CONGENITAL HEART VALVE DISEASE

What 8 Questions Should You Ask?

Cedars Sinai
Guerin Children's

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Introduction:
A Welcome Note from Adam Pick

Dear Patients and/or Parents,

As I personally learned after my diagnosis at seven years old, being told that you, or someone you love, has congenital heart valve disease can bring up a lot of questions and concerns. This guide is designed to walk you through eight of the most important questions so you can feel more confident and prepared in conversations with your medical team.

Throughout this book, you'll find accurate and encouraging information grounded in current medical research — along with practical tips on what to watch for, what to ask, and how to plan for the lifetime management of heart valve disease.

You don't need to read this book cover-to-cover. Each chapter stands on its own, so feel free to jump to the question that matters most to you right now and come back to the others when you're ready.

If you need anything, please do not hesitate to contact me. I'm here to help!

Adam Pick
Congenital Heart Valve Disease Patient
HeartValveSurgery.com Founder

Question #1

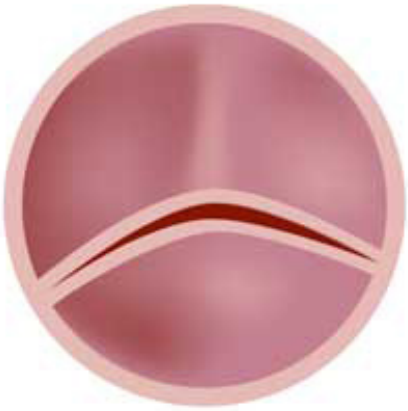
What is Congenital Heart Valve Disease?

Congenital heart valve disease is a condition you are born with, meaning one or more of your heart valves did not form normally before birth. The heart has four valves that act like doors to keep blood moving in the right direction. When a valve is affected, it may be too narrow (stenosis), may leak (regurgitation), or may have an unusual structure — such as a bicuspid valve with two flaps instead of three.

(Source: Adobe)

This condition is more common than many people realize. Overall, congenital heart disease affects about 1 in every 100 babies. Among valve-specific conditions, bicuspid aortic valve (BAV) is the most common — occurring in about 1% to 2% of the population.

Normal tricuspid valve



Bicuspid aortic valve

As shown above, a bicuspid aortic valve is a congenital heart condition in which the aortic valve has only two leaflets (cusps) instead of the normal three, which can affect how blood flows from the heart to the body. Because some valve abnormalities don't cause symptoms early on, many people are not diagnosed until adolescence or adulthood.

Thanks to advances in pediatric cardiology and surgery, about 90% of children born with congenital heart defects survive into adulthood. As a result, the adult congenital heart disease population (ACHD) is rapidly growing. Reports suggest that approximately 2 million adults in the United States are living with ACHD.

“Patients with congenital heart defects were born with it. It’s not something that anybody did or was caused by medications. Heart valve defects are present at the time of birth.”

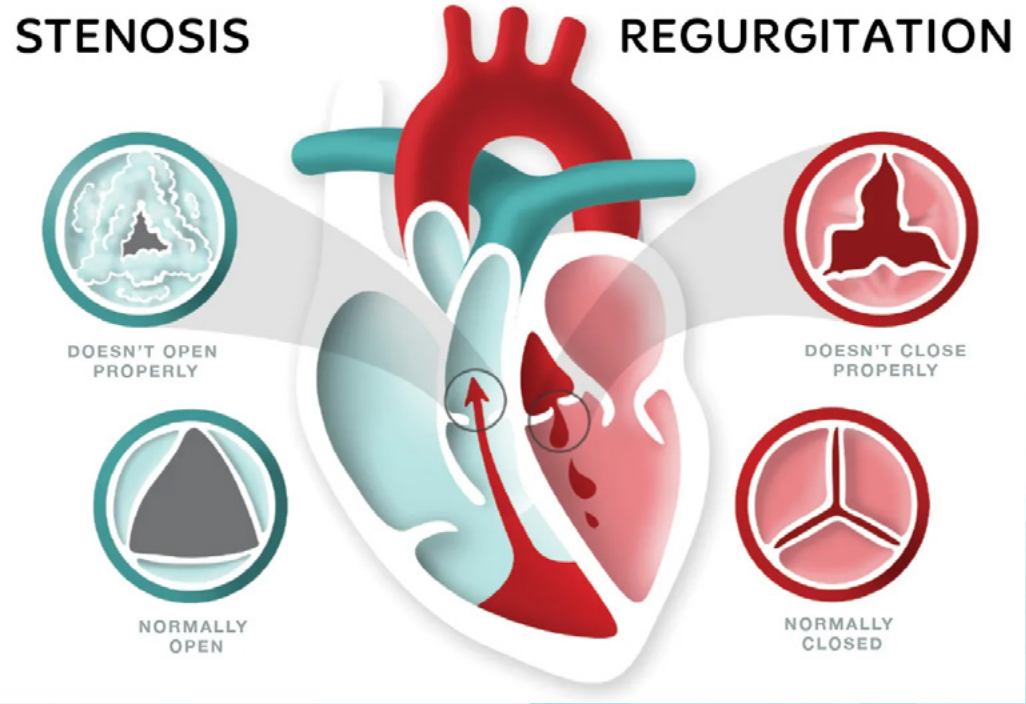
Dr. Melita Viegas
Associate Director, Pediatric and Congenital Cardiac Surgery, Guerin Family Congenital Heart Program, Cedars-Sinai (Los Angeles, California)

Question #2:

What Are the Different Types of Congenital Heart Valve Defects?

Congenital heart valve disease can be very complex to manage and treat. That said, it's very important to understand exactly what type of problem you or loved one may have. In general, most valve issues fall into two main categories: stenosis and regurgitation.

- **Stenosis** means the valve is too narrow and doesn't open fully, which can restrict blood flow.
- **Regurgitation** means the valve is leaky and doesn't close properly, allowing blood to flow backward instead of moving forward efficiently.



(Source: ResearchGate)

In some cases, patients may actually have a combination of both stenosis and regurgitation. Understanding which valve defect you or your loved one has will help you better understand the symptoms the treatment options. For example, stenosis often causes the heart to work harder to push blood forward, while regurgitation can cause the heart to enlarge over time due to extra volume.

Some patients with congenital valve disease may also develop related heart conditions, such as atrial fibrillation, aortic aneurysm, or pulmonary hypertension — which is why ongoing monitoring of the heart is so important.

Severity of Congenital Heart Valve Disease

Congenital heart valve defects vary widely in severity, and clinicians typically group them as mild, moderate, or severe based on how much they disrupt normal blood flow and strain the heart.

- **Mild defects** often involve small structural abnormalities—such as slight valve narrowing (stenosis) or minor leakage (regurgitation)—that do not significantly impair circulation.
- **Moderate defects** cause more noticeable disruption in blood flow and may lead to symptoms like fatigue, shortness of breath, or reduced exercise tolerance. These cases often require closer monitoring and sometimes medical treatment to manage symptoms or prevent complications.
- **Severe defects**, on the other hand, significantly impair the heart's ability to pump blood effectively and can lead to serious consequences such as heart failure, growth problems in children, or life-threatening complications if untreated.

Question 3:

What Are the Symptoms?

Some people with congenital valve disease feel completely normal, while others may develop symptoms gradually. It's important to learn what changes in your body could signal that your condition is progressing or needs attention. Even subtle changes — like feeling more tired than usual or getting winded more easily — can be meaningful and should not be ignored.

Key symptoms to watch for include:

- Shortness of breath (especially during activity or when lying down).
- Fatigue or reduced energy levels.
- Decreased ability to exercise or keep up with normal activities.
- Chest pain or chest discomfort.

- Heart palpitations or irregular heartbeat.
- Dizziness or lightheadedness.
- Fainting (syncope).
- Swelling in the legs, ankles, or feet.

Monitoring is symptoms is not just helpful, it can be life-saving. Clinical research, including findings published in the American Heart Association's medical journal Circulation, shows that once patients with severe aortic stenosis develop symptoms, the disease can become very dangerous. **In fact, approximately 50% of patients with symptomatic severe aortic stenosis do not survive beyond two years without treatment**. This is why recognizing symptoms early — and acting on them — is so critical.

Your doctor should give you clear guidance on when to call. Don't wait if you have concerns. Early communication can prevent complications and ensure you receive timely care when it matters most.

Question #4:

When Do Patients Get Treatment?

Not every congenital heart valve condition requires immediate treatment. Many patients are safely managed with a **"watchful waiting"** approach, which means your care team closely monitors your heart over time to determine if and when intervention is needed. This strategy helps avoid unnecessary procedures while still protecting your long-term health.

To monitor your condition, doctors use several important diagnostic tests that provide detailed information about your heart valves and overall function. The most common and important test is the echocardiogram (ultrasound of the heart), but other imaging tools may also be used depending on your condition. These tests help your care team track changes and make timely decisions.

(Source: Cardiac X)

Common diagnostic tests include:

- Echocardiogram (Echo): The most important test; uses ultrasound to evaluate valve structure and blood flow.
- Transesophageal Echocardiogram (TEE): A more detailed ultrasound performed through the esophagus.
- Cardiac CT Scan: Provides high-resolution images of the heart and surrounding structures.
- Cardiac MRI: Offers detailed information about heart function and blood flow.
- Electrocardiogram (ECG/EKG): Measures the heart's electrical activity and rhythm.
- Stress Testing: Evaluates how your heart performs during physical activity
- Cardiac Catheterization: Invasive test used in certain cases to measure pressures and assess severity.

Regular annual imaging, also known as yearly surveillance, is critical because valve disease can change — even if you feel fine. For many patients with congenital valve disease, annual echocardiograms are the cornerstone of care. **Yearly checkups allow your doctor to detect subtle changes early, often before symptoms develop, so that treatment can be timed appropriately.**

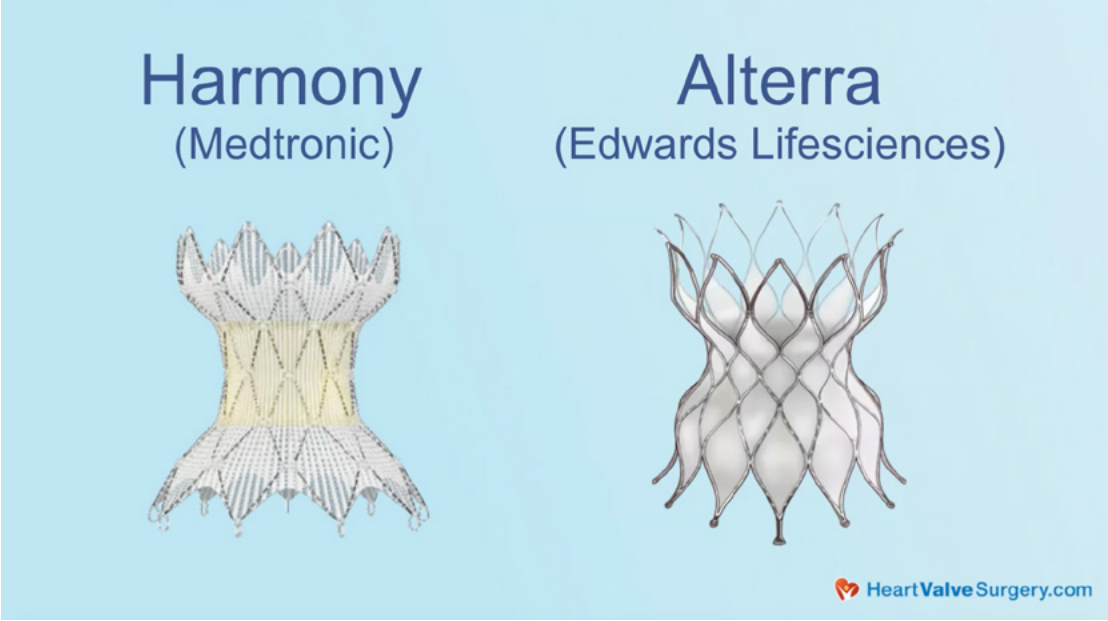
Question #5:

What Are the Different Treatment Options

If treatment becomes necessary, there are generally two main approaches to treating congenital heart valve disease: surgical procedures and transcatheter (minimally invasive) therapies. The right option depends on many factors, including your age, the specific valve involved, how severe the condition is, and your overall health. Your doctor will help guide you, but understanding the differences can help you feel more confident in your decision.

Surgical treatment has been the traditional approach for many years and remains the best option for certain patients — especially younger individuals or those with complex anatomy.

Surgery may involve repairing your existing valve (which is often preferred when possible) or replacing the valve with a mechanical or biological valve. For patients with aortic valve disease, one specialized surgical option is the Ross Procedure, which uses your own pulmonary valve to replace the diseased aortic valve. This approach can offer excellent long-term outcomes in select patients, particularly younger individuals, because it uses living tissue and may avoid the need for lifelong blood thinners.



New Transcatheter Pulmonary Valve Replacement Devices

Transcatheter therapies (as shown above) are less invasive procedures performed through a small tube (catheter), usually inserted through a blood vessel in the leg or chest. These approaches — such as transcatheter valve replacement or repair — do not require opening the patient's chest and often result in shorter recovery times.

In many cases, doctors can also treat other heart conditions at the same time as your valve procedure. For example, conditions like atrial fibrillation or aortic aneurysm may be addressed during the same operation, which can improve overall outcomes and reduce the need for additional interventions later.

Understanding your options, and why one is recommended over another, is key to making the best decision for your health and future.

"It's a really exciting time to be a congenital interventional cardiologist. There are new types of transcatheter devices that are now available for patients."

Dr. Stephen Nageotte
Director, Congenital Catheterization Lab in the Smidt Heart Institute and the Guerin Family Congenital Heart Program, Cedars-Sinai (Los Angeles, California)

Question #6:

Should Patients Seek Treatment At a Specialized Heart Valve Center?

Congenital heart valve disease can be complex, especially for patients who were born with heart conditions and are now navigating care as adolescents or adults. Specialized centers — often called **adult congenital heart disease (ACHD) or heart valve centers of excellence** — have teams with deep expertise in diagnosing and treating these conditions. These teams typically include cardiologists, cardiac surgeons, imaging specialists, and other experts who focus specifically on valve disease.

One of the most important reasons to consider a specialized center is better outcomes. Research consistently shows that hospitals performing a higher volume of heart valve procedures tend to achieve lower complication rates and lower mortality. For example, studies have demonstrated that higher-volume cardiac centers have lower 30-day mortality rates compared to lower-volume centers (approximately 2.66% vs. 3.19%).

In addition, large national data analyses have shown that mortality after aortic valve replacement decreases as hospital experience and procedural volume increase. In simple terms: teams that do more of these procedures tend to get better results

Specialized centers also offer advantages beyond just surgical outcomes. They are more likely to have access to advanced imaging, the latest transcatheter technologies, and multidisciplinary teams that can manage complex or combined conditions. This is especially important for congenital patients, whose anatomy and long-term care needs can be more complicated than typical adult valve disease.

Choosing where to receive care is one of the most important decisions you can make.

"It is critical for patients and parents to find a cardiac center that is very familiar with the unique nuances of treating congenital heart valve disease."

Dr. Richard Kim
Director, Pediatric and Congenital Heart Surgery
Guerin Family Congenital Heart Program
Cedars-Sinai (Los Angeles, California)

Question #7:

What Are the Risks and Long-Term Outcomes of Treatment?

Understanding the risks and long-term outlook of treatment is an important part of making an informed decision. In the short term, your doctor should explain the possible risks of surgery or a transcatheter procedure, including bleeding, infection, stroke, rhythm problems, or the need for another procedure. For the long term, the discussion often shifts to durability — how long the repair or replacement is expected to last — and whether additional procedures may be needed later in life. These are especially important questions for congenital patients, because many are diagnosed young and may need lifelong follow-up.

The encouraging news is that long-term outcomes can be very good. For example, a 2023 single-center study of pediatric aortic valve repair reported estimated survival of 90.8% at 10 years, 86.9% at 20 years, and 83.5% at 30 years, supporting a repair-first strategy in many congenital patients. In adults with severe aortic valve disease who underwent the Ross Procedure, a 2021 study found survival of 97.8% at 1 year, 94.2% at 10 years, and 81.3% at 20 years, and the authors concluded that the Ross Procedure restores a normal life expectancy in young and middle-aged adults treated at experienced centers.

It is also important for patients and families to know that some congenital valve patients may have survival similar to the general population, depending on the exact condition, whether the aorta is involved, how early the problem is identified, and how carefully it is followed over time.

A major review on bicuspid aortic valve, the most common congenital valve abnormality, notes that two large contemporary series found that life expectancy in adults with BAV disease was not shortened compared with the general population. At the same time, outcomes are not identical for every congenital heart condition, and some patients still face higher lifetime risks of reintervention, aneurysm, arrhythmias, or other cardiovascular problems — so lifelong surveillance remains essential.

"Our goal is to help patients of all ages - fetuses, babies, children, teenagers and adults - manage their risks and achieve long-term favorable outcomes."

Dr. Jay Pruetz
Associate Director, Guerin Family Congenital Heart Program, Cedars-Sinai (Los Angeles, California)

Question #8:

How Will Congential Heart Valve Disease Affect My Lifestyle and Future?

A diagnosis of congenital heart valve disease can feel overwhelming, especially when thinking about your future. The good news is that many patients are able to live full, active lives with the right care. However, one of the most important concepts to understand is lifetime management — this is not just a one-time issue, but a condition that often requires ongoing attention and planning over many years.

Because you are born with this condition, your heart and your valve may change over time. Even after a successful procedure, many patients will need continued monitoring and, in some cases, additional interventions later in life. In fact, medical research shows that reoperations are common in congenital heart disease — for example:

- **About 1 in 5 adult congenital patients (20%) require heart surgery later in life, and**
- **Of those surgeries, approximately 40% are repeat (reoperative) procedures**

This is why it is so important to think beyond the immediate treatment and develop a long-term, lifetime care strategy with your medical team. Planning ahead can help ensure that each decision made today supports your health in the future.

Your doctor can help you build a personalized roadmap that considers your lifestyle, goals, and the likelihood of future procedures. With thoughtful planning, many patients are able to stay active, pursue careers, travel, and even start families. The key is staying engaged in your care and thinking long-term.

“With lifelong care, proactive planning, and careful monitoring, most of my patients with congenital heart disease lead full, active lives, and many of my female patients go on to have successful pregnancies.”

Dr. Prashanth Venkatesh
Associate Director, Congenital Echocardiography Laboratory, Cedars-Sinai (Los Angeles, California)

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For more information, please visit:

www.HeartValveSurgery.com

To find a medical team that specializes in congenital heart valve disease, please contact Cedars-Sinai Guerin Children's at (310) 340-6578

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