MO053 GITELMAN'S SYNDROME AND PREGNANCY: TWO SUCCESSFUL CASE REPORTS

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BACKGROUND AND AIMS: Gitelman's Syndrome (GS) is a rare autosomal recessive hereditary salt-losing tubulopathy characterized by hypokalemic metabolic alkalosis with hypomagnesemia and hypocalciuria. Pregnancy in women with GS often aggravates hypokalemia and hypomagnesemia. However, there are few reports of pregnancies in GS. Here, we report the course of two Chinese women who were diagnosed as GS during pregnancy in 2019 and 2020 respectively.
METHOD: Case 1: A 21-year-old woman was referred to our hospital at 9 weeks gestation of her first pregnancy. She had complained of muscle weakness and cramps for one year. Before the referral she was diagnosed as hypokalemia and treated by oral potassium supplementation. However, her symptoms became severer after pregnancy. Case 2: A 20-year-old woman was admitted to the hospital because of elevated plasma glucose level and hypokalemia at 27 weeks gestation of her first pregnancy. The woman was asymptomatic and denied history of chronic diseases. The laboratory examinations were taken after admission. Genetic testing was conducted for pathogenic mutations in SLC12A3 (GS) and SLC12A1, KCNJ1, CLCKNB and BSND (Bartter syndrome 1-4).
RESULTS: Case 1: Initial biochemistry examinations revealed hypokalemia (2.3 mmol/L, normal range 3.5-5.3 mmol/L) with inappropriate renal potassium wasting (urine potassium 254 mmol/24h, normal range $<$ 20 mmol/24h), alkalosis (arterial blood gas pH 7.49), hypomagnesemia (0.55 mmol/L, normal range 0.67-1.04 mmol/L), hypocalciuria (urine calcium 1.6 mmol/24h, normal range 2.5-7.5 mmol/24h) and elevated renin (276 pg/ml, normal range 4-24 pg/ml) and aldosterone (825 pg/ml, normal range 10-160 pg/ml) levels. The blood pressure was normal-low (97/68 mmHg, 12.9/9.0 kPa) and the renal ultrasound was normal. Homozygous mutations [c.179C>T (Thr60Met)] were identified. The woman's father and sister had a heterozygous c.179C>T, but had no electrolyte disorders. After the treatment of oral potassium supplementation (KCl 3g tid) and spironolactone (40mg bid), her serum potassium level increased to 3.4-4.0 mmol/L and muscle weakness was relieved. The woman delivered a healthy female infant weighing 2600 g at 39 weeks gestation via cesarean section. Maternal serum potassium level remained normal and no symptoms reoccured after delivery. Case 2: Initial biochemistry examinations identified hypokalemia (2.3 mmol/L, normal range 3.5-5.3 mmol/L) with inappropriate renal potassium wasting (urine potassium 81 mmol/24h, normal range $<$ 20 mmol/24h), hypomagnesemia (0.49 mmol/L, normal range 0.67-1.04 mmol/L), hypocalciuria (urine calcium 0.3 mmol/24h, normal range 2.5-7.5 mmol/24h) and elevated renin (54 pg/ml, normal range 4-24 pg/ml) and aldosterone (834 pg/ml, normal range 10-160 pg/ml) levels. The blood pressure and renal ultrasound were normal. Heterozygous mutations [c.179C>T (Thr60Met), c.658G>A (Gly220Ser)] were identified. The woman was treated by oral potassium supplementation (KCl 3g tid) and her serum potassium level maintained normal during pregnancy. She had a normal delivery of a healthy female infant weighing 3050 g at 40 weeks gestation. After delivery she discontinued oral potassium supplementation and her serum potassium level ranged from 3.0-3.4 mmol/L without symptoms.
CONCLUSION: The outcome of mother and fetus of GS pregnancies appears favorable. Intensive monitoring of electrolyte levels and sufficient electrolyte supplementation are advised during pregnancy.