southwestern medical school - graduate school of biomedical sciences - school of allied health sciences

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******Dallas researcher discovers first evidence of a genetic factor in rheumatoid arthritis.

DALLAS--Rheumatoid arthritis typically strikes when the victim--most often a woman--is young or middle-aged. But it also attacks children, who as a result may be crippled for the rest of their lives with painfully swollen joints.

More than two million Americans have rheumatoid arthritis; more than 200,000 of them are children.

The cause of this tragic disease remains a mystery. But now a Dallas researcher has discovered that victims of rheumatoid arthritis have a certain genetically determined immunological makeup--in other words, an inborn predisposition to develop the disease.

Dr. Peter Stastny, associate professor of internal medicine at The University of Texas Southwestern Medical School at Dallas, says there always has been "an undercurrent of suspicion" that a genetic factor is involved in rheumatoid arthritis, but until now the evidence seemed to be against it.

"A well-known study of the Pima and Blackfeet Indian tribes found no evidence of a genetic factor in RA (rheumatoid arthritis)," he says. "That study was highly publicized and it had been generally accepted that perhaps genetics doesn't have much to do with RA after all.

"I think our study reverses that trend. I think from this we can say for sure that there is evidence of a genetic factor in rheumatoid arthritis."

What Dr. Stastny discovered was that certain "genetic markers" (molecules on the surface membrane of the body's cells) occur in a significantly higher percentage of those with RA, as compared to those without the disease. The presence of these markers is determined by an area on a particular chromosome known as the "HLA region," which has a great deal of control over the human response.

"It really is not surprising to find an association between the chromosome that codes for the immune response and a disease such as rheumatoid arthritis, because in RA there are all kinds of funny things going on that involve immunity," Dr. Stastny says. "It's something that one would expect, but this is the first time that it has indeed been found."

first add rheumatoid arthritis

Fifty-eight percent of the adult RA patients Dr. Stastny tested had the HLA-determined marker known as "DW4", while only 16 percent of a control group of disease-free adults had it. He also found that another HLA-determined marker, "LD-TMO," occurred in 22 percent of juvenile RA patients, compared to only one percent of a control group. One subset of juvenile patients, those with persistent inflammation in just a few joints, showed an even higher occurrence of the LD-TMO marker--50 percent.

Since Dr. Stastny reported his findings in the scientific journals, they have been confirmed by several independent investigators. One of those investigators, Dr. Hugh McDevitt of Stanford University, has suggested that the discovery may lead to tests that can identify people at risk of developing rheumatoid arthritis.

But that, unfortunately, remains far in the future, Dr. Stastny notes. "We're a long way from clinical application," he says. "DW4 typing is unlikely ever to be a useful diagnostic test, but it is possible that we will find other genetic markers associated with the HLA region that show a bigger differential between the patients and the controls (which is what is needed to make it a useful diagnostic test)."

The discovery does open up new possibilities for performing experiments, Dr. Stastny says. "Presumably it will help us unravel the development of the disease-how and why it appears in certain people. That, most likely, is its importance."

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