Research supports broader screening for sudden cardiac death

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Around one in 500 Swedes carry a genetic mutation which can cause sudden cardiac death. The diagnosis can lead to major lifestyle changes, but quality of life can be maintained with the right advice and support, reveals a new study from the Sahlgrenska Academy at the University of Gothenburg, Sweden.

The most common cause of sudden cardiac death in children and adolescents is the heart muscle disease hypertrophic cardiomyopathy. The condition is hereditary, and if one family member is affected the Swedish National Board of Health and Welfare recommends that the whole family is screened. This can lead to major upheavals in family members' lives: people who see themselves as healthy can suddenly get a diagnosis that necessitates big lifestyle changes and sometimes even lifelong medical treatment.

Physical effects of screening

In her thesis, Ewa-Lena Bratt, a paediatric nurse at the Queen Silvia Children's Hospital and doctoral student at the University of Gothenburg's Sahlgrenska Academy, looks at the physical effects of family screening and weighs them against the psychosocial effects and impact on quality of life. The study of parents and their children aged 7 to 25 at the Queen Silvia Children's Hospital reveals that the diagnosis itself rarely has a negative effect on quality of life.

Children and parents grateful

"Despite learning that they have a chronic and potentially life-threatening heart disease, both children and parents were grateful that the disease had been picked up before any serious complications arose, and that they could now call on medical treatment and advice," says Bratt. "The lifestyle changes, on the other hand, have a greater negative impact, not least for older children and adolescents who are already doing sport and now have to avoid unsuitable sporting activities."

Greater risk for adolescents

The risk of serious psychosocial effects is greater among adolescents, many of whom are at a sensitive stage and have built up a social network through their sport. It can therefore help to be diagnosed earlier in life. "Younger children can be guided by their parents towards other leisure activities which they can pursue during their youth without running any risks," says Bratt.

Not informed of the risk

One problem is that many adults diagnosed with hypertrophic cardiomyopathy are not informed of the risk of the disease being inherited, with the result that their children are not referred for screening. There is room for improvement here, Bratt believes.

"Our most important conclusion is that newly diagnosed children and adolescents given the right support and information can maintain their quality of life and look confidently to the future," she says. "I hope that my thesis will spur further improvements in the way these patients are looked after."

Good physical performance

Many people with hypertrophic cardiomyopathy are treated with beta-blockers, but their use in those who are not symptomatic is controversial, as it has been speculated that this type of treatment affects physical performance to such a degree that it can be disabling. Bratt's thesis shows, however, that physical performance is as good in those treated with beta-blockers as in those who only make lifestyle changes.

"My thesis lends support to broader screening for hypertrophic cardiomyopathy," says Bratt.

More information: The thesis "Screening for Hypertrophic Cardiomyopathy in Asymptomatic Children and Adolescents. Psychosocial consequences and impact on quality of life and physical activity" was successfully defended on Oct. 28 .

Provided by University of Gothenburg