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Researchers crack the genetic code of a sudden death cardiac killer

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Researchers in Newfoundland have cracked the genetic code of a sudden death cardiac killer.

As a result, they have developed a unique prevention program in which people with no symptoms, but with a suspect gene and a family history, are being implanted with internal cardiac defibrillators (ICDs) which can restart their hearts if they stop.

"Our discovery has led to a targeted genetic screening and individualized therapy that is significantly improving survival rates," Dr. Sean Connors told the Canadian Cardiovascular Congress 2010, co-hosted by the Heart and Stroke Foundation and the Canadian Cardiovascular Society. "It's allowing people with the condition to live normal, longer lives. Individualized genetic therapies like this are the future of medicine."

The excitement among cardiologists concerns a rare genetic condition – arrhythmogenic right ventricular cardiomyopathy (ARVC).

"Newfoundlanders likely have the highest incidence in the world of this disease," Dr. Connors, a cardiologist and associate professor of medicine at Memorial University in Newfoundland, told the Congress.

The term arrhythmogenic refers to deadly cardiac rhythms that can be triggered by electrical impulses within the heart. Cardiomyopathy is a worsening condition where heart muscle is slowly replaced by scar and fat tissue.

The combination of the two is lethal, Dr. Connors says.

"People who are at risk often have no symptoms, so the first time we know they have this disease is when they die."

Genetic profiling?

The surest sign that a disease is genetic in origin is when it is manifests itself in family histories, showing up in generation after generation.

"Our diagnostic testing showed that some members of these families have a specific, genetic, electrocardiogram (ECG) mutation – ARVD5," said Dr. Connors. There is a 50 per cent chance that children of those with the condition will also be carriers of the gene. It is considered the second-most common cause of sudden cardiac death in young people.

The mutation causes premature sudden cardiac death in males: 50 per cent die by age 40 years and 80 per cent by 50 years. For women the rate is five per cent and 20 per cent.

Given those figures, Dr. Connors realized nothing would be lost by implanting ICDs in asymptomatic patients with ARVD5 to maintain normal heart rhythms.

Earlier this year his team reviewed the data of the prevention program which they started in 1999. They concluded that implanted defibrillator treatment for primary prevention in both sexes, and secondary prevention in males significantly improves survival.

According to Heart and Stroke Foundation spokesperson Dr. Beth Abramson, there are as many as 40,000 sudden cardiac arrests every year in Canada.

"What's fascinating about this study is that they show that preventive intervention works," says Dr. Abramson. "This treatment is not only prolonging lives; it's giving families peace of mind and hope for the future."

She also notes, "This is exciting information for physicians working with these rare cases but is also important to remember that the most common causes of heart disease relate to lifestyle. It is important that all of us, regardless of family history, take care of our hearts: basic things like smoking cessation, following a healthy diet, and physical activity go a long way in protecting our health."

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