

An unusual cause of deranged liver function tests: From paracetamol to Pompe's

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Case report

We would like to draw to your attention an unusual cause of deranged liver function tests. A 42-year-old woman was admitted following drainage of a dental abscess under general anesthesia. This was uncomplicated but she had accidentally taken a staggered overdose of 10 g paracetamol for pain. She denied any symptoms and examination was unremarkable.

Blood tests revealed an aspartate transaminase (AST) of 72 iu/dL and a paracetamol level of 0.06 mmol/L but were otherwise normal. Twenty four hours later there was no change in the liver function tests and the INR remained 1.1. Hepatitis screen including ferritin, ceruloplasmin, autoantibodies, antimitochondrial antibodies, Hepatitis A, B and C serology and abdominal ultrasound were normal.

She was followed in the hepatology clinic. The AST remained high but there was no change in the other liver function tests. Two years later serum AST isoenzymes were requested. However, these were not routinely performed in the laboratory and the consultant in clinical biochemistry performed creatine kinase (CK; 523 iu/dL) and alanine transaminase (ALT; 44iu/dL) instead.

This suggested that the serum AST originated from muscle rather than the liver. A history of mild generalized weakness was elicited, and examination revealed slightly reduced power (4±5) and absent reflexes in all four limbs.

Muscle biopsy revealed vacuolation of several myocytes. Periodic acid schiff staining demonstrated glycogen stored within the vacuolated myocytes. The diagnosis of acid maltase deficiency (AMD) was confirmed on enzyme assay.

Discussion and conclusion

AMD (glycogen storage disease type 2) or Pompe's disease is an autosomal recessive disorder with an estimated incidence of approximately 1:40,000 [1,2]. There is a deficiency of the enzyme α 1,4 glucosidase which breaks down glycogen in cell lysosomes. Glycogen accumulates within lysosomes and disrupts cellular function. This affects skeletal muscle including the diaphragm. The presentation is usually insidious with gradually increasing peripheral muscle weakness or respiratory failure. Prognosis is highly variable and depends on the gene mutation and enzyme activity. As no cure is available, supportive therapies such as noninvasive ventilation and genetic screening of family members are the cornerstones of management. However, enzyme replacement therapies have been developed and the results of initial studies are promising [3,4].

References

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