Case of Pseudo-Bartter's Syndrome: An atypical presentation of cystic fibrosis

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Abstract

A three months infant who in the beginning had disease cystic fibrosis was diagonosed with pseudo-bartter's syndrome. The disease began with coughing, diarrhoea, vomiting and weakness. Investigation revealed; electrolytes showin hyponatremia (110 mmol/L) and hypokalemic (2.6 mmol/L) and hypochloremic (63 mmol/L) metabolic alkalosis (HCO_3=43 mmol/L).

Key words: Pseudo Bartter's Syndrome, Cystic fibrosis, Metabolic alkalosis.

Introduction

Pseudo-bartter's syndrome is a rare syndrome of electrolyte depletion, metabolic alkalosis,and failure to thrive. Hypokalemic metabolic alkalosis, encountered in variety of disease without renal tubular pathology will ultimately be corrected once the underlying disease is identified and treated. Any corrective fluid and electrolyte will therefore be a part of basic disease treatment. (PBS) can be differentiated from barter s syndrome where sweat electrolyte loss is normal and the electrolyte disturbance is due to detective renal electrolyte handling.

Case history

A three months old female, born to consanguineous parents was hospitalized with a cough since the previous 10 days, there had been no prior respiratory symptomatoms, the pregnancy including the perinatal period was eventful.

There was histoty of intravenous fluid administration during the first month of her life because of weakness and poorfeeding. She was being breastfeeding two weeks before being admitted to the hospital. She then developed frequent loose stools, non bilous vomiting, abdominal distension and failure to thrive (FTT).

On examination the child weighed 4300 grams with a length of 51 cm and head circumference of 37 cm (which was in the 5th centil for the age). Her vital signs revealed a temeprature of 37.2 C, Respiratory Rate of 60/min, Pulse rate was 120/min, Blood Pressure was 70/50 mmHg. There was no facial dysmorphism and had no localizing signs on neurological examination. The systemic examination was otherwise normal.

She was resucitated with dextrose saline, intravenous antibiotics (cephalothin and amikacine), Creon 500iu/Kg (pancreatic enzyme), Vitamins and supplements (Zn, Fe, B complex, Vit A, Vit D, Vit E) and formula caprinol (MCT oil), Chest physiothrapy, Bronchodilator as nebuliser.

She was discharged after 10 days in good condition but still without full correction of her alkalosis (PH=7.5, HCO_3=38meq/L, PO_2=95mmg, PCO_2=45mmg, O_2 sat=98%).

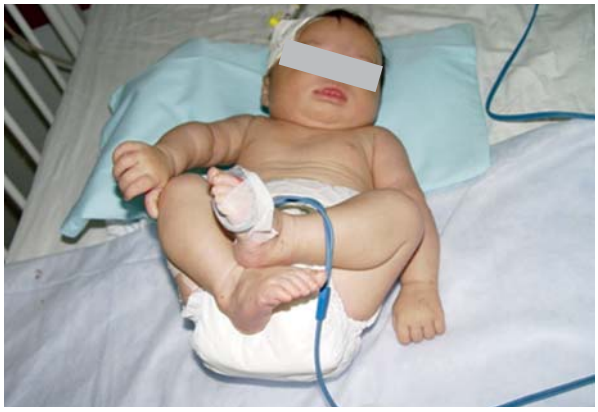

Fig 1: Photograph of the baby with pseudo-bartter's syndrome

Laboratory Examination Reports

Sr No.	Investigations	Results
1.	Hemoglobin	10.5gm/dl
2.	White Blood Cells	12000/mm^3
3.	Neutrophils: Lymphocytes: Monocytes	42.4%, 39.4%, 18.2%
4.	Platelet	488000/mm^3
5.	Red Blood Cell Counts	435000000/mm^3
6.	Hematocrit	32%
7.	Blood Suger, Erythrocyte Sedimentation Rate (ESR), C-Reactive Protein (CRP), Liver Function Tests.	All within normal limits.
8.	Creatinine	1.4 mg/dl (0.5-1.5)
9.	Sodium (Na)	110 mmol/L(135-145)
10.	Potassium (K)	2.6mmol/L(3.5-5.5)
11.	Chloride (Cl)	63 mmol/L(90-110)
12.	Bicarbonate (HCO_3)	43meq/L(24-28)
13.	Blood Culture, Stool Microscopy and Stool Culture, Urinalaysis and Urine Culture and Lumbar puncture.	No Abnormality Detected (NAD)
14.	Metabolic profiles (ammonia, lactate, pyruvate, LDH, Reducing Substance of Serum and Urine).	All normal and within normal limits.
15.	24 hours Urinary Volume	900 cc
16.	Urinary Chloride	20meq/L
17.	Urinary Potassium	18meq/L
18.	Chest X Ray	Hyperairation of lungs
19.	Abdominal X Ray	Distention of intestinal loops
20.	Abdominal Ultra-Sonogram	Normal
21.	Sweat test×3 times	Cl=130meq/L, Na=110meq/L

Discussion

Cysyic fibrosis (CF) is an autosomal recessive multisistemic disease affecting 1 in 2500 newborns among Caucasians (though rare among Orientals; 1 in 90000). The disease was described first by Anderson in 1938 as cystic fibrosis of the pancrease to the point of the pancreatic exocrine function[1,2]. In 1953 Di sant Agnese and et.al. demonstrated that excessive salt loss occurs in the sweat of CF patients. This finding led to the use of sweat electrolytes measurement as a diagnostic tool. The major clinical characterstics of CF are pancreatic insufficiency and progressive lung disease, caused by thick and dehydrated airway mucus frequently infected with pseudomonas and staphylococcus leading to respiratory failure and CF mortality. CF is typically present in infancy with combinations of FTT and steatorrhoea and respiratory symptoms[3,4].

Pseudo-bartter's syndrome (PBS) is a rare atypical presentation of CF with electrolyte depletion, alkalosis and FTT[3,4].

Investigations of serum electrolytes in our patient showed hyponatremia (110mmol/L) and hypokalemic hypocholermic metabolic alkalosis (K: 2.6mmol/L, Cl: 63mmol/L, HCO_3 : 43meq/L, PCO_2 : 47.3). Pseudo-bartter's syndrome is often difficult to distinguish from bartter's syndrome.

Bartter's syndrome is an inherited renal tubular disorder characterized by hypokalemia, hypochloremic metabolic alkalosis, hyper-reninemia, hyper-prostaglandinism, normal blood pressure with an increased urinary loos of Na, Cl, K and prostaglandin[3,4].

The history of anamnesis of Bartter's syndrome in our patient was not evident and clinically the above characteristics of the barter syndrome and Gitelman's syndrome. (Gitelman's syndrome is another clinical type of Bartter's syndrome that characterized by hypomagnesemia, hypocalciuria, pseudo-hyperaldostronism (hypertension with no evidence of increased secretion of mineralocorticoids) and pseudo-Bartter's syndrome due to an administration of high dose of prostaglandin E1[5,6].

Nutritional requirements in CF will differ depending on: age, sex, efficacy of small intestinal absoption, respiratory status and activity level. Improved nutrition benefits growth, respiratory muscle strength and immunological status. Nutritional intervention should begin as soon as the diagnosis is made in order to prevent or resolve malnutrition. Early diagnosis and prompt treatment of CF reduce long-term morbidity and promote psychological and social adaptation to the condition[5,6].

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