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Gitelman's syndrome as an incidental finding in a 60-year oldmale

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Abstract

Gitelman's syndrome (GS) is an inherited benign salt-losing tubulopathy characterized by hypokalemic metabolic alkalosis, hypomagnesemia and hypocalciuria and is often diagnosed in adolescents and middle-aged population. GS features include electrolyte imbalance, symptoms such as severe fatigue, cramps, polyuria and nocturia, and hypertension. Asymptomatic GS have also been reported and would present as an incidental finding.

Gitelman condition (GS) is an autosomal latent kidney tubule issue portrayed by low blood levels of potassium and magnesium, diminished discharge of calcium in the pee, and raised blood pH.[2] The confusion is brought about by hereditary transformations bringing about ill-advised capacity of the thiazide-touchy sodium-chloride symporter (otherwise called NCC, NCCT, or TSC) situated in the distal tangled tubule of the kidney.[2] The distal tangled tubule of the kidney serves a negligible job in salt ingestion and a more prominent job in dealing with the discharge of electrolytes like magnesium and calcium to deliver increasingly focused urine.[3]

Hereditary changes along the sodium chloride symporter, lead to lacking vehicle of numerous electrolytes along this channel, for example, sodium, chloride, calcium, magnesium, and potassium. The net impact is an electrolyte unevenness reliable with thiazide diuretic treatment

Gitelman condition was some time ago viewed as a subset of Bartter condition until the unmistakable hereditary and sub-atomic bases of these disarranges were recognized. Bartter condition is additionally an autosomal passive hypo kalemic metabolic alkalosis, yet it gets from a change to the NKCC2 found in the thick climbing appendage of the circle of Henle.[4]

Introduction

Gitelman's syndrome (GS) is a rare genetic disorder affecting about 1-10 per 40,000 and is the most common inherited tubulopathy. It is caused by a mutation in the SCL12A3 gene encoding the thiazide-sensitive sodium-chloride transporter (NCC), with a minority of cases reported also having a mutation in CLCNKB gene encoding the chloride channel C1C-Kb. More than 140 gene mutations of NCC have been identified. Symptoms usually develop late childhood and is usually diagnosed during adulthood.

Case Report

Methods

A 60-year old male,hyper tensive for more than 5 years, with good control, usual BP120-130/60- 80 maintained on Losartan 50mg once a day and Spironolactone 25mg once day, known case of Chronic Obstructive Pulmonary Disease (COPD) on Doxofylline 400mg twice a day, came in for bilateral swelling of feet and legs with associated shortness of breath, dyspnea, allegedly increasing abdominal girth and undocumented febrile episodes. Venous compression test was negative for deep venous thrombosis in the bilateral lower

extremities. 2D echo and ECG were doneandyieldedunremarkableresults.

Results

He was referred to Gastro where a one-organ ultra sound and whole abdominal CT scan were done, still with un-remarkable results. Liver elastography showed F2 (significant fibrosis). Urinalysis showed pyuria (WBC 8) and bacteriuria (Bacteria 1244).At this time, the patient is being managed as a case of complicate durinarytract infection and was started on piperacillin-tazobactam. Urine culture and sensitivity showed Acinetobacterbaumanii sensitive to the current antibiotic. Other diagnostic tests were requested where an incidental finding of abnormal electrolytes led to a referral to a Nephrologist. Patient had hypokalemia (K 3.3), hypo magnesemia (Mg 1.3), no hypo calcemia (Ca 7.9, corrected Ca 8.86). Correction of these were started. Patient occasionally has complaints of cramping of legs, otherwise, denies any fatigue, polyuria, nocturia. After correction of electrolytes, a renal panel was requested, with the following results: potassium 3.2, magnesium 1.8, calcium 7.7 (corrected Ca 8.6). Correction was again started for potassium and magnesium. Repeat electrolyte check was done showed potassium 3.3, magnesium 1.7. Another cycle of correction was done; repeat potassium 3.5, magnesium 1.5; patient now has hypocalcemia 6.4(correctedCa7.3).Correction was again started. Hewaswork- up forother causes of hypokalemia. Doxofyl line dose was decreased. Thyroid function tests were normal. Urine studies were requested: urine osmolality 453mosm/kg, urine sodium 38mmol/L, urine potassium 31mmol/L, urine creatinine 70.65mg/dL, urine calcium 5.5mg/dL; serum osmolality 293mosm/kg. Transtubular potassium gradient (TTKG) was 5.73. BP remained within normal range. Arterial blood gas(ABG)showed metabolic alkalosis. Urine chloride was 85mmol/L and urine chloride/calcium is computed at 0.06.Following the algorithm for hypo kalemia, weare left with two differentials: Gitel man's syndrome and thiazide diuretic use. Exclusion of other causes for hypo kalemic metabolic alkalosis, such as surreptitious vomiting and surreptitious diuretic use, was done. However, no genetic testing was done. Levels of renin and aldosterone were not checked during his stay in the hospital.

This patient was maintained on potassium and magnesium supplementation when he was discharged until follow up.

Discussions

Gitelman's syndrome is an auto somalrecessive traitest imated to beoccurring~1% in Caucasian populations. GS is often discussed with Bartter syndrome and should be taken as a spectrum of disease rather than distinguished them by their individual features. In Bartter syndrome, the primary electrolytes which are affected are sodium, potassium and chloride. Hypo magnesemia is not found in Bartter syndrome; how ever some studies (Jiang, 2014), haves howed that normal levels of magnesium can still be present in some patients with GS. Hypocalciuria has previously been used to distinguish Bartter from GSinonestudy done by Berryet.al.(2013), finding so flow urinary calcium were variable, attributed to the degree of hypomorphism of mutantalleles.

GSc an present as a symptomatic or can have chronic symptom so fmuscle weakness and cramps. Vomiting and diarrhea may also be present and hypo kalemia may be attributed to gastrointestinal losses. Surreptitious vomiting can be suspected if urine chloride is less than 10 meqs/L. Patients may also experience polydipsia, polyuria and nocturia and may have salt- craving. Blood pressure may be abnormally low, however, elevated blood pressure may develop later in life as this may be a compensate for initial low blood pressure. According to Berry et. al. (2013), despite obligate salt-wasting in GS, hypertension is present in nearly half of patients, more prevalent in the older males but was not always a feature of advancing age.

Most asymptomatic patients with this syndrome remain untreated. They may undergo ambulatory monitoring annually. Diagnosis of GS depends of symptoms and

blood tests. Urine electrolyte measurement can also help in the diagnosis. Molecular genetic testing, although rarely done, confirms the diagnosis of GS.

Lifetime supplementation of magnesium and potassium is recommended. Cardiac work-up should be offered to screen for risk factors of cardiac arrhythmias from significant electrolyte imbalance. Sudden cardiac arrest have been reported.

All GS patients are encouraged to maintain a high-sodium and potassium diet. In general, the long-term prognosis of GS is excellent.

Conclusion

Mainstay of treatment for Gitelman's syndrome is high potassium diet and magnesium supplementation. The goal of treatment for GS patients is to alleviate symptoms rather than to balance the electrolytes.

References

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