ORIGINAL RESEARCH PAPER

PEDIATRICS

A CASE REPORT OF GITELMAN SYNDROM

KEY WORDS:

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INTRODUCTION

Gitelman syndrome is a rare autosomal recessive cause of salt losing tubulopathy characterized by renal potassium wasting, hypokalemic hypochloremic metabolic alkalosis , with distinct features of hypocalciuria and hypomagnesemia. Patient of gitelman syndrome typically present in late childhood or early adulthood. Gitelman syndrome also referred to as familial hypokalemia – hypomagnesemia. Prevalence is 1:40000.

Pathogenesis

The biochemical features of Gitelman syndrome resemble those of chronic use of thiazide diuretics, which act on sodium chloride cotransporter present in distal convulated tubule. Mutation in solute carrier family 12 , member 3 gene SLC12A3 Which encode thiazide sensitive NACL cotransporter.

Diagnosis

The key clinical complaints and manifestations suggesting a diagnosis of GS include the following: salt craving, muscle weakness, fatigue, limited sport performance, episodes of fainting, cramps, tetany, paresthesia, carpopedal spasms; growth retardation, pubertal delay, short stature; thirst or abnormal drinking behavior; episodes of abdominal pain. Dizziness, vertigo, polyuria, nocturia, palpitations, joint pain, and visual problems may be reported in adults.The diagnosis of Gitelman syndrome is suggested in late childhood or early adulthood presenting with hypokalemic hypochloremic metabolic alkalosis , hypomagnesemia and hypocalciuria with renal potassium wasting.normal or low blood pressure; normal renal ultrasound with absence of nephrocalcinosis or renal abnormalities.

Case Presentation

6 year male child admitted in PICU K T Children hospital Rajkot with complain of decreased oral intake with difficulty in breathing, muscle weakness , vomiting , normal blood pressure. On blood investigation patient having metabolic alkalosis, hypochloremia, hypokalemia ,hypomagnesemia, hypocalciuria. According to this investigation patient diagnosed as Gitelman syndrome.

Treatment & Prognosis

Therapy is directed at correcting hypokalemia and hypomagnesemia with supplemental potassium and magnesium . After clinical diagnosis life long supplement of magnesium , potassium and sodium started. In the presence of hypomagnesemia, magnesium supplementation should be considered first, because magnesium repletion will facilitate potassium repletion and reduce the risk of tetany and other complications.Prognosis is excellent.

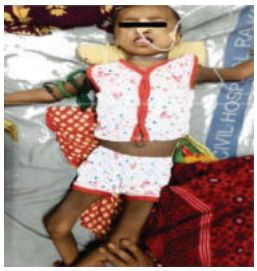

Figure1 - Patient Photo

PARAMETER	PARAMETE	PARAMETER	PARAMETER
PH	7.8	Alkalosis	PH 7.35-7.45
HCO3	70.4	metabolic	22-26 meq/l
SERUM SODIUM	144	normal	136-145 meq/l
SERUM POTASSIUM	2.2	hypokalemia	3.5- 5.1 meq/l
SERUM CHLORIDE	79	hypochloremia	98-107 mmol/l
SERUM MAGNESUIM	1.1	hypomagnesemia	1.7-2.3 mg/dl
URINE POTASSIUM	19.48	potassium	20-40 meq/l
URINE CALCIUM	1.2	hypocalciuria	100-200 mg/dl
URINE CHLORIDE	85	low	110-250 meq/l

Figure2 – Biochemical Change Of Patient

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