Adults with CHD: Common Mistakes

From www.medscape.com, September 22, 2014

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Who Are the Adults With Congenital Heart Disease?

Sunil V Mankad, MD: Greetings. I'm Dr Sunil Mankad, associate professor of medicine and a cardiologist at Mayo Clinic, Rochester, MN. Today we will be convening a roundtable review about adult congenital heart disease. I am privileged to be joined by my colleagues Dr Carole Warnes, director and founder of the Congenital Heart Disease Clinic here at Mayo Clinic Rochester and professor of medicine, as well as Dr Naser Ammash, also professor of medicine and Practice Chair of Cardiology at Mayo Clinic.

Let me start by asking, what type of patients are we talking about? Are these people who have survived a malady as a child and perhaps had surgery, or were they discovered later in life to have congenital heart disease (CHD)?

Carole A Warnes, MD: We are actually talking about both kinds of patients. It comes as a surprise to some patients to discover that they have CHD at age 40 or 50, because a heart murmur was heard in childhood and they didn't realize that it had to have an explanation. No further studies were done, and so they lived their lives with a heart murmur and never knew what it was. Sometimes the murmur was missed. You can imagine the surprise for patients to find that they have CHD when they are adults. It is partly because clinical examination has slipped in recent years, so it is easy to miss little murmurs that are quiet, and physicians often don't think that this patient could have CHD. It can be missed, and those patients can have quite complex anomalies, even in adulthood.

But the larger population includes those who had surgery in childhood. Remember that cardiac surgery began in the 1950s, in terms of operating on the heart itself. Thanks to that surgical success, we now have a new population of adults with CHD. Fifty years ago, 25% of babies born with CHD would die in infancy, and about 50% would die before they reached adulthood. Surgery, imaging, intensive care, and everything that we do to make an early diagnosis has advanced, so we now have a population of patients, not only in North America but around the world. In North America, a million adults are living with CHD—more adults than children.

There are two discrete populations. The second group, those who have had surgery, is the larger group, and this group is growing at a rate of about 5% per year, so physicians and cardiologists are going to be seeing these patients in their practices.

Repair Doesn't Mean Cured

Dr Mankad: What are some of the common conditions in this larger group who have survived surgery as children, and what pearls can you pass on?

Naser M Ammash, MD: Among the patients who have never had an intervention, there are two groups: those whose CHD was missed during childhood and early adolescence, and those who had something like a small hole in the heart, an atrial septal defect (ASD) or a ventricular septal defect (VSD), that might not become apparent until later. In our clinic, we see patients before and after their operations. We see a lot of ASDs and VSDs. We see patients with a big right ventricle that they don't know the cause of for a long time, and then we identify it as unusual. Shunt lesions are common among patients who have not been operated on before, but we also see patients who come in with heart failure or atrial fibrillation in whom we find more complex CHD, such as Ebstein's anomaly or those with congenitally corrected transposition of the great arteries who are known to survive into adulthood without an intervention for a long time. Sometimes, when they come to us, it is too late to operate, and this is why it is very important to see them earlier.

On the other hand, we see patients who have had an intervention or surgery, and these patients are all unique. Patients who had a previous repair of tetralogy of Fallot are common, as well as patients who have had a repair of transposition of the great arteries using the atrial switch procedure and lately, the arterial switch procedure. We see a lot of patients who have had shunt surgery.

So, we see quite a big variety of simple and complex CHD in patients who come in after an intervention and surgery. The nice thing about our practice is the variety that we see.

Dr Warnes: A fundamental misconception about patients who have had surgery in childhood or adolescence is that these patients are corrected. Even today, the operation is sometimes called "total correction," but there is almost nothing that is totally corrected by a surgeon, even the world's most fantastic surgeons.

There are some simple lesions, such as VSD surgery or ligation of a patent ductus arteriosus, for example, that can be reasonably said to be corrected, as long as the surgery was good. But among most patients and particularly the ones that Naser mentioned, who have had complex surgical definitive repairs, all will have residual problems. It is an important concept for physicians and patients to understand, because if patients think that they have been totally corrected, they won't see a cardiologist, and they don't realize that lifelong follow-up is essential.

You can imagine the resentment, surprise, and difficulty that patients face when they discover that there are other issues they have to face in adulthood. They don't always follow endocarditis guidelines, for example. The fundamental concept that they are totally corrected is quite flawed

and, for many reasons, I think we should stop saying that. These patients need lifelong follow-up, and those with moderate and complex lesions need to be seen in special centers that have the training and expertise to serve these patients better.

Dr Ammash: One of the messages that we always give when we are teaching about CHD is that there is really no cure. There is only repair, and that is why it is so important to follow these patients, and why it is so important to follow the guidelines that have been established.

The Need for Lifelong Follow-up

Dr Mankad: We are going to talk about the guidelines, but let me ask you first, what are some of the common mistakes that you see? Dr Ammash mentioned some of the common diseases (ASD, VSD, tetralogy of Fallot). What common mistakes are made by general cardiologists?

Dr Warnes: In terms of patients who haven't had previous surgery, a common mistake is missing the diagnosis—not being able to interpret the physical signs, not being able to interpret a chest X ray or an echocardiogram and not even thinking of the diagnosis. Patients with a simple ASD are often asymptomatic until age 50 and they go into atrial fibrillation. Suddenly they become symptomatic, and somebody reads the chest X ray as abnormal, so an echocardiogram is done and a big right ventricle or ASD is found.

You might have a patient who has been followed for years with hypertension, but no one has ever palpated the femoral pulses. The patient starts having chest pain, and a cardiac catheterization is done to look at coronary disease. Oops, the patient has a coarctation of the aorta. Nobody ever thought that this patient might have a coarctation, despite having hypertension. It can easily be overlooked on an echocardiogram or a chest X ray. Some of those relatively simple lesions can be overlooked.

With respect to the patient who has already had surgery, coarctation is a great example. Coarctation repair has been done since the 1940s. You cut the two ends of the aorta, remove the coarctation, put the ends together, and the patient is corrected—end of story. Of course, we know that these patients have many problems, and persistent hypertension is very common (75% at 30-year follow-up). They have abnormal aortas, which are vulnerable to dilatation and dissection; the higher the blood pressure, the greater the risk. They are also at risk for stroke and premature coronary disease. All of those things require lifelong follow-up. It is easy for physicians and cardiologists to consider those patients cured and the proper follow-up is not established.

Early Referral Is Imperative

Dr Ammash: There is also a group of patients who come in to see us, not for surgery, but to see the transplant team for heart transplantation, thinking that they have CHD that is not amenable to repair, so they are sent for heart transplant or heart/lung transplant. When they come here, they have congenitally corrected transposition of the great arteries, and under the right conditions we can replace the valve and they do well. In patients who have had tetralogy of Fallot repair and have severe pulmonary and tricuspid regurgitation, or in those with Ebstein's anomaly, we could intervene earlier, especially with pulmonary regurgitation. Some adult cardiologists don't see pulmonary regurgitation on echocardiogram as well as cardiologists who specialize in CHD. It can be missed. If they haven’t seen it before, they are not going to be able to identify it. The patient comes in with severe pulmonary regurgitation after tetralogy of Fallot repair, but by the time they get to us, it might be too late to operate.

Dr Mankad: I have learned a lot about pulmonic regurgitation and certain VSDs, as well, and the threshold for operation.

Dr Warnes: Late referral is very important, because patients with CHD are very different from the typical cardiology patients. A person with an infarct and ventricular dysfunction will suddenly notice a change in exercise capacity. Patients with CHD who have never known what normal is may wait until they develop symptoms. The cardiologist or primary-care provider who follows these patients may think they are doing just fine, but patients who do not have normal exercise capacity might have gradually slowed down without noticing a difference. If we put them on a treadmill at the time of referral, we discover that their exercise capacity may be stopping at 60% of predicted. Some patients believe that they are okay or that it's just age. They can't do as much as they used to, but they had the surgery and it was fine. They don't realize that there are sequelae and residual effects from the surgery that was done in infancy or childhood and that these sequelae may need to be repaired. It's heartbreaking to have the patient referred too late, when they are symptomatic and can't even walk across the room. They may have severe right ventricular dilatation or dysfunction, and it doesn't matter whether the pulmonary valve is replaced now or whether the surgery goes well. The right ventricle will never recover. So it's not only where the surgery is done, but the timing of the surgery is absolutely key. You want to get to patients before they have irreversible ventricular dysfunction. Whether it's on the right side where the right ventricle is pumping to the lungs, or in the systemic circulation (as with the the transpositions) and it's pumping to the body, you want to get to those patients early so that you can hopefully prevent irreversible ventricular dysfunction.

If there is one fundamental mistake in those postoperative patients, it's the failure to recognize that they are symptomatic and referring them to a tertiary-care center too late, when the risks associated with surgery go up. Their dysfunction might be 3% to 10% if they are referred when the heart is very large, whereas had they been referred 10 years earlier, their dysfunction might be only 1%. Early referral is very important.

New Guidelines on the Way

Dr Mankad: You were involved with the 2008 guidelines.[1] Is it time for new, overall global guidelines? The tetralogy of Fallot guidelines[2] that you were involved with, as well, recently were published. But how about an overhaul of the entire guidelines?

Dr Warnes: The new guidelines were begun this year, and hopefully they will be ready next year. The emphasis will continue to be that patients need to be followed for life. When patients reach age 18 or 21, their care doesn't stop; they should be sent to congenital heart centers for adults where they can be taken care of. Some new discoveries have been made since 2008 when those guidelines were published. Some of these discoveries relate to early surgery and how to intervene early.

Dr Ammash: Those guidelines are very important. There was a recent study from Canada[3] looking at the impact of guidelines on outcome of patients with adult CHD. As soon as the guidelines were published (ours were published in 2008, but the Canadian guidelines came out a bit earlier), there was an increase in the number of referrals of patients with complex CHD to specialized centers, with a significant improvement in survival in those patients. There is a link between appropriate care by the trained physician following established guidelines and improved survival in patients with moderate and complex CHD.

Dr Mankad: Thank you so much for these interesting insights on this great topic. To our readers, we hope you will continue to follow our roundtable review series where we bring you the best of the Mayo Clinic on www.theheart.org on Medscape.

References

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