

Boston University

OpenBU

<http://open.bu.edu>

BU Publications

BU School of Medicine News: Press Releases

1991-07

Boston University School of Medicine news: July 1991

<https://hdl.handle.net/2144/28514>

"Downloaded from OpenBU. Boston University's institutional repository."



Boston University School of Medicine

Department of Public Relations
Office of Media Relations

Elizabeth E. Russell, Director

80 East Concord Street
Robinson 7 (B-7)
Boston, MA 02118-2393
(617) 638-8491
Fax: (617) 638-8044

NEWS

For Immediate Release
July 1991

Contact: Patti Jacobs or
Gina DiGravio
(617) 638-8491

SEARCH FOR GENE THAT CAUSES WILSON'S DISEASE NARROWS,
IMPROVING TESTING FOR DISEASE

Boston, Mass.--Researchers have narrowed the location of the gene that causes Wilson's Disease--a finding that will allow earlier and more accurate diagnosis and treatment of the disease. The study, published in the July 1991 issue of Neurology, was conducted by researchers at Boston University School of Medicine (BUSM) and Stanford University School of Medicine.

Wilson's Disease is an inherited disorder that affects about one in 30,000 people worldwide and is perhaps the most common cause of chronic liver disease in children. People with Wilson's disease are unable to rid their bodies of copper, which, although needed for normal metabolism, is very toxic in excess amounts. In Wilson's disease, excess copper is deposited in the liver and the brain. Left untreated, it can cause serious neurological or liver damage and eventually death.

Because the symptoms of the disease often mimic other illnesses and aren't evident until at least age five, early testing to prevent irreversible liver and brain damage is vital. Traditional biochemical tests that detect copper in organ tissues and invasive liver biopsies cannot be done under the age of two.

The researchers of this study believe their new findings will enable very early detection of the disease. In the study, these scientists--who previously identified the chromosome that contains the gene for Wilson's disease--came closer to finding the gene's precise location by studying genetic markers on chromosomes of members of 50 families throughout

-more-

the world with histories of Wilson's disease. They also found that the location of the Wilson's disease gene was the same in all the families, indicating that there is only one single gene that causes the disease.

"With this information we have developed a blood test that can detect the potential for Wilson's disease at a very early age, even prenatally," says Lindsay Farrer, Ph.D., the principal investigator of the study and assistant professor of neurology at BUSM.

This means, he says, that physicians will be able to let parents know very early on if their child might be a candidate for this disease. Thus, treatment with drugs that have been shown to bind with copper and deliver it from the body can be offered earlier, thereby preventing onset of the disease.

Farrer emphasizes that this test can only be done on people whose families have a history of the disease and requires examination of tissue samples from several family members, including at least one affected individual.

The next step, according to Farrer, is to find the gene itself, which is now feasible, given that its location has been pinpointed to a small region of the chromosome. "Now that we are working within such a tiny area of the chromosome, we can use powerful molecular biology techniques to zero in on the gene," he says.

"Once the gene has been identified," he adds, "it would allow us to understand what the basic defect is, which is currently unknown. This, in turn, would enable us to do direct screening of people in the population for this mutant gene and potentially effect a cure."

This international study was performed in collaboration with investigators from Tel Aviv University in Israel, Albert Einstein College of Medicine in New York, the University of Cagliari in Italy, the University of Toronto in Canada, Karl Marx University in Germany, Yale University School of Medicine in Connecticut, and Washington University in Missouri.

Boston University School of Medicine is located in the South End of Boston between two of its principal teaching hospitals--The University Hospital and Boston City Hospital.