

# Recurrent Rhabdomyolysis due to Peroxisomal Alpha–Methylacyl-CoA Racemase (AMACR) Deficiency Associated with Antidepressants



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## Introduction

- Alpha-methylacyl-CoA racemase (AMACR), an enzyme involved in fatty acid metabolism deficiency is a rare disorder. This rare disorder presents with variable phenotype ranging from cholestasis to late onset sensory motor neuropathy.
- There has been little to no data collected on rare cases of recurrent bouts of rhabdomyolysis that led to the discovery of AMACR deficiency.
- Understanding patient history, physical findings, laboratory values, and treatment of the disease is beneficial to further developing treatment and preventative care strategies to reduce morbidity and mortality.

## Aim

- Describe all aspects of case report on patient with AMACR deficiency and cross-reference data with other studies to highlight trends.

## Case History/Description

- A 33-year-old female with a history of depression presented with complaints of acute diffuse proximal muscle aching.
- She recently started the antidepressant sertraline.
- Similar episodes occurred in the past associated with the use of fluoxetine and bupropion, however that was resolved after discontinuation of these medications.
- Her laboratory results showed a significantly elevated creatinine kinase level of 12,769 unit/liter, transaminitis, and elevated inflammatory markers.
- Due to progressive muscle pain and weakness an electromyography was performed, which was consistent with chronic proximal myopathy.
- Panel of myositis-specific autoantibodies, including anti-Jo1 antibody were negative.
- Subsequently, a muscle biopsy displayed necrotizing myopathy consistent with rhabdomyolysis.
- Genetic testing showed elevations of pristanic acid and bile acid studies diagnostic of alpha-methylacyl-CoA racemase deficiency.

## Discussion

- This is a novel case of recurrent rhabdomyolysis due to AMACR deficiency in the literature triggered by antidepressants.
- This case also highlights the consideration of extended work up to include pristanic acid and/or bile acid intermediate concentrations to screen for this rare metabolic disorder in patients with unexplained relapsing symptoms of myopathy.
- We have a multi-disciplinary approach including genetic, nephrologist, neurologist, rheumatologist with special focus on dietary measures to offer a potential treatment for the disorder.

## Conclusion

- Although this is a rare case, special interest should be given to medications and finding other triggers of symptoms in patients with AMACR deficiency.
- Future studies should analyze larger populations of patients with AMACR deficiency and compile a list of associated triggers of symptoms such as rhabdomyolysis.

## References

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