University of Minnesota Physicians Heart
Adult Congenital and Cardiovascular Genetics Center
When the young man met with Renuka Jain, MD, Director of the Cardiovascular Genetics Clinic at University of Minnesota (UM) Physicians Heart at Fairview, he brought his sister’s autopsy records.

“Heart failure killed my sister and my dad,” he told Dr. Jain. “How do I prevent this from happening to me?”

Historically, congenital heart disease has been the purview of pediatrics. Few patients survived into adulthood. But advances in surgical options, genetic testing and cardiac care have gradually improved patient outcomes. In 2005, for the first time, there were more adults than children living with congenital heart disease. Today, there are an estimated 1 million adult patients with congenital heart disease in the United States. This new patient demographic requires care that cannot be provided in the pediatric environment and has spurred the development of a subspecialty: adult congenital heart care.

With a 60-year history distinguished by pioneering treatment in heart care, the University of Minnesota Physicians Heart Adult Congenital and Cardiovascular Genetics Center offers the most comprehensive heart care in the Twin Cities for the changing face of congenital heart disease.

“The University of Minnesota performed many of the firsts in congenital heart disease,” explains Cindy Martin, MD, Co-Director of the Adult Congenital and Cardiovascular Genetics Center. “The first heart-lung machine, the first successful ventricular septal defect repair, the first atrioventricular canal repair, the first tetralogy of fallot repair, the first wearable transistorized pacemaker; all of these and others were initiated here at the university. We provide our patients with a substantial depth and breadth of congenital expertise.”

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“Are there estimated to be fewer than 50 formally trained adult congenital subspecialists in the country,” explains Dr. Martin. “Board certification for this training is still in progress, but data already indicates that when adult congenital surgery is performed by an adult congenital-trained surgeon, outcomes are significantly better.”

One of these formally trained specialists, Kimara March, MD, is a member of the UMPhysicians Heart adult congenital heart team. Both pediatric and adult cardiologists further enhance the team’s expertise. Pediatric cardiologists bring experience with congenital heart disease to the table. Adult cardiologists enhance congenital knowledge with skills in assessing and treating adult coronary artery disease and inherited cardiomyopathies. Finally, the UMPhysicians Heart program is the only one in the Twin Cities with a cardiac geneticist and certified genetic counselor on staff. This depth of expertise draws patients from across the upper Midwest.

Congenital disease often requires collaborative treatment with specialists in electrophysiology, structural catheterization, heart transplantation and pulmonary hypertension. Congenital heart disorders...
also affect kidneys, pulmonary status, contraception options, pregnancy and psychosocial conditions. The university medical setting provides the adult congenital heart program with access to these additional specialists to address the entire spectrum of adult congenital cardiac complexities.

Most first-time patients have a joint consultation with a pediatric physician and a physician who treats adults. “We feel it is important for a new patient to meet two members of the team to establish familiarity and continuity of care,” Dr. Martin says. “Information gleaned during the initial exam is very important during subsequent visits. Joint exams enable us to pool our pediatric congenital and adult cardiology knowledge. Finally, a pediatrician’s patient interaction is very different from that of an adult cardiologist. Pediatric patients don’t make medical decisions—their families make the decisions. Our adult patients may not be fully aware of their medical history through childhood and are also confronting a range of new choices regarding contraception and pregnancy.”

Normal heart disease benefits from treatment guidelines determined by extensive trial data in very large patient populations. There is less clinical data to guide the treatment of patients with congenital heart disease and many inherited cardiomyopathies because the patient group has only recently emerged, and the number of patients with each specific condition is small.

“We have guidelines based on expert consensus for certain adult congenital conditions,” explains Dr. March, Director of Adult Congenital Heart Disease Clinics at UMPhysicians Heart. “In some situations, there are treatment nuances for specific conditions that are a part of formal adult congenital training. But clinical trials and basic clinical research are absolutely part of our program. We are also creating patient registries with long-term follow-up to better understand outcomes.”

One of the services distinguishing the comprehensive care offered by the Adult Congenital Heart program is an emphasis on looking beyond the individual patient to assess family members as well. When a patient is diagnosed with a medical problem related to the heart, physicians might not be aware that the patient’s condition may be heritable, possibly impacting the health of other family members. This is where genetic testing can be helpful.

“Awareness of genetic issues affecting family as well as the patient is not yet widespread,” Dr. March explains. “For instance, there’s a 20 percent chance of heritability in first-degree relatives for patients with a bicuspid aortic valve. That inherited trait could manifest in a valve problem or enlargement of the aorta, known as an aneurysm. Aneurysms are silent killers and need to be found before it is too late.”

Recently, studies have shown 30–40 percent of basic or idiopathic heart failures (whose cause was previously unknown) are actually familial cardiomyopathies caused by genetic issues. Physicians in the program bring this awareness to gathering family history information.

“There are a lot of questions to be asked in addition to finding out whether someone else in the patient’s family has a heart problem,” explains Dr. Martin. “We ask about deaths that may have arisen from a sudden cardiac event, such as a drowning, a single motor vehicle accident or a fall. Any of these could signal an undiagnosed genetic cardiac problem. Unless you ask the question specifically, you may not recognize the potential problem. If an aunt developed heart failure, it’s important to know her age. A 75-year-old aunt with heart failure reveals a very different story than a 30-year-old aunt with heart failure. Families may have very valuable information, but it takes specific awareness, focused questions and the luxury of
time to tease out the familial component. When patients do have genetics-based issues, their families are also at risk, and it’s recommended that all first-degree relatives be screened.”

Another feature of the UMPhysicians Heart Adult Congenital and Cardiovascular Genetics Center is cardiovascular genetic testing.

“Our genetic testing serves the larger patient population born with inherited cardiomyopathic conditions that either cause a cardiac issue or make these patients more susceptible to developing heart problems,” says Dr. Jain. “Gene testing has evolved significantly during the last decade, but results are not always black and white. We’ve identified some genetic mutations, but a negative test isn’t necessarily conclusive — there may be mutations that testing does not yet identify. Other genetic conditions pose a predisposition that may not be a patient’s presenting problem but place him or her at risk for morbidity or mortality. Our specialists understand the risks and know how to screen for them. We have the expertise to interpret nuanced test results and provide education for the patient and family.”

Like the congenital cardiology program, the cardiovascular genetics program includes meticulous detective work into the patient’s family history.

“As soon as we receive a call from a referring physician, and often before our first meeting with a patient who has a possible familial cardiomyopathy, I look at medical histories, autopsy reports and slides, and recreate a comprehensive family history,” Dr. Jain explains. “Compiling a patient history is the best part of my job. Healthcare professionals are so pressed for time, and many of our patients are unaware of the history of their pediatric care and its implications. It is our priority to sit down with patients and review their entire histories. Patients feel really appreciative when we are able to talk with them not only about their own conditions, but also about how to help their family members.”

Addressing congenital heart conditions and inherited traits can prompt anxiety, trepidation and guilt in patients and their families. The team’s certified genetic counselor is critical in supporting families and addressing their concerns.

“Our entire team supports each patient, and our genetics counselor is an invaluable resource,” explains Dr. Jain. “It’s important to counsel patients before they make a decision about gene testing, and they need counseling afterward as well. Our counselor is formally trained in meeting with families and is able to explain the underlying genetics and how they translate into a patient’s condition.”

Some of the greatest risks for this patient population arise from misinformation. The UMPhysicians Heart adult congenital cardiology team emphasizes that congenital heart disease is never “cured.” These patients require lifelong follow-up.

“The transition of care from childhood to adulthood is very important,” notes Dr. Martin. “It is well-documented that these patients are most frequently lost to medical follow-up in the teen-to-early-20s age range, when responsibility for medical care shifts from their parents to them. Particularly if these patients believe their childhood issues were cured, they discontinue follow-up only to later run into trouble that could have been prevented with regular monitoring.”

Even with advances in treatment and testing, the average age of survival for a patient with congenital heart disease is less than 40.

“If we diagnose these patients early enough, we really can improve outcomes,” says Dr. March. “The guidelines specify that every congenital patient should be seen at least once by an adult congenital specialist. Twenty years ago, many of these patients were told as children that they were cured. That is not the case. We now know that congenital heart disease must be monitored for the life of the patient. We work with referring physicians to gather the patient’s records and operative reports, confirm the patient’s entire medical history, and ensure he or she is being properly managed and things are not missed. Educating patients about their original anatomy and how they were repaired is also an important component of the visit, as this empowers them to take control of their health.”

“Working closely with referring physicians is a reflection of the University of Minnesota Physicians Heart mission,” states Dr. Jain. “We are here as a resource to answer questions and to offer the full spectrum of subspecialty congenital heart care. When patients come to us early, like the young man with his sister’s autopsy records, we can’t always prevent heart failure, but frequently, we can help these patients to avoid a crisis.”

“We see our role as partnering with referring physicians to help provide specialized care to these patients,” adds Dr. Martin. “We are intimately familiar with the complexities of congenital cardiology patients, and we offer a breadth of subspecialty resources to improve their health care and outcomes. It can be a huge comfort to these patients to be treated in a center where they feel a sense of normalcy. Together, our goal is to help these patients and to support their families.”