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UVM Health HEMATOLOGY ELEVATED FERRITIN Referrals/Consult

Background:

- Ferritin is an acute phase reactant and is elevated for a variety of reasons.
- Cirrhosis can cause minor iron loading in the liver which is not treated with phlebotomy.
- Hemochromatosis refers to pathologic iron loading in organs, an elevated ferritin may or may not represent iron loading and a risk for end-organ damage.
- Acquired hemochromatosis is caused by blood transfusions or iatrogenic iron administration. Inherited iron loading is caused by genetic mutations.
- There are two relatively common mutations associated with hereditary hemochromatosis in the Caucasian population in the HFE gene; C282Y and H63D.
    - Most people who have the mutations for hereditary hemochromatosis do not develop iron loading. Even individuals homozygous for C282Y only have a 10% chance of iron loading in their lifetimes. Risk factors for hemochromatosis include an increased iron saturation and a 1st degree family member with hemochromatosis.
- The H63D mutation is common in Caucasian populations (15%) and homozygotes almost never have iron loading, but ferritin levels stable ~600 for their lifetime.

Purpose:

- To identify patients who may have hemochromatosis to expedite the referral process.
- To allow a treatment plan to be put in place at the first Hematology visit.

Laboratory studies (within the prior 3 months):

- Ferritin elevated at two timepoints at least 1 month apart
- Blood work in every patient: Complete Blood Count with differential, comprehensive metabolic panel, hemoglobin A1c, **FASTING** serum iron, serum iron binding capacity, transferrin saturation
- Work up elevated liver function testing as appropriate (hepatitis serologies, alcohol and medication history, etc).
- Transferrin saturation relies on a serum iron concentration which reflects iron ingestion in the past few hours.
- HFE gene mutation testing (C282Y and H63D) – may need prior authorization.

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If elevated liver function tests and/or ferritin >1000 (exam must be scheduled but does not need to be done prior to hematology visit):

- MR Abdomen without contrast. Indicate in comments section of order: Elastography AND Ferriscan. This test is done at UVMMC only.
- If the elastography demonstrates fibrosis or cirrhosis refer concurrently to hepatology.

Please call Hematology consult attending through PAS (802 847-2700) to expedite a referral if there is concern, such as unexplained moderate to severe anemia, hypercalcemia, or renal failure, or patient is systemically ill with unexplained illness.