

# Stanford Hospital Health Notes

A community health education series from Stanford Hospital & Clinics

## Deadly Heart Condition Yields to Screening and Prompt Treatment

High school senior James Cooper and his mother laughed at first when, in the middle of a sports physical, the examiner mentioned that the young man's arm span was suggestive of Marfan syndrome, a connective tissue disease that can seriously affect the heart. Cooper was otherwise in the peak of youthful fitness, working out four to five hours a day and playing several competitive sports.

"But a couple of weeks later my mom said, 'Maybe we should get you checked out, just to be safe,'" Cooper said. This exam, with a cardiac specialist, included an ECG, a non-invasive test that can identify potentially dangerous heart conditions by measuring the heart's electrical activity. When his mother, a former U.S. Navy medic, saw the look of that ECG and the physician's scrutiny of it, she knew something was wrong. "That's when my heart dropped," she said. Cooper did not have Marfan's, but the specialist was fairly certain he did have a genetic heart condition called hypertrophic cardiomyopathy, sometimes shorthand as HCM.

The physician recommended Cooper and his mother go to Stanford Hospital

& Clinics, where Euan Ashley, MD, PhD, heads the Hypertrophic Cardiomyopathy Center, one of a few clinics in the world devoted to the diagnosis and treatment of the condition.

*"Thank God we found out. I thought of all the families who have no idea, and sudden death is how they realized their son or daughter had this condition"*

— Paulette Cooper, mother of patient at Stanford's Hypertrophic Cardiomyopathy Center

"From the moment we first met Dr. Ashley at Stanford," said Paulette Cooper, "I felt we were sitting in a room with a doctor who really cared – a really gentle person, a really caring person who was not rushing us through, not treating us like he had a waiting room full of other people he had to see. His staff was amazing, too. I felt we were in a really good place."

Ashley, himself an athlete as a youth, developed an early clinical and research interest in heart health, including that specific to the kind of activity athletes ask of their bodies.



James Cooper has had to give up the vigorous athletic conditioning that once filled his life, but his enthusiasm for life and his future is undiminished.

"I've always been fascinated by the heart as an organ," he said. "It's a phenomenal thing that can power Olympic athletes but holds an 80-year-old woman to her chair, not able to walk."

He suspected Cooper had HCM, but also thought some of the behavior of Cooper's heart might be the result of his heavy athletic training, so he ordered the young man to take a break.

That enforced inactivity was a major blow to Cooper. Sitting on the bench and watching his classmates playing team sports was the complete opposite of a path he had followed since he was six years old and ran his first 10k. "I wasn't feeling too good about myself," he said.

As such a change might affect any young athlete, "that just about took the rug out from under him," said Cooper's mother. "This was a kid who worked out four to five hours a day, without a coach. It was hard for him just to suddenly stop. Everybody's saying, 'I'm sure you're fine,' and James is sitting there saying, 'I hope so, but there's something happening.'"

### Recognizing the unusual

Stanford's center is a place where hypertrophic cardiomyopathy is understood as a condition that can be found, as it was in Cooper, in the most athletic of patients. It is caused by genetic mutations that change the structure

of the heart's muscle cells, thickening them and disrupting the flow and force of blood through its chambers. It is the most common cause of sudden death in young people and the most common form of inherited cardiovascular disease.

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Cooper's mother knew her grandmother had been in and out of the hospital, treated for congestive heart failure. And there had been others in the family who had died young, stricken with sudden heart attacks. But she had had no clue that that history might be reflected in her son's heart health.

Often, the symptoms HCM chest pain, fainting, palpitations – can be misdiagnosed. Genetic testing is still evolving, but only at a few places, like Stanford, is it available.

The center has had long experience with the surgery sometimes performed to counteract the obstruction to blood flow caused by the condition. Few surgeons specialize in that surgery, Ashley said. It calls for judgment and experience. "You might only make a small



Once James' mother, Paulette, knew something was wrong with her son's heart and that it could be inherited, she began to recall the heart ailments and early cardiac deaths of other family members.

# What To Know About Hypertrophic Cardiomyopathy

- Hypertrophic cardiomyopathy is a disease of the heart muscle which causes certain areas to enlarge and obstruct blood flow. Typically, it's inherited and can affect both children and adults.
- Symptoms don't always appear in the early stages of the disease, but later can include dizziness, shortness of breath, fatigue and swelling of legs, ankles and feet.

- Treatments include medication to alter how the heart muscle acts, surgery to remove a portion of the thickened heart muscle or implantation of a defibrillator, which starts the heart if it stops.
- Anyone with a family history of unexplained early cardiac death should think about screening and genetic testing.
- Euan Ashley, MD, recommends that young athletes be evaluated by a physician before they begin to do sports. Adults with family history of heart issues should see a physician to address their risk factors. The disease can show itself in adults into their 40s and 50s.

For more information, visit [stanfordhospital.org](http://stanfordhospital.org). Video talks by Dr. Ashley can be viewed at [healthlibrary.stanford.edu](http://healthlibrary.stanford.edu). Contact the Hypertrophic Cardiomyopathy Clinic at 650.736.1384.

number of cuts, but they make a very big difference," he said.

Even after cutting back on his strenuous physical workouts, Cooper's heart still showed the disturbing abnormalities and Ashley presented Cooper with his choices. "He said I could continue living my life as I had before and have the possibility of suddenly dropping dead. Or, I could get a defibrillator or we could talk about medications like beta blockers."

## The next steps

Cooper chose the defibrillator. Compared to his pre-diagnosis lifestyle, his physical activity now is quite restricted – he's not allowed to push his body in ways that significantly raise his heart rate. That means no marathons. And no more competitive sports. But Cooper's personality fights against the restrictions.

People with this heart condition face "something they're going to be dealing with for the rest of their life," said Heidi Salisbury, a nurse who has worked with Ashley at the center for several years. "We encourage our patients to learn as much as possible about their condition, to make the necessary changes in behavior and lifestyle and then to live a high quality of life."

Cooper is both the worst and best case scenario, she said. "He epitomizes a young man, playing basketball, who could have died of cardiac arrest. He had no idea he had this disease or the severity of the disease. He had a higher chance of death than others. But he was a save."

"I've been living with it for awhile now, it's not something I dwell on," Cooper said. "I'm in a good place now. It's not something I fear."

"For a long time, I went through a lot of difficult feelings," said his mother. "On one side, I thought, 'This isn't fair. Here's a kid who wanted to be a firefighter since he was four years old – why does it have to be this? If this was a kid who played videogames all day, it wouldn't have had the impact. On the other side - thank God we found out. I thought of all the families who have no idea and sudden death is how they realized that their son or daughter had this condition."

"James has gone through some hard times, being angry and disappointed. It's been hard for him, but once he had the defibrillator put in, his goal was to help other people like him. He wanted to reach out and talk to others and help them."

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– James Cooper, patient at Stanford Hospital Hypertrophic Cardiomyopathy Center

Cooper took part in last year's Stanford HCM Patient Day. He ran on a treadmill in front of the 75 attendees, demonstrating the importance of remaining active while exercising within the necessary restrictions. His internal defibrillator, about the diameter of a



Norbert von der Groeben

Since his diagnosis, James Cooper is only slightly less busy than he was before. He's completing his undergraduate degree, holds a part-time job and sings in a church choir, along with his fiancée, Breanna.

can of shoe polish, was visible below the surface of his skin. "That visual of seeing him run was really inspiring to the audience," Ashley said.

His internal defibrillator is routinely monitored, but doesn't need much maintenance except for its battery, which lasts for an average of 10 years. The technology is improving continually, Salisbury said. His medical team monitors Cooper's activity remotely, so they know when he's pushing the limit. "It's hard to change your behavior when you're young and vital," she said. "But we are here to support every patient and their family for the long run. This is a process and James is living proof of our mission."



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Euan Ashley, MD, director of the Hypertrophic Cardiomyopathy Center, sees several new patients each week who come for treatment of the thickened heart muscle that can cause sudden death even in young athletes.

Stanford Hospital & Clinics is known worldwide for advanced treatment of complex disorders in areas such as cardiovascular care, cancer treatment, neurosciences, surgery, and organ transplants. Consistently ranked among the top institutions in the U.S. News & World Report annual list of "America's Best Hospitals," Stanford Hospital & Clinics is internationally recognized for translating medical breakthroughs into the care of patients. It is part of the Stanford University Medical Center, along with the Stanford University School of Medicine and Lucile Packard Children's Hospital at Stanford. For more information, visit [stanfordmedicine.org](http://stanfordmedicine.org).

