning Objectives:-

Infant presenting with failure to gain weight inspite of adequate feeding practices, and combination of hypoglycaemia, hyponatremia, hyperkalaemia along with metabolic acidosis is likely to be suggestive of adrenal insufficiency, most common being the CAH. Treatment with hydrocortisone gives an excellent response.

Emergency management of salt wasting crisis is to be considered as arrhythmia's resulting from hyperkalaemia is a fatal complication.

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