

Ataxian Spring 2004
Dr Julie Greenfield reports

Ataxia with Vitamin E deficiency - a treatable form of ataxia

If you were diagnosed with Friedreich's ataxia many years ago and have not had a genetic test to confirm the diagnosis it may be worth asking your doctor to check your vitamin E levels. This is because you may have been misdiagnosed. You may have Ataxia with Vitamin E Deficiency, a rare form of ataxia that is treatable with vitamin E.

People with this form of ataxia usually display symptoms similar to those of Friedreich's ataxia, but their ataxia is linked to low levels of vitamin E and it can be treated with vitamin E supplements. Studies have shown that a number of patients' neurological symptoms have stopped progressing when they were treated with daily vitamin E supplements; some even saw some improvement. Early diagnosis and treatment is therefore very important.

The first case of Ataxia with Vitamin E deficiency was described in 1981 and it was originally thought to be very rare. However, from the early 1990s onwards, more patients have been identified with this condition. In a review published in 2000 one ataxia researcher suggested that Ataxia with Vitamin E Deficiency is not as rare as originally thought. He stressed the importance of diagnosing this treatable type of ataxia and of giving treatment promptly. As it is similar to Friedreich's ataxia, some cases have been reported where people have been misdiagnosed. One Ataxia UK member, Dianne, describes her experience:

'Many years ago (I am now 46 years of age) my parents were told I had Friedreich's Ataxia. I always refuted that diagnosis, and was trying to find out what my real problem was. Last year I went for CT scans, a spinal tap and numerous blood counts in Aberdeen. Only then did my doctor diagnose a deficiency in vitamin E. I, of course, have been 'gobsmacked' at such a simple sounding problem.'

Ataxia with Vitamin E deficiency is a type of ataxia that is inherited recessively, meaning that parents of the person with the condition could be carriers of the condition without being affected with ataxia themselves. The ataxia is caused by changes in a gene. There are a number of different changes that all lead to the same condition; some gene changes lead to more severe forms of the condition than others. The age of onset ranges from two to 52 years old. Some people have much more severe symptoms than others, but everyone has a deficiency in vitamin E - an antioxidant that protects cells from damage by free radicals. Because they are not sufficiently protected, they are damaged by the free radicals.

It is now recognised that everyone displaying symptoms suggesting Friedreich's ataxia should have their Vitamin E levels tested. If you have a genetically confirmed diagnosis of ataxia (such as Friedreich's ataxia), or you know you have a dominantly inherited ataxia, testing is not relevant. (For more information on how ataxia is inherited go to www.ataxia.org.uk or contact me at the Office.) However, if you have not had a genetic test that confirms the type of ataxia you have and you have no family history that suggests that it may be inherited dominantly, you may wish to ask your neurologist to check the level of vitamin E in your blood, to avoid the situation that Dianne and others have found themselves in.