

Date: January 19, 2016
To: All Zones
From: Genetic Laboratory Services (GLS)
Re: Changes to HFE-related testing for Hereditary Hemochromatosis

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Key Messages:

- As of February 15, 2016, GLS will no longer offer routine testing for the HFE-related hereditary hemochromatosis (HFE-related HH) p.S65C variant in the HFE gene. Molecular testing for the p.C282Y and p.H63D variants will continue to be performed.

What is HFE- related hereditary hemochromatosis (HFE-related HH)?

- HFE-related HH is an autosomal recessive, adult onset genetic disorder characterized by iron accumulation in organs, primarily the liver, which can potentially result in life-threatening organ impairment. Initial symptoms are often non-specific.
- HFE-related HH is a relatively common disease in Caucasians but is rare in non-Caucasian populations.
- HFE-related HH molecular testing should be considered for individuals with increased transferrin saturation in two or more independent screens.
- Early treatment for HFE-related HH is effective and simple: regular phlebotomy to normalize biochemical parameters

Why this is important:

- p.S65C is a normal variant in the European population and has also been described as a susceptibility allele in patients with iron overload, particularly in association with heavy alcohol consumption, but there is no evidence to support a causative role in HH.
- Current clinical practice guidelines recommend neither testing nor reporting the presence of p.S65C in the diagnosis of HFE-related HH.
- Patients being investigated for HFE-related HH in Alberta will only be tested for the p.C282Y and p.H63D variants.

Action Required:

- Please refer to the Toward Optimized Practice website for information on how to screen patients for HFE-related HH and when and how to order genetic testing
- MDL requisitions and information sheets are available at <http://www.albertahealthservices.ca/lab/page8667.aspx>

Inquiries and feedback may be directed to:

GLS North Genetic Counsellors Phone: 780-407-1015
GLS South Genetic Counsellors Phone: 403-955-3097

This bulletin has been reviewed and approved by:

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