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Double filtration plasmapheresis for pregnancy with hyperlipidemia in glycogen storage disease type Ia: A case report

Jie Wang, Yi Zhao, Pan Chang, Bin Liu, Rong Yao

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Abstract

BACKGROUND

Glycogen storage disease type Ia (GSDIa) is an autosomal recessive inborn error of carbohydrate metabolism that is caused by deficiency of the enzyme glucose-6-phosphatase (G6Pase), leading to disturbed glycogenolysis and gluconeogenesis. Patients with GSDIa show severe fasting hypoglycemia, hyperlipidemia, hyperlactacidemia, and hyperuricemia, which are associated with fatal outcomes in pregnant women and fetuses.

CASE SUMMARY

Herein, we report the case of a 24-year-old female who on her first visit to the hospital, presented with pregnancy combined with extremely high hyperlipidemia and hyperlactic acidosis with anemia, and frequent hypoglycemia occurred during the treatment. Genetic tests revealed a mutation in the G6Pase gene (*G6PC*) at 17q21, the patient was finally diagnosed with glycogen storage disease type Ia for the first time after 22 years of inaccurate treatment. She has been treated with a continuous double filtration plasmapheresis (DFPP) strategy to remove blood lipids, and a cornstarch diet therapy. The patient did not develop pancreatitis during the course of the disease and a healthy baby girl weighing 3 kg was delivered.

CONCLUSION

Patients with GSDIa may be misdiagnosed as epilepsy. DFPP can be used to control hyperlipidemia in GSDIa patients during pregnancy.

Key Words: Glycogen storage disease type Ia; Pregnancy; Hyperlipidemia; Double filtration plasmapheresis; Case report

Core Tip: Glycogen storage disease type Ia (GSDIa) is a glucose metabolic disorder caused by the deficiency of the enzyme glucose-6-phosphatase. The gold standard for diagnosis is genetic testing, and the main treatment is a corn starch diet. Specific risks are associated with GSDIa patients during pregnancy. Herein, we described a woman who suffered from GSDIa but had been misdiagnosed with recurrent seizures for 22 years. She developed anemia and extreme hyperlipidemia during pregnancy. After genetic testing and double filtration plasmapheresis lipid-lowering treatment, the patient was accurately diagnosed and eventually gave birth to a healthy baby.

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INTRODUCTION

Glycogen storage disease type I (GSDI) is a glucose metabolic disorder caused by the deficiency of the enzyme G6Pase. The incidence of GSDI is approximately 1/100000, of which glycogen storage disease type Ia (GSDIa) accounts for about 80% of cases[1]. GSDIa patients usually experience severe hypoglycemia, hyperlactatemia, hyperlipidemia, hyperuricemia as well as growth and development retardation mainly because the last step of glycogenolysis and gluconeogenesis is affected. A rounded "doll-like" face is a typical feature of this disease due to the deposition of fat. Also in rare cases, repeated hypoglycemic seizures affect neurological development. If it remains untreated, complications from GSDIa can lead to a failure to thrive, an enlarged liver, abdominal swelling, and delayed motor development[2].

The chances of a successful pregnancy are still possible in patients with GSDIa, but specific risks are associated with GSDIa which mainly include prematurity, retarded growth, macrosomia, hypoglycemia, and fetal death[3-5]. These pregnancies need to be monitored closely because they are associated with a risk of worsening kidney problems and increased risk of bleeding, thereby requiring satisfactory metabolic control for the fetus. Among GSDIa mothers, most newborns are delivered by a cesarean section[2]. As more and more patients reach adult age, pregnancy is becoming an important problem among female patients with GSDIa.

In recent years, double filtration plasmapheresis (DFPP) has emerged as a new type of plasma exchange technology accompanied by the development of membrane separation technology[6]. DFPP is a semi-selective method where a filter separates the whole blood into cells and plasma. Then the plasma is allowed to pass through a second filter, which does not allow the passage of high-molecular-weight molecules including lipid-binding proteins. Thus using this technique, smaller proteins can be reinfused into the patient's bloodstream together with blood cells that were previously separated from the plasma.

In this report, we describe a case of a pregnant woman who presented with extreme hyperlipidemia and seizure and was diagnosed with GSDIa during treatment. She delivered a healthy baby at 37 wk of gestation.

CASE PRESENTATION

Chief complaints

A 24-year-old Chinese pregnant woman presented to the emergency department with complaints of anemia for 3 mo, gingival bleeding for 1 mo, and hematuria for 2 wk.

History of present illness

She was 24 wk pregnant and presented to the emergency department complaining of anemia for 3 mo, gingival bleeding for 1 mo, and hematuria for 2 wk.

History of past illness

Before pregnancy, she had a history of chronic seizures for 22 years that occurred about every 2-3 mo. The patient was on a long-term regimen of levetiracetam tablets (1000 mg per day) but stopped taking antiepileptic drug 1 d before admission.

Personal and family history

The patient had no family medical history of lipid disorders.

Physical examination

Although she had a normal physique (height: 1.58 m, weight: 61 kg; body mass index: 24.4 kg/m²) with no impaired intelligence, she was anxious with a characteristic round “doll-like” face.

Laboratory examinations

The biochemistry of this patient was found to be abnormal as indicated by various biochemical tests (Table 1): hemoglobin: 67 g/L, extremely elevated triglyceride: 5483.5 mg/dL (normal value < 150 mg/dL), cholesterol: 836.6 mg/dL (normal value < 200 mg/dL), elevated lactic acid: 13.3 mmol/L (normal value 0.5-1.7 mmol/L), slightly elevated uric acid: 412 μmol/L (normal value 89-360 μmol/L), albumin: 33.6 g/L, qualitative urine protein: 0.3 (1+) g/L, urinary erythrocyte++++/haptoglobin, and elevated thyroid-stimulating hormone: 8.080 mU/L (the recommended value for pregnant women is less than 2.5 mmol/L). The patient's enzymes such as amylase and lipase were in the normal range.

Imaging examinations

Ultrasound examination showed abnormal enlargement of the liver where the maximum oblique diameter of the right lobe was found to be about 17.2 cm. Two stones smaller than 0.5 cm were found in the left kidney. The patient had been undergoing regular prenatal check-ups after pregnancy and the nuchal translucency thickness, as well as Down's syndrome screening, were normal. Obstetric ultrasound examination at 24 wk of gestation revealed a live single fetus in the uterus with a thickened and sail-shaped placenta, which needed to be closely monitored during the pregnancy.

Further diagnostic work-up

After eating, the patient's blood glucose returned to normal levels (6-9 mmol/L) after two episodes of intermittent fasting hypoglycemia (minimum 2.66 mmol/L). We highly suspected that she was suffering from GSDI. To confirm this diagnosis, we treated the patient with cornstarch diet therapy and performed genetic testing using samples of her blood and fingernails. The genetic test results showed that there was a gene c.648G>T mutation on chromosome 17q21, indicating that the patient had glucose-6-phosphatase (G6Pase) deficiency.

FINAL DIAGNOSIS

Based on her medical history and examination, the patient was diagnosed with GSDIa (OMIM232200), combined with 24 wk gestation, dyslipidemia, moderate anemia, hypoproteinemia, proteinuria, hematuria, epilepsy, hypothyroidism, and left hydronephrosis with kidney stones. This was the first time she had been diagnosed with GSDIa after 22 years of inaccurate treatment.

TREATMENT

The patient was treated with a DFPP strategy intended to rapidly lower the elevated blood lipid levels (Figure 1), followed by infusion of a red blood cell suspension to correct the anemia. Furthermore, the DFPP therapy was supplemented with thyroid hormone to control hypothyroidism. Levetiracetam tablets (1000 mg per day) were also added to her prescription to control a seizure. No further seizures were observed during the subsequent hospitalization period. Following DFPP therapy for 4 d, her triglycerides were reduced to 1288 mg/dL and then she was switched to oral fenofibrate for lipid-lowering. In Table 1, the biochemical data of the patient during treatment are shown. We treated the patient with cornstarch diet therapy after she developed hypoglycemia during hospitalization.

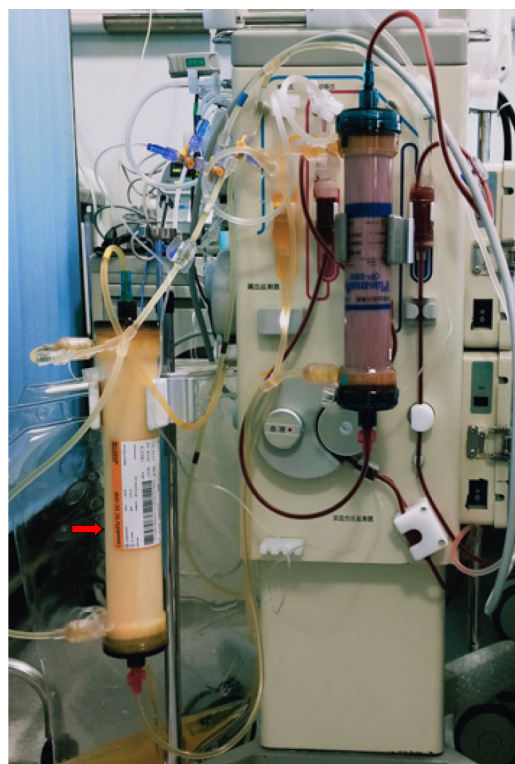
OUTCOME AND FOLLOW-UP

At 26 wk of gestation, she was discharged, cornstarch diet therapy and oral fenofibrate were continued, and her blood cholesterol levels were regularly monitored. At 37 wk + 5 d gestation, a healthy baby girl weighing 3 kg was delivered *via* cesarean section.

Table 1 Treatment/pregnancy course and biochemical data

| | Day 1 (24 wk) | Day 2 | Day 3 | Day 4 (25 wk) | Day 5 | Day 6 | Day 8 | Day 11 | Day 14 | Day 17 | Day 27 wk | Day 29 wk | Day 30 wk | Day 32 wk | Day 33 wk | Day 34 wk | Day 36 wk |
|---------------------|------------------|-------|-------|------------------|-------|-------|-------|--------|--------|--------|--------------|--------------|--------------|--------------|--------------|--------------|--------------|
| DFPP | | | | | | | | | | | | | | | | | |
| CHOL in mg/dL | 837 | 645 | 558 | 348 | 339 | 296 | 322 | 324 | 308 | 289 | 293 | 324 | 339 | 433 | 340 | 304 | 251 |
| TG in mg/dL | 5484 | 2932 | 1352 | 1256 | 1269 | 1069 | 1727 | 1297 | 1297 | 1111 | 1263 | 1432 | 1476 | 2080 | 1532 | 953 | 1138 |
| Uric acid in μmol/L | 412 | 393 | 396 | 384 | 361 | 321 | 413 | 387 | 337 | 346 | 326 | 364 | 481 | 508 | 438 | 408 | 448 |
| Creatinine in mg/dL | 30 | 22 | 21 | 22 | 23 | 22 | 29 | 32 | 27 | 32 | 23 | 22 | 24 | 25 | 27 | 28 | 28 |
| Lactate in mmol/L | 13.8 | 10.1 | 12.8 | 12.1 | 11.3 | 9.3 | 8.2 | | | | | | | | | | |
| Glucose in mmol/L | 6.11 | 6.71 | 5.90 | 5.86 | 6.0 | 4.14 | 2.66 | 2.83 | 3.69 | 3.54 | | | | | | | |
| Hemoglobin in g/L | 67 | 50 | 65 | 75 | 72 | 74 | 70 | 65 | 72 | 73 | 71 | 72 | 74 | 74 | 70 | 71 | 69 |
| TSH in μmol/L | | 8.1 | | | | | 5.3 | 3.4 | 3.6 | 2.7 | | | | | | | |
| Proteinuria | + | - | | | | | +/- | | + | | | | | | | | |

Days 1-17 were the days of hospitalization; weeks 24-37 were the weeks of pregnancy. DFPP: Double filtration plasmapheresis; CHOL: Cholesterol; TG: Triglyceride; TSH: Thyroid-stimulating hormone.



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Figure 1 Image of the patient undergoing double filtration plasmapheresis therapy. Red arrow indicates lipemic plasma sample during double filtration plasmapheresis therapy.

DISCUSSION

In this case, the patient presented with typical symptoms of GSDIa including hypoglycemia, hypertriglyceridemia, hyperlactatemia, hyperuricemia, and liver enlargement. This GSDIa patient,

however, was unusual. A large European study found that the median age at which GSDIa was diagnosed is 6 mo, with 80% diagnosed within < 1 year[7]. In this case, it is worth noting that the patient had never been diagnosed with GSDIa in her 24 years and she was largely untreated due to a lack of symptoms as well as poor compliance. Furthermore, she developed extreme hyperlipidemia (triglycerides > 1000 mg/dL) during pregnancy, with a triglyceride level of 5483.5 mg/dL that had never been reported for patients with GSDIa.

Hyperlipidemia among GSDIa patients usually results from both increased synthesis from an excess of acetyl-coenzyme A *via* malonyl-coenzyme A (the first step of fatty acid synthesis) and decreased lipid serum clearance[2]. For pregnant patients, the elevated estrogen levels enhance lipogenesis and suppress hepatic lipase activity, while elevated human placental lactogen causes insulin resistance leading to a decrease in lipoprotein lipase activity and increased lipolysis in adipose tissue[8], and these gestational changes may pose an additional pregnancy-related risk for GSDIa patients who are already predisposed to hyperlipidemia[9].

The metabolism of excessive triglycerides by pancreatic lipase leads to acute pancreatitis in about 15%-20% of women with severe hypertriglyceridemia during pregnancy[2]. During pregnancy, anti-inflammatory factors increase to counteract stresses/inflammation-like states occurring in pregnancy along with favoring maternal immune tolerance toward the fetus to result in a successful pregnancy outcome[10]. Hypertriglyceridemia-induced acute pancreatitis, however, is often accompanied by inflammation during pregnancy and if inflammation persists, it can result in abortive events, preeclampsia, and placental abruption[11]. We carried out DFPP in this patient to rapidly lower the triglyceride levels and significantly reduce the risk of adverse events for maternal and infants.

Compared with traditional plasmapheresis, DFPP requires only 10%-15% of all plasma each time, which can be replaced with 4%-5% of albumin when the plasma supply is insufficient. A study by Lu *et al*[12] found that patients with hypertriglyceridemic pancreatitis who received DFPP early had a rapid and efficient reduction in triglyceride levels compared to those without DFPP. DFPP not only effectively and accurately reduces serum triglyceride levels but also reduces allogenic plasma infusion, thereby minimizing blood transfusion-related complications, making it an optimal method of choice for this patient to achieve lipid removal rapidly.

Seizure is a relatively rare symptom among GSDIa patients. Cerebral tissue is generally found to be highly sensitive to energy metabolism, and severe hypoglycemia can lead to hypoglycemic encephalopathy. Imad *et al*[13] study found that generalized seizures might relate to a glucose level of < 2.0 mmol/L. Severe hypoglycemia that starts during infancy may have been the primary cause of her seizures instead of structural abnormalities of the brain, and her seizures masked her actual diagnosis of GSDIa. This might explain why she continues to have seizures despite taking levetiracetam to stabilize her condition.

GSDIa occurs in an autosomal recessive pattern. Both parents of an affected child are heterozygotes. In each pregnancy, there is a 25% chance of recurrence[2]. It may be possible that her infant is a genetic carrier. Identification of the human G6Pase gene (*G6PC*) mutation in the patient enables the diagnosis of potential heterozygotes in the family.

CONCLUSION

In summary, we described a case of a woman who was having GSDIa but had been misdiagnosed as a patient with a recurrent seizure only for 22 years. During her pregnancy, she developed anemia and extremely high hyperlipidemia. After DFPP lipid-lowering treatment and genetic testing, the patient was accurately diagnosed and eventually gave birth to a healthy baby.

FOOTNOTES

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