

Tips for Getting a Proper Diagnosis of an Autoimmune Disease

Do your own family medical history.

Take an inventory of your family's health problems, expanding your research beyond your immediate family to cousins, aunts, uncles, and other relatives. Since current research points to a genetic component in most autoimmune diseases, you should know the health histories of your first-degree relatives, including grandparents and cousins, if possible. In a family with a history of autoimmune disease, an individual has a higher risk of developing autoimmune diseases in general, rather than a specific autoimmune disease. Once you know your family history, you can communicate it effectively to your doctor who can then assess the possibilities with a degree of accuracy and order appropriate tests.

Keep a symptoms list.

People with autoimmune diseases often suffer from a number of symptoms that, on the surface, seem unrelated. In addition, they may have suffered from other seemingly unrelated symptoms throughout their lives. It's important, therefore, to make a list of every major symptom you've experienced so that you can present it clearly to your doctor. List and date the symptoms in the order of concern to you. Don't leave out any symptoms that you think are insignificant. Keeping a daily journal of symptoms can help to pinpoint what they are, how long they have been bothersome, and the time of day they usually occur.

When the nurse calls you into the exam room, review your list—or, better yet (in most cases), give the list to the nurse to record in your chart. When you see your doctor, at the very start of your visit, be sure to mention the symptom that bothers you the most. This is often the problem to which your doctor will pay the most attention. Unless your problem list is lengthy, or the doctor notes a serious problem which takes higher priority, she/he usually will be able to answer all your questions. *Know in advance what questions are truly important to you. In this way, you will not spend a lot of time discussing things that confuse the issue of your current needs.*

Seek referrals to good physicians.

Talk to your family and friends. If you're having trouble getting a proper diagnosis, see whether someone you know and trust can recommend an internist in your community who is also a good diagnostician. It's always good to ask around. Check your community resources—attend health agency meetings and community health meetings sponsored by local hospitals, and talk with the health care professionals at those meetings and elsewhere. Because there is no medical specialty called "autoimmunology," it can be difficult to determine the type of doctor you may need to see. One thought is to identify the medical specialist that deals with your major symptom and then check with a major medical center for a referral to that specialty department. A number of agencies dealing specifically with autoimmune diseases maintain referral lists.

Inquire about the experience of both the hospital and the physician in treating autoimmune disease.

All patients want to receive the highest quality treatment, but it is difficult to evaluate physicians and hospitals to whom they entrust their care. It's a good idea to ask the physician whether he or she treats patients with the specific disease that has been diagnosed. Generally speaking, the larger number of patients with a particular autoimmune disease treated by the physician, the better. Also, a specialist should be adept at managing the therapies used to treat a particular autoimmune disease.

Obtain a thorough clinical examination.

Tests vary for different autoimmune diseases, and a diagnosis is usually reached through careful analysis of laboratory test results combined with a patient examination and history. When facing test situations, a patient might ask: What is the purpose of this test? Are there any alternatives? Is this an outpatient or inpatient procedure? Can I anticipate any pain, discomfort, or claustrophobia; and if so, can I take medication to make me more comfortable? How much does the procedure cost, and is it covered by my health insurance? Can I get a copy of the test results? What will they tell me about my condition? (You will need a copy of lab test results to give to other doctors if you are going to seek a second opinion.) The patient needs to understand that, although diagnostic criteria define a disease, they are sometimes uncertain.

Get a second, third, and fourth opinion, if necessary.

Sort out your options for treatment at the beginning before symptoms worsen, but check first to see whether your insurance will pay for a consultation. Since autoimmunity has just begun to be recognized as the underlying cause of some 100 known autoimmune diseases and because symptoms can be vague and not visibly apparent, many doctors don't think to test for autoimmune diseases initially. If a doctor doesn't take your symptoms seriously, dismisses them as stress-related (when you do not feel as though you are under any excess stress), or refers you to a psychologist, find another doctor. You know you're not feeling well--don't be intimidated. When trying to get a correct diagnosis, it's important to be assertive.

Partner with your physicians to manage your disease.

Once you have settled on your treatment plan, keep in mind that your health is best managed through a partnership—you and your medical team. If more than one medical specialist is treating you, select one of those physicians to be your "main" provider to manage your medications. This physician needs to agree to take the lead role. Establish a dialogue, a give-and-take on a mutual respect basis. Don't be afraid to ask questions:

- ◆ What are the treatment options?
- ◆ What are the advantages and disadvantages of each?
- ◆ How long will the treatment last?

Learn to deal early on with the long-term effects of autoimmune disease.

The complicated process of obtaining a diagnosis and developing an appropriate plan of treatment may mean that you will be subjected to a great deal of uncertainty. Accept that patients with autoimmune disease and their families very likely will need to adapt to a somewhat different lifestyle.

Sharing your situation with others can have enormous benefit, including eliciting the kind of emotional support that is so necessary for people with undiagnosed autoimmune diseases. Some people are using the Internet as a way to communicate their symptoms with others. Doing so can help advance your own personal research, may provide clues to what disease you are experiencing, and can be a means of uncovering good resources for finding a good diagnostician. Because the Web can also be a source of misinformation, be sure to evaluate what you read. Be especially cautious when using online chat rooms because they can be full of misleading and inaccurate medical information.

If you don't have your own computer, many libraries provide access to the Internet through their computers. Ask your librarian whether this is available to you, and don't hesitate to ask for help if getting "on-line" isn't clear to you.

Also, contact your hospital community education department, a nurse at the health department, or a nonprofit disease-specific organization to find out about support groups.

Getting a diagnosis can be a challenging journey.

For people with autoimmune diseases, getting a proper diagnosis can be one of the most difficult challenges they face. AARDA conducted a survey of autoimmune disease patients and found that the majority of those eventually diagnosed with serious autoimmune diseases had significant problems in getting a correct diagnosis. Many were incorrectly diagnosed with a variety of conditions that have no specific blood test to confirm the diagnosis. Many were told that their symptoms were "in their heads" or that they were under too much stress. Furthermore, the survey revealed that 45 percent of autoimmune disease patients had been labeled as chronic complainers or were told that they were overly concerned with their health in the earliest stages of their illnesses. On average, autoimmune patients see four different doctors over a four-year period before a diagnosis is made.

Since many autoimmune disease have confusing and unrelated symptoms, AARDA urges patients to follow the preceding steps to arrive quickly at an accurate diagnosis.

Do you know your family AQ?

The following article is an AARDA press release that was designed to spread autoimmune awareness, especially during May, which was "Autoimmune Diseases Awareness Month." This information formed the basis for a PSA featuring Kellie Martin, AARDA's spokesperson, which has been picked up by TV stations across the country. Watch for it!



The American Autoimmune Related Diseases Association (AARDA) wants to help you learn your family's AQ. AQ is a play on IQ and stands for **Autoimmune Quotient**. It's about knowing how likely you or a loved one is to develop an autoimmune disease, based on the prevalence of these diseases and your family history.

AARDA offers the following advice to help you determine your family's AQ.

Understand that autoimmune diseases constitute a major U.S. health crisis.

According to the National Institutes of Health (NIH), there are *23.5 million Americans* who suffer from autoimmune diseases and the *prevalence of these diseases is rising*. In comparison, cancer affects up to 9 million and heart disease up to 22 million. Collectively, autoimmune disease is *one of the top 10 leading causes of death* in children and women under 65 and represents some *\$100 billion in annual direct health care costs*. Yet, *fewer than 6 percent of Americans* surveyed in a recent AARDA/Roper poll could identify an autoimmune disease.

Get educated.

There are more than *80 known autoimmune diseases* and an *additional 40 diseases that are suspected to be autoimmune-related*. The diseases themselves can affect almost any part of the body, including the kidneys, skin, heart, liver, lymph nodes, thyroid and the central nervous system. As a result, they cut across various medical specialties, such as endocrinology, neurology, dermatology, rheumatology, gastroenterology and hematology, among others. Autoimmune diseases include multiple sclerosis, myasthenia gravis, scleroderma, polymyositis, vasculitis, lupus, Sjögren's disease, idiopathic thrombocytopenic purpura (ITP), type 1 or juvenile diabetes, Crohn's disease and Graves' disease.

Autoimmunity is the underlying cause of these diseases. It is the process whereby the immune system mistakenly recognizes the body's own proteins as foreign invaders and begins producing antibodies that attack healthy cells and tissues, causing a variety of diseases.

Visit www.aarda.org for more detailed information and a complete disease list.

Be aware that autoimmune diseases target women.

Women are more likely than men to be affected; some estimates say that 75 percent of those affected are women. These women are usually in the childbearing years. In the past several years, autoimmunity has begun to be recognized as a major women's health issue, with the Office of Research on Women's Health at NIH recognizing it as such and the Society for Advancement of Women's Health Research naming it as one of 10 diseases that most disproportionately affect women.

Know that autoimmune diseases run in families.

Current research points to a *genetic component in autoimmune diseases*. However, autoimmune diseases are not typical genetic diseases like, for instance, sickle cell anemia, where there is a specific gene mutation. With autoimmune diseases, multiple genes are involved that collectively increase vulnerability or susceptibility. Thus, what is inherited is not one specific gene but several genes that increase risk. As a result, autoimmune diseases tend to "cluster" in families - not as one particular disease, but as a general tendency to the autoimmune process and, consequently, different autoimmune diseases. For example, one family member may have autoimmune hepatitis; another, celiac disease; another, rheumatoid arthritis.

Do your own family medical history.

Given the family connection, knowing the health histories of other family members is critical. For example, if your grandmother or father or sister or uncle has an autoimmune disease, you could be more susceptible to developing one yourself. Take an inventory of your family health problems, expanding your research beyond your immediate family to include grandparents, aunts, uncles, cousins and other relatives. Once you know your family history, share it with other family members and your doctor who can then assess the possibilities with a degree of accuracy and order appropriate tests.

Keep a "symptoms" list.

People with autoimmune diseases often suffer from a number of symptoms that, on the surface, seem unrelated. In addition, they may have suffered from other seemingly unrelated symptoms throughout their lives. It is important, therefore, to make a list of every major symptom you've experienced so that you can present it clearly to your doctor. List the symptoms in the order of concern to you.

Realize that getting an autoimmune disease diagnosis is often challenging.

An AARDA study of autoimmune patients found that the average time for diagnosis of a serious autoimmune disease is 4.6 years. During that period, the patient typically has seen 4.8 doctors (Kellie Martin's sister had seen seven doctors before being diagnosed); and 46 percent of the patients were told initially that they were too concerned about their health or that they were chronic complainers.

One of the factors that makes getting a correct autoimmune disease diagnosis so difficult is that symptoms can vary widely, notably from one disease to another, but even within the same disease. Also, because autoimmune diseases affect multiple systems, their symptoms can often be misleading.

The medical community's lack of knowledge of autoimmune disease compounds the problem. Even though these diseases share a genetic background and tend to run in families, most health questionnaires at doctors' offices do not ask whether there is a family history of autoimmune disease.

Hold the power to protect your family's future health and well-being in your hands.

Congratulations! By working through these steps and doing your homework, you now have the knowledge to determine whether you or your loved ones could be at risk for developing an autoimmune disease.

American Autoimmune Related Disease Association
22100 Gratiot Avenue, Eastpointe, MI 48021
www.aarda.org