Hemochromatosis - Unusual Presentation

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Introduction

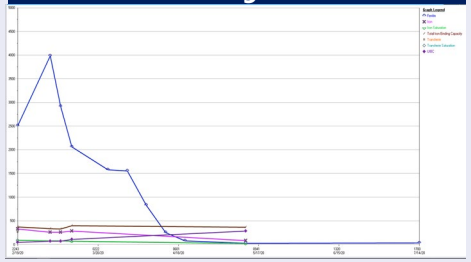
Hereditary hemochromatosis is a common autosomal recessive iron storage disease. The classic clinical triad of liver cirrhosis, hyperpigmentation, and diabetes is nowadays rare, most likely due to early recognition[3]. Usually, the homozygous C282Y mutation in the HFE gene is responsible for most cases of hereditary hemochromatosis .[1]

Case Report

We present a unique case of a 31-year-old healthy female with no significant past medical history. She presented to the Emergency Department with complaints of abnormally dark-yellow discoloration of her urine followed by yellow discoloration of her eyes, headache, nausea and vomiting.
She also had abnormally high liver enzymes with AST (Aspartate aminotransferase) and ALT (Alanine aminotransferase) as high as the 2000's with an associated conjugated hyperbilirubinemia.
Family history was negative for liver disease or autoimmune disorders.
She denied any intravenous drug use, alcohol use, excessive non-steroidal anti-inflammatory drugs use or recreational medications. All laboratory workup for infections as well as autoimmune etiology were negative.
Imaging was also unimpressive with ultrasound of abdomen and magnetic resonance cholangiopancreatography only showing gallbladder wall thickening.
A liver biopsy was performed and it demonstrated subtotal hepatic necrosis with a small collection of iron in the liver.
PCR (polymerase chain reaction) of blood identified a single mutation C282Y.
She is being treated with phlebotomy and her liver enzymes levels improved significantly.

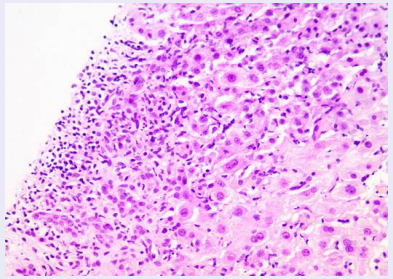
Dates	2/19	3/18	4/20	5/12	7/14
AST (U/L)	1,605	1,007	697	156	101
ALT (U/L)	1,865	742	461	112	53
Alk-Phos (U/L)	301	262	275	261	245
Bilirubin (mg/dL)	5.3	3.7	3.2	1.4	1.2
Ferritin (ng/mL)	2,916	1,579	78	25	32

Trend of Liver Enzymes and Iron Studies During-Treatment

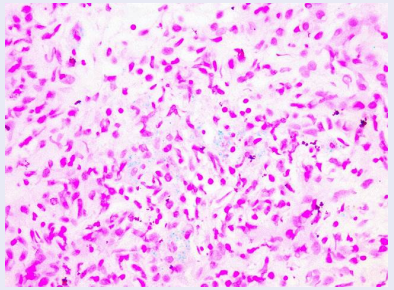


Date Range

Liver Biopsy Results



Individual cell apoptosis appreciated in lobules.
No granulomas or fibrosis appreciated.



Positive Iron Stain.

Conclusion / Discussion

To our knowledge, this is the first case of its kind of hemochromatosis in which the patient presented with symptomatic hepatic failure without any positive family history in the setting of a single C282Y gene mutation[2].
Consulted teams including Gastroenterology and Oncology physicians were hesitant to accept this level of iron overload as secondary to hemochromatosis being caused by just the single gene involvement [5].
Obstructive causes were the initial suspected etiology with the most common causes being ruled out via imaging and laboratory results.
We conclude that individuals who may be heterozygous for C282Y gene who have a coexisting insult to the liver, associated with the use of medications (specifically in this patient case, she used negligible amount of ibuprofen for pain after sustaining a fall while ice skating), may still present with overt manifestations of iron overload.[4]

References

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