Investigators identify gene mutations in patients with Becker muscular dystrophy

January 13 2010

Investigators in The Research Institute at Nationwide Children's Hospital have identified a link between specific modifications of the dystrophin gene and the age of cardiac disease onset in patients with Becker muscular dystrophy (BMD). This information could help clinicians provide early cardiac intervention for BMD patients based on genetic testing results performed on a blood sample. These findings are a result of analysis of the largest number of BMD patients to date and are published in the December issue of the journal *Circulation: Cardiovascular Genetics*.

Becker muscular dystrophy is a genetic disorder that usually begins in adolescence causing progressive muscle weakness of the legs and pelvis. Most patients - more than 70 percent - will also develop cardiac disease that is likely to go unnoticed until it has reached an advanced stage. To date, clinicians cannot predict when cardiac disease will occur and which patients would most benefit from early heart screenings.

"Our study findings revealed areas of gene mutation most associated with early onset of heart disease," said the study's lead author, Rita Wen Kaspar, BSN, RN, a PhD student at The Ohio State University College of Nursing who conducted this research at the Center for Gene Therapy in The Research Institute at Nationwide Children's Hospital. "By identifying which dystrophin mutations are most likely to cause early-onset heart conditions, our research could help clinicians identify at-risk patients, provide early intervention and ultimately prolong patient survival."

Investigators collected data from 78 patients with BMD or X-linked dilated cardiomyopathy from Nationwide Children's Hospital, The Ohio State University, the Utah Dystrophinopathy Project, the Leiden Open Variation Database and published case reports. They then correlated genetic mutations with the onset age of heart disease.

Federica Montanaro, PhD, the study's corresponding author and a principal investigator in the Center for Gene Therapy at Nationwide Children's, described the study as an important example of collaboration between basic scientists and clinicians.

"The results from this study are important at two levels," explained Dr. Montanaro, also a faculty member of The Ohio State University College of Medicine. "First, as genetic screening becomes more widely available, clinicians will now be able to use this information to deliver more personalized care to BMD patients. Second, our findings provide new clues as to the functions of dystrophin in the heart. These clinical findings are now being brought back to the research laboratory to help design more effective treatments for heart disease in BMD patients as well as in children that suffer from the more severe form of this disease known as Duchenne Muscular Dystrophy."

Provided by Nationwide Children's Hospital