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Very Long-Chain Acyl-CoA Dehydrogenase Deficiency Presenting as Rhabdomyolysis

A. Ahmed, C. de Buitleir, N. Elsheik, M. Sweeney

Emergency Department, Sligo university Hospital, Sligo, Ireland.

Abstract

Presentation

A 20 year old female attended the Emergency Department by ambulance following a collapse at a concert. On arrival she was complaining of generalised muscular pain. She had not eaten for over 12 hours and had been dancing for approximately 6 hours. The patient was known to have Very-long-chain acyl-CoA dehydrogenase deficiency (VLCAD). She had a normal exam, and normal vital signs.

Diagnosis

A diagnosis of rhabdomyolysis was made after her creatinine kinase (CK) was found to be >100000 units/litre (Normal range < 170U/L). Her urine was dark brown with urinalysis positive for blood.

Treatment

The patient was admitted to the high dependency unit, where she was treated with intravenous fluids. Her urine output and renal function were closely monitored. She made a full recovery and was discharged home four days later.

Conclusion

(VLCAD) is an inherited, autosomal recessive, metabolic disorder caused by mutations in the ACADVL gene.

Management includes treatment of manifestation, primary prevention of manifestation, and prevention of secondary complications.

Introduction

Rhabdomyolysis is a syndrome characterized by muscle necrosis and the release of intracellular muscle constituents into the circulation. Creatine kinase levels are typically elevated.

There are multiple potential causes of rhabdomyolysis. These can be broadly divided into three categories: traumatic or muscle compression (e.g., crush syndrome), non-traumatic non-exertional (e.g., drugs or toxins, infections), and non-traumatic exertional (e.g., metabolic myopathies which account for less than 10% of presentations).[1]

The metabolic myopathies represent a very small percentage of cases of rhabdomyolysis overall but are relatively common causes among patients with recurrent episodes of rhabdomyolysis after exertion. Very-Long-Chain Acyl-CoA Dehydrogenase (VLCAD) Deficiency, a rare autosomal recessive condition usually diagnosed on newborn screening that affects the oxidation of fatty acids is one such cause.[2]

We present the case of a 20 year old female, known to have VLCAD, who presented with rhabdomyolysis after a period of prolonged fasting and exertion.

We describe the presentation, management, and subsequent clinical course. We also discuss the pathophysiology and the pattern of disease manifestation.

Case report

A 20 year old female attended the Emergency Department by ambulance following a collapse at a concert. On arrival she was complaining of generalized muscular pain, weakness, and an unquenchable thirst. She also reported that her urine was a very dark colour. She had not eaten for over 12 hours and had been dancing for approximately 6 hours. The patient stated that she had a history of VLCAD deficiency.

On examination, she was fully conscious and oriented. The vital signs were as follows: heart rate per minute 99, blood pressure 90/50 mmhg, respiratory rate 22, and temperature 36.5 Celsius. Her power was reduced globally to 3/5.

Investigations: White cell count 18.26 with neutrophilia (04.00-11.00 10 9/l), aspartate aminotransferase 3650(0-32 U/l), alanine aminotransferase 1107 (0-33 U/l), with normal ALP, GGT, bilirubin, total protein, albumin, and globulin. Lactate was 2.2 (< 1.5mmol/L). Creatinine Kinase (CK) was >100000 (< 170U/L). Both renal function and blood glucose were normal. Urine was dark brown with urinalysis positive for blood.

A diagnosis of rhabdomyolysis was made. Intravenous fluids were commenced to maintain a urine output of 2ml/kg/hr, and she was admitted to a high dependency ward for four days. She made a full recovery and her CK and liver function tests normalised prior to discharge.

Discussion

VLCAD deficiency is an inherited, autosomal recessive, metabolic disorder caused by mutations in the *ACADVL* gene. Incidence varies from one in 40,000 to one in 120,000 worldwide.[3]

VLCAD comprises three subtypes: Severe, hepatic/hypoketotic hypoglycaemic and later onset episodic myopathic. Severe typically presents in early childhood with a high associated mortality. Hepatic typically presents in early childhood, with later onset often presenting during adulthood, with potentially only muscle related symptoms.[4] VLCAD should be suspected in such cases of intermittent myopathy/rhabdomyolysis associated with exercise intolerance.[5]

Diagnosis is established with biochemical testing and/or identification of biallelic pathogenic variants in ACADVL on molecular genetic testing. Specialized biochemical testing using cultured fibroblasts or lymphocytes may be needed for confirmation of the diagnosis.[6] Management of VLCAD includes treatment of manifestations, prevention of primary manifestations, and treatment of secondary complications.

Adult onset VLCAD deficiency is a rare cause of rhabdomyolysis. The keystone of diagnosis is the recognition of the typical clinical course: recurrent fasting, exercise, or infection induced rhabdomyolysis. Adjustments in lifestyle and diet are crucial in the prevention of recurrent episodes of metabolic decompensation.[7]

Declaration of Conflicts of Interest:
There are no conflicts of interest to declare.

Corresponding Author:
Ahmed Ahmed,
Sligo University Hospital,
Ireland.
E-Mail: ahmedmarahmed@rcsi.com

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