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Case Introduction

- 31-year-old female G1P0 at 34 weeks 5 days gestation presented with nausea/vomiting and decreased fetal movement.
- Pregnancy complicated by anxiety/depression and restless leg syndrome (controlled with iron infusions).
- Patient with normal BMI, non-smoker, no alcohol use in pregnancy, and no other drug use.

Hospital Course

- Hypokalemia on initial presentation; K of 2.8 mmol/L.
- Symptom resolution with medications and potassium repletion.
- Discharged home next day.
- Readmission 7 days later with recurrent symptoms, hypokalemia and new onset hypertension.
- Potassium repletion with both IV and PO potassium chloride.
- Nephrology consulted when hypokalemia persisted despite treatment.

Diagnosis and Outcome

- Nephrology consult suggested Geller Syndrome after the treatment team ruled out preeclampsia, congenital adrenal hyperplasia, Liddle syndrome, primary hyperaldosteronism, and syndrome of apparent mineralocorticoid excess.
- Refractory hypokalemia (Figure 1) prompted recommendation by Maternal Fetal Medicine for delivery of the infant.
- Due to breech presentation, a healthy infant was delivered via cesarean section at 36 weeks 4 days.
- Five hours post delivery, potassium levels normalized (3.9 mmol/L) and blood pressure stabilized.
- Potassium levels remained stable postpartum with oral supplementation (40 mEq daily).
- Three weeks postpartum, potassium supplementation was discontinued and repeat serum levels remained normal thereafter.
- Furthermore, the patient remained normotensive after delivery, in the postpartum period, and at subsequent clinical visits.

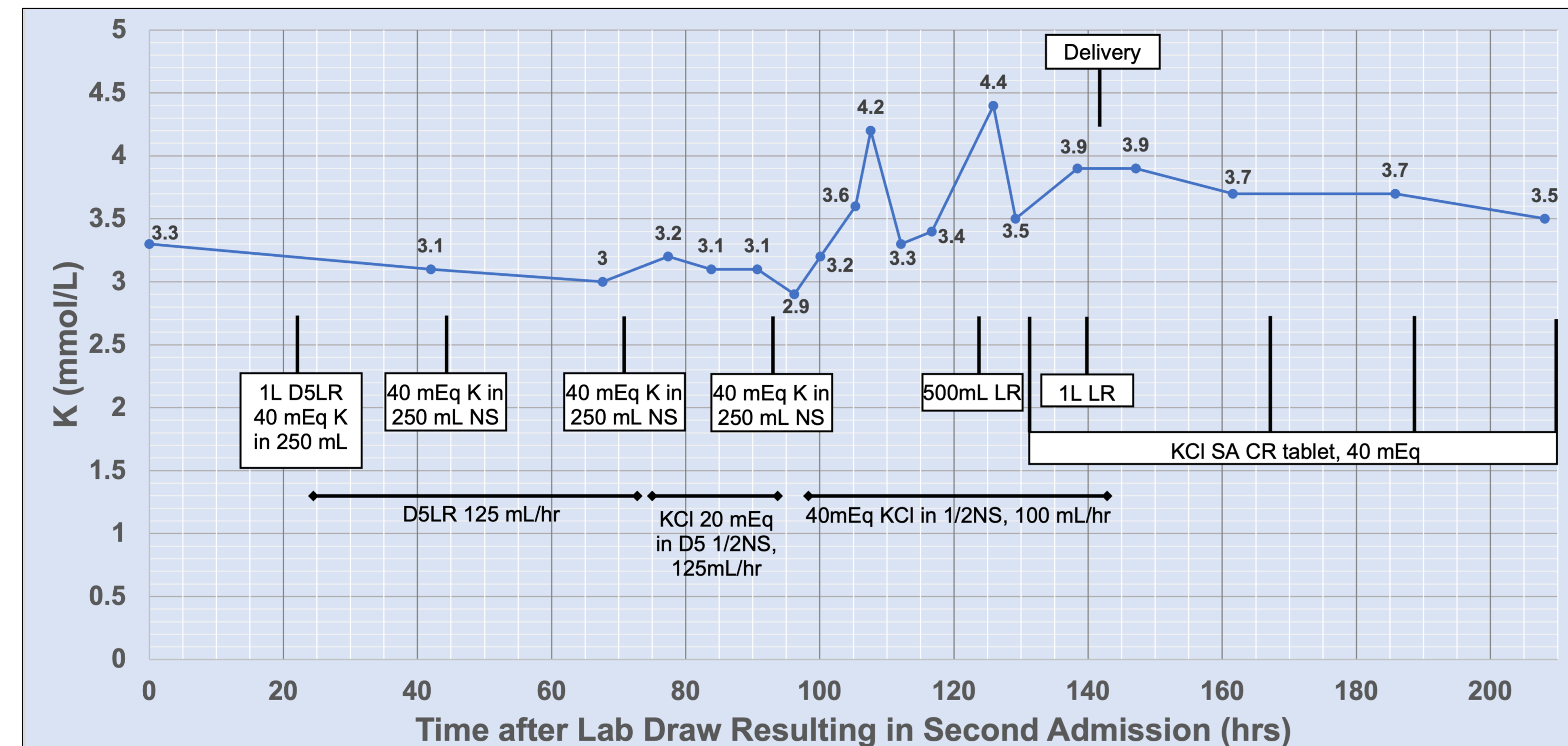


Figure 1: Potassium levels (mmol/L) measured since the time of the patient's second hospital admission. Time is measured as hours after the outpatient lab draw which resulted in this second admission, including delivery on hospital day six. Electrolyte correction measures attempted are shown. Potassium repletion was attempted through infusions, continuous maintenance fluids, and oral tablets. The first documented episode of hypokalemia (potassium of 2.8 mmol/L, treated with 2 IV potassium infusions, 40mEq each) six days prior to this admission is not included on this graph.

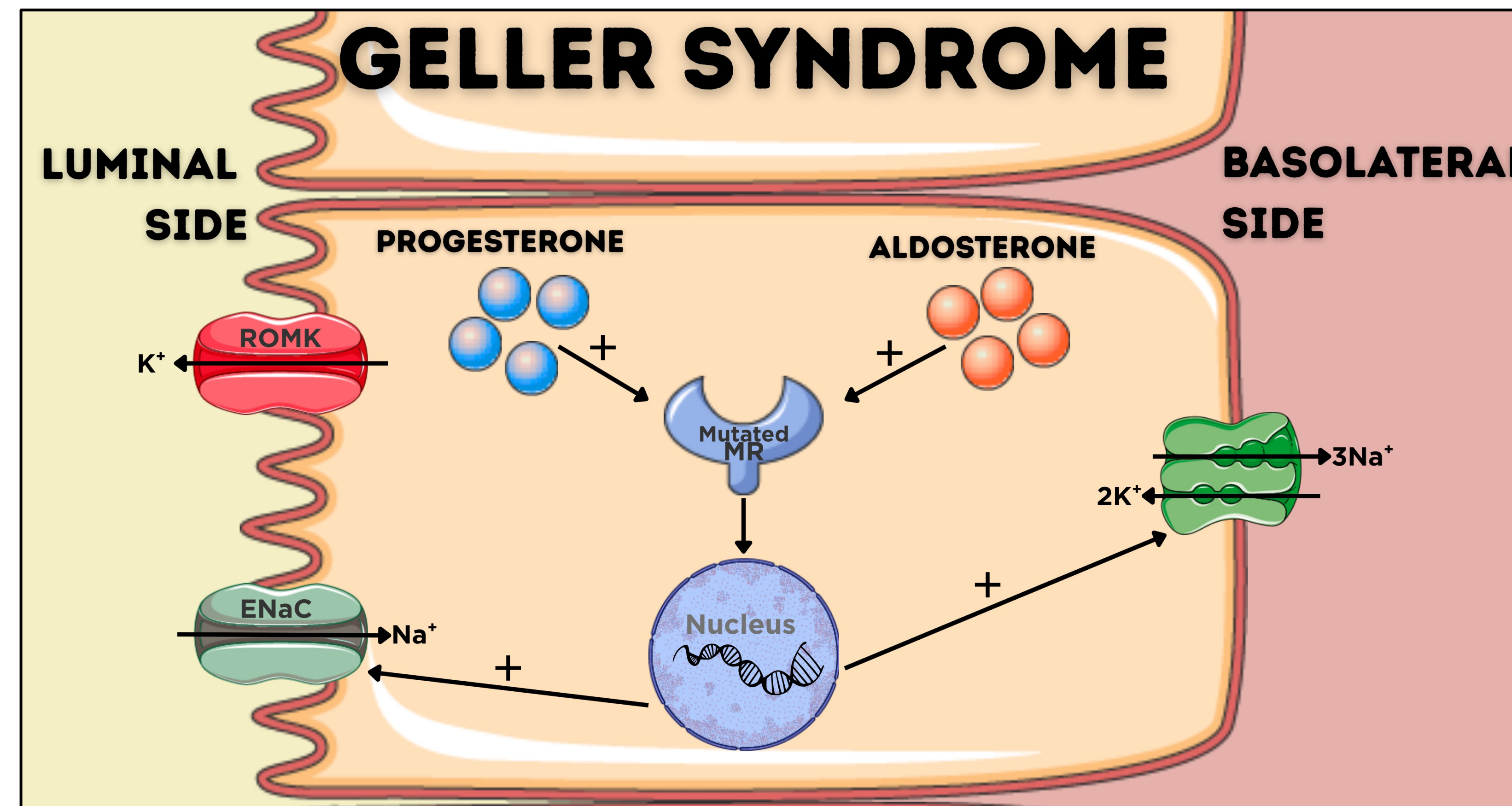


Figure 2: Geller Syndrome's effect within the renal cells, specifically, the principal cells in the collecting ducts. Normally, progesterone has an antagonistic effect on the mineralocorticoid receptor (MR). However, the mutated mineralocorticoid receptor (MR) is activated by both aldosterone and progesterone.³ Through this activation, more epithelial sodium channels (ENaC) are created and inserted into the cell membrane. Because there is more sodium (Na⁺) being reabsorbed into the cell, potassium (K⁺) is then excreted into the lumen through the renal outer medullary potassium channel (ROMK).³⁻⁴ This helps maintain cation balance within the cell. Image adapted from Servier Medical Art (<https://smart.servier.com/>), licensed under CC BY 4.0 (<https://creativecommons.org/licenses/by/4.0/>)

Discussion

- There has been a global rise in the incidence of hypertensive disorders of pregnancy.¹ Therefore, clinicians need to be aware of potential causes when evaluating patients.
- Geller syndrome is a rare autosomal-dominant condition characterized by persistent hypokalemia and hypertension (HTN) during pregnancy due to a mutation in the mineralocorticoid receptor (MR) (S810L mutation on chromosome 4q31).²⁻³
- The mutated MR is activated by progesterone (Figure 2), inducing potassium wasting and HTN without other signs/symptoms of preeclampsia.²⁻⁴
- Management is supportive, utilizing potassium repletion along with monitoring of the patient and fetus. Symptoms generally improve after delivery.⁵
- Existing case reports have found amiloride to be beneficial in maintaining potassium levels, while also lowering blood pressure.⁶

Conclusion

- Geller syndrome, though rare, should be part of a broad differential diagnosis when encountering hypertensive disorders of pregnancy. Especially, in cases of otherwise unexplained hypokalemia alongside hypertension.
- Early diagnosis helps prevent complications like arrhythmias, muscle weakness, constipation, acute kidney injury, stroke, and preterm delivery due to uncontrolled hypertension.
- Future research should focus on raising awareness, improving diagnostic methods, and exploring optimal treatments, including pharmacological interventions tailored to pregnancy.
- A deeper understanding of the genetic and molecular mechanisms underlying Geller syndrome is key to improving patient outcomes and guiding clinical practice.

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