"HEREDITY HEMOCHROMATOSIS"

THE HIDDEN DANGER OF ELEVATED IRON LEVELS

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Most physicians diagnose only a few cases of hereditary hemochromatosis in their practice because they do not routinely test for iron overload.

Primary (inherited) and secondary (acquired) conditions of iron overload both have similar consequences, which make it essential to determine the cause of iron overload for a complete diagnosis and commencement of therapy. Iron overload is confirmed with a combination of clinical and laboratory findings often prompted by abnormal routine blood work or the patient's complaint of symptoms.

Though the symptoms of iron overload are not specific, chronic fatigue is among the first complaints of patients with iron overload, whether the cause is primary or secondary. Besides chronic fatigue, individuals with iron overload due to hereditary Type 1 hemochromatosis are often seen with arthralgia and impotence initially with liver disease, diabetes and heart trouble later on. By comparison, patients with thalassemia major display heart trouble and hypogonadism initially, and liver disease, and diabetes thereafter.

Symptoms, findings, or diseases associated with iron overload include arthritis, diabetes, mellitus (especially type II), heart trouble or arrhythmia, liver disease, and mildly evaluated liver enzymes – especially ALT. Also associated with iron overload are amenorrhea, anterior pituitary failure, impotence and loss of libido, inappropriate increase in skin pigmentation, depression, hypothyroidism, infertility, viral hepatitis, liver cancer, NASH, and porphyria cutanea tarda(PCT).

Clinical Features of Patients with Hemochromatosis

Symptoms

Asymptomatic

- **Abnormal serum iron studies on routine screening chemistry panel**
- **Evaluation of abnormal liver tests**
- **Identified by family screening**

Identified by population screening

Non-specific, systemic symptoms

Weakness, fatigue, lethargy, apathy, weight loss

Specific Organ-related Symptoms

Abdominal pain secondary to hepatomegaly
Arthralglas
Diabetes
Amenorrhea
Loss of Libido, Impotence
Congestive heart failure, arrhythmias

Signs

Asymptomatic

Hepatomegaly

Symptomatic

Liver
Hepatomegaly
Cutaneous stigmata of chronic liver disease
Splenomegaly
Portal hypertension (ascites, encephalopathy)

Joints

Arthritis
Joint swelling

Heart

Dilated cardiomyopathy
Congestive heart failure

Skin

Increased pigmentation

Endocrine

Testicular atrophy
Hypogonadism
Hypothyroidism

In conditions where iron overload and anemia occur simultaneously, as in thalassemis major, or for individuals who are transfusion dependent, life expectancy is generally shortened. Iron-induced cardiac disease remains the main

cause of death in patients with thalassemia major, despite conventional chelation therapy.

Most patients with classic type I hereditary hemochromatosis that are diagnosed prior to severe organ disease such as diabetes mellitus or cirrhosis will have a normal life expectancy.

Elevated serum iron can be an indicator that iron overload is present, but serum iron alone is not a reliable way to determine excessive tissue iron. Serum iron is influenced by many factors, such as time of day, and for this reason must be done while fasting for accuracy. Fasting does not affect serum ferritin, but ferritin is an acute-phase reactant and can be elevated if inflammation is present. Inflammation can occur as a result of taking certain medications, such as hormone replacement therapy, and in the presence of chronic disease and infection.

Both the fasting transferrin iron saturation percentage and serum ferritin will be elevated in persons with classic hemochromatosis (type 1) if tissue iron is excessive. However, in about 30-50- percent of patients with acute viral hepatitis and alcoholic liver disease, both serum ferritin and transferrin iron saturation percentage can be elevated.

Individuals with non alcoholic fatty liver disease (NAFLD), also called non-alcoholic steatohepatitis (NASH) can have an elevated serum ferritin with a normal or only slightly elevated transferrin iron saturation percentage. Along with iron overload, NASH patients will exhibit hyperinsulenemia induced by insulin resistance, two factors significantly associated with NAFLD in obese patients.

High serum ferritin can occur in the absence of elevated iron or of inflammation. For example, patients who have hyperferritinemia-cataract syndrome are not iron loaded and should not be phlebotomized. Hyperferritinemia-cataract syndrome is an inherited condition of early onset cataracts. Though the serum ferritin is elevated, it is not due to tissue iron overload but some defect resulting in overproduction of ferritin. Many times these patients are inappropriately bled, with unpleasant results. Patients are usually young, male, have elevated serum ferritin, but normal serum iron and complain of eye pain when subjected to bright lights. An ophthalmologist can confirm diagnosis.

A physical examination to ascertain hepatomegaly or splenomegaly is important, albeit somewhat subjective. If either organ is palpable a liver biopsy may be indicated, especially if liver enzymes are abnormal.

Dr. Badanek's office looks at sickness and disease at a totally different perspective. We address the cause of all conditions, test for them, and treat the cause not just the symptoms. It is a totally new paradigm shift of conscientiousness for the new patient. To input knowledge to the patient, which is most lacking today in our health care delivery system, is empowering the patient to be successful with their health care challenges presented.

Please schedule an appointment to consult with your health challenges. We offer a courtesy consultation for your first visit to meet Dr. Badanek. Dr. Badanek has been in private clinical practice for 36 years working in the field of Integrative/Functional Medicine.

Dr. Badanek's website: Dr.Badanek.com will give you an idea of what his facility has to offer the sick and health challenged.

To schedule an appointment, please call 352-622-1151.