A Peculiar Case Of Limb-Girdle Muscular Dystrophy Masquerading As Elevated Transaminases

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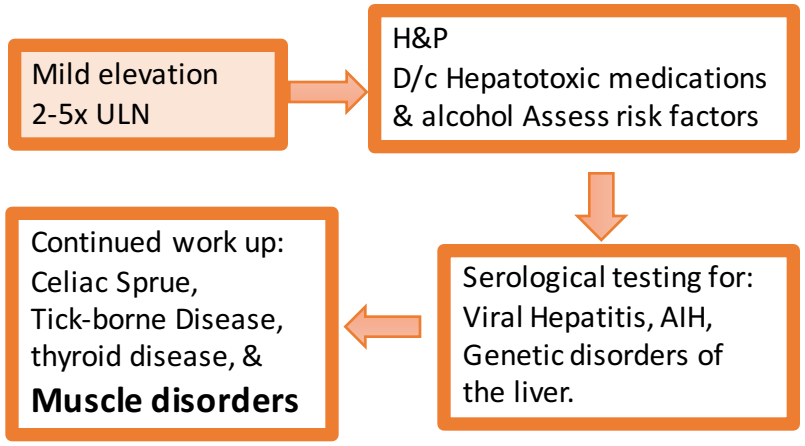
Learning Objectives

- A disproportional elevation in aspartate transaminase (AST) and alanine transaminase (ALT) is often considered a marker of hepatocellular injury.
- The differential for chronically elevated liver chemistries should include hepatic and nonhepatic etiologies such as skeletal muscle damage, but the workup should follow an algorithmic approach.
- Elevation of liver chemistries could be the herald finding in a patient with limb-girdle muscular dystrophy (LGMD).

Case Presentation

- 19-year-old adopted male presented to gastroenterology clinic for asymptomatic elevation in transaminases. AST was 133 IU/L and ALT was 223 IU/L. Total bilirubin and alkaline phosphatase were normal.
- He denied alcohol use, prescribed medications, herbal supplements, or weightlifting supplements. BMI was normal.
- Workup for viral hepatitis, hemochromatosis, autoimmune hepatitis, Wilson's disease, alpha-1 antitrypsin, celiac disease, and thyroid disease were normal.
- Liver biopsy revealed reticulin collapse with complete regeneration without inflammation.



Adapted from: Kwo et al. AJG 2017

Follow Up

- Subsequent follow-up over years demonstrated persistently elevated but stable elevations.
- At the age of 24, he began to endorse proximal lower extremity muscle pain and weakness.
- Creatine kinase (CK) was elevated at 11,937 U/L; aldolase was elevated at 31.4 U/L; myositis workup was normal.
- Right thigh biopsy revealed a myopathic process.
- Genetic testing confirmed mutation in the CAPN3 gene consistent with calpainopathy, a subtype of LGMD.

Discussion

- LGMD is a rare, genetic disorder resulting in loss of ambulation within 20 years after disease onset.
- Concurrent liver disease is rare in LGMD but elevations in transaminases can be found because of severe elevations in CK.
- Our patient was found to have calpainopathy, a subtype associated with a recessive mutation in the CAPN3 gene.

Conclusion

- Our case demonstrates that algorithmic evaluation of abnormal liver chemistries should not inconclusive evaluation of abnormal liver chemistries should include a CK level to further investigate nonhepatic causes, specifically a myopathic process.
- Although there is no cure for LGMD, interventions focused on muscle conditioning are more beneficial with early detection to mitigate the devastating effects of this disorder.

References

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