

O·PIN·ION

Living with the Celtic curse

Mary Sue Daniels

My dear mother's most loving and steadfast heart stopped beating at the age of 53. She was inexorably brought to this untimely and unseemly end by a liver that suddenly betrayed her. Her torso swelled, her limbs shrunk and her skin turned yellow, while her sad, astonished sunken eyes reflected the bewilderment and the guilt that lay beneath. Adding to this burden of physical misery was shame. Her doctor had told her that the cirrhosis scarring her liver was the result of alcoholism — that she was drinking herself to death. So after several weeks in a hospital she was admitted to a rehab facility (this was prior to Betty Ford speaking about her struggle with addiction; at that time treatment was not fashionable nor talked about openly) and she returned home optimistic and determined to halt the process. Though she plausibly read the Twelve Steps and Twelve Traditions during long lonely nights of fitful sleep her health continued to deteriorate, and it was only three short years before her body gave in to the inevitable. I was her only child — 15 years old, angry, confused and utterly bereft.

Thirty-two years later her youngest sister, the woman who filled a large part of the enormous void left by my mother's absence, died in much the same manner. My aunt rarely, if ever, drank alcohol, but she too found herself in middle-age with rapidly falling health. When her liver enzymes tested off the chart, her doctor repeatedly told her to "come back after not drinking for a while and those numbers will come down." After her daughter, a medical tech, read her test results she realized that something was terribly wrong, and after additional blood tests and a second opinion a diagnosis was reached — hemochromatosis, a disorder where the body absorbs too much iron and that excess iron becomes toxic. My aunt began to think that perhaps my mother also had suffered from this condition and suggested that I monitor my health. But when I spoke to my doctor she disagreed, saying that yes it was indeed a genetic condition, but that it was passed only through males. I asked her to please keep an eye on my liver function anyway, and it was never mentioned again.

By the time my aunt was diagnosed, her liver was more than half cirrhotic, but the good news seemed to be that with diet and regular phlebotomies the iron counts could be lowered and additional organ damage could be kept minimal. And following treatment, her health was restored for a time. However the damage to her liver and pancreas was significant enough that several years later she was unable to fully rebound from the trauma, medications and reconstructive surgeries that followed an unfortunate accident. She developed diabetes and in the end succumbed to liver cancer.

Watching my aunt during her final months was a vivid reminder of my mother's death, and I became convinced that their health issues were too similar not to be related. I then learned of a genetic blood test that identifies the gene mutation indicating a predisposition for the disease. After I

took the blood test, it was confirmed that I, too, have the mutation and early signs of iron overload.

Often overlooked, especially when diagnosing women, hereditary hemochromatosis is a genetic condition that causes the digestive system to absorb too much iron. This excess iron is stored in vital organs and can lead to life-threatening conditions such as cancer, heart problems, liver disease and lung damage. Women with this disease usually don't start accumulating excess iron until after menopause, which has led to the misconception that it is primarily a male disease. Women with this condition simply get sick at a later age than their male counterparts. As I am on the cusp of menopause, with this early diagnosis and a good maintenance program, I probably will never develop organ damage. And luckily for me, I stopped smoking and drinking years ago. These nasty habits are particularly deadly for someone with this condition because in addition to their advertised health risks, they also actually increase the body's ability to absorb iron. It also is lucky that I have been a regular blood donor. Giving blood is not only a great way to help people and contribute to the community — it also has the added benefit of keeping some of that excess iron from building up in your system.

This all started me thinking about my maternal grandfather. He died at the age of 67 after years of being an insulin-dependent diabetic and suffering several debilitating heart attacks. He had only one sibling, a sister who also developed diabetes. Could she have been iron loading? What about my mother's other two sisters? Could they have had a connection to this disease too? An alarming sequence of middle-aged bad health had decimated my close-knit family and caused irreparable emotional damage to the survivors, especially the children. I started to see a pattern in what had once seemed to be a random coincidence of bad luck and heartbreak.

HFE (for High Iron Fe) the gene for classic type 1 hemochromatosis was discovered in 1996, and a genetic test became commercially available in 1997. There are two major mutations that are attributable to iron loading; the one most common among people of Northern European descent is the C282Y mutation. Also known colloquially as "The Celtic Curse," it is estimated that this mutation occurred over 45,000 years ago in Ireland and was actually beneficial to people living with an iron-deficient diet. It then spread with the Irish Diaspora, first to Scandinavia with the Vikings and then to the British



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Isles, America and Australia. My mother's family was Irish and my father's family of Irish and Scottish descent. I am classified as having a homozygous C282Y mutation, which means I have two copies of the mutated gene, inherited from each parent. My aunt's children are heterozygous, which means they have one copy from their mother and thus they are carriers of the gene.

It seems clear to me now that my grandfather had this disease and I am starting a campaign to educate and persuade my reluctant relatives into getting tested. We may never know the full story of our family's medical history, but we now have the information to end needless suffering from too much iron in the future.

July is Hemochromatosis Awareness Month and with the rich Irish heritage in Anaconda it seems like the perfect time to start educating our community. The statistics show that 1 in 8 people of Northern European ancestry have this mutation. Around the world, millions of lives are permanently damaged or prematurely ended by untreated hereditary hemochromatosis, and yet most people know nothing about the disease. According to <http://CelticCurse.org> there are three important things to consider:

- Testing for hereditary hemochromatosis is relatively inexpensive.
- Treatment to prevent the adverse effects of hereditary hemochromatosis is relatively inexpensive, if started early enough.
- The medical, personal and social costs of treating the damage caused by untreated hereditary hemochromatosis? Huge, crippling, ruinous.

Although the disease is present at birth, symptoms usually don't appear until later in life: between the ages of 30-50 for men and after 50 for women. Some of the early symptoms include joint pain, fatigue, abdominal pain and impotence. (Ernest Hemingway had hemochromatosis, and it is now widely believed to have been a factor in his suicide.) If you have a family member with this disease or suspect that you may have some of the symptoms, I urge you to speak to your doctor and inquire about the genetic test, which is the only reliable way of diagnosing this condition.

Here are a few web sites, which have been helpful to me:

- Iron Disorders Institute: <http://www.irondisorders.org/>
- hemochromatosis
- American Hemochromatosis Society: <http://americanhs.org/celtic.htm>
 - The Hemochromatosis Information Center: <http://www.hemochromatosis.org/montana/>
 - Iron Overload Diseases Association: <http://www.ironoverload.org/>

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