Genetic causes found in nearly 1 in 5 patients with dilated cardiomyopathy heart failure

March 27 2012

(Medical Xpress) -- Researchers have identified genetic causes in nearly 1 in 5 patients who suffer a type of heart failure called dilated cardiomyopathy.

Carolyn Jones, MD, PhD, of Loyola University Medical Center, is co-author of the study, published in the *Journal of* Cardiac Failure. First author is Neal Lakdawala, MD, of Brigham and Women's Hospital.

Researchers did genetic testing on 264 patients with dilated cardiomyopathy and found that 17.4 percent had gene mutations associated with the disease. Pediatric patients were more likely to have the mutations than older patients.

The findings will help in the development of new treatments, Jones said. "By understanding the genes involved in dilated cardiomyopathy, we possibly will be able to circumvent the defect."

Also, if a genetic test shows a patient has an inherited form of the disease, it would indicate that other family members also should be tested, Jones said.

Dilated cardiomyopathy is a condition in which the heart becomes weakened, enlarged and unable to pump efficiently. It is the leading reason for heart transplants. In addition to genetic causes, there are environmental causes, including alcohol abuse, atrial fibrillation (irregular heartbeat) and autoimmune diseases such as lupus.

Earlier studies involved genetic testing on carefully selected research subjects. The new study, by contrast, involved genetic testing in real-life clinical practices. Jones was among the physicians in the study who saw patients, obtained their family histories and arranged for their genetic testing, which was done at the Laboratory for Molecular Medicine at the Partners HealthCare Center for Personalized Genetic Medicine in Cambridge, Mass.

The study included an ethnically diverse sample of patients ranging in age from newborn to 71 years. The average age was 26. Children with dilated cardiomyopathy frequently tested positive for mutations, even if they did not have a family history. Conversely, no patient over age 40 had mutations, unless they also had a family history.

Jones is director of Clinical and Cytogenetics and an associate professor in the departments of Pediatrics and Pathology of Loyola University Chicago Stritch School of Medicine.

Provided by Loyola University Health System