Ask an expert: What is sarcoidosis?

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Q: My sister was just diagnosed with sarcoidosis in her lungs. I've never even heard of it. Could you please tell me about this disease, treatments available and if it's an inherited condition?

A: Sarcoidosis is an uncommon inflammatory disease that most frequently involves the lungs and lymph nodes, but can affect virtually any part of the body. Normally, our immune system defends our body against infections and other environmental dangers, but sarcoidosis is associated with abnormal activation of the immune system in the absence of an identifiable cause.

Sarcoidosis causes a typical inflammatory pattern in affected tissues called “granulomas”. Granulomas are clusters of inflammatory cells that often form around foreign objects or in response to certain infections, such as tuberculosis. Thus, all other causes of granulomas have to be considered before the diagnosis of sarcoidosis is rendered.

Symptoms
In many cases, sarcoidosis causes minimal symptoms and requires no specific treatment. However, serious complications can arise when sarcoidosis becomes very active in the brain, eyes, heart or lungs and in individuals who develop disfiguring skin manifestations.

A common symptom of sarcoidosis is fatigue, which often reflects how active the disease is. But fatigue is a very nonspecific symptom. Therefore, it is often overlooked as a manifestation of sarcoidosis.

Treatment
Patients with complications threatening the function of vital organs, such as the lungs, heart, brain or the eyes or with disfiguring skin involvement should be treated. Most people with sarcoidosis respond to appropriate therapy. Some patients have such severe fatigue that treatment to reduce the amount of inflammation is necessary for the sake of improving their functional status.

It is often difficult to predict who will eventually require treatment or how long treatment will be necessary. For this reason, patients with sarcoidosis should see their doctor at least once a year, even if they are feeling well, and more frequently if they are experiencing significant symptoms or if they are receiving treatment.

Based on the fact that sarcoidosis is an inflammatory disease, the treatments are designed to suppress the immune system. The most established treatments include corticosteroids and methotrexate, but many other treatments or treatment combinations have been used, and new ones are currently being tested.

Drugs that block a specific mediator of inflammation, called “tumor necrosis factor,” recently have shown encouraging results in those who are resistant to standard treatments. Decisions relating to if and when to start treatment, and how to adjust these medications are best made by physicians with experience caring for
sarcoidosis patients.

Research and inheritance

While sarcoidosis remains a mysterious disease, researchers are making progress toward understanding its cause. Evidence for genetic causes of sarcoidosis is growing.

It has long been recognized that sarcoidosis runs in certain families and is more common in certain ethnic groups. For instance, sarcoidosis is much more common and tends to be more severe in African-Americans than in whites, suggesting that genetics plays an important role. In this regard, certain genetic mutations appear to influence the likelihood of developing sarcoidosis, while others seem to influence the severity of the disease.

In addition to genetics, some studies indicate that sarcoidosis may represent an abnormal reaction to common environmental exposures that are difficult to detect by common laboratory tests. Considered together, most experts would agree that sarcoidosis is influenced by a combination of genetic and environmental factors.

What to expect if you have sarcoidosis

Most patients who have sarcoidosis will live a normal lifestyle, including a normal lifespan, and will require little or no treatment for their disease. For these individuals, a healthy lifestyle, including regular exercise, a balanced diet and the avoidance of smoking and excessive alcohol intake, and regular follow-up with a doctor are most important for maintaining optimal health.

For those who require ongoing treatment, consultation with a physician having experience caring for sarcoidosis patients is recommended to optimally manage the disease. A small minority of sarcoidosis patients will experience progression of their disease despite medical treatment. In those cases, organ transplantation has emerged as an effective treatment for those who are eligible, and for whom an organ donor becomes available.