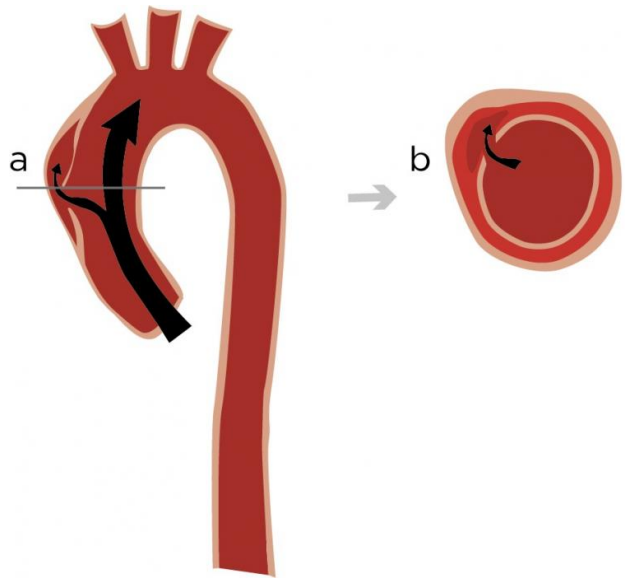
Genetic mutation linked to aortic dissection in the chest

11 May 2017



An illustration of an aortic dissection in the length axis (figure A) and in a cross section (figure B). Aortic dissection is caused when a sudden rupture in the inner wall of the vessel causes blood to flow into the aortic wall, resulting in structural weakening. Credit: Mattias Pettersson/Umeå University

Researchers at Umeå University in Sweden have discovered a genetic mutation that can cause dissection of the thoracic aorta, which is the body's main artery. The mutation leads to an impaired function of the smooth muscle cells of the aortic wall and thus an increased predisposition for aortic dissection.

Aortic dissection, or rupture, is the bursting of an aortic aneurysm and can occur both within the chest (thoracic) and in the abdomen. A ruptured abdominal aortic aneurysm is usually preceded by aortic calcification (atherosclerosis), while aortic dissection in the chest cannot be associated with calcification of the main artery. In Sweden, about 400–600 people per year are affected by thoracic aortic dissection, a number which is likely higher due to immediate and high mortality and the fact that the right diagnosis is never determined in many cases. The disease is hereditary in about 20–25 per cent of cases.

"Aortic dissection in the chest is a life-threatening condition that requires immediate care, usually emergency surgery," says Matias Hannuksela, medical doctor at the University Hospital of Umeå Heart Centre and author of the doctoral dissertation.

"The aorta usually dilates before a dissection, but people carrying the genetic mutation that we have discovered can have a dissection even with a mildly dilated aorta."

Matias Hannuksela, who works as a researcher at the Department of Surgical and Perioperative Sciences, has also studied thoracic aortic function in patients with a dilated versus a normal sized aorta. According to the research, patients with a hereditary predisposition for aortic dissection and a dilation of the aorta also appear to have an increased aortic stiffness.

Matias Hannuksela comes from Vaasa, Finland. He studied medicine at Umeå University and earned his medical degree in 1995. Matias Hannuksela is a specialist in anaesthesia and intensive care and currently works as a consultant at the thoracic surgery unit within the Heart Centre at the University Hospital of Umeå (NUS). Credit: Daniel Harju/Umeå University

"In the future, an increased aortic stiffness could, just as a measurement of the aorta's size, be used as an indication of when preventive surgery is needed," explains Matias Hannuksela.

Together with research colleagues, Matias Hannuksela has also shown that screening, usually in the form of ultrasound examination, can be used to discover previously unknown aortic dilation in relatives of affected patients. The researchers therefore recommend screening of close relatives to people affected by thoracic aortic dissection.

"I order for us to focus screening efforts on individuals who are at risk of aortic dissection because they are carriers of the disease-causing mutation, more research on the genetic causes of aortic dissection is needed. Why some people without a genetic mutation can also be affected by this disease is still unclear. The genetic component is probably more common than we think," says Matias Hannuksela.

Provided by Umea University