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Case Report: A Rare Presentation of Quadriplegia as Gitelman Syndrome

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Abstract: ***Gitelman syndrome is an autosomal recessive disorder caused by the mutation of SLC12A3 or CLCNKB which encodes renal Na – Cl co transporter channels it causes imbalance of potassium magnesium and calcium metabolism. Gitelman symptoms are similar to thiazide diuretic-abusers with salt wasting. Gitelman syndrome, the defect resides at the distal convoluted tubule and rare cause of quadriplegia***

Keywords: gitelman, quadriplegia, potassium

1. Introduction

Gitelman syndrome is an autosomal recessive disorder caused by the mutation of SLC12A3 or CLCNKB which encodes renal Na – Cl co transporter channels it causes imbalance of potassium magnesium and calcium metabolism. Gitelman symptoms are similar to thiazide diuretic-abusers with salt wasting. Gitelman syndrome, the defect resides at the distal convoluted tubule (DCT). (1)

2. Case Report

A 58 years old male named Ramangouda came to our hospital with complaints of weakness of both upper and lower limbs since 3 days, which was insidoius in onset and gradually progressed in 3 days. O/E patient has hypotonia with 0/5 power in all 4 limbs with the absence of deep tendon reflexes with mute plantar without involvement of any cranial nerves, sensory and autonomic system examination was normal, investigations wer showing potassium- 1.7 with low magnesium and elevated bilirubin. ABG showing metabolic alkaosis with USG showing Ascites and Hepatomegaly. MRI shows old infracts urine potassium was more than 15mmol. TTTKG more than 4 with low BP and metabolic alkalosis with high chloride in urine with calcium/ creatinine is less than 0.15. there was no history of usage of any Thiazides and patient was improved with potassium and magnesium supplementations.

Investigations

- Hb - 14.5 g/dl (14-18)
- TLC - 7.3x1000/cu mm(4.3-10x1000/cu
- MCV -76.2 fl (77-93 fl)
- MCH -27.2pg(26-32pg)
- MCHC -31.4gm/dl (32-36gm/dl)
- Platelet -3.0(1.4-4.0 lakhs/ml)

Bilirubinin-
Total : 4.1 (upto 1.0mg/dl)
Direct : 1.9 (upto0.1-0.3mg/dl)
Indirect : 2.2 (upto0.2-0.7mg/dl)
SGOT : 164(upto 40 U/L)
SGPT : 86(upto 50 U/L)
ALP : 56(upto 25-140IU/L)
LDH : 320(440)
CPK : 45(26-140)
LIPID PROFILE : NORMAL
S.Calcium : 7.6(8.5-10.5mg/dl)
S.Total protein : 4.1(6.2-8.5gm/dl)
S.Albumin : 2.3(3.5-5.5gm/dl)
S.Electrolytes
Sodium - 136(135-145mmol/l)
Potassium - 1.7(3.5-5.5mmol/l)
Chloride - 101(95-105mmol/l)
S.Urea - 17(15-45mg/dl)
S.Creatinine - 0.5mg/dl(0.7-1.5mg/dl
Urine Routine - Normal.
ABG
pH - 7.52(7.35-7.45)
pCO2- - 28(35-45)mm/Hg
pO2 - 94(80-100)mm/Hg
Na+ - 136(135-145)mEq/L
K+ - 1.6(3.5-5.5)mEq/L
HCO3- - 36(22-26)mEq/L
SpO2 - 98%
ABG showing metabolic alkalosis with hypokalemia.

Differential Diagnosis-

Brainstem and Cervical Cord Lesion With Spinal Shock
Gullian Barre Syndrome
Mysthenia Gravis
Metabolic Causes

	POINTS IN FAVOUR	POINTS AGAINST
BRAIN STEM AND CERVICAL CORD LESION WITH SPINAL SHOCK	ACUTE ONSET AREFLEXIA IN ALL 4 LIMBS	NO H/O TRAUMA NO SENSORY INVOLVEMENT NO BOWEL BLADDER INVOLVEMENT NO RESPIRATORY INVOLVEMENT MRI- NORMAL
GULLIAN BARRE SYNDROME	ACUTE ONSET AREFLEXIA IN ALL LIMBS NO BOWELBLADDER INVOLVEMENT NO SENSORY INVOLVEMENT	NO ASCENDING PATTERN AND SIMULTANEOUS ONSET IN ALL LIMBS NO RESPIRATORY INVOLVEMENT NCV NOT SUGGESTIVE OF GBS
MYSTHENIA GRAVIS	LMN QUADRIPARESIS	NO DIURINAL VARIATIONS NO PTOSIS
HYPOKALEMIA PARALYSIS	ACUTE ONSET AREFLEXIA SERUM POTASSIUM LOW AND IMPROVEMENT IN WEAKNESS WITH POTASSIUM SUPPLEMENTATIONS	NO H/O DRUG EXPOSURE NO H/O EXCESSIVE STRAIN, EXERCISE OR CARBOHYDRATE CONSUPTION THYROID - NORMAL

3. Discussion

In gitelman syndrome the mutation of SLC12A3 or CLCNKB which encodes the renal thiazide sensitive Na-Cl cotransporter predominantly located in distal convoluted tubule. Defect causes imbalance of electrolytes mainly potassium,magnesium and calcium. It is relatively a benign condition. Described by GITELMAN in 1966 in three adult patients with intermittent episodes of tetany , hypokalemia,and hypomagnesemia, but with no polyuria or growth retardation.It is usually diagnosed during late childhood or adulthood.

Sodium chloride enters the cell via the apical thiazide – sensitive NCC and leaves the cell through the baso-lateral Cl channel(ClC-Kb),and the Na+/k+-ATPase.(2)Studies Recently identified magnesium channel TRPM6 in the apical membrane and a putative Na/Mg exchanger in the basolateral membrane,These transport mechanisms play a role in familial hypokalemia-hypomagnesemia or Gitelmans syndrome. (3)The urinary calcium/creatinine ratio was calculated The ratio was Ca/Cr ratio - 0.01mmol/gl. If Ca/Cr ratio is >0.2,it would favour hypercalciuria and thus BARTTER'S SYNDROME

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