Mutation in gene causing heart failure identified

Researchers have uncovered a major genetic risk for heart failure — a mutation affecting a key muscle protein that makes the heart less elastic.The mutation increases a person's risk of dilated cardiomyopathy. This is a form of heart failure in which the walls of the heart muscle are stretched out and become thinner, enlarging the heart and impairing its ability to pump blood efficiently, a new international study has revealed.

The mutation causes the body to produce shortened forms of titin, the largest human protein and an essential component of muscle, the researchers said in background information.

In this study, researchers studied more than 5,200 people, including both healthy people and people suffering from dilated cardiomyopathy. The researchers performed genetic sequencing on all these people, examining the specific gene that the body uses to create titin.

They uncovered a specific type of titin mutation that occurs in families and appears to greatly increase the risk of dilated cardiomyopathy (DCM).

Up to now, genetic testing for heart failure has been difficult because it's been hard to interpret which mutations might lead to heart disease, [study author] Roberts said. These findings could better help doctors figure out which people are at greater risk for heart failure — especially those who have a family history of the disease.

Read full, original article: Scientists Spot Mutation Behind Genetic Form of Heart Failure