Acute presentation of Multiple Acyl CoA Dehydrogenase Deficiency in third trimester with multiple organ dysfunction

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INTRODUCTION

Fatty acid oxidation disorders are a group of rare metabolic disorders commonly presenting in childhood or detected on neonatal screening. Acute presentation in adulthood has a high mortality rate and is often precipitated by a period of stress or starvation [1]. Multiple Acyl CoA Dehydrogenase Deficiency (MADD) is an autosomal recessive disorder of fatty acid metabolism caused by an enzyme defect involved in the mitochondrial respiratory chain, resulting in a disorder in fatty acid metabolism [1]. We present a case of a pregnant woman with gross metabolic disturbance requiring emergency caesarean section, and life-threatening multi organ failure.

CASE DEMOGRAPHICS

Age: **Gestation:**	34 years 32+0 weeks
Past obstetric history:	2 previous emergency LSCS – x1 for post-dates, x2 category 2 for fetal disress
Past medical history:	Fit and well
Drug history:	No known drug allergies No regular medications
Family and social history:	No significant family history, no consanguinity. No illicit substances, no alcohol, non-smoker

REFERENCES

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CASE REPORT

A 34 year-old woman (G3P2) 32 weeks gestation attended hospital with a four-week history of oedema, malaise, nausea and vomiting. Recent development of a dry cough and reduced urine output. Previous caesarean section for foetal distress, with uneventful recovery.

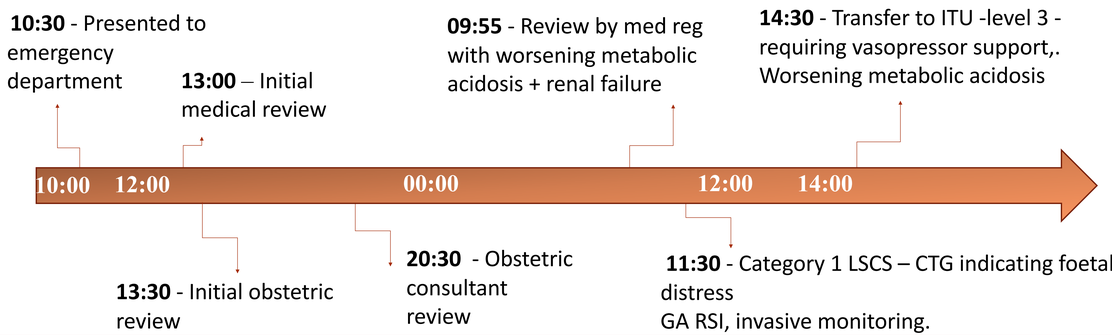
Initial tests noted a raised anion gap metabolic acidosis and serum lactate (3.1mmol/L), acute kidney injury (AKI) with raised creatine kinase (1185U/L) and hyponatraemia. Initial impression was of renal pathology combined with possible atypical pre-eclampsia.

Foetal cardiotocography demonstrated distress and decision was made for category one caesarean section under general anaesthetic. Uncomplicated intra-operative findings, but baby was acidaemic at delivery. Patient had unresolving hyperlactataemia and cardiovascular failure required transfer to the intensive care unit where rhabdomyolysis, AKI requiring haemodiaylsis and acute liver injury ensued.

Supportive treatment including intravenous dextrose and pabrinex for presumed starvation ketosis was provided but no unifying diagnosis identified. No cause for presentation noted on CT Thorax, Abdomen, pelvis, echocardiogram or urine toxicology screen. Urinary organic acids and metabolic screen were requested, suggesting MADD. Started on riboflavin supplementation.

Successfully discharged after 13 day admission to ITU. Baby had uncomplicated admission to NICU. Follow-up investigations under the metabolic team ongoing.

TIMELINE



DISCUSSION

MADD in adulthood can present as acute metabolic decompensation with liver failure, myalgia and rhabdomyolysis [1], precipitated by a period of increased stress; in this case pregnancy due to increased metabolic demand and concurrent viral illness. It is important to consider MADD in adults presenting with unexplained metabolic disturbances after periods of fasting or hyper-metabolic states. Points which would raise clinical suspicion include previous intolerance to fasting, association with intercurrent infection or family history of sudden child death. Obstetric anaesthetic considerations in patients with known MADD include: prevention of prolonged fasting times, regular blood glucose monitoring and IV glucose infusion preoperatively [2]. Riboflavin and carnitine supplementation are also essential treatments specifically for MADD [2][3].

LEARNING POINTS

- **Metabolic team should be involved in pre and post-natal care of pregnant women with known metabolic conditions**
- **Metabolic disorders can often present with rhabdomyolysis/ metabolic/keto acidosis triggered by stress – always check ketones!**
- **MDT approach to care essential – O&G, paediatrics, clinical biochemist, intensive care all involved in treatment**

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