Detecting the Often Hidden Threat of Adult Congenital Heart Disease

Neither the 41-year-old woman nor her primary care physician connected the irregularity that turned up in a routine checkup to the woman’s coarctation of the aorta, which surgeons had corrected when she was a toddler. However, cardiologist Elyse Foster, MD, had seen these cases before, as had her team at the UCSF Adult Congenital Heart Disease (ACHD) Clinic, which Foster led for many years.

“The woman had a large aneurysm at the repair site, about 7.8 centimeters in diameter, way beyond something we just follow,” says team member and heart surgeon Tara Karamlou, MD. Karamlou partnered with UCSF Chief of Vascular and Endovascular Surgery Michael Conte, MD, to excise the segment and insert an interposition graft that significantly reduced the woman’s risk for sudden cardiac death.

“These sequelae are more common than many physicians realize,” says Foster.

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This issue of *UCSF Heart and Vascular News* powerfully illustrates how to improve the health of entire populations by joining the finest clinicians with world-class science and technology.

For example, two stories on our adult congenital heart disease clinic showcase work that helps unsuspecting adults — who falsely believe their heart disease is a childhood memory — prevent adult symptoms from becoming lethal; pregnant women are particularly at risk. At UCSF, a team of renowned cardiologists, pediatric heart surgeons and vascular surgeons deliver the very best care from a full range of treatment options.

A second story examines the challenge of diagnosing and treating women with peripheral artery disease (PAD). Because women tend to be asymptomatic or present without typical symptoms, experts here partner with community physicians to detect PAD as early as possible; our UCSF Center for Limb Preservation can offer alternatives to amputation that others cannot.

Another story highlights our ambitious Health eHeart study, which uses mobile and Internet technology to monitor and collect unprecedented amounts of data on people with heart disease or at risk for it. The massive study is a unique opportunity to leverage technology for new discoveries that will enable more precise, efficient and effective care.

Finally, we discuss the value of a cardiovascular genetics program, which uses scientific and technological advances to help families more precisely understand their risks for sudden cardiac death, take preventive measures and, occasionally, receive more tailored treatments.

Ongoing advances depend on these ongoing collaborations — including those with you, our community partners. As always, we appreciate your referrals.

*Jeffrey Olgin, MD*
*Chief of Cardiology*

*Michael Conte, MD*
*Chief of Vascular and Endovascular Surgery*

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**Screening Essential, Especially for Women Considering Pregnancy**

Due to surgical and medical advances, at least 1.3 million adults in the United States are living with congenital heart disease. Although many of these patients live healthy adult lives, others develop significant late complications. They may be unaware that the condition they believed was healed has silently grown into a life-threatening condition.

Any adult who had a congenital heart problem as a child, even a minor one, should have at least one screening visit at a center that specializes in this condition, says cardiologist Ian Harris, MD, who now directs the ACHD Clinic. “If you’re not an adult congenital cardiologist, you may not be in tune with what symptoms or signs to look for.”

Aneurysms, abnormal heart rhythms, heart failure or deterioration of the original repair are among the possibilities. Those who required surgical repair as a child are especially at risk, as are women considering pregnancy.
The types of sequelae we see after surgery may be asymptomatic for a long time, but once pregnant, the increased load puts these women at risk for problems, which can be catastrophic,” says Foster. “We work with perinatologists here to evaluate patients before pregnancy, help anticipate problems, provide contraceptive counseling and manage patients through the pregnancy.”

Diverse Expertise, Experience and Technology
The UCSF ACHD Clinic brings together expert cardiologists, heart and vascular surgeons and other health professionals with deep experience diagnosing and treating adults with congenital heart disease. The clinic also offers:

- Advanced imaging techniques
- Diagnostic and interventional catheterization
- Open, percutaneous and hybrid surgeries
- Heart and lung transplantation

“Management for all patients depends on the complexity of the lesion, but our goal is to help our community colleagues by following patients closely where necessary and providing consultative services and advice wherever possible,” says Harris. ✧

WHEN TO REFER
According to current guidelines from the American Heart Association and the American College of Cardiology, when patients transition from their pediatric cardiologist, they should be referred to a regional adult congenital heart disease center, such as the one at UCSF. As per the recommendations, we partner with primary care physicians and referring cardiologists to provide care for these patients.

To contact Dr. Foster or Dr. Harris, call (415) 353-2873.
To contact Dr. Karamlou, call (415) 476-3501.
When he was born in New Jersey in the 1950s, Mark Willey was a 3-pound preemie with congenital pulmonic stenosis and numerous holes in his heart. He spent a good portion of his early childhood in and out of various hospitals. Then, at age 7, he had what at the time was a rare open-heart surgery to fix his heart defect. No one expected him to live past his late 20s.

Instead, Willey became a high school athlete and, later, moved to California, where he remained physically active and asymptomatic even as he became CEO and managing partner of a general aviation company.

Symptoms, Stroke and Referral to UCSF

In Willey’s early 40s, however, symptoms appeared and physicians discovered an arrhythmia. After a failed ablation attempt, the medication flecainide seemed to work until one day, while driving, Willey suffered what he believes was a small stroke. He managed to pull over and stumble into a convenience store, where aspirin and rest eased the symptoms.

When his local cardiologist ordered an echocardiogram, the technician was shocked by Willey’s enlarged heart and other abnormalities. Subsequently, the cardiologist and Willey’s primary care physician helped Willey research centers that specialize in adult congenital heart disease and, ultimately, recommended UCSF. There, Willey met with cardiologist Ian Harris, MD, who runs the UCSF Adult Congenital Heart Disease (ACHD) Clinic.

“He had severe regurgitation through his pulmonic valve that was overloading his right ventricle,” says Harris, who notes that lesions like Willey’s are surprisingly common. “They slowly progress, and if nobody notices, they become symptomatic and potentially lethal.”

Treatment

Today, the common first-line treatment for Willey’s condition is percutaneous valve replacement with the Melody valve, or perhaps a hybrid approach. Research indicates that surgeons and interventional cardiologists with expertise in congenital heart disease and access to advanced imaging services generally achieve the best outcomes.

The team at UCSF recommended catheterization, and once in, they discovered more than a leaky valve: they found a significant hole in the heart’s upper chamber. A team of experts reviewed the case and recommended that pediatric heart surgeon Anthony Azakie, MD, perform an open-heart procedure.

“Because of the scar tissue, it was a long and difficult surgery,” says Willey. “But ultimately, Dr. Azakie and his team sewed the hole and replaced the valve.”

Today, except for a slight murmur, Willey’s arrhythmia is gone, he’s off his arrhythmia medication (except aspirin), and his enlarged heart has shrunk. “I don’t have the stamina or body strength I had,” he says, but he still runs five to seven miles, three to five days a week and is back at work full time.

“People who’ve had heart surgery early in life should be followed by a specialized clinic,” says Willey. “I realize I’ll face other hurdles, but for this one the expertise of Drs. Harris and Azakie kept me alive…. And it’s more than what they do; it’s who they are – the whole staff at UCSF are truly amazing and caring.”

From left to right: Mark Willey, his daughter Ariana, son Steven and wife Jeanette

To contact Dr. Harris, call (415) 353-2873.
WOMEN AND CARDIOVASCULAR DISEASE

Heart disease is the number one killer of women in the United States. Yet as with PAD, many women – and some physicians – remain unaware of this fact.

“For both men and women, the most common symptom of a heart attack is the same: chest discomfort, usually described as tightness, heaviness or a pressure sensation. However, women have some risk factors and symptoms that may be somewhat different from those in men,” says Anne Thorson, MD, co-director of the UCSF Center for Prevention of Heart and Vascular Disease at Mission Bay and director of its Women’s Cardiovascular Care Program.

Women are more likely to experience:
- Shortness of breath
- Nausea
- Vomiting
- Pain that goes to the neck or jaw or radiates through to the back
- Weakness
- A heartburn sensation related to exertion rather than to eating

Thorson says physicians should refer at the first appearance of these warning signs and that increasing awareness of the symptoms – along with careful monitoring of risk factors and making important lifestyle changes – are the keys to reducing the death rate and improving these women’s lives.

To contact Dr. Thorson, call (415) 353-2873.

Late Diagnosis of PAD in Women Increases Risks

Though the American Heart Association and others have called for raised awareness, too many physicians remain unaware that the prevalence of peripheral artery disease (PAD) in women is equal to or higher than that in men, says vascular surgeon Jade Hiramoto, MD, of the UCSF Center for Limb Preservation (www.ucsfhealth.org/limbpreservation).

She adds that because the condition tends to be asymptomatic in women or present differently than it does in men – most notably, women are less likely than men to present with typical symptoms such as claudication – women are typically diagnosed at an older age, when the disease is more advanced and the patient more likely to require costly and invasive procedures.

“That’s why early detection is so important,” says Hiramoto, who is performing research to better characterize women’s unique risk factors and symptomology.

“If there is any reason to suspect PAD, physicians should refer these women for screening, because as with any disease, if we catch it early, treatment is more effective,” says Hiramoto.

Managing the Increased Risk for Amputation

Without early detection, she says, it’s not uncommon for the first presenting symptom in women to be pain at rest or a nonhealing foot wound. “Too often, this necessitates invasive treatment, and women, for unclear reasons, tend to do worse after revascularization compared to men.”

If revascularization is not an alternative or fails, amputation is the next option, though Hiramoto says it is more avoidable than commonly believed.

“By bringing together a team of experts – including vascular surgeons, podiatrists and other specialists – we can often offer alternatives for maintaining a functional limb that others cannot,” she says.

To contact Dr. Hiramoto, call (415) 353-2357.

To make a referral to the UCSF Center for Limb Preservation, call nurse practitioner Joanna DeLong at (415) 353-2256.
Today’s pervasive use of mobile technologies holds the potential for heart disease patients to take better charge of their care while speeding and enhancing research efforts, says Jeffrey Olgin, MD, chief of cardiology at UCSF Medical Center. Olgin, UCSF cardiologist Greg Marcus, MD, and UCSF cardiovascular epidemiologist Mark Pletcher, MD, lead an online study that uses mobile and Internet technology to monitor and collect unprecedented amounts of data on people with heart disease or at risk for heart disease.

Olgin believes that by enrolling 1 million Americans in the Health eHeart Study, which he calls a “Framingham 2.0,” the country has a unique opportunity to harness so-called big data to generate new discoveries about the country’s biggest killer while delivering more precise, efficient care that improves outcomes and decreases costs.

**Research and Enhanced Disease Management**

Health eHeart is a UCSF initiative that transforms clinical research by eliminating the need for in-person visits required for the more typical brick-and-mortar research project. To optimize its impact, the study needs community physicians to encourage enrollment from a wide range of patients, with or without heart disease.

Participants use mobile technologies and sensors, including apps developed for the study, integrated with commercially available sensors (such as activity monitors, Bluetooth-enabled blood pressure cuffs and scales, electrocardiogram electrodes built right into smartphone cases and new ones under development). By leveraging a simple, engaging approach to collecting survey data, the study will collect real-time and real-life measures that will contribute terabytes of data that researchers can analyze to glean more specific insights than ever before into heart disease risk factors and the efficacy of prevention and treatment techniques.

“One of the aims of the study is to develop tools that let patients better understand when they’re sick or at risk, eventually leading to more timely and targeted prevention and treatment; think of how many heart failure patients are readmitted because they miss early-warning signs,” says Olgin. “We also hope it will help get patients to change unhealthy behaviors.”

Another aim of the study is to develop new disease management strategies, allowing for more cost-effective and better health care delivery. By developing scalable approaches to monitoring with the now ubiquitous smartphone, clinicians may be able to restrict office-based appointments to those patients most in need.

Recently, Olgin and the Health eHeart Study have teamed up with the American Heart Association to spread the word and leverage the AHA’s expansive network and influence to enroll participants.

**Developing and Validating mHealth Technologies**

UCSF clinicians and scientists are also developing and validating the technologies used in the study, including a powerful, data-secure platform, new sensors, and web and mobile applications.

“Most of what we know about predicting and treating heart disease comes from older studies,” says Olgin. “This is a new type of clinical research. It’s cheaper, allows us to respond quicker and study many more people and factors – and help patients at the same time. I strongly encourage physicians to ask their patients to enroll.”

For more information or to join the study, visit www.health-eheartstudy.org. To contact Dr. Olgin, call (415) 476-1325.
Cardiovascular Genetics Advances Clinical Care

By leveraging ongoing scientific discoveries and technological advances, a cardiovascular genetics program can clarify the risks and diagnoses associated with sudden cardiac death in ways that clinical screening alone cannot.

“In turn, patients and families can take proper preventive measures and, on occasion, receive more tailored treatments that can reverse or slow their disease,” says Melvin Scheinman, MD, of the Cardiovascular Genetics Program at UCSF Medical Center, one of only a few such centers on the West Coast.

“The spectrum of disease within a given genotype has broadened,” says UCSF cardiologist Rahul Deo, MD, PhD. “A center like ours, which offers extensive genetic testing and counseling, can widen the search to clarify clinical ambiguities.”

“The familial nature, variability and treatability of these conditions make it essential to screen patients with inherited heart disorders and their family members as early as possible,” says genetic counselor Julianne Wojciak.

Genetic Arrhythmias

In the last few years, clinical research and improved understanding of the connection between life-threatening arrhythmias and complicated genetic mutations have changed some approaches to diagnosis and treatment. For example:

- The discovery of new genes associated with long QT syndrome (LQTS) has made it reasonable to retest people in whom previous testing did not identify the responsible genetic mutation.
- Flecainide has been found to be effective for controlling difficult arrhythmias in catecholaminergic polymorphic ventricular tachycardia (CPVT).
- The discovery that early repolarization syndrome has a genetic basis makes its treatment different from those for other conditions that produce sudden cardiac death.
- Based on encouraging results in animal studies, UCSF is testing whether decongestive drug regimens for arrhythmogenic right ventricular cardiomyopathy (ARVC) can decrease the risk for an enlarged ventricle.

“Centers like ours use this information for enhanced decision support,” says Scheinman. “For example, if we have somebody who is typically classified as being in an intermediate risk range, but we know the genotype is less responsive to beta-blockers, we might feel more compelled to offer an ICD [implantable cardioverter defibrillator].”

Familial Cardiomyopathy

Cardiomyopathy is another key focus, especially as parents have become more aware of it as a factor in the death of many young athletes.

CONTINUED ON BACK COVER
PHYSICIAN LIAISON SERVICE
Tel: (800) 444-2559   Fax: (415) 353-4395
www.ucsfhealth.org

Physician liaisons visit referring physicians and practice representatives to learn more about their referral needs and to provide information about the services available at UCSF Medical Center.

TRANSFER CENTER
Tel: (415) 353-9166   Fax: (415) 353-9172

The UCSF Transfer Center is staffed 24/7 by a specialized team to evaluate the clinical needs of your patient to ensure the most appropriate medical care is provided and to coordinate transfer and transport from hospitals throughout the region.

HEART AND VASCULAR CENTER EMERGENT TRANSFER PROTOCOL

Possible diagnoses for emergent Heart and Vascular Center transfer (priority bed):

- Acute aortic syndromes: dissection, symptomatic abdominal or thoracic aneurysm
- Acute visceral or limb-threatening lower-extremity ischemia
- Cardiogenic shock or severe heart failure requiring evaluation for ventricular assist device or heart transplant
- Severe lung failure requiring evaluation for extra-corporal life support (ECLS) or lung transplant
- ST-segment elevation myocardial infarction
- Ventricular tachycardia

For questions about the Emergent Transfer Protocol, or to transfer a patient, call the UCSF Transfer Center at (415) 353-91663.

To reach the practices directly, please call the numbers listed below:

- Cardiothoracic Surgery (415) 353-1606
- Center for Limb Preservation (415) 353-2256
- Electrophysiology (415) 353-2554
- General Cardiology (415) 353-2873
- Heart Failure (415) 514-8866
- Vascular and Endovascular Surgery (415) 353-2256

CARDIOVASCULAR GENETICS
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The program at UCSF combines arrhythmia monitoring, exercise testing and genetic testing to assess the risk of sudden cardiac death.

“We can often identify the pathologic variant responsible,” says Deo. “This enables us to identify additional family members at risk and focus prevention efforts, whether it’s modifying sports choices or counseling about risky behaviors.”

In conjunction with UCSF perinatologists, the center also helps women with congenital heart disorders manage their high-risk pregnancies. For patients whose symptoms and/or disease are worsening, the center is a gateway to a full range of potential therapies, from alcohol septal ablation and septal myectomy to transplant.

Finally, the center has an active, wide-ranging research program that supports its clinical efforts.

To contact the Cardiovascular Genetics Program, call (415) 476-3202.