More tangible results from genetic testing: A new way to predict heart disease

By ACSH Staff — December 19, 2013

Many of us are aware of the mutation of the BRCA gene which leads to an 85 percent chance of a woman with this mutation developing breast cancer in her lifetime. The best known example of this occurred earlier this year, when Angelina Jolie revealed that she had the mutated gene and underwent a prophylactic bilateral mastectomy currently the only way to prevent this type of breast cancer from developing.

Now, another, recently-discovered mutant gene [1] seems to be associated with a 38 percent increased risk of having a heart attack, in men at least. And the gene was found in about one-eighth of those men tested, making this quite an interesting and potentially highly important risk factor.

The authors of the PLoS paper call this dysmorphic (abnormal) gene, the heart attack gene. The mutation is much less common in women.

Here's a bit more detail, for those interested: the authors are a group at the Duke University Medical Center, and they found that a single mutation in the 5HTR2C gene (5HT is 5-hydroxytryptamine, aka serotonin, and R2C refers to receptor that binds the serotonin) can have a profound effect on the development of cardiovascular disease. They believed that the effect on the heart was mediated by an increase in the production of the stress hormone cortisol (aka hydrocortisone), which is a known factor in heart disease. The study authors conclude that the presence of this gene is as harmful as smoking or obesity.

Lead author Beverly Brummett, said: The exciting part to me is that is genetic trait occurs in a significant proportion of people with heart disease. If we can replicate this and build on it, we might be able to find ways to reduce the cortisol reaction to stress and reduce deaths from heart attack.

ACSH's Dr. Josh Bloom comments, Although disease diagnosis and treatment using genetic information has led to some important medical advances, I'm guessing that this field is still in its infancy, and that we will see further significant advances in diagnosing and treating numerous conditions based on genetics in other words, improved personalized medicine.