Gitelman syndrome (GS) is an inherited cause of metabolic alkalosis due to salt losing tubulopathy. It is a rare autosomal recessive renal disorder resulting from mutations in the gene SLC12A3, which encodes for the thiazide-sensitive sodium chloride co-transporter in the distal convoluted tubule.

- It is classically associated with hypokalaemia, hypomagnesaemia, hypocalciuria, metabolic alkalosis, and normal or low blood pressure.
- We report a case of GS with on-going episodes of symptomatic hypokalaemia even post miscarriage, requiring multiple hospital admissions for intravenous electrolyte replacement. (1)(2)

**CASE HISTORY**

A 27 years old lady who was known to have hypokalaemia since she was 17 years of age, presented to hospital in December, 2017 with symptomatic hypokalaemia complaining of tingling and numbness around her mouth and weakness of both lower limbs. This was her first admission in hospital related to hypokalaemia and the potassium was 2.2 mmol/L and magnesium was 0.6 mmol/L, she was found to be pregnant at this admission. Prior to this admission she was just on oral potassium citrate syrup 5 ml BD.

- She received intravenous potassium and magnesium infusion and was discharged on oral supplements. Later in January 2018 she had miscarriage at 15 weeks. In December, 2018 she had another miscarriage at 11 weeks.
- Between February to July 2018 she had monthly at least once hospital admission for iv potassium and magnesium infusion. After July 2018 she required weekly intravenous potassium infusion about 80-160 mmol of KCL per admission and still continues to have admission twice a week for electrolyte replacement.

Following informed consent, DNA analysis revealed two heterozygous pathogenic SLC12A3 variants c.334G>T, p.(Glu112Ter); c.2883+1G>T, confirming a molecular genetic diagnosis of GS.

**INVESTIGATIONS**

- Potassium levels 2.8-3.0 average
- Magnesium levels 0.6 average

The priority should be to keep patient safe, improve quality of life and prevent complications. Physicians need to be aware of such challenging cases of GS which could help in novel treatment options.

**DISCUSSION**

- After pregnancy or miscarriage the symptoms of GS often improve and patients have stable levels of potassium. Our case presents an unusual example where patient had resistant severe symptomatic hypokalaemia which persisted beyond her pregnancy/miscarriage, even on maximum tolerated therapy.

- There is no literature reporting such cases, as majority of them get better post partum. We suggest that further studies and in-depth analysis are required to understand the pathophysiology of such cause.

- A randomized controlled study (3) was performed in patients with GS providing evidence of efficacy of treatment with three drugs (Indomethacin, amiloride and eplerenone). Although these drugs have different mechanisms of action, they all increase plasma potassium concentration on the short term basis. Additional studies are needed to address the long-term efficacy and tolerability of these treatment options.

- In general, GS is a chronic condition that is usually manageable. However, as in our case severity of GS may seriously hamper daily activities and effect quality of life. The main aim is to correct electrolyte abnormalities, improve quality of life and prevent life threatening complications like ventricular arrhymnias and cardiac arrest.

**CONCLUSION**

- Physicians need to be aware of such challenging cases of GS which could get worse post pregnancy or miscarriage.
- The priority should be to keep patient safe, improve quality of life and prevent complications.
- In this particular case patient wanted to continue to try to conceive, making it more challenging to manage.

**PATHOPHYSIOLOGY**

- Gitelman syndrome is an autosomal recessive pattern of inheritance.

**Common symptoms of GS**
- Weakness and fatigue
- Tingling and numbness, particularly face and hands
- Muscle cramps
- Feeling more thirsty
- Muscle twitching
- Palpitations