

Following the Trail of Broken Hearts

A congenital cardiovascular abnormality has become a leading killer of young athletes in the U.S. So why isn't more being done to save those who have it?

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By David Epstein

The kid's playing with me, William Batts figured. One second 17-year-old DeCarlo Polk had gone up confidently for a jumper, his chiseled 5' 9", 237-pound frame rising just beyond the free throw line in their game of one-on-one. The next, he had tumbled to the Nashville blacktop like a piece of laundry at the end of a dryer's cycle. Now, on the afternoon of June 27, DeCarlo lay flat on his back in the simmering heat. Real funny, the 37-year-old Batts thought. But then he took a step closer and saw that DeCarlo's pupils had rolled back into his skull, and he knew it was no joke.

Somewhere mid-jump the electrical signals that cued DeCarlo's heart to pump had misfired terribly. Rather than flexing and contracting rhythmically, his heart trembled like jelly in a shaken jar. His left ventricle, the chamber that takes oxygenated blood from the lungs and sends it through the body, stopped working, causing a circulatory traffic jam. As blood piled up in the lungs' capillaries -- vessels so small that red blood cells have to pass through them single file -- water in the bloodstream breached the capillary walls and settled into the air sacs of his lungs, spaces normally reserved for oxygen. DeCarlo had stopped breathing and started drowning.

Batts called his boss -- DeCarlo's uncle, Jimmy Boulton -- at his paint shop three doors down, and Boulton came running with Rick Jones, a coworker. Jones became DeCarlo's lungs, blowing oxygen-rich air into his mouth. Then he became his heart, pushing down on his chest to force the oxygenated blood through the lungs and into the rest of his body. But CPR could only buy time.

Don't let this happen, Boulton prayed, not now. On June 8, DeCarlo had graduated from Hillside High in Durham, N.C., where he had been an honor student, homecoming king and star defensive lineman. Football scholarship to Division II St. Augustine's College in nearby Raleigh in hand, he was baptized two days later, then jumped in Boulton's truck for his annual summer visit to Nashville, his hair still pearled from the baptismal dip. Within a week of his arrival DeCarlo became the proud cousin of Jimmy's third child, James Kerrigan Boulton Jr.

It took at least 10 minutes for the rescue workers to arrive; already DeCarlo's oxygen-deprived brain cells were dying in droves. The paramedics applied defibrillators to DeCarlo's chest, trying to fix the deadly rhythm that had begun in his left ventricle. Throughout his heart electrical signals were ricocheting like pinballs, spurring spastic muscle twitches. With each jolt from the paddles DeCarlo's powerful body would lift and then flop limply to the ground. His 17-year-old heart was broken.

DeCarlo was one of an estimated 600,000 Americans with hypertrophic cardiomyopathy (HCM), a genetic disease that causes the walls of the left ventricle to enlarge, usually during adolescence; the thickened ventricle does not relax fully, inhibiting the flow of blood into the heart. While many will never exhibit a symptom, some 6,000 will die from HCM each year, more than from asthma and Hodgkin's disease combined. In those HCM



Herrion (above) dropped dead after a preseason game, while Collier died at home in the middle of the night.

victims, the left ventricle's muscle cells, rather than stacking up flush like bricks in a chimney, are arranged at odd angles, all askew, as if the bricks had been tossed into a Dumpster. With the cells in such disarray, an electrical signal crossing them is liable to travel in an erratic path and fatally misfire.

"Hypertrophic cardiomyopathy is the most common cause of sudden death in young athletes," says Dr. Barry Maron, director of the Hypertrophic Cardiomyopathy Center at the Minneapolis Heart Institute Foundation and one of the top experts on HCM. "And it's the most common cause of [natural] sudden death in young people in the general public. But it's still a little-known disease."

For those who suffer from some of the nation's most common health problems, such as diabetes, high cholesterol and coronary artery disease, exercise is a potent remedy. But for those with HCM it is the most active individuals -- the young (usually teenaged) paragons of fitness -- who are at higher risk of sudden death because of their athletic pursuits. At least every two weeks an athlete with HCM will die during or immediately after exertion, when his or her abnormally thick heart, triggered by strenuous exercise, will start beating in a lethal cadence.

Some of the victims are famous, such as San Francisco 49ers offensive lineman Thomas Herrion, 23, who collapsed in the locker room minutes after a preseason game in August 2005, and Jason Collier, 28, the Atlanta Hawks center who died in the middle of the night two months later. But the majority are less well-known, their lives just beginning, such as 16-year-old Adam Litteken, who went lifelessly sliding across a hockey rink after he collapsed during an October practice in St. Peters, Mo., and DeCarlo Polk.



Curry refused to undergo screening that might have revealed HCM, so the Bulls traded him to New York.

And Kevin Richards. During my senior year at Evanston (Ill.) Township High, Kevin, a precocious freshman, became my friend and training partner in track. After I went to Columbia in August 1998, Kevin helped Evanston win its first 4X800-meter state title the following spring. By his junior year Kevin, the son of Jamaican immigrants, was on his way to becoming the first member of his family to attend college, most likely on a track scholarship. Indiana was high on his list.

On the afternoon of Feb. 12, 2000, Kevin was locked in a tight race in the indoor mile. The bell hollowly signaled the final lap as Kevin kicked to the shoulder of Dan Glaz, a top Illinois distance runner from Amos Alonzo Stagg High in Palos Hills. During that lap Kevin's heart struggled to function. Still he kept coming, finishing second by a meter. After crossing the finish line Kevin walked three steps, dropped to his knees and flopped over on his back. It's entirely normal for a bone-weary runner to sink to the ground after a hard race, but never Kevin. Yet there he was, quaking and heaving as he died on the track.

Each of these victims had HCM etched into his genetic code. Despite their apparent good health, they were destroyed by one mutation in the three billion base pairs -- the chemical compounds that form the rungs of the twisting DNA ladder -- that make up the 25,000 or so genes crammed into the nucleus of a human cell. That's the equivalent of a single typo in 60 full sets of The Encyclopedia Britannica.

How can a disease about 20 times more prevalent in the U.S. than Lou Gehrig's disease kill in relative anonymity? Perhaps it is because HCM, unlike ALS, isn't always deadly, and when it is, it often strikes without notice. There is no progressively weakening immune system, no final battle embodied by a courageous ballplayer. "Right now we really don't know most of the time who is at risk of sudden death," says Dr. Paul D. Thompson, a cardiologist at Hartford Hospital. The first diagnosis is often made by a medical examiner, if it is made even then. With few live patients to examine, clinicians have had trouble getting a fix on the disease since British pathologist Donald Teare first compared HCM to a tumor of the heart in 1958. The disease would acquire more than 80 different

names, from apical hypertrophy to subvalvular aortic stenosis, until becoming widely known as hypertrophic cardiomyopathy within the last decade.

While an enlarged heart might serve as a conspicuous sign of trouble, it is also characteristic of a highly conditioned athlete. It is no surprise to anyone who saw DeCarlo race sideline to sideline for a sack that he had, literally, a lot of heart. A normal, fully mature human heart weighs about 300 grams, or two thirds of a pound. DeCarlo's heart -- perhaps enlarged from working out, and certainly enlarged with HCM -- weighed 600 grams. (Ryan Shay, 28, who died 5 1/2 miles into the Olympic marathon trials in New York City last month, and Toronto Blue Jays pitcher Joe Kennedy, 28, who died at his in-laws' home three weeks later, also reportedly had enlarged hearts. Their autopsies are pending.)

If, during high school, DeCarlo had been given an electrocardiogram, or ECG, a \$50 test that takes a few minutes and records the electrical signals in the heart, a cardiologist might have noticed a suspicious pattern indicative of an enlarged ventricle. The doctor might have followed up with an echocardiogram -- or echo, for short -- an ultrasound that gives a real-time picture of the heart and costs \$1,000 to \$2,000. To a cardiologist who had seen HCM before, an echo could determine whether the heart was merely enlarged by exercise (in which case both the muscle of the left ventricle and the chamber it surrounds would expand) or whether it was afflicted by HCM (in which case the muscle walls would grow but the chamber would not). The doctor might have recommended that DeCarlo trade his cleats for golf clubs, tools of a less vigorous sport that he loved. He could have lived a long, normal life with a surgically implantable cardioverter-defibrillator (ICD) standing sentinel inside his chest. The titanium-encased, computerized device is about the size of a matchbox and is programmed to give the bearer's heart a 750-volt shock when it detects an abnormal rhythm.

DeCarlo would have undergone at least an ECG had he grown up in Italy, where all competitive athletes, from grade-schoolers to pros, must, by law, have their hearts undergo government-subsidized screening. When an American child's heart fails, crestfallen parents often, and understandably, speak out in favor of a nationwide screening program that includes ECGs. If Italy can do it, why can't the U.S., the richest and most sporting of nations?

For one, the U.S. has 37% fewer doctors per capita than Italy, a nation that opened the doors wide to its medical schools after World War II. Some Italian doctors have spent almost their entire careers screening athletes. With an American citizenry that is far more geographically and genetically diverse, and that has more than twice as many high school athletes (7.3 million) as Italy has high-school-age people, marshaling enough experts to do quality, uniform screening is impractical, with the costs prohibitive. "The U.S. health care system does not have the mechanisms to pay for an ECG for every athlete," says Lisa Salberg, who has HCM and founded the Hypertrophic Cardiomyopathy Association. "Nor do we have enough trained professionals to evaluate the results. There would be a lot of athletes placed on alert for no reason, and a lot of missed diagnoses."

And yet, immediate steps can be taken given the hereditary nature of HCM. "Asking whether there is anyone in the family who has died before the age of 50 of cardiovascular causes is incredibly valuable," says Dr. David Glover, a Warrensburg, Mo., physician and expert in presports participation screening, "and that doesn't cost a nickel."

Although Glover has seen improvement in preparticipation screening questions, he has also documented a dangerous trend. In 1997 there were 11 states that allowed chiropractors or other nonphysicians to perform the exams that deem a high school athlete fit to play. By 2005 that total increased to 18 states that sanctioned, Glover says, "practitioners with little or no cardiovascular training" to conduct athletic screening. "You need somebody that listens to hearts every day," Glover says. "A trained examiner knows that if a heart murmur is very loud, or if it occurs during the relaxation phase of the heart, or if it gets louder when you stand up, it raises suspicion that a person could have HCM."

There is a sleek, steel-and-glass building about a mile away from Boston's Fenway Park where the men's and women's rest rooms are labeled with diagrams of chromosomes. On its second floor is Harvard's Seidman Lab, run by the husband-and-wife team of Jonathan and Christine Seidman. "Looking at a patient's genes can tell us

that somebody has HCM or is going to develop HCM as they get older," says Christine. The lab is putting mice genetically bred to have HCM through the first HCM drug trial, with the hope that, Christine says, "in a decade or two, the [manifestation of the] disease may be preventable."

Perhaps those future patients will have had HCM diagnosed with absolute certainty through their genes at the nearby Harvard-Partners Center for Genetics and Genomics, where each week researchers identify a new HCM mutation, of which more than 800 are currently known. For \$3,000 the center can pop the blood of a living person (or a deceased one, if a medical examiner is astute enough to freeze a sample) in what looks like an oversized gray microwave and find out if that individual got HCM from any of the currently known mutations.

And once a mutation is identified in one member of a family, other members can be tested for the same mutation - at \$250 apiece. That's what Jimmy Kogut's three younger siblings are doing. A 21-year-old junior at Pittsburgh, Jimmy spent his leisure time playing basketball and lifting weights until last December, when tests revealed that he had HCM; eight months later genetic screening found that he has one of the most common HCM mutations, on a gene that helps to regulate heart contraction. Jimmy's mother, Eileen, had long known that something dangerous ran in her family. Her brother Joe, then 15, died at the dinner table in 1978 while horsing around with Mark, his older brother. Mark died 20 years later, at age 37, while running on a treadmill.

Because of her family history, Eileen had Jimmy's heart checked regularly from the time he was three, and everything seemed fine. At seven Jimmy complained of shortness of breath and was told that he had asthma. Fatigue and shortness of breath, even fainting, are common symptoms of HCM, and a misdiagnosis of asthma is also common. This can be especially hazardous because asthma inhalers can cause irregular heartbeats in people with HCM.

Jimmy now knows that he never had asthma. He has HCM, and he got his gene mutation from his mother, along with his hazel eyes and freckles. An HCM mutation follows a hereditary pattern known as autosomal dominant: In other words, it has a 50-50 chance of passing from parent to offspring. Those who inherit the gene will each have a 50-50 chance of passing it on to their children, and so on down the family tree. Those who do have it may, like Jimmy, undergo surgery, have a defibrillator implanted and give up intense exercise for good. "I can still do some light weightlifting," he says, "but nothing over my head or that stresses my left side too much so that it might damage the ICD." As more families submit to genetic testing and the list of mutations grows, the Harvard-Partners Center is keeping an eye toward categorizing which are deadly (and demand an ICD and cessation of vigorous sport) and which are relatively harmless (and might permit life as usual).

But will people feel safe to seek that knowledge? In September 2005, six months after Chicago Bulls center Eddy Curry felt his heart skipping beats, the team added a genetic testing clause to the one-year, \$5 million contract offer it had on the table. If the tests showed that Curry had HCM, the team would not let him play, but it still promised to pay him \$400,000 a year for the next 50 years. Fearing that the public disclosure of the results might jeopardize his career, Curry refused, and the Bulls traded him to the New York Knicks. "As far as DNA testing, we're just at the beginning of that universe," Alan Milstein, Curry's attorney, told the Associated Press. "Pretty soon, though, we'll know whether someone is predisposed to cancer, alcoholism, obesity, baldness and who knows what else.... Hand that information to an employer and imagine the implications."

The Genetic Information Nondiscrimination Act (GINA) would prevent employers from requesting employees' genetic information, and employers and insurance companies from discriminating against a person based on the content of their genetic code. In effect, it would give people like Curry the freedom to submit to a potentially lifesaving test without fear of reprisal. Last April the House of Representatives voted 420-3 to pass the bill, and President George W. Bush has already said that he will sign it should it clear the Senate. But Senator Tom Coburn (R., Okla.), a physician who voted for a similar version of the bill that passed the Senate 98-0 in 2005 (but never made it to a vote in the House), has placed a "hold" on the latest GINA, forestalling a vote. Coburn initially argued for specific language ensuring the rights of the fetus. The language of the bill was altered to that end, but his hold persists.

Coburn says that he supports a genetic nondiscrimination law, but that the current version of GINA does not provide enough protection for employers. "What if an employee files a form to take family leave from work and they write on the form that they have to take care of their mother who has breast cancer?" he says. Because breast cancer can be hereditary, "the employer may have gathered genetic information accidentally, and they are vulnerable in a lawsuit. We need protection for [employers and insurance companies] who don't mean to discriminate, but have accidentally collected information."

As far as Christine Seidman is concerned, the passage of GINA, which was first introduced in 2002, is overdue. "Americans have to support this," she says. "I have Alzheimer's and breast cancer in my family. You may have schizophrenia. We all have something. I believe that families have a right to know everything they can."

But should all patients know everything about their genome? Even experts aren't certain. "I see some kids, and they don't have a family history of death and they don't have symptoms or a very thick heart, and I don't think a lot of them are at great risk," says Thompson, a former marathoner who competed in the 1972 Olympic trials. "I usually say to them, 'I don't think you're at great risk, but I have to sleep at night, and I can't take a chance with you, so I'm prohibiting you.' For some acne-stained 17-year-old who's accepted at that high school because he's a good linebacker, to tell him that's gone is a load."

The most contentious case of HCM in sports is the one that Nick Knapp is sure he never had. When Knapp rises from behind his desk in Peoria, Ill., where he works as a financial consultant, it's not hard to imagine him having once been one of the top high school basketball players in the state. Even now, eight years removed from his playing days, he is, at 6' 5", an erector set of a man, his sharp jaw line leading to a tautly muscled neck that tapers to his bulging shoulders.

In September 1994, Knapp was on his way to breaking the state record for career three-pointers when his heart stopped during a pickup game at Woodruff High in Peoria. He was revived, had an ICD placed inside his chest and two months later spurned more prestigious programs to accept a basketball scholarship to Northwestern. When he arrived in Evanston, however, the school's doctors, concerned that he might have a fatal heart condition, declared him ineligible. Knapp's left ventricle was slightly enlarged, and cardiologists were divided on whether he had the disease. Knapp himself doubted that he had HCM and sued the university for the right to play. Keeping him off the court, Knapp argued in his lawsuit, violated the 1973 Rehabilitation Act, which protects "otherwise qualified individuals" from discrimination based on a disability.

Knapp won in federal court, but Northwestern got the ruling overturned by the U.S. Court of Appeals for the Seventh Circuit. The court acknowledged that Knapp could play college ball, but that Northwestern also had the right to heed its doctors and to prevent him from potentially risking his own death at its facilities. Though the university honored his scholarship, Knapp, who sat out for two seasons during the litigation, transferred to Northeastern Illinois, then to Division II Ashland (Ohio) University. He still ponders what might have been. "I've played against guys who went to the NBA, like Brian Cardinal and Stephon Marbury," Knapp says, "and I was at that level."

Since Knapp's ordeal, the use of implanted defibrillators has become more common in the population at large, and doctors have permitted some athletes with ICDs to compete. In January 2006, Washington guard Kayla Burt, who has Long Q-T syndrome, a genetic heart disease that, like HCM, is treated with an ICD, was sitting on the bench in a game against UCLA when her device fired. "She thought someone came up behind and punched her in the head," recalls team trainer Jenn Ratcliff. The shock "usually gets a 10 out of 10 on the pain scale from patients," says Dr. Mark Estes, an electrophysiologist and director of the New England Cardiac Arrhythmia Center. Burt, who had nearly died on New Year's Eve 2002, when she went into cardiac arrest, gave up playing for good that night. "The device worked, which is good, but not exposing yourself to potentially lethal cardiac arrhythmias is a good choice," Estes says, adding that an ICD's wires are not made to withstand the strain of sports. "The leads can be damaged," he says. "I had one 20-year-old woman who destroyed a lead just doing aerobics."

How well implanted defibrillators hold up during sports is one of the many questions surrounding HCM. There are still mutations to find. There is still much that is not known about the progression of the disease -- when and

how the cellular disarray develops, for example -- largely because a live heart can't be put under a microscope. There is still no explanation for why Eileen Kogut's two brothers died but she, with the same HCM mutation, leads a normal life. Says Salberg, "Somewhere between screening every kid in America for a couple of billion dollars and not doing anything lies the right answer."

DeCarlo Polk's mom, Tommie (Lady) Polk, knows a lot about HCM now, but she does not have any more children to lose. DeCarlo was her baby, and she clings to his memory as if trying to keep a balloon from drifting into the sky. There are mementos around every corner in her yellow clapboard house in Durham: in the family room DeCarlo's first football shoes, burgundy-and-yellow Redskins sneakers that fit in the palm of Lady's hand; in his bedroom the folded New Orleans Saints replica jersey with Reggie Bush's number 25 stitched on the back that he never got to wear. She keeps the bedroom door locked and the fan continually running. "I can smell him when I'm in here," she says.

If only she had known about HCM, Lady Polk says. If only the woman down the street had spoken up when her son had died from the disease, Lady would have gladly paid for the tests to make sure her baby was safe. As for his football scholarship? She never counted on that anyway. "DeCarlo could have carried water for the team," she says. "He wasn't born to play sports."

DeCarlo was more than Lady's only son; he was her guide in a noisy world. In the last few years Lady's hearing has deteriorated; nearly deaf, she had to leave her teaching job. "I couldn't keep up with kids how I knew I should," she says. Nick Knapp had to adjust to life without basketball, and Lady is just beginning life without something far more precious. Most of the time she is alone, in her silence.

<http://sportsillustrated.cnn.com/2007/more/12/04/broken.hearts.1210/index.html>