# Heart failure linked to gene variant affecting vitamin D activation

December 1 2009

Previous studies have shown a link between low vitamin D status and heart disease. Now a new study shows that patients with high blood pressure who possess a gene variant that affects an enzyme critical to normal vitamin D activation are twice as likely as those without the variant to have congestive heart failure.

"This study is the first indication of a genetic link between vitamin D action and heart disease," says Robert U. Simpson, professor of pharmacology at the University of Michigan Medical School and one of the authors of the study in the journal *Pharmacogenomics*.

"This study revealed that a critical enzyme absolutely required for production of the vitamin D hormone has a genetic variant associated with the development of congestive heart failure," Simpson says. "If subsequent studies confirm this finding and demonstrate a mechanism, this means that in the future, we may be able to screen earlier for those most vulnerable and slow the progress of the disease." Such a screening test would be years away.

Study co-authors Russel A. Wilke of the Medical College of Wisconsin and Catherine A. McCarthy of the Marshfield Clinic Research Foundation in Marshfield, Wis., analyzed the genetic profiles of 617 subjects from the Marshfield Clinic Personalized Medicine Project, a large DNA biobank. They looked for variants in five candidate genes chosen for their roles in vitamin D regulation and hypertension. One-third of the subjects had both hypertension and congestive heart failure,

one-third had hypertension alone and one-third were included as healthy controls.

The results showed that a variant in the CYP27B1 gene was associated with congestive heart failure in patients with hypertension. It is already known that mutations that inactivate this gene reduce the required conversion of vitamin D into an active hormone.

"This initial study needs to be confirmed with a larger study that would permit analysis of the full cardiovascular profile of the population possessing the gene variant," Simpson says. A future study also would need to include people of more diverse origins than this study's population of mostly European ancestry, the authors say.

More information: *Pharmacogenomics*, (2009) 10(11):1789-97

Source: University of Michigan Health System (news : web)