Celtic thunder! Ironing out the most common genetic disease in Caucasians

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Hemochromatosis is the most common genetic disease in Caucasians and is due to iron overload in persons who are homozygous for the HFE gene mutation C282Y. It is more common in those of Celtic ancestry. This poster will highlight the strategies of recognizing this worldwide disorder, which can be clearly viewed as an important clinical applied toxicology threat. Symptoms of hereditary hemochromatosis are nonspecific and typically absent in those earliest phases of this disorder. Heightened awareness is therefore the key to diagnosis. Often the only clue is finding of minimally elevated level of liver function tests, with no other obvious causes being found. The disorder is sometimes found when a family member is diagnosed with hemochromatosis, as first degree relatives of patients with classical ZHFE related hemochromatosis should be screened as well as those with abnormal iron studies. Occasionally those afflicted may present only fatigue or abdominal pain as their complaints. Physical exam findings can be protean, to include a bronze discoloration to the skin in those in the late stages of this disorder. Most critical to the diagnosis of hemochromatosis is a heightened level of suspicions and awareness of how prevalent this condition is. How the clinician may suspect this disorder, as well as confirmatory laboratory studies will be reviewed. Hemochromatosis will be shown to be a minimizable worldwide clinical toxicological threat.

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