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Case Report

Management Challenges of Maple Syrup Urine Disease in Pregnancy

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Abstract

Maple Syrup Urine Disease (MSUD) is an inherited metabolic condition, with an incidence of approximately 1/185,000 infants worldwide. We discuss the case of MSUD and the management challenges that are posed by this rare condition.

A 31 year old woman presented for Maternal Medicine review in the early first trimester of her first pregnancy. She had an infant diagnosis of MSUD, which also affected two of her female siblings. She had previously attended for pre-conceptual counselling with obstetrics, metabolic medicine and anaesthesiology. Her pregnancy was complicated by hyperemesis gravidarum, posing a risk of metabolic decompensation. Dietetic input was essential in the successful management of this. Throughout pregnancy, she had frequent metabolic and antenatal visits. A male infant was delivered at xx weeks gestation owing to poor maternal weight gain and a fetus that was small for gestational age, who was not affected by MSUD.

Keywords: *Maple Syrup Urine Disease (MSUD); Pregnancy; Branched Chain Amino Acid (BCAA)*

Background

Maple Syrup Urine Disease is a genetic defect related to the lack of an enzyme needed to absorb isoleucine, valine, and leucine in the body. Due to metabolic failure, the three amino acids abnormally accumulate together with other toxic products [1]. This defect may become severe if not well treated at an earlier stage since there is always a plasma concentration increase within a few hours of delivery [3]. It is often triggered by frequent stress. Some of the common signs of this defect include irritability, fatigue, and lack of appetite, among others [6]. Failure to seek medical assistance may lead to severe effects. The defect is usually diagnosed through screening [9]. Pre-natal tests are always advisable to detect this defect hence taking precautions to prevent the inborn from severe effects.

Case Report and Discussion

A 31 year old Caucasian woman presented at 4 weeks gestation in her first pregnancy with nausea and vomiting of pregnancy. She was a patient known to Metabolic Medicine services, owing to a history of classical MSUD which was diagnosed at birth. She had a Body Mass Index of 29 kg/m^2, was a non-smoker and had normal intellectual function. She had a peripheral neuropathy in a glove-and-stocking distribution, and mild pigmentary retinopathy. She had two affected older sisters; one which died from complications of MSUD at 6 weeks of life.

She attended the maternal medicine service for pre-pregnancy counselling 6 months prior to conception. Here, the limited data on MSUD in pregnancy was discussed and possible maternal and fetal complications. Risks of metabolic decompensation and the importance of strict dietary control in pregnancy was discussed, with particular attention to periods of prolonged fasting as a result of hyper-

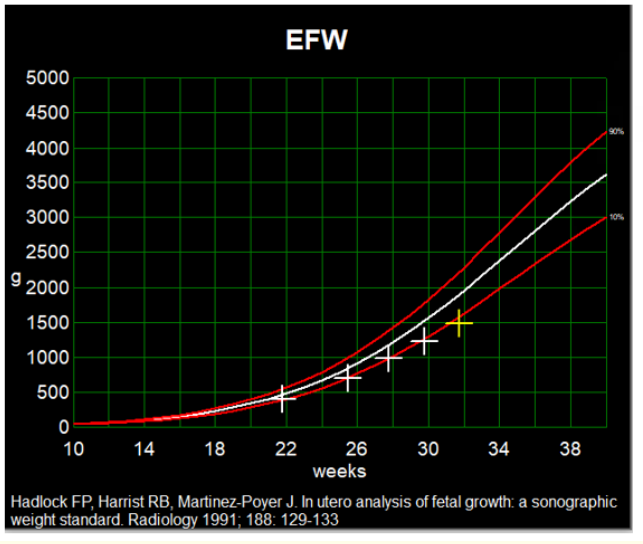
emesis, labour or delivery. In order to manage the MSUD and changes in protein requirements, she had a strict caloric daily intake, with supplementation of essential fatty acids and thiamine. These were determined by weekly branched chain amino acid (BCAA) levels. Pre-implantation genetic diagnosis was also discussed. However, in Ireland, *in vitro* fertilisation services are privately funded and are costly. The couple decided not to persue this, but were referred to a geneticist. The patient had a heterozygous mutation in the dihydrolipoamide branched chain transacylase E2 (DBT) gene, the partner was not found to be a carrier, all children will be carriers of the DBT gene mutation.

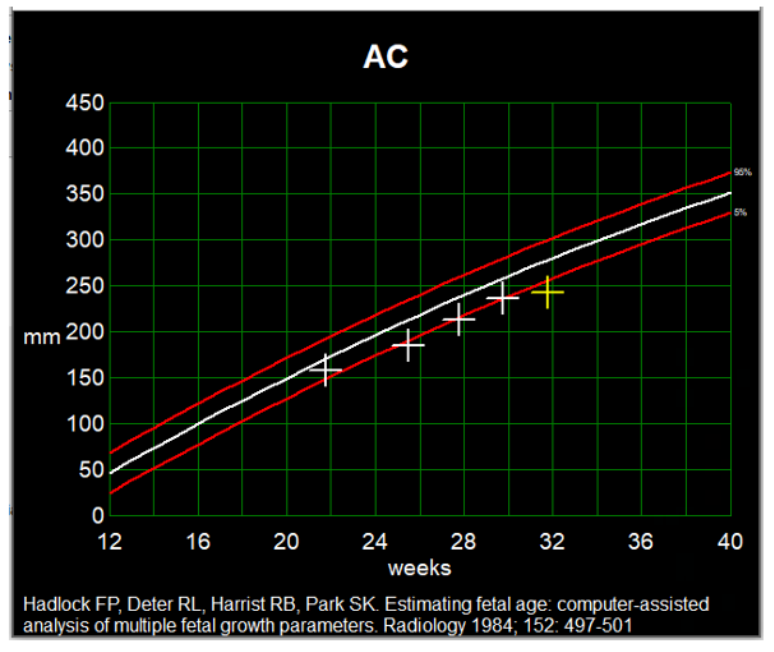
During the first trimester, from 4 weeks to 12 weeks, she suffered from nausea and vomiting of pregnancy, which was managed with oral prochlorperazine. During this period, her diet needed adjustment; caloric intake was increased to 120%, with a reduction in synthetic protein intake.

She attended for antenatal care at fortnightly intervals, in liasing with the metabolic medicine team on a weekly basis for BCAA levels. She also attended anesthesiology and dietetic services on a regular basis. During the second trimester, vascular access was obtained with the insertion of a Peripherally Inserted Central Catheter (PICC) due to poor venous access, ongoing nausea and the risk of metabolic decompensation. Parenteral iron was administered at 25 weeks gestation to treat iron-deficiency anemia (hemoglobin level of 102 g/dL).

An anatomy scan at 21 weeks gestation was reassuring. Further biometry at 25 weeks revealed an infant that was small for gestational age (estimated fetal weight 10th centile, 693g. Abdominal circumference < 5th centile). Serial fetal growth monitoring was performed and growth remained at less than the 10th centile, but never reached < 3rd centile.

She had admissions at 28, 31 and 33 weeks gestation with nausea and dehydration, where she was cared for in the High Dependency Unit (HDU). Intravenous hydration therapy and electrolyte replacement were administered, with adherence to an emergency unwell plan. By 33 weeks gestation, this lady had lost 9% of her body weight, and the decision was made to deliver her infant by Caesarean Section at 33+5 weeks gestation owing to a small for gestational age fetus and poor maternal weight gain. A male infant was delivered, weighing 1.7 kg. Newborn screening confirmed that the baby was unaffected by MSUD. The woman had an uneventful recovery, reverting to her pre-pregnancy diet post-delivery, and was discharged on day 4 post-partum.





Conclusion

In conclusion, this case has expounded more on possible ways of dealing with this disease among pregnant women. It has also given ways of preventing the unborn baby from getting the disease. Maple Syrup Urine Disease can be prevented if diagnosed at an earlier stage. The victims, especially pregnant women, can observe the doctor's prescriptions and advice while taking care of their pregnancy. By strictly adhering to the advice, the mother will prevent the unborn child from getting this related hereditary disease. Some unnecessary deaths can be prevented, especially when an individual starts experiencing some of the disease's signs. Sometimes it may be challenging to deal with pregnancy-related cases. Expectant mothers should be more careful with their health to prevent harming the unborn child. There is also a need to regularly attend clinics for counselling to be regularly examined to ensure they are healthy as they nurse their pregnancy. It is also good to prevent risks that may happen during and after delivery by being well informed of personal health.

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