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Medicine

GITELMAN SYNDROME NAVIGATING THE CHALLENGES IN DIAGNOSIS AND MANAGEMENT

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KEYWORDS :

BACKGROUND

Gitelman syndrome is a rare autosomal recessive inherited disorder that affects the kidneys.

GS is more prevalent in Asia, with estimates of 1.7 per 1,000 people.

Diagnosed in adolescence or adulthood,most commonly in second and third decade of life.

The ratio of men to women affected by Gitelman syndrome is roughly 1:1.

It is a type of **renal tubular disorder.**

Caused by mutations in specific genes that encode for sodium-chloride co-transporter proteins in the kidney tubules, most commonly the **SLC12A3** gene

•This defect impairs the kidney's ability to reabsorb salt and causes changes in various electrolyte concentrations as well as contraction of extracellular fluid volume (thus causing symptoms of dehydration).

• Specifically important electrolytes like

– **Sodium,**

– **Potassium,**

– **Magnesium,** and

– **Calcium**.

leading to disturbances in electrolyte balance.

Often manifests as muscle cramps, muscle weakness fatigue, tingling or numbness, hypotension, gastrointestinal problems such as abdominal pain, nausea and vomiting, polyuria, polydipsia .

Diagnosis is typically made through laboratory tests, which show

– Hypokalemia

– Hypochloremia

– Hypomagnesemia

– Hypocalciuria

– Hyperreninemia

– Hyperaldosteronemia

– metabolic alkalosis)

AIM AND OBJECTIVE

• To report a rare case of GITELMAN SYNDROME.

• To discuss about the treatment plan.

CASE REPORT

A 47 year old male patient, presented to medicine opd of PRAVARA RURAL HOSPITAL, Loni, with complaints of

- tremors in both the upper and lower limbs since 3 days,
- bilateral upper limb weakness since 3 days(left>right) ,
- unable to supinate b/l upper limb since 1 day.
- Loose stools 4-5 episodes since 2 days.

All the symptoms were sudden in onset and gradually progressive in nature,

• On examination

– power at the level of wrist 3/5 in both upper limbs

– 4/5 at the level of elbow in both upper limbs

– Plantar reflex was decreased in both limbs.

Patient was provisionally diagnosed as ? CVA, hence was admitted and all necessary investigations were done.

Blood picture was as follows

ELECTROLYTE	VALUE	REMARK
POTASSIUM	2.2	HYPO
SODIUM	136.67	NORMAL
CHLORIDE	95.45	NORMAL
MAGNESIUM	1.6	HYPO

- Serial blood repeats showed persistent Hypokalemia Values being- 1.81 meq/l, 1.98 meq/l, 2.4mEq/L
- Urinary electrolytes were as follows calciuriaggesting severe Hypo

ELECTROLYTE	VALUE
U. NA	163.90
U. K	23.72
U. CL	175.72
U. CA	0.6

- Nerve conduction studies were done as well which was normal.

TREATMENT

- Patient was given corrections for hypokalemia along with dietary changes with potassium rich diet.
- Inj. Mgso4 1 amp in 100 cc ns over 2 hours, for 3 consecutive days for hypomagnesemia.
- Along with adequate hydration and other supportive medications.
- After 3 days of electrolyte correction there was a drastic improvement in patient symptoms, his power has increased to 5/5, and loose stools have stopped.
- His serum potassium was 4.1 meq/l
- Hence the patient was clinically diagnosed with **Gittleman syndrome.**
- **Now patient is on regular follow up for close monitoring, and is doing well.**

CONCLUSION

- Early diagnosis, based on clinical symptoms and understanding the genetic and physiological underpinning of this disorder, is crucial for effective management.
- Treatment focuses on correcting electrolyte deficiencies through supplementation and dietary adjustments, with regular monitoring to prevent complications such as arrhythmias and kidney damage.
- With appropriate care, individuals with Gitelman syndrome can lead a normal life and avoid severe long-term effects.