

New explanation for cardiac arrest

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Researchers have discovered a new disorder linked to heart problems that stems from a genetic defect in the protein glycogenin. In a worst case scenario, disruption of this protein's function can lead to cardiac arrest, which is exactly what happened to the young man whose case triggered the investigation at Sahlgrenska University Hospital at the University of Gothenburg, Sweden, that led to a brand new diagnosis.

Published today in the revered [New England Journal of Medicine](#), the study details how a young man suffered a cardiac arrest but survived thanks to the work of the ambulance paramedics. An investigation at Sahlgrenska University Hospital led to the discovery of not only a new disorder but also how a defect in the protein glycogenin can lead to an energy crisis in the muscle cells.

This protein's job is to initiate the build-up of glycogen that constitutes the muscle cells' carbohydrate reserves. The glycogenin starts the actual process by building up a short chain of around ten [sugar molecules](#), which can then be turned into glycogen with the help of other enzymes. During strong muscular work the sugar molecules in the glycogen are used to create energy.

"The disorder is characterised by an inability to form the initial chain of sugar molecules," says Anders Oldfors, who headed up the research team and is a professor at the Sahlgrenska Academy and consultant at Sahlgrenska University Hospital. "This leads to a shortage of glycogen and an energy crisis in the muscle cells that can result in cardiac arrest."

The study also reveals how muscle cells that have a severe congenital defect can adjust and find other ways of sourcing energy, though it may not be sufficient in all situations.

"We're hoping that our continued research in the field will provide answers to how the change in the glycogenin causes an inability to start accumulating carbohydrates in the [muscle cells](#)," says Oldfors.

Clinically, the discovery means that this disorder must be considered as a diagnosis when investigating [heart problems](#). For patients, a correct diagnosis means that there is preventative treatment available, though no cure is on the horizon at present. As the cause of the disorder is a genetic defect, it is hoped that in the future patients can be given a customised treatment, or gene therapy, for it.

"But we don't yet know how common this disorder is," says Oldfors.

"This is something that the future will hold now that we are in a position to make the correct diagnosis."

Cardiac arrest occurs when the blood suddenly stops pumping out of the heart. This leads to unconsciousness, and the breathing stops on account of an inadequate supply of blood. It is one of the most common causes of death and accounts for 11-18 per cent of all deaths in Sweden. Many old people are affected, with the trigger frequently being a heart attack. Cardiac arrest is very rare in young people and is generally caused by some form of hereditary heart muscle disorder. Cardio-pulmonary resuscitation and defibrillation can save patients who have suffered [cardiac arrest](#). Preventative treatment takes the form of medication, and a surgically inserted defibrillator can also be used as protection. According to the Swedish Resuscitation Council 300-400 people a year are revived after [cardiac arrest](#) outside hospital and roughly 1,000-1,500 people in hospital.

Provided by University of Gothenburg

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