

New gene test flags risk of serious complications in sarcoidosis

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(Medical Xpress)—Researchers at the University of Illinois Hospital & Health Sciences System have identified a genetic signature that distinguishes patients with complicated sarcoidosis, an inflammatory lung disease that can be fatal, from patients with a more benign form of the disease. The gene signature could become the basis for a simple blood test.

Their findings are reported online in the journal *PLOS ONE*.

In sarcoidosis, tiny clumps of abnormal tissue form in organs of the body. These clusters of immune cells, called granulomas, cause inflammation. Sarcoidosis can occur in the lymph nodes, liver, eyes, skin or other tissues, but almost always also in the lungs. The cause of the disease is unknown. African Americans are at higher risk for the disease and for more severe cases.

"One of the perplexing aspects of this disease is that two thirds of the people who get sarcoidosis get better with only minimal therapy," says Dr. Joe G.N. "Skip" Garcia, vice president for health affairs at the University of Illinois and principle investigator on the study.

But one third of [patients](#) go on to develop complicated sarcoidosis—neurologic sarcoidosis, cardiac sarcoidosis and progressive lung disease, Garcia said. Complicated sarcoidosis can leave patients with lung damage, and in a small percentage of cases the disease can be fatal.

The challenge, Garcia says, is that there is no difference in the clinical presentation between patients with simple sarcoidosis and those who will go on to develop more serious disease.

The researchers took blood from patients with simple and complicated sarcoidosis as well as patients without the disease to look for a pattern of gene expression unique to complicated sarcoidosis.

They were able to identify a distinct 20-gene pattern of gene expression that could reliably identify those most likely to progress to complicated sarcoidosis.

A 31-[gene expression](#) signature had been identified previously, but a smaller panel of genes makes the new test less expensive and more useful clinically, said Garcia.

"We are dedicated to looking for new insights as well as new therapies for sarcoidosis and hope to someday be able to identify people at risk for it ahead of time," Garcia said.

UI Health has partnered with the Bernie Mac Foundation to form the Bernie Mac Star Clinic for Sarcoidosis, where the researchers hope to further validate use of the genetic signature.

They hope the [genetic signature](#) could someday be the basis for a biochip that could identify patients most likely to progress to life-threatening disease.

Provided by University of Illinois at Chicago

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