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BY PAUL TAYLOR
Medical Reporter

TORONTO — The gene responsible for a disease that causes copper poisoning of the liver and brain has been discovered by researchers at the Hospital for Sick Children.

The finding, reported today in the scientific journal Nature Genetics, should pave the way to a "simple and conclusive" genetic test for Wilson's disease, the hospital said.

The gene contains several different mutations, said Diane Cox, a geneticist and head of the research team. "When we get all the mutations identified, we will have a reliable test. That should take about a year."

In people with Wilson's disease, copper is not properly taken out of the liver. Over time, the copper reaches toxic levels, causing liver damage and neurological problems that include speech difficulties and loss of movement control ranging from tremors to staggering. Some patients are misdiagnosed as having psychiatric disorders.

They can be treated with a drug that removes the excess copper. But if the disease is left to progress too far, the only treatment is a liver transplant.

Liver disease — sometimes displayed as severe liver failure — will usually appear in childhood or early adulthood. But existing tests for the illness take several weeks or months and can be inconclusive. "It can be difficult to tell whether it's Wilson's disease or another liver disorder," Dr. Cox said.

She added that a lack of awareness of Wilson's disease is a serious problem. One in every 30,000 Canadians is believed have the defective gene. In other areas of the world, the incidence is higher. It is one of the most common genetic disorders in Japan, China and parts of Italy.

A complete genetic test would eliminate the guesswork of diagnosis and lead to quicker treatment, Dr. Cox said. For the past six months, the Hospital for Sick Children has been using the mutations identified to date to screen families with a history of the disease, enabling treatment before signs of the illness arise.