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Rhabdomyolysis induced acute kidney injury due to long chain acyl-CoA dehydrogenase (VLCAD) deficiency

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A 40 year old female presented to Accident and Emergency with a short history of bilateral loin pain, muscle cramps, difficulty walking and reddish brown urine. This started shortly after she had been for a coastal walk. She described similar symptoms, as well as a poor exercise tolerance, intermittently over the past 20 years particularly around times of dehydration, which had been noted when she had fasted for Eid.
Her past medical history included hypothyroidism, treated with levothyroxine, and fertility issues including two first trimester miscarriages and two failed attempts at IVF. She also presented 18 years previously with an acute kidney injury and a creatine kinase of 17,000u/l though this was coded as acute renal failure of unknown aetiology. She denied taking any over the counter medications or recreational drugs. She had no family history of renal failure though her parents were second cousins.
Initial investigations showed 1+ of blood and protein on urinalysis. Her admission serum creatinine was 216umol/L and creatine kinase 22923u/l. The patient was treated with IV fluids (4 litres NaCl 0.9% and 9 litres Hartmann's in 7 days) and both creatinine and creatine kinase improved prior to discharge on day 11 (Figure 1). Creatinine continued to improve to a baseline of 68umol/L.
During admission a full renal screen was requested and she was seen by multiple specialties including renal, rheumatology and neurology; the latter of which suggested testing for serum acylcarnitines. This showed elevated C14:1 and C14:2 acylcarntines, with increased C14:1/C12:1
and C14:1/c2 ratios consistent with very long chain acyl-CoA dehydrogenase deficiency. Genetic testing showed a homozygous mutation at the ACADVL gene confirming the diagnosis.
This case illustrates a genetic predisposition to rhabdomyolysis, exacerbated by periods of fasting, in a patient with consanguine parents. The resultant homozygosity for the recessive ACADVL gene led to recurrent episodes of rhabdomyolysis and acute kidney injury, which was diagnosed at her second hospital admission with the help of multiple specialties. Treatment involved the appropriate dietary measures focused on maintaining a high carbohydrate intake and avoidance of fasting.