

IRON OVERLOAD DISEASE -- HEMOCHROMATOSIS

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Dedicated to John, my brother, who was misdiagnosed for decades and then diagnosed too late to recover from multiple organ damage. He lives with the ravages of this easily controlled disease. Listen up, and take charge of your health.

This article was first posted two years ago in April. However, I have been reminded of and "realarmed" at the extent of ignorance regarding an often misdiagnosed, easily preventable but debilitating and even deadly disease — for the lack of two blood tests.

LOOSE IRON — A PLAYGROUND FOR DISEASE

Many of you are aware of the recent report that it was a case of undiagnosed hemochromatosis (iron overload disease) that made it possible for genetically weakened and otherwise safe laboratory *plague bacterium* to thrive and cause the sudden death in Sept. 2009 of Dr. Malcolm Casadaban, a genetics and cell biology professor at the University of Chicago, who, after working with the bacterium, became ill, checked into hospital and died within hours. Lab results were positive for the plague, and the university's "biosafety fire alarm" was triggered.

Dr. Casadaban's liver and blood were awash and swimming with a free heavy metal — unbound iron. Early diagnosis could have been made with an iron panel when routine blood tests were being done, something every man over 40 and every woman after menopause should request. How is it that a scientist with degrees from M.I.T., Harvard, and Stanford in Biology, Genetics, and Cell Biology did not know to be tested for this genetic disease? He may have never taken it seriously as a threat to himself. Certainly the family care physicians don't bother with it. Why is it not a routine screening test, especially for those of the right age with a northern European heritage?

Iron overload disease weighs heavy with the need for as many as possible to know. Most of you at CrossFit are young; certainly from my elderly perspective that includes all of you! The men among us won't need to be concerned with this inherited mutated gene until they are 40 and the females until after menopause, although in some, earlier than that. This is important enough, common enough, and devastating enough for you to

understand it now for yourself and your loved ones. Iron overload is a destructive disease – a sleeper that slowly compromises internal organs and joints, seducing one into thinking "it's just age catching up" — until years later it has taken over, trashing hopes, dreams and goals; family life, love-life and love of life, and finally – life itself. All for lack of two simple diagnostic blood tests! How incredibly unacceptable!

CrossFit can build physical strength, help to keep us focused and healthy, strengthen bones, and for some it has been shown to restore and maintain blood pressure and cholesterol levels within normal range, but no number of life-time workouts can prevent the progress of iron overload disease. This article would not even be necessary if there were no treatment – no way to control it — there would be no point.

So here we go – all you never thought you wanted to know about hemochromatosis.

THE NORMAL

Iron in our diet is essential to health. Most of us absorb about 10 percent of the iron in the foods we eat. When our iron stores are adequate, the body protects us from iron overload by reducing the amount of iron absorbed by the intestines. However, once absorbed, the body cannot rid itself of excess iron except through bleeding or pregnancy.

IRON OVERLOAD

An inherited genetic mutation, called *Iron Overload Disease*, or *Hemochromatosis*, causes loss of the body's rigorous control over intestinal absorption of unneeded iron, allowing it to continue to enter the blood stream from the intestines unabated. Over the years, 5 to 20 times as much iron as normal may be stored, causing damage to major organs such as the liver, heart, pituitary gland, thyroid gland, pancreas, joints, and the retina (macular degeneration). In the retina, excess iron can cause retinal toxicity through the generation of oxygen free radicals. Iron overload in the brain is seen in people with Alzheimer's disease, early onset Parkinson's disease, epilepsy, multiple sclerosis, and Huntington's disease.

PREVALENCE

Hemochromatosis is the most common genetic disorder in the western world. In the United States, Europe, Australia, New Zealand and other western countries there is approximately 1 case in 300 persons, mostly of northern European origin (British, Irish, Dutch, German, French.)

AGE AT FIRST MANIFESTATION

The first sign of hemochromatosis may go unnoticed for decades because it requires a blood test showing high iron blood levels that show up in men over 40 and in women after menopause. Later, when physical symptoms of organ or joint damage begin, even then, you will be lucky indeed to have a physician who orders an iron panel to screen for

hemochromatosis. File this for later reference and never be afraid to bring up the possibility of iron overload; insist on the test.

My brother, to whom this article is dedicated, was diagnosed by accident. He had an uncontrollable nosebleed that sent him to the ER. The physician stopped the nosebleed and sent blood to the lab asking for an iron panel because of concern for the blood loss. That was his first test ever for iron levels. They sent him home with a supply of iron tablets to take every day. The next day they called and told him to stop taking the iron. His serum iron levels were off the charts. He had been accumulating iron with no means of off-loading for years. It was too late to repair the damage to his body. After decades of concatenating symptoms and seeing many physicians, any one of whom could have ordered a lab test for iron levels, he was finally diagnosed by accident. A scenario that is very difficult to live with. When caught early and treated effectively, damaged organs can heal, especially the liver, which is amazingly regenerative.

EARLY SUBJECTIVE SIGNS AND SYMPTOMS

This disease is difficult to recognize by its physical signs and symptoms because of the number of organs and functions involved. Early signs and symptoms include:

- Joint pain (44%), including hand, wrist, knees, feet, back and neck
- Chronic Fatigue (74%)
- Impotence (45%)
- Lack of normal menstruation
- Abdominal pain
- High blood sugar levels
- Low thyroid function (hypothyroidism)

BLOOD TESTS

Hemochromatosis is diagnosed at any stage with two fasting blood tests. Testing is crucial, as iron can build up to damaging levels in your body before symptoms appear.

- **Transferrin Saturation %.** This test is the ratio of serum iron (SI) to total iron binding capacity (TIBC) multiplied by 100, a more useful indicator of iron status than just iron or TIBC alone. It measures the amount of iron bound to a protein (transferrin) that carries iron in your blood. Transferrin saturation values greater than 45 % are considered too high.
- **Serum ferritin.** This test measures the amount of iron stored in your body, (whereas serum iron measures the level of iron in your blood.) The Iron Disorders Institute lists the normal range of Ferritin for adult males as up to 300 ng/mL and for women up to 200 ng/mL.
- **Note.** It is likely that you will not be told to fast for the blood draw. Remember to do so. Additionally, for one week prior to the blood test, you should not be taking iron supplements or vitamin C, which enhances intestinal absorption of iron.

TREATMENT

This is a manageable condition. Treatment involves off-loading the iron by giving blood. If your iron panel shows elevated Ferritin and Transferrin Saturation %, your physician will hopefully start a regimen of treatment. However, my experience has been that many physicians are uninformed about this genetic condition. So, for your own information and personal guidance, there are treatment and maintenance protocols on www.irondisorders.org or www.ironoverload.org.

DNA TEST

DNA (deoxyribonucleic acid) codes genetic information for identification and for the transmission of diseases. If there is a family history of hemochromatosis, get a DNA test even if you are too young to have symptoms. If you have inherited this gene mutation, you can then be on the alert for an increase in iron levels above normal and start treatment before it becomes a burden. A significant number of people with the genetic mutation for hemochromatosis do not have elevated blood iron levels. They are not symptomatic, but they are carriers, therefore their children and grandchildren should be alerted. For the details of the genetics of inheritance, www.irondisorders.org has excellent charts.

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