

Researchers target vascular disease linked to cancer-causing gene mutation

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Researchers have discovered how a genetic disease known mainly for its life-threatening tumors also can cause sudden death from cardiovascular disease in children, and are mounting a clinical trial to develop treatments for the problem.

Scientists from the Indiana University School of Medicine found that the mutation that causes [neurofibromatosis type 1](#) disease leads to arterial inflammation and damage that is similar to the long-term damage that can occur as people age. They reported their findings in the March issue of the [Journal of Clinical Investigation](#).

Neurofibromatosis results from mutations in a gene called NF1, which causes tumors to form in the cells that make up the protective sheaths around nerves. In humans, NF1 mutations resulting in neurofibromatosis occur in one in 3,500 births, making it the most common genetic disease in humans that results in a predisposition to cancer.

However, cardiovascular disease in children with neurofibromatosis is a significant but under-recognized problem for which the patients are rarely tested, said David Ingram, M.D., associate professor of pediatrics and of biochemistry and molecular biology and principal investigator of the research team. Moreover, he said, "It's often a silent killer with no symptoms or warnings in advance of a catastrophic event - the children present with a heart attack or stroke."

A 2001 analysis of death certificates by Jan Friedman, M.D., Ph.D, of

the University of British Columbia in Vancouver, found that the median age of death of NF1 patients was 15 years younger than the general population. NF1 patients who died at age 30 or younger were more than seven times as likely as normal patients to have been diagnosed with a cardiovascular problem.

Using genetic experiments in mice Dr. Ingram and his team were able to narrow the cause of the cardiovascular problems down to the [inflammatory cells](#) delivered to the site of the damaged blood vessel, ruling out potential effects from NF1 gene mutations in the blood vessel muscle cells and the cells that line the inside of the [blood vessels](#).

In addition, they compared blood samples from a small group of human patients with and without the NF1 mutation and found that the neurofibromatosis patients had significant levels of inflammatory cells and other compounds that pose a higher risk of cardiovascular disease.

The IU researchers, in collaboration with Dr. Friedman in Vancouver, have begun a pilot clinical trial to evaluate potential diagnostic tests, including blood pressure monitoring and ultrasound tests of carotid arteries, that might enable physicians to discover and treat neurofibromatosis patients who are developing [cardiovascular problems](#).

"We think that if we can demonstrate this association with vascular effects and the ability to diagnose them we could proceed to an intervention clinical trial. Statins have anti-inflammatory effects and there are other agents that could potentially be used," Dr. Ingram said.

Provided by Indiana University

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