BETR BOARD MEMBER PUBLISHES IN MEDICAL HYPOTHESES

By Donna Beales, MLIS

My paper *Biome Depletion in Conjunction with Evolutionary Mismatches Could Play a Role in The Etiology of Neurofibromatosis 1* was published in the journal Medical Hypotheses on January 24, 2015. The date is fitting; I celebrated my 34th wedding anniversary on the same day.

There are two types of neurofibromatosis (NF), NF1 and NF2. My spouse and members of his family suffer from NF1. It can be painful and disfiguring. It's considered a genetic disorder, passed through families 50% of the time, causing tumors to grow anywhere nerves are present in the body. The movie *The Elephant Man* brought NF to public consciousness several years ago.

Not in my wildest imaginings did I ever think I would have anything to do with NF research. I'm a bit like Emily Dickinson*, "I'm nobody, who are you? Tell me, are you nobody too?"* I'm not connected with a major university; I don't work in a research lab; I'm a Medical Librarian by profession, which gives me some rudimentary research background along with a little undergrad background in human experimentation, but I'm no scientist.

I have, however, been exploring biotherapeutics since 1998, especially helminthic therapy, or the use of live organism intestinal worms to control autoimmune diseases. My interest first started with a news interview with Joel Weinstock, M.D. and a group of researchers who were having people drink a "worm cocktail" to treat their inflammatory bowel diseases. *Eeeeeww!!,* I thought. *But really, really cool!* The idea behind their research seemed to make sense to me. Worms dial down immune response in order to trick the body into thinking they aren't there, a little like Obiwan Kenobi in *Star Wars* (*these aren't the droids you're looking for...).* The immune system seems to need to be challenged by worms and infections in order to learn what to do. We got rid of worms starting in the late 1800's, along with other common but serious infections, and by doing so, we may have inadvertently caused the immune system to malfunction, creating the current epidemic of autoimmune and allergic diseases.

My interest grew into a passion, and in some circles I even began to be known as the "Worm Grrl." What I learned over time kept cropping up in what I continued to learn about NF. There were tantalizing pieces of information that seemed as if they might fit together, about mast cells, and inflammation, and the way NF tumors form, but nothing unifying.

In early 2011, when I was first thinking about all of this, everyone studying helminthic therapy was looking at it from a "drug model" perspective— you get an autoimmune disease, and you treat the disease by introducing helminths into the body, like a drug. At the time, so was I, and consequently, I was asking the question, *could worms help people with NF?* So I kept wiggling the puzzle pieces around, but really couldn't seem to make them fit together.

Then in 2012, I became aware of the work of the Parker Research Lab at Duke University. They were writing works on "biome depletion," or the removal of key organisms from the inner human ecology (biome), and how it might be a serious problem. William Parker, PhD, and his colleagues completely

blew out the walls of my thinking. Helminths, they seemed to be saying, are so integral to the immune system that almost everyone needs to be colonized with them. Parker and his colleagues were hot on the trail of evidence that helminths could possibly even prevent autism. Their writings included eerily prescient projections of a future time in which people might go to their doctors not only for their regular vaccinations, but also for doses of helminths to ward off diseases of inflammation. Jaw-dropping, heady stuff for a "worm grrl!"

By this time, I had nearly forgotten about the question of NF, that is, until I went with my husband to an appointment at the *Neurofibromatosis Clinic* at Massachusetts General Hospital, where we saw Scott Plotkin, M.D. Dr. Plotkin is patient and extremely knowledgeable. As he examined my husband, he kept up a running dialogue with us about NF and what is known about the disease. In passing, he mentioned a fact I had known for more than 30 years, but had somehow forgotten. NF, he said, arises without any family history whatsoever in almost 40% of cases.

In a flash of insight that almost caused me to faint in the middle of his office, I suddenly understood that all the time I had been looking at the problem *I had been asking the wrong question.* The question wasn't, *could worms help people with NF?* The real question was, *could worms prevent NF from ever happening at all?*

The puzzle pieces began to assemble. I wrote a first draft and submitted it, but the paper was rejected. All during the summer of 2014, I continued to write and revise, and at times, the puzzle pieces seemed to almost be moving together by themselves. The experience of writing was like swimming deep under cool, silent water, refreshing and invigorating. Eventually, the work was accepted.

I believe, and research is beginning to corroborate, that the human body is an ecosystem, and we need our resident fauna in order to be healthy human beings. Can I say I'm right about my theory? Truthfully, no I can't. What I hope to gain from publishing the work is to open an initial dialogue about another way of looking at genetic disorders, as not arising spontaneously out of nowhere, but instead arising because our bodies appear to be full of inflammation as a result of lack of organisms that "train" our immune system.

I'm really asking the ultimate scary question, *can biome depletion even affect our genes?* Only time, and research, will tell.