Children's Cardiomyopathy Foundation™

Answers from the Experts: CCF Cyberguests

A Compilation of Q & A Pediatric Cardiomyopathy Listserv Sessions
January 2012 - December 2013

Q&A

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A compilation of Q & A listserv sessions on pediatric cardiomyopathy

The Children's Cardiomyopathy Foundation (CCF) offers several support services including an online resource known as the "CCF Forum." The CCF Forum is a private listserv that offers registered members the opportunity to correspond with other families affected by pediatric cardiomyopathy. The e-mail discussion group, which includes members from the U.S. and abroad has become an important and valuable resource. It allows parents to keep in touch, exchange information, and provide emotional support to each other in an easy and informal manner.

From time to time, CCF schedules professionals (cyberguests) to address specific topics related to living with pediatric cardiomyopathy. These guests volunteer their time and expertise to answer questions posted by CCF Forum members. To serve as an additional parent resource, CCF has edited and compiled transcripts of all the question and answer sessions starting from 2006. Each topic is covered in a broad sense with questions asked most frequently by parents of a child with pediatric cardiomyopathy. CCF hopes that the information provided from these experts will assist families in better understanding pediatric cardiomyopathy and encourage them to seek more specialized information and/or recommendations from their child's physician and healthcare team.

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Psychological Concerns of Children with Cardiomyopathy

Victoria Norton, Psy.D – January 2012

Dr. Victoria Norton is a pediatric psychologist at the Rocky River Behavioral Pediatrics Clinic in Rocky River, Ohio. Dedicated to the needs of children and adolescents, Dr. Norton has extensive experience in the evaluation and management of psychological concerns of children, such as developmental and behavioral concerns, parenting, depression, anxiety, socialization or relationship difficulties, and family stress.

Question: As children and teens grow and develop, things about them can change – their personalities, their habits, how they respond to different situations and relationships. The same can be true when a child or teen has cardiomyopathy. Is there a way to tell the difference between typical "growing pains" and something more serious? Is there a way to tell between typical child/teen growing pains and typical child/teen living with cardiomyopathy growing pains? Is there a way to tell between either of the above and something more serious?

Answer: There are differences between the growing pains of children/teens with and without cardiomyopathy, but unfortunately I cannot name them specifically. Living with a chronic illness has implications on every aspect of development. Depending on the type of cardiomyopathy, symptoms and treatment plan, the impact may be minimal or very great. What has been interesting to notice is that for some children minor disruptions to normal daily living can be highly bothersome, while other children accept and embrace life managing hospitalization, frequent appointments, restricted activity and being "different" from peers.

I think that increased distress during adolescence and young adulthood can be expected, even for the child who has previously done fine and not been bothered by aspects of illness (i.e. taking medication, educating teachers, telling peers, attending appointments). This can be confusing for the parent who has had a child who has always coped well with aspects of care, and then all of a sudden things are different!

When are growing pains more than just that and indicative of a need for more attention and intervention? If parents notice changes in the way the child is interacting and behaving — for instance a decrease in academic performance or less interest in spending time with friends — this may be a signal. From a teen's perspective, their job is school and friends. When these areas are impacted by growing pains, then it may be time for some additional assistance. Often, the kids that I meet are dealing with emotional distress that is secondary to challenges of living with cardiomyopathy or other chronic illness, rather than a primary mental health disorder. This distinction is an important one in terms of finding a provider that can best assist in the journey to improve overall health.

Question: I have five daughters ranging in age from 16 years old to 15 months old. My 15 month old has left ventricular non-compaction (LVNC) which is stable right now, and two of my other daughters have the characteristics of LVNC and are being monitored. What is the best way to address the issue of cardiomyopathy or any other illness or disability with children and teenagers in a way not to scare them but to make them aware?

Answer: Parents of children with cardiomyopathy and other chronic medical conditions often struggle with when and what to say to their child about their illness. A common response is to say less or nothing at all in an effort to protect the child and prevent distress. However, what we know is that this can actually create more confusion and send the message that it is not safe to talk about or ask questions about the illness. Instead, it is best to create an "open-door," and let the child lead the way in terms of communication. This can vary greatly from child to child. Offer the basics in age-appropriate terms in a simple, direct and honest manner. Let the child decide if they have additional thoughts or questions at that moment and, if not, share your openness to talking about things at a later time.

Like you said, awareness is important so that children can fully know about their health status, what they can or cannot do, and how to help themselves manage the difficulties of living with cardiomyopathy.

Question: I have a son who is now 27 years old who has obstructive hypertrophic cardiomyopathy (HOCM). He has had two myoectomies and two defibrillator implants. He is very angry regarding his condition. One of his recalled defibrillator causes phantom shocks. He escapes with drugs (OxyContin) and has experienced problems in behavior and drug addiction from medications from past surgeries. He seems to like the drugs and is very angry at the world for his HOCM defect. Is this behavior common or is it just our son who is going through this? We have tried counseling and nothing seems to resolve the anger issues. We also lost a son to HOCM — his brother at 11 years old.

Answer: What a difficult journey! That sounds incredibly frustrating for you and your son. You asked if your son's reaction of anger and hatred towards the world and difficult behavior was common. My response is for some, yes. This is a not an uncommon response to illness and/or grief. Your son has had to deal with both. There is no question that your son has been faced with an unique set of challenges that make it easy to feel sorry for oneself and ask "why me?" Some kids and young adults get stuck in this spot and continually struggle to accept the cards life has dealt them. This becomes a means for justifying undesirable behavior and poor choices, which are seen as a result of the illness or complications of the surgery rather than how one thinks about these events ("It is not fair," "My life is terrible," "I am never going to be normal"). The addition of pain medication further complicates this and sets up an external locus of control and of being stuck.

I rarely find children and families benefitting from traditional “counseling.” Often I meet patients after they have seen one or many providers without success. The key is to work with someone who understands the impact of medical illness on development (even when the patient is 27). Notice I did not say that they understand cardiomyopathy. While this would be fabulous, it is unlikely unless you happen to live near a major heart center.

Another key factor is your son’s motivation at the time of intervention. His interest in finding a different approach is critical to effective treatment. Previous negative experiences with “therapy” and beliefs that it will not be helpful add to the challenge. I fully believe that there is hope, and it is possible for your son to find his light and shine.

Question: My son was recently diagnosed with bipolar disorder in April 2011. He is 20 years old and has had cardiomyopathy his entire life with no symptoms. He has been on several different medications over the years but is currently only taking enalapril 2.5 mgs twice a day for mild dilated cardiomyopathy. His episode leading up to the bipolar diagnosis was very severe and required almost two weeks of hospitalization because his heart rate jumped up to around 200 with the medications being prescribed to level out his mood. His counselor told us that if he had sought medical help earlier when he was feeling “manic” then he would not have been so sick.

This all happened the beginning of his second semester of freshman year away at college. He was not sleeping well, admittedly anxious over some events he was trying to manage at the time and was not taking his medication regularly. On the day it all came crashing down, he said he felt his heart racing and thought he was having a heart attack. He took about 13 of his enalapril over a period of an hour and his friends ended up taking him to the emergency room because he was talking so fast. The hospital sedated him and then released him several hours later. He went back to the hospital that afternoon in a psychotic state where they admitted him and gave him Ativan.

Does this sound like bipolar disorder or does it sound like a bad reaction to the enalapril? He has not had another episode since his release and is now taking 100 mgs of Lamictal along with the enalapril. He did go back for his fall semester and is now about to start the spring semester. Your thoughts and advice are very much welcomed as I am still having “flashbacks” of his horrible ordeal nine months ago. I can’t seem to stop worrying that it will happen again.

Answer: Your question was in regards to whether your son’s behavior was consistent with a bipolar disorder or if it was related to the large dose of enalapril. Based on your son’s symptoms and presentation, it does not seem consistent with

what would be expected of enalapril (low blood pressure, headache, dizziness, lightheadedness, possible back pain if kidneys were compromised). I do not think this explains his talking fast or psychotic state. Not knowing more, I am not sure if bipolar is accurate or if these symptoms are more consistent with severe panic attacks.

It is wonderful to hear that he has been stable over the past nine months and that he has returned to school! Whether it is due to the Lamictal or if he would have returned to school on his own regardless, I cannot say. However, given that he is doing so well, I would be reluctant to modify his treatment without clarifying that bipolar disorder is not present. The future may not include another episode like he experienced last month, but if it does, you have information to help you understand what is going on and how to prevent the escalation of events. Whether it is related to cardiomyopathy, bipolar or any other illness, the challenge is the same... living in the moment and enjoying the journey.

Question: My son is 13 and has had dilated cardiomyopathy his entire life. We have always kept a very open line of communication and he is very open about his disease. We live in a fairly small town (population 8,000), and most of the people he comes in contact with know about his disease. He goes through periods where he is full of rage. Other times he is as sweet as can be. It all started around puberty, and I know some of this is normal teenage stuff (even though my older boys were nothing like this). He goes through these periods of “My life is terrible,” “Everything bad happens to me,” “It isn’t fair.” We try and talk through it, going over all the great things he has in his life and all he has to look forward to. His behavior has gotten much better over the past couple of months, but we still wonder if he would benefit from talking to someone. Anytime I bring it up he gets upset and says,“I don’t want to have anything else wrong with me! No more doctors!” so we let it drop. One of his older brothers goes to a therapist, partly to deal with his younger brother’s disease and for some other reasons. He has told his brother how much it has helped him. My question is: should I continue to bring it up occasionally or just drop it and wait until he mentions it?

Answer: I am so glad that you posted this question because I am certain that other parents are wondering the same thing. Your son’s perception is much like many other children and teens that I meet — “I am not crazy,” “Nothing is wrong with me,” or or “A psychologist can’t help me get rid of my cardiomyopathy.” My job in the hospital setting is easy because I get to meet kids when they are captive and I can present a very different perspective than what they imagine in terms of talking with a psychologist. I can also “sell them” on how they might find it beneficial to learn how to deal with the frustrations of x, y and z. In my private practice setting, it is often more challenging for parents because the introduction of the idea of a psychologist implies that there is something else wrong. The last thing they want to do is spend more time attending another doctor’s appointment or perceiving themself as having another problem.

When a parent calls me, I say that if enough is going on that they found me and called, then it is worth making an appointment for a consult. They then have the tough job of getting their child to agree to come once. The rest is up to me or the child. I encourage parents to present it as coming from a place of wanting to help your child be as happy as they can be in all areas of their life, and you can tell that certain things are not as good as they could be. My approach is to meet with kids alone, find out what they enjoy and do well, and use this as a metaphor for doing well in other areas that are more difficult. I teach kids what I do, how some kids find it beneficial and then let them know that they are the boss in determining if it makes sense to meet now or at another time in the future. I do all this before parents come in the room. Then, parents join and what has been decided on is discussed. Parents then share their perspective. I think this is critically important because from the beginning it places kids in charge. After all, who is more in charge of their body, thoughts, beliefs and emotions than they are?

In your case, I would suggest you continue to push. I would present the idea behind why you think it may be a good idea. Maybe even go so far as to find the right provider, talk with them and let your son know about them and that this is available if and when he would like to go. Then, leave it up to him as long as there are no significant behavioral concerns or emotional or academic issues involved. Some parents find it helpful to get some guidance, which you may wish to consider too. I believe that kids know what they need to do and when it is best to do it, but the perception of a psychologist and who goes to one is skewed by what is seen in the media. When kids find out that I only see children with medical illnesses, they are surprised and relieved. Educating your son that kids go for all sorts of reasons, not just because they are crazy or something is “wrong,” may help increase his openness.

Question: My daughter is only 2 years old and had a heart transplant at 6 months old. Over the past few years there has been a lot of attention placed on her heart transplant and all of her medical issues. We are very careful about germs, diet and her medication schedule. At times we feel isolated because we do not see as many people. It is partly because we are careful and partly because others are so afraid to get her sick. This has kept her well and she is doing phenomenal with no rejection so far.

Over the past few months I have been thinking a lot about her future. I know we have a few years until school starts, but I am trying to find a healthy way to deal with our concerns. I don’t want her to ever feel like an outcast or for people to act scared around her and call attention to her condition, but I want her to remain as healthy as possible. I know she is going to have some issues with everything eventually, but I want some advice on what are the important things to think about as she gets older and is more socially involved. I try to remember that she had this transplant for a reason and that is to live a fulfilling life.

What are some of the things transplant kids face in elementary, middle and high school? What can I do to help her adjust and teach her to take care of herself? I also fear that she will become anxious about death, as a lot of people with chronic health issues I know develop anxiety. What can I do to help her? Do we talk about it or leave the topics open for her to talk about? When she starts to have school friends, do we tell the other children or do I quietly mention it to the parents? At the same time, I do not want to get into a pattern of making it all about her.

Answer: Your thoughtfulness to the future of your daughter is valuable. Recognizing that there will be more to think about as she develops that goes beyond physical health is important. When children are young, parents are able to protect them from infection with isolation and at the same time largely meet their needs. As they enter toddlerhood, parents begin to question when and how much socialization to allow and the pros/cons of allowing these interactions that bring with it the risk of infection. As the parents on this listserv can attest to, this struggle continues as children become school age and only intensifies during pre-teen and teen years when children desire more time with peers and are less satisfied with spending Friday night with their parents! I have worked with many families who have struggled with allowing their child to go to school. I recall one mom whose 12 year old daughter endured a transplant and desperately wanted to return to school. The mom was terrified of the risk of infection and insisted on home-schooling, which left the girl emotionally distressed and socially isolated. These are very difficult decisions that do not have a “right” answer in my opinion. Rather it becomes about picking the best option for your child and family. I tend to assist in this decision-making process by looking at the whole child, which includes their physical and emotional health, and deciding what choice best allows for optimal functioning of the child (not just their heart). You said it beautifully when you commented that your daughter had a transplant so that she could “live a fulfilling life.” Let that philosophy be your guide as you face tough challenges ahead.

You asked about specific issues that children face as they develop and how to best help your daughter at these times. There are a number of difficulties that come up, ranging from not being able to take gym class, feeling that they are “not normal” or “different,” not liking themselves, worrying about being treated differently, not being liked by peers and missing school. For some kids, this intense emotional distress can be so great that they struggle to find happiness or peace.

As your child grows, teach her about her body and how she is unique, as well as what this means for her. Focus on what she can do, even if it is different. Her heart transplant makes her very special, and there may be points in her life where she embraces this and is proud. There may be other points in her life where she hates it. When it is hard, do your best to listen to her. It sounds simple but often my little friends share with me that they wish their parents would do that better. If she does worry, help her learn how to control her worry rather than let worry control her.

This is where it may be beneficial to have some support, depending on the degree of worry. When worry goes away because a child gets to avoid whatever is the source of the worry, then the worry wins and the child does not learn how to manage it. Living with cardiomyopathy means that there are going to be things to worry about, so the sooner kids learn how to be the boss of their worries, the better!

In terms of how much to talk to her about her health, talk a little and let her ask the questions. Focus on her as a whole person, as she is much more than a kid who had a heart transplant. Kids are so different in regard to the amount of attention they want to devote to talking about their illness. In general, most kids prefer to talk about everything else and give as little attention as possible to their condition.

During her early years, the education of friends and parents will fall on you. As she gets to school age, I suggest helping kids develop answers to the questions that they might get asked by others about their health. Some kids develop very short and simple responses; others kids offer more elaborate details. Kids are wonderfully creative and can come up with responses with just a little help. Once in school, giving some information to school personnel is essential, but how much is told and in what capacity can vary. I think it is best to let the child be part of this process to the degree that they want to be. Some children want what makes them different to be private, and when this can be respected, it makes sense to do so. I am not generally a fan of sharing information secretly. As the parent, you want to always maintain your child's trust and be honest with her, especially when it is about her.

Question: I have a 6 year old boy with hypertrophic cardiomyopathy (HCM). He is asymptomatic at this time. He is finally able to say "hypertrophic cardiomyopathy" and knows he has a form of heart disease. He knows that we have an automatic external defibrillator (AED) to help if something goes wrong with his heart. He is very familiar with all the cardiac tests, and although he gripes about them leading up to the appointments he does cooperate when the time comes to do the tests. He was very hesitant two weeks ago with the holter monitor because he did not want to have it at school. After he realized how small the unit was (his last one was much bigger), he realized it would not be seen at all. Many of the other comments parents posted hit home in our household too. Basically, he complains that it is not fair he has a special heart and has to do all the testing. We have not gotten to the point of it not being fair because he is not allowed to play sports, but I expect that will be coming in the future.

At this point we have not gone into details about HCM and the risks associated with the disease. We have discussed all the symptoms in great detail because he knows he is supposed to let us or an adult know if he experiences any of them. Also, he is not one to share a lot; we have to ask him. Even then he does not always like to talk about things. I have a feeling as he matures, this could become worse and he might bottle up some of his concerns, fears and feelings regarding his HCM diagnosis. These are my questions:

1) At what age do you recommend fully disclosing the facts and possibilities he may face in the future?

2) Do you recommend counseling for all young children/adolescents with a chronic illness such as cardiomyopathy?

3) If yes, what age do you recommend starting?

4) If no, then how do you know when it is important to start?

As I said, I tend to be proactive so I would rather not wait until it becomes a huge problem for him. I had asked his cardiologist about this two weeks ago and he said to discuss it with his pediatrician for a referral. My thought was to start earlier before he comes to understand the negative perception that is associated with seeing a counselor or psychologist. Although another problem for us is that we move often being military and he would not be able to stay with the same professional. It could be frustrating to him to have to build a rapport with a new person each time we move.

Answer: It sounds like your approach up to this point has been ideal – sharing what makes his heart unique and the symptoms that he may experience and what to do if he does. When you disclose more will depend on him and his body. By that I mean if he asks you questions or if he begins to develop symptoms, it may naturally lead to having conversations that include more details.

To answer your questions, I do not have a chronological age that I think is best to disclose information. Kids vary so greatly in their openness, interest and maturity. These are factors to consider when deciding when and how much to say. Given that your son tends to prefer to say less (as do many boys), use that as your guide. There are times that you are going to have to talk about his health at a doctor's appointment, and it sounds like during these times he handles it just fine. When you can focus on other topics, do so. He will be more likely to listen if it is not something that is "preached," but rather something that is an ongoing dialogue.

I do not think that all children with cardiomyopathy need to be involved in treatment. That said, they do need a forum for addressing the unique challenges that they face living with a chronic illness. In the last decade what I have learned from parents is that all of them benefit from having access to a pediatric psychologist for guidance in dealing with these unique challenges (often as a one-time deal, which is different from going to their own "counseling").

I am sad, but not surprised, to hear the response of the cardiologist because it means that there is not a provider that he works closely with to refer you to. Given your situation, I would not recommend starting now, unless you have

specific concerns. It sounds like you are doing wonderfully helping your son manage what he needs and that there is no need to work with a professional at this time. I agree that there are benefits to working with the same provider over time. I have kids that I have seen for many years, and they pop in when necessary, usually during a medical event or when there is increased distress. This is comforting for families and fun for me to watch kids grow and develop.

Since I do not think all kids need to go for counseling, I recommend starting when you notice changes in academic, social or emotional functioning that you cannot seem to get a handle on and does not resolve over time. How much time depends on the degree of change and impairment.

I fully agree about being proactive, and it is why I fought to develop a program where pediatric psychology/behavioral medicine was part of the pediatric intensive care unit and the heart center. I wanted to meet all families before a crisis develops. Luckily, I worked with a group that was receptive to my initiative and allowed me to build a program that worked for the physicians and the families. Unfortunately, this is not the case in most institutions and that creates a greater challenge for most parents. I think you can remain proactive by doing what you are doing right now.

Question: As you can see by the flood of questions for you, so many of us cardiomyopathy parents have to deal with ongoing emotional and psychological issues with our kids. Given this, my question (which has two parts) is more about the way this aspect of care is approached within cardiology:

1) What kind of training do pediatric cardiologists receive to help them deal with the psychological aspects of the disease and the ways those aspects affect the entire family?

2) Are there any efforts underway that you know of to integrate psychological treatment into cardiology treatment of pediatric cardiomyopathy beyond the usual recommendation to make an appointment with the hospital social worker, psychologist or one's own therapist? In your opinion, what would such a truly integrated care look like?

I am a parent, who has come out the other side of my teen's depression and am still dealing with all the repercussions on our family, as well as a medical humanities teacher devoted to humanizing medical practice and education. I would like to move us beyond the lonely and often desperate scramble for psychological help that many of us go through. I would love to hear your ideas about this.

Answer: I will share my thoughts on this subject with the disclaimer that this is not something I have formally assessed, and as a non-cardiologist, this is not

something that I know from first-hand experience. I am not sure what the standard protocol is across medical schools and residency/fellowship training programs. However, I can speak from my experience working with physicians (residents, fellows, attendants) in different children's hospitals. I have been in four major medical centers as a resident, fellow or on medical staff.

My experience is that there is limited training in regards to the psychological, emotional and behavioral needs of healthy children during residency training with little-to-no training in regards to the unique aspects of development for children with chronic illness. My teaching with residents was limited to a one-time, one-hour meeting, which meant highlighting the most basic elements of what I had learned in four years of college and seven years of graduate school, residency and fellowship! Essentially, it meant providing information on when to refer to a psychologist.

While psychology as a science has been around a long time, the subspecialty area of "pediatric psychology," which focuses on the unique aspects of caring for children with a medical condition, is relatively new (within the last 50 years). What that means is that most medical providers are unfamiliar with how pediatric psychologists differ from general child clinical psychologists. Referrals to psychology are seen as warranted when there are behavioral issues, anxiety or depression, rather than to promote development, prevent emotional upset, increase compliance, decrease distress related to a medical regimen, teach pill swallowing, or address how to handle peers and promote social development.

Having worked as the psychologist for a pediatric ICU for many years in a children's hospital with a large heart center, the integration of behavioral medicine/ pediatric psychology made an essential difference to the medical team as well as to the families. I was able to work closely with staff so that they could help identify which families I should meet during rounds. I was able to meet with families easily during an admission and then determine what, if any, follow-up after discharge was warranted. Being a member of the medical team helped minimize the stigma associated with psychology for the child and family. It also allowed for collaboration between medical providers and served to enhance the relationship between the child and family and the medical team. This type of integration creates the optimal care for the child. I believe that this is becoming more the norm across medical institutions. Hospitals are a business, and psychology is not a money-maker for a medical institution. While there are a lot of "therapists" out there, there are not many pediatric psychologists. These two factors make creating this integrated care a challenge.

Question: I have an almost 3 year old who was diagnosed with dilated cardiomyopathy (DCM) at 7 weeks old and had a heart transplant at 5 months. At what age should we start her in counseling? I don't know what is too young or if there is such a thing. She has started talking about "hospital" and "doctors"

and is often very traumatized at her appointments (even getting measured and weighed). I have not had any counseling, but have made this one of my resolutions because I feel so much anxiety and am incapable of focusing since her problems started.

She has been healthy, although we recently spent four days in the hospital because she had pneumonia. I was able to stay with her the whole time she was in the hospital, so she was not alone. Since then she has been very clingy and unlike herself when I try to go to work or go out to do things. About a month after she got out of the hospital, we had her babysitter of a year come over so we could go out shopping and she cried so hard that she threw up and we had to come back home immediately. She had loved this babysitter previously, but now if I even say her name she starts saying "No...No [babysitter's name]." She has been like this with others who have taken care of her in the past and has really only been okay with grandparents and my sister. I feel like she should be past her separation anxiety stage by this time, so I am wondering if it has to do with being in the hospital.

I also do not know how much I should "push" her towards independence or if I am being too "easy" on her. I know there must be some issues that develop due to her constant medical appointments, testing, hospitalizations and catheterizations. I am unsure when to treat her like a "regular" kid and when to indulge her as far as independence and anxieties are concerned. I am also unsure how to deal with the crying so hard that she throws up, which tends to happen whenever she gets very upset.

Answer: There is a value in bibliotherapy (reading books). There are a variety of topics related to medical procedures and hospitalization available by Magination Press. This is a publisher that is exclusive to works by psychologists for children, and each book includes a "note to parents" section. You can review the list of available books by going to www.apa.org and clicking on the link for Magination Press. These are a great resource for ages 3-8.

In addition, as parents, it is essential to maintain a sense of control with the process of the illness and around medical appointments and hospitalizations. You are a model for your daughter and she will sense your uneasiness, even at age 2. You may find it beneficial to talk with someone to figure out how you get yourself to a place to do this more easily.

In terms of when to treat your daughter like a "regular" kid and when to indulge her, my advice is to treat her like a regular kid all the time. Reason is, a child with a chronic illness has to learn to live with a chronic illness. It does stink, but it is reality. Going easy on her now will only make it harder for her and for you later. It is in her best interest to learn how to self-regulate and deal with upsets sooner, rather

than later, in regards to appointments, procedures or separation. I think that kids have to be taught how to do that, rather than just “made” to do it. You can teach her this by preparing her for an appointment or separation, by giving her something tangible to hold (favorite stuffed toy or something of yours), distraction and practice. Each time she has to deal with her upset, she learns that she can do it on her own. When you leave, do so confidently and minimize your good-bye, calmly state that you love her and you will be back soon. Doing so sends the message that she is safe and you trust that she can do it! Crying (even if it leads to her vomiting) should not bring you back home. Some kids do this and the temptation is to prevent crying by giving in, but this is not effective long term. It can lead to greater behavior problems, and it prevents the development of the self-regulation skills and confidence that she needs.

Parenting is hard work, and when the complexity of managing a chronic illness is added it can feel impossible! The good news is that children are amazingly resilient and can tolerate their parent’s mistakes. As hard as it is, the challenges of cardiomyopathy create an opportunity for many valuable lessons to be learned by parents and children. These can serve as great tools for dealing with the natural ups and downs of life in the future.

Question: My 3 year old was recently started on enalapril. Since then, we have noticed “ups and downs” a lot. It could be a few hours or the whole day. She goes from bouncing off the walls and out of control (totally uncharacteristic of her) to super moody, grouchy and tired. I can not tell if this is medication related or just a new stage.

Answer: Before responding to your question, I want to address medication questions more generally. In the state of Ohio, psychologists do not have prescription privileges. Psychologists who work for the military and those who practice in a handful of states can prescribe with the appropriate supplemental training. While I have trained in pharmacology, I want to clarify that at this time, making recommendations about medication is outside my scope of practice. I will share my opinion, with this disclaimer in place.

To my knowledge, the behavioral side effects that you described are not associated with enalapril. Given how it acts on the heart (dilates vessels, allows blood to flow more smoothly, decrease blood pressure), it would not make sense that this is causing the shifts in energy and mood that you are seeing with your daughter. This is not a complaint that I have heard from other parents. I would suggest sharing this concern with your cardiologist to be sure.

Having a child myself that is almost 4 years old, I understand the “ups and downs“ that you describe. What I would say is when behaviors are uncharacteristic, it is smart to be mindful and be sure that these are not signs of medical changes.

Whether these behaviors are or are not related to underlying medical causes, you and your husband want to establish what the expectations are in terms of behavior and help your daughter to control her emotions and body, even when feeling tired, grumpy or excited. Children with medical illnesses have to learn how to tolerate more than other kids, and these lessons are easier taught young even though as parents we tend to be more tolerant and understanding when children are younger. This stage of development is wonderful as little ones are discovering who they want to be and showing their will. Some children naturally live life more out loud, meaning that they experience all of their feelings fully! It is wonderful in many ways, but it can be challenging for family members because of the unpredictability and intensity. With maturity, sometimes this settles. For other kids, they gain more control but their strong spirit stays the same. I tend to view this kind of energy as a "gift" for kids with chronic illness, although there may be moments where it is not experienced as a "gift" by parents.

Question: I have a question to ask you about medications for anxiety. My son is 16 years old and has hypertrophic cardiomyopathy. He had a cardiac arrest and three aborted cardiac arrests. Also, his implantable cardioverter defibrillator (ICD) lead fractured and he underwent a brutal surgery to extract the fractured lead and receive a new ICD. He was very angry that his lead fractured, as he obeyed all his doctors orders, limited his physical activities and quit playing drums in the marching band. It was a manufacturer's defect and had nothing to do with anything he did.

Recently he went into a very fast rhythm – atrial fibrillation with rapid ventricular response. He got out of the hospital four days later and went into atrial fibrillation again the next evening. This time the rate got so high that he was shocked by his defibrillator. He was in the hospital another five days, got out and was back in the hospital that night. We finally got out of the hospital and so far he has not had any more atrial fibrillation attacks. We have added several cardiac medications, one being amiodarone. The doctors sent him home with Xanax. My son asks me for Xanax quite frequently, as he is anxious. We went from taking one medication since he was 7 years old to seven medications this month.

While the episodes of ventricular fibrillation and the shocks he received for ventricular fibrillation were much more dangerous, these bouts of atrial fibrillation and the shock he received a few weeks ago have had a much more adverse affect on him mentally. With his episodes of ventricular fibrillation, he was always doing something physical and knew he was going to pass out and get shocked. When he woke up, his heart would be beating normally. With these atrial fibrillation attacks, they happen out of the blue when he is sitting down at home doing nothing. The atrial fibrillation rhythms can last for hours and cause shortness of breath and chest pain. He worries that he will receive an inappropriate shock at any time. He says he feels like he is walking a tight rope between two skyscrapers.

He never had any type of premature ventricular contraction (PVC) or atrial fibrillation before his lead extraction surgery. The doctors think it is the trauma to his heart from the surgery that is causing these atrial fibrillation runs, and they think it will resolve in eight weeks. He has not been in school for a month and a half, and his replacement ICD and lead extraction were over a month ago. I promised I would not send him to school until he can go two weeks without a run of atrial fibrillation.

He was in counseling before all this happened. I have taken him to a psychiatrist as well, as the counselor is not able to write prescriptions. His electrophysiologist is an adult cardiologist and said that he gives his older patients Lexipro, but since my son is 16, he was uncomfortable giving him a prescription for it. My son's psychologist wrote him a prescription for Lexipro and Klonopin. I have not started either of these two medications yet. I do not want to start him on something that I will have to wean him off of. He really likes the Xanax, but I know this is highly addicting. What would be the best thing for his anxiety as far as medication goes? Is Klonopin basically a long acting Xanax?

Answer: While medical challenges are never easy, for toddler and young children they tend to live in the moment and be protected from their lack of understanding. For a teen, they can cognitively grasp the gravity of the situation. To answer your question — "What do I think is best for anxiety in terms of medication?"— I am not a fan of the benzodiazepines (Klonopin and Xanax). As you said, they are habit-forming and cannot be used long-term. While they are highly effective in that they are sedating and cause the muscles to relax, this is only true for the brief period that they are in the system. It is essential that children learn how to create a state of relaxation on their without the benzodiazepines. There are also many negative side effects to the benzodiazepines such as drowsiness, light-headedness, headache, irritability, difficulties with mental clarity and memory, and the list goes on.

Lexapro, on the other hand, is an selective serotonin reuptake inhibitor (SSRI), which is a medication that increases serotonin and facilitates mental balance. The SSRIs do not act quickly but rather take weeks to build up in the system. They do offer the same level of relief. However, they are safe to use long-term and effective at aiding in the treatment of depression and generalized anxiety.

I know that you asked about medication, but I firmly feel that medication be used as an adjunct to cognitive-behavioral treatment and the learning of self-regulation skills. The goal is to get as many tools as possible in your son's toolbox so that he can help himself deal with the stressors that will continue to arise. Solely using medication keeps an external locus of control. I think it is so valuable when we can help kids feel an internal sense of control and confidence to manage themselves.

I am well-trained in the use of self-hypnosis, and I have found it highly effective in helping kids use their mind to control their anxiety, mood, pain and other

difficulties. I would encourage folks to look into providers who are certified in the use of self-hypnosis. This is not a hypnotist; it is a physician or psychologist with advanced training in using hypnosis to treat what they are already trained to treat. I suggest referencing the Association of Clinical Hypnosis website (www.ASCH.com) to learn more and find a provider in your area. I also use a biofeedback program with children and teens called Emwave. This is available and affordable and a great way for kids to learn and practice self-regulation skills (www.hearthmathstore.com) and the connection between their mind and body through their heart.
Kids love it!

I hope these ideas are helpful and that your son gets the balance and coordination he needs to walk that tight rope with ease!

Question: My son had difficulties and was neglected at a local hospital where he arrested and almost lost his life. We were lucky because he was medi-vaced to a children's hospital where he got a heart transplant and survived. It took almost two months for him to get well enough to even list for a heart transplant. He missed a year of school and his memory was affected because of the trauma. We recently had him go through a neuropsychological test, which he did not enjoy. We found a practitioner through his school and he seems really nice, but the whole process is one my son really does not like.

We feel very lucky to have our son, but there are black days when he says, "Why me?" Before we found the neuropsychologist we are using, we tried to find a psychiatrist for this evaluation. Our experience was that some of the people we saw were not beneficial for my son. One place treated patients like a factory – one person saw them when they came in, another on their visits and a third did the neuropsychological evaluation. The first intake person we saw as a family and the second person my son saw on his own. She was dour with no smiles, had a real down attitude, was late for our appointment and asked him questions from a computer, which made my son really angry. In his typical humorous manner, it made him wonder if he should answer her questions falsely ("Did he want to kill anyone? His parents, his teachers?"). Needless to say, we did not go back. A second psychiatrist I pre-interviewed before taking my son. Even though I had asked for someone who could do a neuropsychological exam, after the interview I was told he did not do neuropsychological exams. I am really glad I did not drag my son to that interview.

You suggested that the child interact with the psychiatrist without the parents at first, but I really think that the parents need to interview the psychologist and get a good idea of who they are and how they conduct their business first. It is hard enough convincing a teen to see a psychiatrist. If you have one who is not beneficial, it will make it that much harder to get your child to go to another. It is really hard to find somebody who has an idea of what it is like to live with a chronic

disease, go through the trauma of hospitalization, have to take medicine your whole life and return to the hospital for visits. My son is not actually seeing a psychiatrist for emotional reasons, but even finding someone to do an evaluation has been a real challenge.

Perhaps you could speak on how a parent can find a psychiatrist with a good fit. What questions do they need to ask, where do they find a list of practitioners, and how do they ensure that the psychiatrist cares more about their patients then their bottom line business?

Answer: I wish that your story was not unique, but I am sure that many parents on the forum can relate, as it is consistent with what I have heard from many patients and families. While I sit with children and teens without parents, what I did not mention on the listserv is that I speak by phone with all parents before the first appointment. I like to be sure that I am the best provider and also to explain my style and reasons for it. If parents prefer we handle it differently, we do, which means sometimes my first meeting is with the parents only or sometimes it is with the parent and child. I strongly agree that as the parent you want to do the screening. I recommend that parents ask to speak with the provider or go independently at first. Your son is like others, and each bad experience makes it that much harder to have a good one, even once you find a person whom you think is a good fit!

So how do you find someone? This is tricky in the world of kids with medical complications. In addition to the American Psychological Association, there are also state associations for psychologists (i.e. Ohio Psychological Association). These can be a starting point as they have referral lists on their websites. I also recommend referencing the American Society of Clinical Hypnosis. The website also has a referral list. Your insurance company can give you a list of providers that are in-network and this may or may not be helpful. I went out of network for all panels due to difficulties getting insurance companies to reimburse; this allows me to be more flexible with families because I am not under contract and I do not have to add a psychiatric diagnosis in order to justify appointments. It may also be helpful to talk with a pediatric psychologist on staff at the closest children's hospital. I routinely help families get referred into the community because I could not meet demand. Perhaps the neuropsychologist who you found for the testing may also be a resource. I routinely consult just for the testing and then refer kids out for treatment.

It is hard enough to have a child who needs assistance and so frustrating when you can not find help despite your best efforts! I hope that this provides you with some ideas and that it leads you to the person who can work well with your son!

Occupational Therapy and Sensory Integration

Stormy Hill, M.D., OTR/L – March 2012

Dr. Hill is a licensed occupational therapist and the owner of Busy Bee Therapy Services in Utah. Dr. Hill has a medical degree and a master's degree in occupational therapy with an emphasis in pediatrics and sensory integration. She treats children with a wide spectrum of disorders that have fine and gross motor limitations that hinder their academic and home based functioning. Dr. Hill's experience includes work in school-based settings, private practice, hospitals and a pediatric mental health clinic.

Question: What are some signs or indications that a child may benefit from occupational therapy (OT) and/or sensory integration?

Answer: Your child might benefit from OT services, either clinic based or school based or both, if they are demonstrating delays in fine motor skills, visual motor skills, handwriting challenges, difficulties with "play" or challenges in self-help skills.

In addition, your child may be exhibiting signs of Sensory Processing Disorder. If this is the case, an OT trained in sensory integration framework would be able to work with the child to help integrate the sensory system. Sensory Processing Disorder is a condition in which a child has difficulty taking in certain sorts of sensory input, making sense of the information, integrating that information and executing an adaptive response. Signs of Sensory Processing Disorder often represent being hypersensitive or hyposensitive to one or more of the sensory systems. The seven senses that OT's trained in this area work with are:

1) Tactile: the sense of touch

2) Vestibular: the sense of movement; input from the inner ear about equilibrium, gravitational changes, movement experiences and position in space

3) Proprioception: the sense of position; input from the muscles and joints about body position, weight, pressure, stretch, movement and changes in position

4) Auditory: input relating to sounds

5) Oral: input relating to the mouth

6) Olfactory: input relating to smell

7) Visual: input relating to sight

According to Sensory Integration International (SII), a non-profit corporation concerned with the impact of sensory integrative problems on people's lives, the following are some signs of sensory integration disorder (SID):

1) Oversensitivity to touch, movement, sights, or sounds

2) Under activity to touch, movement, sights, or sounds

3) Tendency to be easily distracted

4) Social and/or emotional problems that may or may not be related to sensory disorder

5) Activity level that is unusually high or unusually low

6) Physical clumsiness or incoordination

7) Impulsive and lacking in self-control

8) Difficulty in making transitions from one situation to another

9) Inability to unwind or calm self

10) Poor self-concept

11) Delays in speech, language or motor skills

12) Delays in academic achievement

Questions: My son is 14 and currently receives OT. He has attention deficit hyperactivity disorder (ADHD) and he has a pervasive developmental disorder not otherwise specified (PDD-NOS). He is easily distracted by sounds and visuals, which makes it hard for him when doing standardized tests. He has balancing problems so learning to ride a bike is difficult. He has problems with motor-planning. He still uses the donut cushion in math class to stay focused, but I would like him to lose it because he is going to high school in the fall. What therapies have you found successful in treating these issues?

Answer: First, I would wonder if your son has accommodations in his individualized education program (IEP) for test taking (standardized and non-standardized). If not, I would inquire about that because there are many that can be made if warranted.

The challenges with riding a bike may be due to balance but could also be due to challenges in bilateral integration so activities that promote bilateral integration

might be helpful. This might include playing catch, carrying containers of toys when cleaning up, opening packages of snacks and baggies of food, emptying the trash cans, clearing the dinner table, zoom football, trapeze bar swing, holding paper down with one hand and drawing with the other, holding paper with the other hand and cutting, or pushing a laundry basket filled with heavy phone books with two hands on the floor.

The wiggle cushion is providing deep tactile input and proprioceptive input if he is bouncing on it. If you would like to phase the cushion out, I would suggest trying to replace that with other activities that provide those sorts of input, like chair push-ups or pulling out on a theraband (resistance band used in physical therapy) that is tied around the legs of the chair.

If he is distracted by sounds and sights then I would work to minimize the amounts of those inputs or at least to have them be metered and expected when possible. For instance, decrease visual clutter in his workspace, increase the contrast between background and foreground to help organize him, and use cubbies and labels to organize him visually (but limited so as not to overwhelm him). Also, his OT can observe and work with him to determine if he is just sensitive to visual input or if he has challenges with visual-spatial and/or visual-motor aspects as well. If so, there are many activities that can assist him with this.

If his auditory hypersensitivity impacts his function, he can cover his ears when needed to self-soothe and regulate. Otherwise wearing a beanie cap, although often not allowed in school, can muffle sounds. There are programs that work on integrating the auditory system (*Therapeutic Listening, Samonas Sound Therapy, The Listening Program*). I have found these programs to be successful. You can ask his school therapist if she knows of any private practitioners in town that do these programs. It can be challenging to modify sound in a busy school environment, but even being placed in front of the room makes sound from the teacher seem more direct and muffles out other voices.

Question: My 21-month-old has overly-sensitive palms. It is something our OT pointed out to me. She said if we do not work with him regarding this problem he could have greater issues down the road. Do you have any suggestions of techniques I can use on them besides basic massaging?

Answer: Does your child have other areas of the body that seem overly sensitive, such as to clothing tags, textures or temperatures? Does your child have sensitivities with foods and aversion to oral textures? The reason I ask is to investigate whether this challenge is specific to palms or appears to be an overall hypersensitivity to touch? You can ask your OT to look at these other areas if she has not done so already.

It is important to try to work to make the palms functional. If they are too sensitive,

then the child gets into a pattern of avoiding, which leads to less exposure and experiences and then leads to further sensitivity. You see the cycle developing? That is what you want to work at to prevent.

A child learns by exploring the world with its body and its mouth (more so initially). When a child bears weight into his palms and shifts weight to and from them, it is helping to develop the arches in the hand as well as provide tactile input to the palms. You can use various textures on the palms, from soft to sticky. Soft is easier to tolerate. Wet, gooey and prickly are harder, so work up to those and introduce them in small increments. Keep it positive.

Activities that encourage tripod position and crawling are beneficial. In addition, at your son's age, using toys that fill the whole hand and use the palm are good for developing these arches as well as fine motor dexterity.

Pediatric Cardiomopathy and Medications

Elizabeth Blum, M.D. – June 2012

Dr. Elizabeth Blume is currently the medical director of the heart failure and transplant program at Boston Children's Hospital. Her research interests include treatment of end-stage heart disease and heart transplantation, the family dynamic, family-physician communication, and the role of the primary care physician in improving treatment compliance and long-term outcomes.

Question: In the past, people with certain heart conditions would receive antibiotics before getting dental work to avoid possible complications. The American Dental Association (ADA) changed their recommendations, and those with hypertrophic cardiomyopathy (HCM) are in the category of "people who took prophylactic antibiotics in the past but no longer need them" (www.ada.org/2157.aspx). Are there special circumstances that would change this recommendation for the pediatric cardiomyopathy community?

Answer: The revised American Heart Association (AHA) guidelines for SBE prophylaxis come from the concept that trying to prevent endocarditis around dental visits was actually not very effective and lead to more allergies to amoxicillin than endocarditis prophylaxis. There is no indication for SBE prophylaxis for children with HCM, and it may actually tend to breed more resistant organisms.

Questions: I was hoping for some advice regarding mitral valve repair in children with dilated cardiomyopathy (DCM). I know each case is individual, but is it possible to determine how long a mitral valve repair will last?

Answer: There is data in adults that mitral valve repair might help in patients with DCM. It is important to distinguish between mitral valve leak from an abnormal mitral valve versus mitral valve leak from the dilated ventricle pulling apart the valve annulus. It is sometimes hard to determine which is which. In adults with abnormal valves, the repair can allow for improvement of function for a few years.

There is no consensus in children about this procedure, and therefore the issue is addressed on a case by case basis as you suggested. The cardiac surgeon's feeling about the ability to repair the valve or narrow down the annulus (area around the valve) is often our best sense. Success varies greatly and recommendations are based on personal experience.

Question: We have all heard about the medication regimens, infection control protocols and restrictions from many transplant centers around the country. It is interesting and somewhat nerve wracking that the treatment of transplant patients varies so widely from center to center. Why is this, in your opinion? Do you think that this will change over the coming years? And what advancements do you

expect to see overall in transplant in the next five to ten years?

Answer: Luckily there are not hundreds of thousands of children who need heart transplants in which to study our medications and our protocols. Also, the outcomes for children following heart transplant, especially in kids with cardio-myopathy, are very good across the board despite variations in protocols. Trying to find the "best" when most variations work really well is very hard to determine in small populations. When there is no scientific data, we all tend to rely on personal experience to manage the "art" of transplant medicine.

Having said that, there are a number of efforts to try to overcome this. Most of the institutions in North America are part of the Pediatric Heart Transplant Study (PHTS) (www.uab.edu/pht), where patients are consented at the time of listing and followed until they are adults. We pool all of our information from over 30 institutions. There are currently 4,366 children who were enrolled at listing. Besides the scientific papers that come from PHTS, each year we all get a report about what we are doing and how we are doing with respect to not only survival, but also infection and rejection. If one center fell below the general North American group, they would know and would be able to change their protocol. There is a lot of this quality assurance initiatives happening at each institution. So far there are not big differences in these outcomes across centers despite some differences in medications, duration or use, and restrictions. Therefore, seemingly large variations in practice do not correspond to large variations of success or complications.

As for the future, the National Institutes of Health has committed to learning more about the pediatric transplant world and has funded a large trial called the Clinical Trials in Organ Transplantation in Children. The first five years allowed seven centers to agree on a common protocol and to study sensitization and development of donor-specific antibodies. Contrary to what you might think, it was actually not that hard to get all of the sites to agree to common drugs, levels, infection protocols and biopsy schedules, so maybe the future will include common protocols. Another round of funding is coming soon.

Lastly, foundations like CCF and PHTS continue to help us learn more about the effects of drugs and transplant on the long-term issues for our heart transplant patients. Focus on the future will include maintaining coronary artery health, preventing and treating post-transplant lymphoma, helping guide transition of young adults to adult providers and understanding the changes around adolescence that put grafts at risk. Preventing the need for transplant with heart failure medical research and advancing mechanical support options for children will also be priorities. There are so many directions of focus; these are my own, and I am sure others could add to this list. The pediatric heart transplant community of physicians, nurse coordinators and social workers is small, and we talk a lot about these issues frequently.

Question: My daughter has just celebrated her one year heart transplant anniversary. My daughter was in advance stages of dilated cardiomyopathy (DCM) and required transplantation within three months of us finding out she had a heart condition.

My question is regarding cytomegalovirus (CMV) infection. How likely can a transplant recipient acquire CMV infections after transplantation? Is it easy to know if a person is going through a CMV as opposed to other kinds of infections? My daughter is doing great, except she has been having coughing fits and has had pneumonia which worries me.

Answer: CMV is a widespread, common virus that can infect almost anyone. Almost everyone gets it by the time they are adults. Most people don't know they have CMV because it rarely causes symptoms. However, if you are on immuno-suppression medications like Tacrolimus, Cellcept or steroids and your immune system is slightly weakened by them, CMV can be a cause for concern.

Once infected with CMV, your body retains the virus for life (like other viruses such as chicken pox). However, CMV usually remains dormant if you are healthy. CMV spreads through body fluids, such as blood, saliva or urine. People with weak immune systems have a greater risk of becoming ill from CMV. CMV can cause cough and respiratory illness and can infect almost all your other organs.

All transplant patients and heart donors are tested for previous CMV infection at the time of transplant, so you or your providers know whether you have been exposed before transplant (positive or negative) and the same with your donor. Most programs use medication to prevent CMV infection early post-transplant when the immune system is at its weakest, if the recipient is negative and the donor is positive or if both are positive.

CMV is easily tested for in several ways. First, you can test for antibodies to see if you have been exposed in the past. You can also do a CMV polymerase chain reaction (PCR) test of the blood to see if there is actual virus in the body. Also, you can test a biopsy sample of almost any organ. Most programs do this routinely with any suspicious illness that is unexplained. If the test results are positive, then you are started on gancyclovir, which can decrease the symptoms from the virus. If a symptom is persistent, you should bring it back to the attention of your doctors and have them help sort out the issues.

Question: My son has never had a biopsy. He has had four catheterizations over 12 years and the last one was using ultrasound. He has had no episodes of rejection. For the last six or seven years he has been on a single immunosuppressant (tacrolimus) plus pravastatin, which he has taken since transplant aged 7 months. Prior to this he also took azathioprine.

I would like to know what the current thinking is regarding children that are transplanted under 1 year of age. Is it possible that their body may accept the organ and be less likely to reject in the long term and/or need less immunosuppressants? Am I taking a big risk only using one immunosuppressant? This decision was made due to the high number of viral infections my son contracted.

I am from the United Kingdom and when my son had his transplant for dilated cardiomyopathy in 2000, we were told to expect five years from the organ. I believe they are now telling families to expect 15 years. What is the current thinking and experience in the United States?

As my son is about to move on to high school, the future has come into sharper focus and I wonder what we can hope for. If he has had no problems this far, can that carry on for long or are things about to change? Do you see sudden change this far out of transplant?

I would also like to ask a very specific question regarding my family. A few months ago my 6 year old daughter was diagnosed with a Wilms tumor and is undergoing treatment. I cannot help but look for connections. Both she and my transplanted son were conceived after fertility treatment, and I have an autoimmune disorder called antiphospholipid syndrome. Have you come across anything similar in any families you have looked after?

Answer: Overall transplant outcomes in cardiomyopathy are relatively equal among centers and among different immunosuppressant protocols and surveillance protocols. I only have experience with the one we use, so it is hard to comment on your protocol specifically. I will say that we have moved to a more "personalized" protocol, where each child is managed with his or her own profile in mind. A child with a lot of infectious or autoimmune complications may only be on one medication, whereas a patient with a history of rejection and no infection may be managed differently. This strategy, although seemingly good per patient, makes it impossible to study or learn from since large numbers of children are not on the exact same protocol. I do think it is important though to reevaluate the risks of infection and rejection over time. Infections that are hard to deal with between 2-4 years of age may be much lower risk now that he is 12 years out. I like the idea of reevaluating longer-term strategies over time in these kids. It is not something we are very good at, since if something is working it is hard for the parents and physicians to get the momentum to change it.

We do tell families now that the half-life of grafts is about 15-18 years now. We told them five to eight years when your son was transplanted. That means that half of the grafts are still working well at that time point (not that all grafts only work for that many years). Certainly, there is evidence that the infant transplants may have an ability to accommodate to their graft better than adults.

No matter how we look at it, there appears to be a higher rate of rejection and graft loss in teenagers, no matter when they are transplanted. Whether this is related to their "risk-taking" phase or the way their body uses the medications, we are not sure. It is a tough time for many kids and families. With close follow-up and education of the kids as they get older, most can get through. We start at their 12 year old check-up to discuss what happened to them, their medications and their role in all this. We continue to hope for many more years (there are kids 20-25 yrs out), but watch closely through the teen years and try to engage them in a positive way (since most teenagers think they are indestructible).

Lastly, I hope your daughter is doing well. We may be smarter eventually to see the connection, but right now I do not know of a connection between Wilms tumor and DCM.

Question: We were wondering if there are any guidelines for an implantable defibrillator in dilated cardiomyopathy. Our son went into congestive heart failure at 4 months, stabilized on medication and remained relatively stable for a number of years. He is now 16 and in the past two years has shown loss of function. He has an automated external defibrillator (AED) protocol, but with his age and desire for increasing independence we have been discussing an implantable. How do you determine when the benefits outweigh the potential risks?

Answer: These questions are all so hard, as we try to extrapolate from adult data and balance the pediatric specific risks that might surprise us. Like so many of our decisions, we are comparing the risks of an implantable cardioverter defibrillator (ICD) with the risks of not having one. The adult guidelines are very clear and depend on having the ICD for many years. Adolescence is such a complicated time. We have seen that risks of arrhythmia do seem to mimic adult risk during adolescence. At 16 years, if he met adult criteria, we would probably move forward in a ventricle that is deteriorating. If there is question, we would go to the lab and do the stimulation test. The stimulation test does not always predict, but it gives you another data point in one direction or the other.

Having said that, the psychological implications in teenagers can be life changing. Some of our kids put their cardiomyopathy out of their minds for 99% of their day. If there is suddenly a metal box in their upper chest to 'remind' them of their risks, they can be paralyzed by this. Careful watch with counseling and education around the procedure is critical for most teenagers. As a parent, the peace of mind related with him having an AICD might then let him maintain his independence. This may factor into the plus side.

One exception for me is in patients awaiting transplant. As I mentioned, the benefit of an AICD is factored in over years of device. If the plan is to transplant in the short term, then sometimes we will hold off on the AICD and not take the risk of

the procedure. We generally agree with the strategy that you took for your son and try to avoid prophylactic implant prior to the teenage years.

Question: I am curious as to what your hospital and team do for asymptomatic hypertrophic cardiomyopathy (HCM) children in regards to medication intervention. What is the current thought on starting diagnosed HCM patients on a beta-blocker or possibly a calcium channel blocker prior to the onset of symptoms in hopes that it will preserve a healthier heart function for the individual down the road or even prevent further hypertrophy? Some cardiologists did believe this a few years ago.

We have had the opportunity to be patients at two HCM specialty centers around the country, and I am sure more are to follow down the road as we finish my husband's active duty military career. Both of my son's doctors did not want to start on any medications until he had symptoms to treat and explained there would be no benefit in starting them before it is necessary. Plus we may have to deal with some unpleasant side effects. This topic has been brought up by many HCM families on this forum, and we have all read the various studies regarding thoughts on this. What is current thinking on this? Are there any new studies being done regarding this very issue? Could you possibly give me an idea of what Boston Children's Hospital believes is best for children that fall into this category or possibly even those that are gene positive (like myself and others in my extended family)?

Answer: As you probably know, there is consensus in adults that there is no evidence to start medications prior to symptoms. Large trials have shown that medications do not affect longevity in adults, and the side effects are frequent. There is no clear consensus on this in children. The one study that many folks refer to is the one where one town was on high dose beta blockers and the other town was untreated. The rate of events was less in the treated town. Different family's mutations and HCM disease may be at very different risks for events. This study may be explained by having all the high risk family members in the same town. The other data is that in some animal models, treating animals before they have any muscle changes seemed to decrease the hypertrophy. In those models, if the animals already were hypertrophic, nothing happened. From the kids we see in Boston for a second opinion, I can see that there is a bias, in general, towards treating children.

In general, we do not think there is an indication for treatment in children with HCM that have no symptoms. The exception is for the kids that are diagnosed in the first few years of life. In infants and young kids less than 4 years old, it is difficult to assess symptoms so I assume they might have them and treat them. At Boston Children's Hospital, we prefer to use a calcium channel blocker, but each child is different. Once someone is treated, we usually do not stop treatment,

even in the older kids. Each child is different, and there are certainly exceptions to this strategy. There is also little evidence in either direction.

I do not know of any prospective treatment trials in children for kids with HCM. We are working on a study looking at gene positive siblings and parents that have normal echocardiograms. Animal data suggests that treating the heart prior to changes may alter the hypertrophy. We have just recently completed enrollment in a study of calcium channel blockers in relatives that are gene positive and echo normal to see if this is true in humans. We are hopeful that this study is renewed, and we can start enrolling again — maybe this time with a different combination of medication. We are anxious to see the results of this early work by Dr. Carolyn Ho at Brigham and Women's Hospital and Boston Children's Hospital. We will keep you posted.

Living with Cardiomyopathy

Matthew Protas, CCF Youth Ambassador — July 2012

Matthew Protas was diagnosed with hypertrophic cardiomyopathy (HCM) in 2005, at age 15. He is now a 22 year old pre-med student at the University of Connecticut. Matt has interned with Dr. Carolyn Ho at the Brigham and Women's Hospital in Boston, and with Dr. Barry Maron and Dr. Hauser at Abbot Northwestern Hospital in Minnesota. Matt hopes to eventually train to be a cardiologist specializing in HCM.

Question: Can you walk us through the emotions you felt right when you were diagnosed, and how and when you came to accept your diagnosis?

Answer: To start off I would like to answer the question by attaching the speech* that I gave last year at CCF's golf outing. This tells the story of my diagnosis and how I found the inner strength to move on with my life and pursue a career in the medical field. Highlights of my speech include:

1) "I felt that my life was over."

2) "Football season began and I tried helping the team but I felt left out. I could not relate to my former teammates. The coach never talked to me because I did not exist to him anymore."

3) "It was really hard to stay focused and care about my schoolwork or anything."

4) "I began to realize that while I cannot change the past, the future is in my control."

5) "My family never gave up on me."

Questions: What things were able to help you go from feeling your life was over to realizing and accepting your "new" life?

Answer: One thing that helped me to get over my diagnosis was going to college. At college I was a new person. No one there knew that I was once the kid who excelled at sports but could no longer play. The key is to find something new that you can be passionate about to take your mind off of your diagnosis. After my diagnosis I played recreational basketball. I could stop at anytime so as to not exert myself, but I still got the same thrill out of it as football and wrestling. I also played recreational softball. My friends and I got together every weekend and played in a men's slow pitch league and had a great time. My cardiologist supported this since I was asymptomatic at the time. Golf is another sport to consider.

Different cardiomyopathies come with different limitations so it is a parent's responsibility to understand their child's condition. Ask your cardiologist about your child's limitations and base your judgments on that. My advice to your child is that they have to be careful and to not take risks unless they are mature enough to understand their condition, their limitations and what they are gambling with.

I would like to share with you what doctors told me when I was diagnosed. First, there is so much unpredictability with cardiomyopathies so it is hard to say, for example, that a person cannot play sports because his wall thickness is above 1.3 mm and has minor systolic anterior motion. They have to come up with a specific standard that can be met for any patient who is diagnosed based on research done by experts in the field and having viewed many cases over the years. One thing that I had to understand was that these risks may seem unfair to someone who does not have any symptoms, but being asymptomatic and phenotype positive there is a significant risk for sudden cardiac death. This is not something that one doctor came up with but rather it took a council of top cardiologist to agree to. Doctors told me this many times, but it was only recently that I began to understand this. Your child will probably not like hearing this either. Children think they are invincible and death is a remote concept, so when doctors explain this to them it does not always click. When they mature they will understand this better.

A child with a hereditary heart disease must find a way to get inner strength. Complications can arise at any time during life and bring on the same negative feelings of the initial diagnosis. Once they find their inner strength, they will be able to handle anything the disease throws at them.

Question: What advice could you give to parents whose son or daughter is in the same position now that you were in then?

Answer: One thing that a parent should not do is to smother their child after the diagnosis. I know that you are suffering as well, but give them space! When I say this I don't mean abandon them, which can be worse, but let them find their own way to cope. Frequently pushing coping methods on your child may be worse than doing nothing. Destructive behavior should be a time a parent intervenes. When I say destructive behavior I mean suicidal ideas, self-harming actions, drug use, antisocial behavior lasting more than two months, a drastic drop in grades and any other behavior that you think could harm your child. If your son or daughter wants to spend some time alone, then let them be.

Question: Our son is 13 and he has hypertrophic obstructive cardiomyopathy (HOCM). He was diagnosed shortly after birth with assymetrical septal hypertrophy. He had a myectomy at the age of 6 on his right ventricle (near pulmonary artery). The surgery was successful and, with the exception of being on atenolol, he has done very well post-surgery. We also have a younger son who is 11 years old and he is gene negative.

Our older son has always known from the time he could understand that he would not be able to participate in varsity high school athletics. He is an athlete and that is what he loves! We allowed him to participate in sports throughout grade school (football, basketball, hockey, baseball) with the blessing of our cardiologist and active monitoring from us. We were always involved. We are glad that he was able to participate in these sports, and as our son will tell you, “I am glad I was able to participate for as long as I could than to never have had the opportunity at all.”

A year ago we took him to see Dr. Jeffrey Towbin for a second opinion. He advised us to remove him from baseball, which was the last sport he was participating in, hence ending his sports career. We knew it was coming to an end any way because sports conditioning was becoming a bigger part of the picture. This is of course where the issues lie, but it was and still is hard for our son to accept. To complicate this situation, our younger son is participating in sports.

We did not realize how difficult it would be for our son. It has been an extremely difficult path to travel with a boy whose gift is being physical and then having a younger son who is just spreading his wings in sports, while his brother sits painfully on the sidelines and watches. Until our older son accepts it, he will not be able to move forward, try other things or open new doors. Right now he is just mad, sad, angry, mean, apathetic and just plain difficult. He's tried being the manager of his football team but as he says it, “I don't want to hand my friends a water bottle, when I am as good as them. I should be on the team.” He was chosen to be honorary captain of his baseball team last year. When they asked him again this year, it caught him off guard and he got so choked up he walked away with tears streaming down his face, a big knot in his throat, head down and embarrassed. Later that evening we asked him if he was going to do it; we thought it would be a nice honor again. He said, “if I can’t play then I do not want to be in the dugout!” So it comes and goes; every day is different. He does not want to try new things or experience anything other than sports right now. It is like an obsession. Most of all he just wants to be treated like a normal kid and does not want to talk about it. He says he can do whatever sport he wants; the doctors do not know how he feels or what he can do. He is not a sick kid and you could never tell by looking at him that he has a heart problem. He does have an extremely thick septum and issues with the heat and endurance, but otherwise he has done very well. He does have a lot of friends. He is not a loner and he is very social so that works in his favor as well.

Answer: Your son is going through the worst of it right now; hopefully it will get better. My advice is to not push your son into finding something else that he can enjoy as much as sports. Getting over a diagnosis with sports restriction is similar to getting over being dumped by a girlfriend. Afterwards, friends will usually try to introduce new girls but the person is only interested in the old girlfriend. Eventually the person will stumble upon someone new to date. They won’t forget the good

times they had with their old girlfriend though. The difference between getting dumped by a girlfriend and your son's situation is it will take more time for your son to get over his situation. Presenting your son with new options directly may not work. What you could try is to suggest something to him that he can form a passion for on his own. Take him to a store with something he may enjoy. A good example is to take him into a music store; he may find that he enjoys playing the guitar. Also try to explain to your younger son what his brother is going through so that he can avoid talking about sports in front of him. It is your job to stay strong even though I know it is hard.

Question: Our extended family has a hard time coming to terms with the unpredictability of the disease. Do you have any advice to give our extended family and friends as to how they should treat our affected child? Did you appreciate others asking about your health or did you wish it could go unnoticed? Did people ever go too far to be overly kind to you? Our little girl is only 3 years old, but I am curious to hear your perspective and advise friends and family on how to proceed.

Answer: There is no easy way to come to terms with the unpredictability of the disease. Hopefully one day in the future this problem will be fixed. I generally do not prefer people asking about my condition. It is not something I typically bring up in conversation unless it is necessary. Recently I have been having complications, and it bothers me when I am constantly asked what is going on every time I have a new test. When I answer, people usually do not understand because they have no medical background or misinterpret the results. Asking how I am feeling is fine and sufficient; I understand that they care. People were overly nice to me at first, and I highly suggest that you do not do it. Ask anyone with any sort of disease or disorder and they will tell you that all they want is to be treated like normal.

Heart Transplant Concerns

Anne Dipchand, M.D., F.R.C.P.C. – September 2012

Anne Dipchand, M.D., F.R.C.P.C. is currently an associate professor with the University of Toronto and a staff cardiologist and head of the heart transplant program at The Hospital for Sick Children in Toronto. Dr. Dipchand's research interests include transplant outcomes, pediatric cardiomyopathies and echocardiography.

Question: Can you describe any new or recent developments in pediatric heart transplantation that would be helpful to our community?

Answer: I would say that there are many issues that come to mind when I think of heart transplant developments in children. The ones that come to my mind immediately include:

1) Advances in the understanding and medical management of antibodies before and after a heart transplant. Antibodies are proteins that our bodies make against "foreign" things like donor organs. More and more kids have antibodies before a transplant because of previous surgeries. These antibodies might be against a possible donor heart. The ability to find the antibodies, figure out the chance of them being against a donor heart and to deal with them has changed so much in the last few years that it has impacted our practice in children who have these antibodies. The problem is that these antibodies can cause rejection as early as right away in the operating room. If we try to find a donor that "matches," a lot of patients will die waiting. If we transplant a patient anyway, we worry that their new heart won't last as long. There is a lot of research going on to figure out how to determine if an antibody is important, how strong it is, how to manage it and what it might mean after a heart transplant. There is a unique study being funded by the National Institutes of Health looking at these antibodies in children.

2) Of particular importance to children with cardiomyopathies is the technology that is developing for ventricular assist devices (VADs). Though more frequent in adults, there are children that do and could benefit from these machines which help a poorly functioning heart to supply the needs of the body. The problem is side effects like clots, strokes and infections amongst other things. We need devices that are designed specifically for kids and not just scaled-down versions of adult devices. The Excor Berlin Heart very recently received FDA approval in the US, which is a big development. In addition, the NIH is funding a study to develop pediatric-specific devices, which is almost ready to move into clinical trials in patients — another huge development for the pediatric community.

3) More studies and discussions on the best ways to allocate donor organs are ongoing. For example, recent publications that show equivalent results for babies, whether they receive a compatible (matched) or incompatible (not matched) blood type, are leading to changes in the United Network for Organ Sharing allocation policies which will lead to less children dying on the waiting list and a greater use of donor organs when they become available. This also applies to more research in the understanding of when patients should or should not be listed for a transplant, which is also important to understand.

Questions: My son is almost 5 1/2 months old and he has left ventricular non-compaction (LVNC), Ebstein's anomaly and a ventricular septal defect. They tell me that he will most certainly have to have a transplant, but they do not know when yet. Have you ever seen a case of an infant with left ventricle non-compaction that never ended up needing a transplant? Is it okay for me to let him exert himself in any way, like tummy time or crying? I worry that because of his cardiomyopathy he will tire out and that it may be making things worse. And what do you think is the likelihood of the people who made the Total Artificial Heart, or anyone else for that matter, making one small enough for an infant in the near future?

Answer: Thank you for sharing this. Although I cannot make specific comments about your son's situation, in general, I have seen and followed patients with LVNC who have not yet gone on to require heart transplantation. LVNC is a very challenging problem in that it really describes what the heart muscle looks like on an echocardiogram or through magnetic resonance imaging, but there are likely many causes — just like other types of cardiomyopathy. What eventually happens will depend on the underlying cause to a certain extent, but often we do not know the specific cause. So we are left in a situation where we have to follow a patient clinically for any signs and symptoms of heart failure and follow their response to medications. What happens later depends more on these things. Just having a diagnosis of LVNC does not guarantee a need for a heart transplant — how the child is doing medically is usually the most important consideration. Your medical team would be best poised to give you an idea of what the future holds for your son.

In general, for any young cardiomyopathy patient with heart failure or not normal heart function, it is not realistic to keep them from doing age-appropriate and developmentally appropriate activities like tummy time and even crying. I generally tell my patients that an infant or a toddler doing age-appropriate activities is not going to cause his or her heart to become weaker or worse. In addition, these activities are important from a normal development point-of-view. It is much easier to make activity recommendations for older kids (e.g. no hockey, no weight lifting). Again, for an individual child, it is best to take your cue from your medical team.

Earlier I referred to the research in VADs. There are evolving technologies and

people are working very hard to make devices that are small, able to function in a child, and do not have the complications of the existing devices. I do think that we will have something within the next three to seven years.

Question: My son, who has just turned 8 years old, is currently on the transplant list as status II. My son had open-heart surgery when he was 15 months old. It was supposed to be a normal ventricular septal defect repair but his post-operative course was complicated by tachyarrhythmias requiring extracorporeal membrane oxygenation (ECMO). This resulted in decreased ventricular function and secondary dilated cardiomyopthy with many blood infections.

Do you think that with all the medical problems he suffered before it will affect him post transplant? He also has DiGeorge's syndrome. His ejection fraction has been on a slow steady downfall to 22% with symptoms of fatigue, throwing up and sweating. At the last check up his function was back up to 40%.

Answer: Thank you for sharing your family's story with us. Again I cannot comment on anything specific to your son's case. However, in general, in any patient who might need a heart transplant, the assessment for a heart transplant is very important as it identifies any existing or past medical problems that may affect how things go at the time of a transplant or after the transplant. One example is kidney function. Many patients who have had heart surgery, ECMO or have decreased heart function can have decreased kidney function. Some of this is reversible; some is not. Kidney function at the time of the surgery can affect how things progress in the intensive care unit and the recovery from the surgery. In the time after the transplant, over years, some of the transplant medications can affect kidney function. This is just one example. For your own child, your medical team can talk to you about what to expect at the time of the heart transplant and after the heart transplant based on what they know specifically about your child.

There is not a lot of online information about general concerns after a heart transplant but CCF is partnering with a group called the Pediatric Heart Transplant Study Foundation to make some materials available for parents. I am sure that you will hear about it when it is available in a few months.

Question: Do you know of any strides being made in treating restrictive cardio-myopathy (RCM) that could result in slowing down the progress of this disease and delaying transplantation?

Answer: Unfortunately no. RCM is such a challenging diagnosis. Again it is a diagnosis that is based on clinical findings, imaging and a specific pattern of pressures in the heart. RCM can be caused by many different things. Even when we find the cause, it is usually not treatable. Most of the time we do not find the cause or it is genetic. Our best treatment is to treat the symptoms and signs of

heart failure — not the actual RCM. The progression of the disease is different in each child.

Question: What are your thoughts about the continuation of drug therapy (e.g. beta blockers) for a child with dilated cardiomyopathy (DCM) who has been stable for over two years?

My son was diagnosed at 8 months with DCM and is now almost 7 years old. For the last three to four years he has been taking carvedilol only and has remained stable with an ejection fraction around 55%. Our cardiologist is talking about eliminating the beta-blocker. Is this the recommended action? What is the common practice regarding continuation and discontinuation of drug therapy for children with what appears to be resolved DCM?

Answer: In our cardiomyopathy and heart function clinic, we do eventually wean and stop anti-heart failure medications in patients with cardiomyopathy who resolve. There is no common practice in terms of if, when and how as there is not enough evidence in the literature to say that there is a way that works best. Each physician or program develops their own practice and, of course, it also depends on the circumstances of the individual patient.

Question: My daughter has dilated cardiomyopathy (DCM), which her cardiologist believes was caused by the anthracycline 250 mg/m2 she received when she had leukemia. She is now 9 years old and has been in remission for seven years. She also has Trisomy 21. My questions are:

1) Does chemotherapy increase the risks of complications from a heart transplant?

2) Does having Trisomy 21 impact on the success of a heart transplant?

3) From your experience of children who have anthracycline-related DCM, how does DCM progress over time and are there developments that may delay the need for transplant?

Answer: Chemotherapy in and of itself does not increase the risk of complications from a heart transplant. It really depends on whether the individual patient has any long-term complications of the chemotherapy. The implication of any medical problems can best be explained to you by your medical team. I gave an example of kidney function in my previous response.

It is the same answer regarding the Trisomy 21. Anything that may affect the success of the transplant would be identified at the time of the assessment and the impact explained to your family by the medical team. There are possible things like the risk of cancer, thyroid function problems, lung problems and other things

that may be present in a child with Trisomy 21. But every child is different.

Finally, the progression of heart failure in a child with anthracycline-induced cardiomyopathy is variable. It can happen very quickly at the time of the treatment or over many years. There is not a good way to predict. Also, it can remain stable for many years with heart failure treatment. Though there is nothing new right now to specifically slow down the progression, there is research and increasing experience with medications given at the time of chemotherapy treatment, which may decrease the risk of developing cardiomyopathy in the future.

Question: Have you ever known idiopathic dilated cardiomyopathy (DCM) to reoccur in a transplanted heart? If so what are the implications? Is it more dangerous than DCM in one's own original heart?

Answer: By nature of the fact that the initial cardiomyopathy was "idiopathic" – meaning the cause was unknown, it is highly unlikely to recur in a transplanted heart. It would be theoretically possible if it were a disease that affected the whole body that causes something to accumulate in the heart. Usually in these cases, you have a diagnosis or a suspicion of a diagnosis from the explanted heart or other testing. The most common reason for a transplanted heart to get "dilated cardiomyopathy" is either rejection (of any kind) or transplant coronary artery vasculopathy (coronary artery disease). The implications would depend on why it occurred. Generally it is not a good outcome.

Nutrition and Cardiomyopathy

Tracie Miller, M.D. – December 2012

Dr. Miller is the Associate Chair of Pediatrics for Clinical Research, Director of the Division of Clinical Research and Professor of Pediatrics at the University of Miami Health System in Florida. Her specialties include general pediatrics and pediatric gastroenterology. Her research interests include studying the effects of nutrition and exercise rehabilitation in children with chronic illness, and cardiovascular risk factors in childhood.

Question: Recently there have been articles that have suggested there may be a link between dilated cardiomyopathy (DCM) and celiac disease (CD). In patients with DCM, are there specific symptoms to be aware of that may suggest CD?

Answer: There are reports of people who have both cardiomyopathy and CD, although the association is rare, at least what has so far been reported. Many of the reports are of just one or a few patients. However, this year there was a large epidemiological study from Sweden where they have a national registry of 29,000 people with biopsy proven CD. They compared these patients with CD to 144,000 patients who did not have CD and then determined the prevalence of DCM in each group. They looked at how many of each group developed DCM. Of the 29,000 patients with CD, 17 developed DCM and of the 144,000 patients without CD, 52 developed DCM. The patients with CD were at a higher risk of developing DCM, but this did not reach statistical significance. This is a very large study – one that would be hard to replicate in the US. It does show a suggestion of a possible modest likelihood, but this is a modest risk at best. One has to be careful in interpreting these associations, considering CD is prevalent in 1 out of 133 people. If one mixes a fairly common illness with a rarer one, the associations can be “true, true and unrelated.” That is why research is so important.

Celiac disease can cause very severe nutritional deficiencies, but it can also be very quiescent. Nutritional deficiencies alone can cause cardiac dysfunction (selenium, carnitine, iron, etc...). The classic symptoms of CD include abdominal pain, bloating, gas, growth issues and constipation. However, many patients are asymptomatic. For instance there is a fairly common association between type 1 diabetes and CD. Many kids that I diagnose with CD, who are referred to me because they have diabetes, are totally asymptomatic.

I would like to advise some caution on this. I would not advocate putting a child with DCM, or even any child, on gluten-free diet until appropriate testing has been done for celiac disease. The elimination of gluten is a major dietary change and may predispose the child to take in less calories than he or she should (because their favorite foods are taken away or gluten-free foods may not taste as good). In a child who may be predisposed to growth problems because of their heart

problems, this could cause a serious decrease in calories and result in even worse growth failure.

Questions: Our 13 year old has hypertrophic obstructive cardiomyopathy (HOCM). He is on 75 mg of atenolol a day. How does digestion affect the cardiomyopathy heart? He has never been a good eater. He eats small meals all day long. He does not like to be full and he tends to not feel very well if he overeats. The atenolol seems to diminish his appetite after he takes it.

He tends to not eat breakfast or he eats a lot late at night before he takes his next dose of Atenolol. We have basically let him eat the way he wants. He likes all foods and has a good diet. He is not a big meat eater but he eats a lot of pasta. He says meat makes him feel sick. I tend to chalk that up to the fact that meat is a much harder food to digest and it taxes the heart and digestive/blood system during its digestion. Do you have any suggestions on eating or how to help him?

I am also assuming that this is how our son's eating will be throughout his life and it is best to let him eat the way he feels best.

Answer: Digestion or gastrointestinal (GI) symptoms can be a problem for children who have heart conditions. As you know, the heart circulates blood to vital organs, including the intestinal tract. If the blood supply is compromised, this can cause problems in absorbing nutrients as well as causing the intestine not to move as well as it should. I am sure your cardiologist is optimizing his heart function. Children with heart conditions can also have gastritis, which can cause pain and problems with the stomach emptying. It is hard to tell with your son; it depends on how impaired his heart function is. If your child is growing well, that suggests that his intestine is absorbing nutrients well. Growth is the best measure of a functioning and healthy GI tract. He may have an element of slow stomach emptying which can cause decreased appetite. Eating small frequent meals that are relatively low in fat (if he is growing well) might be best. Also, thinking about a trial of an acid blocker like Zantac may be reasonable. I would check with your cardiologist and primary doctor about this as you want to make sure that the medicines he is on do not interact adversely with a new medicine. You also need to discuss with your doctors about any side effects from medications he is on. Some can cause nausea or other GI symptoms. If warranted by his healthcare team, an evaluation by a gastroenterologist may also be valuable.

Question: I am curious about the link between beta-blockers and hypoglycemia. Our son is almost 4 years old and takes 10 mg atenolol twice daily. He was diagnosed with hypertropic obstructive cardiomyopathy (HOCM) at 3 months and started on propranolol right away. He had other issues with propranolol so we switched him last November to atenolol. He is a very big 3 1/2 year old — 50 pounds and 3 1/2 feet tall with frequent premature ventricular contractions — so that is why

he is on a high dose of atenolol. I have read and heard other people talk about beta-blockers causing hypoglycemia but his cardiologists do not seem to think it is anything to worry about.

Our son had his first seizure this past year and the emergency room (ER) doctors called it a febrile seizure, but he had no fever before the seizure and his temperature was 102 degrees after the seizure. He has spiked from normal to almost 105 degrees in the past with no issues. The ER doctors were not able to find anything wrong except “possibly a mild ear infection” and his temperature returned to normal after about an hour. They did urine culture, blood cultures, and chest and abdominal x-rays. He did vomit everything he ate the night before and then his breakfast the next morning at school. After we got something in his stomach he returned to normal. He also had what I think was a hypoglycemic episode about a month ago after not eating a very good dinner the night before. He was complaining of stomach pain when he woke up and then he turned pale, sweaty and started shaking (not seizing). I gave him a drink of sweet tea and his symptoms improved. Then he ate breakfast and seemed normal other than being a tad lethargic for about an hour after eating.

I also need to mention that both of my grandfathers are diabetic, both of my husband's grandfathers were diabetic, and my husband's sister was diagnosed with juvenile diabetes at age 9 and is insulin dependent still at age 25. Do you think that these episodes could just be random or should I push for some kind of appointment either for medication issues or diabetes screening?

Answer: It is hard to know exactly what is going on. Certainly, some of the symptoms you describe are consistent with hypoglycemia, but they are not very specific either — meaning they can be caused by a number of other things. You should bring your concerns to both your cardiologist and primary care doctor. I have not heard that hypoglycemia is that common when using those medications, but it has been reported. Also I am not a cardiologist, so I do not have as extensive experience in this area as your child’s doctor.

Question: Our son is two months old and was diagnosed with non-compaction cardiomyopathy and a severe mitral and tricuspid regurgitation at two weeks old. He is now under medication (Lasix, Capoten, and digoxin). He is having difficulty breathing and has not gained weight. Do you think that replacing his valves would make him better?

Answer: It sounds like your son is pretty fragile now with some difficulty breathing, poor weight gain and is very young. You should have him reevaluated by his cardiologist as soon as possible to make sure they are doing all they can to help his heart function the best possible way. As I mentioned before, growth is an excellent sign of health. You want to make sure he is getting all of the calories he

needs. With children who have heart conditions, they can need up to 50% more calories per day than a child who has normal heart function. Write down what he is getting in every day for a few days and review this with his doctor. His doctors and nutritionists can often recommend ways to get more calories into him.

Question: Our 4 year old son with dilated cardiomyopathy had his six month evaluation a few weeks ago. He measured and weighed the exact same as he did 6 months prior. His heart function and size are within normal ranges and he's on enalapril. We are starting to think there might be something else going on that we haven't diagnosed yet such as a connective tissue disorder. Our son is a picky eater probably due to his age. What supplements can we entertain giving him to boost his caloric intake? What about Duocal or Ensure shakes?

Answer: No growth in six months deserves some attention. First you need to document exactly what he is taking in and based on that information, a good plan can be put in place to hopefully provide him with more calories. Pediasure is an excellent high calorie supplement. Please keep in mind that a tablespoon of butter or oil has 100 calories! This can be easily disguised in most foods several times per day. It is a pretty high caloric "bang for your buck." Again, growth is a measure of health in children, so make sure you discuss with your cardiologist if there are any concerns about his heart function. If everything is fine with his heart, you can see a pediatric nutritionist for guidance as well.

Pediatric Cardiac Social Work

Rachel Justus, L.M.S.W. and Anna Zelig, L.M.S.W. – February 2013

Both Rachel Justus and Anna Zelig are pediatric social workers at the Kravis Children's Hospital at Mount Sinai Hospital. They both work with children and families followed by the pediatric cardiology, cardio-thoracic surgery and cardiac transplant services. In the past Ms. Justus worked with general pediatric patients and the pre-surgical preparation program. Ms. Zelig also worked with general pediatric patients and is on the Family Advisory Council at Mount Sinai.

Question: What kinds of general services are offered by a pediatric social worker for children with cardiomyopathy and a heart transplant?

Answer: The pediatric cardiology social workers work closely with the multi-disciplinary team to provide support to patients and their families in a variety of ways. Having a child with a cardiac condition can be a stressful and challenging experience for the whole family. Some of the primary roles of the social workers are to provide developmentally-appropriate emotional support, guidance and counseling to the patient and family members before, during and after the child's hospitalization. The social worker uses a family centered approach to assisting each family, adjusting to the challenges of both acute and chronic cardiac illness.

Social workers collaborate with other members of the medical team to make sure that each child is discharged safely from the hospital with necessary supplies and services. In addition, the social worker can help connect families to important resources in the community. Because of the diverse population of patients treated at Mount Sinai Hospital, the social worker can arrange translation services for any family, if needed.

We also offer intensive services to pre- and post-transplant patients and their families. Attached is a brochure that we distribute to families detailing the psychosocial programs available.

Question: My question has to do with the appropriateness of and/or need for therapy for a child who had a transplant at four and a half months old, another open heart surgery at three years old (to fix a valve), has just turned four and seems relatively well adjusted. I know that there are a lot of problems with tweens and teens becoming resistant to taking their medications. I figure that working through some issues before they become problems may help.

Should you wait until there are signs of problems or struggles before you take a child to see someone or is it better to do it earlier? At what age would you do that if there

are no apparent issues? Should we seek out someone at our transplant facility that works with “cardiac kids” specifically or is just a general therapist in our home city acceptable? We live about four hours away from our transplant hospital.

Answer: It sounds like you have experienced a lot. We believe that the transplant experience is stressful for each and every member of the patient’s family. Therefore, we strongly recommend seeking supportive counseling to help the entire family cope with life post-transplant. As kids (both patients and siblings) enter new developmental stages, fresh questions might come up regarding the transplant. Having a therapist in place can help make sure any questions or issues that arise are dealt with in a timely, healthy and appropriate manner. My suggestion would be to speak to a bunch of therapists in your home city and assess their comfort level in dealing with families where there had been a medical issue. I would ideally look for someone within your insurance plan, as mental health expenses can add up. It sounds like your child is already doing a great job adjusting and hopefully will continue to thrive!

Question: Our son was in the intensive care unit for four months as a baby (from four months old to eight months old). He had a very tumultuous time with lots of nterventions (intubation, Broviac line implantation, sepsis, spinal tap, peripherally inserted central catheter lines in his scalp), a few emergency room visits and a few other hospitalizations a week long (gastrostomy tube placement, pneumonia).

He is almost five years old now and is starting to show more signs of terror around certain types of routine but necessary things that are painless such as visiting the dentist and getting a haircut. The reactions seem to be getting more intense the further away we are from the hospital time. Haircuts result in a total and complete freak out, with me holding him down and the hairdresser trying to cut as fast as she can without cutting any of us. Within one or two minutes of it being over, he is happy, cheerful and wanting to give the hairdresser a hug. It is the same with the dentist.

My wife, who is a school age social worker, thinks it is getting “worse” because he is becoming more aware of himself and his own body. He definitely checked out of his body a couple of times as a baby (sleeping during really stressful things), and it has been a slow process to get him back in it. His gross and fine motor skills are pretty low, and he is still not potty trained, in part because he does not even seem to notice a wet or dirty diaper.

I am not sure how to best help him. Should we 1) keep working towards getting back in his body and owning that it is his or 2) get through these kinds of appointments without traumatizing him and/or the providers? We try to do lots of talking in advance of whatever the appointment is, try to work it out through play at home, but when it comes to the moment, he is completely out of his mind with terror. It is awful for everyone. Do you have any thoughts on how to help make these kinds of interactions less terrible?

Answer: Your son's reactions seem really appropriate given his medical history. I agree with your wife – as he is getting older, he is probably becoming more aware of not only his body, but also his past medical experiences and the feelings associated with them. I would strongly suggest trying to connect to a professional in your community who is adept at working with kids in a variety of modalities. For example, we have found that at your son's age, talking therapy is not always the most helpful way to process emotions and reactions. Sometimes art, play or music therapy can be more effective depending on the child. Regarding current stressors, such as haircuts or dentist visits, we have had a lot of success in helping kids cope with anxiety-provoking experiences using hypnotherapy techniques such a deep breathing, guided visualization, blowing bubbles, etc. Portable electronic devices such as iPhones, DVD players or handheld games can also be useful distractions during these challenging times. You may ask your son which of these techniques he thinks might be most helpful next time he needs a haircut or has a dentist visit. There's also nothing wrong with giving him a treat after he has successfully completed his difficult task – you can decide whether the best reward would be ice cream, a cuddle or some stickers.

Question: Any advice on what kinds of activities are safe for our children? I have mixed reviews from doctors but am eager to keep my son physically and mentally fit. He is a former hockey and lacrosse player so we have had an extremely difficult time with this. Right now, he has mild hypertrophic cardiomyopathy but it's not uncommon for HCM to grow during puberty. We are scared to death!

Answer: We checked with our medical team and they emphasized that every single patient is unique and you really need to consult with your child's specific team about their individual medical situation so you can help figure out what physical activities might be appropriate. We recommend bringing a list of questions to each medical appointment. This would be a really appropriate topic to discuss at your child's next scheduled visit.

Question: I know my son is very lucky to have a mild case of left ventricular non-compaction cardiomyopathy, as many face much greater challenges. Nonetheless, I worry about the day when we have to tell my son he cannot play sports because of his heart condition. In the whole scheme of things this is a minor setback, but my husband and I both grew up playing sports. At our particular schools everyone was into sports so the social aspect was very important. While I hate for him to miss out on anything, I worry the most about the social impact it will have on him. We grew up going to smaller schools so I have considered making sure he goes to a big school in the hopes that the greater number of kids in his class and available opportunities will help minimize any feelings of isolation. We are going to try to help him find other hobbies and do things that the entire family can do together, but I want to make sure he has the healthy, happy, social experience every kid deserves. Any

suggestions you can provide for parents who worry about the social impact of heart disease would very much be appreciated. I played sports all through high school not knowing I had the condition, which makes accepting this even harder, but it just is not worth the risk. The first bad heart rhythm can be the last.

Question: I would like to ask your opinion on how to deal with the situation where there are active, healthy kids in the family who want to play sports. How do you keep everyone happy? We are trying to direct our five-year-old son with dilated cardiomyopathy to sports such as golf, tae kwon do, fishing, etc. He always wants to do what his nine-year-old brother is doing, which is basketball and football. For now this is fine because he sits down when he can't go anymore, but this won't last. Plus, he has a twin brother, and I don't want his twin to miss out on any opportunities.

Answer: It sounds like a lot of families with active, school-aged kids are dealing with the same challenges. At our hospital, we implemented a peer mentoring program called Heart-to-Heart, which matches more newly-diagnosed cardiac patients with patients and families with a similar diagnosis who are further along in their journey. We have found that for the kids (and their parents) being connected to someone else in a similar situation, who can relate firsthand to athletic restrictions challenges, can be incredibly validating. In the words of one family, who was recently matched with a peer mentor, making this connection helped them understand that they are not alone. Knowing that other people out there are going through the same thing is comforting. Perhaps you can speak to your medical team about being introduced to another pre-screened family in your geographic area that has kid(s) that are appropriate age(s) to be matched with your family. Our adolescent patients love being considered "experts" and have really tried to use their own experiences to help other kids! In addition, speak to your medical teams about what physical activities are permissible. Based on that, you can compile a list of options and show that to your kids. Let them choose what sounds cool and fun to them. While golf may not be appealing, your son may absolutely love the idea of archery. Making them active participants in the selection process can help them feel empowered and special. Maybe your other kids will even want to join their siblings in their new athletic endeavors. For families with multiple siblings, combining non-competitive physical family activities that everyone can do together, such as hiking or biking, allows each child to have something that is unique to them and helps everyone feel good. Whatever the decision, always check with your child's medical team before starting any new physical activity or if you have any questions about athletic restrictions.

Left Ventricular Non-Compaction (LVNC)

Stephanie Ware, M.D., Ph.D., F.A.C.M.G. – April 2013

Dr. Ware is an Associate Professor of Pediatrics at the University of Cincinnati College of Medicine as well as the Co-Director of Cardiovascular Genetics at the Heart Institute and Director of Research and Development at the Heart Institute Diagnostic Laboratory. Dr. Ware's research interests include the genetics and development disorders of heart structure and function, and her research laboratory has made significant contributions in the areas of congenital heart defects and cardiomyopathy. At Cincinnati Children's Hospital, Dr. Ware evaluates and manages patients with genetic disorders and has specific expertise in cardio-myopathy and syndromes with cardiovascular disease.

Question: How and why did left ventricular non-compaction (LVNC) become recognized as a form of heart disease?

Answer: LVNC was first recognized over 80 years ago in association with congenital heart disease. The American Heart Association increasingly recognized LVNC in the 1990s as being associated with heart muscle disease, and the World Health Organization classified it as a form of cardiomyopathy in 1996 and in 2006. So, LVNC is a relatively newly recognized form of cardiomyopathy and it has variable features. Therefore, not everyone with LVNC has the same symptoms, findings on imaging or prognosis.

Question: Our son was diagnosed with LVNC, three years ago, at the age of 11. He has no other heart defects, so as of now he just has yearly appointments and takes a baby aspirin every day. We have seen different doctors and they all seem to think differently about LVNC as far as our son's limitations. One doctor said he was to play no sports whatsoever, but then another doctor said he should play sports and be active. Is this because there just is not enough information yet about LVNC and what the long-term effects can be? We want to keep our child safe but the difference of opinions are alarming and do not give us a straight forward answer to what our child can do or cannot do. To us there is just not enough information about LVNC.

Answer: LVNC can have a variety of clinical outcomes. The term LVNC describes the way that the heart muscle is formed or looks on imaging but does not necessarily imply that the heart muscle function is not normal. There are different "flavors" of LVNC. In some cases, it takes a dilated form where the heart muscle does not pump well. In other cases, it is associated with a hypertrophic form, where the heart muscle is too thick and does not relax well. In some cases, it may alternate between the dilated and hypertrophic forms. In some cases, the non-compaction is present, but there is no evidence that the heart muscle does not function normally (benign form). Each of these LVNC forms carries a different set of symptoms and prognosis – and it

can lead to confusion for families and different answers from physicians. You are correct that there is not as much information about LVNC as the other cardiomyopathies, because it was described relatively recently.

Question: My son was diagnosed with LVNC last year at age 17 and currently plays high school football. We have been to several cardiologists and have been given conflicting information about his limitations. He currently has no other health problems and but we were told that he could continue to play sports. Another doctor was against him continuing to play. Is it safe for a child diagnosed with LVNC with no other health issues to play recreational sports?

Answer: In my last response, I described the different forms that LVNC can take. It will be important to discuss all of the specifics of your son's diagnosis with his physicians. At our center, we do not generally restrict activity for patients with the benign form of LVNC, in which there is no evidence of heart muscle dysfunction after required testing. This must be customized to each patient and family.

Question: What percentages of patients develop irregular heart rhythms over time as they age? I ask because I was diagnosed with LVNC at age 27 upon the birth and diagnosis of my son. Following my diagnosis and a Holter test, it was recommended that I have an ICD/pacemaker implanted. Unfortunately the first time anyone recorded my heart rhythms was at age 27 so I do not know if I have always had irregular heart rhythms or if they developed as I aged. My son who is now 2 1/2 years old has had regular annual Holter readings. What evidence do you have for the likelihood that my son would continue with regular heart rhythms versus developing irregular heart rhythms over time which would need to be addressed like my situation.

Answer: That is a great question. Unfortunately, we do not yet have good longitudinal data on large numbers of patients with LVNC. Irregular heart rhythms are commonly associated with LVNC and the fact that you have an abnormal heart rhythm means that your son is also at higher risk. However, we do not have the ability to predict when or if that might occur.

Question: Last week we got the results back from the Gene DX panel we had done on my 11 month old son who has dilated cardiomyopathy. The results were "hemizygous for a novel frameshift mutation in the TAZ gene, consistent with a genetic form of cardiomypathy or Barth syndrome." We are just in the beginning phases of figuring out what this means. We have met with our two cardiologists, and neither has said that he definitely has Barth Syndrome, but that he most likely does. My questions are:

1) What would determine if he does in fact have Barth Syndrome? We are waiting for the results of my blood test that we did last week to see if I am a carrier.

2) From my understanding, his echocardiogram shows that he does not have LVNC. Could he have isolated LVNC that is presenting itself as a severe case of DCM, even though his echocardiogram does not show it?

3) From your experience, how many kids have you seen with Barth Syndrome, and is there a general outcome or trend you have found relating to their cardiomypathy?

Answer: I would suggest that you also see a geneticist if you have not already done because genetic testing results could be interpreted in the context of his signs and symptoms. Based on the information you provided, it is very likely that he has Barth syndrome but I would have to see the exact genetic testing results. If this is indeed his diagnosis, then he will require care from additional specialists and that should be initiated as soon as possible. I see the posts from the others with children with Barth syndrome, and I would recommend contacting the Barth Syndrome Foundation.

With regard to your questions:

1) The genetic testing results in combination with the physical findings determine whether he has Barth syndrome. Your carrier testing will not influence this, although it will have significant implications for your recurrence risk if you are planning on having more children.

2) LVNC is commonly seen with DCM and in a very dilated heart, the trabeculations may be more difficult to appreciate. In addition, children with Barth often have an undulating phenotype, where the appearance of the heart, including presence of LVNC, appears to change over time.

3) I have seen several children with Barth clinically, and many more at the Barth Syndrome Foundation meeting. With regard to the cardiomyopathy, a general trend is that the first year of life is difficult, and at some point after that many of the toddler age children and beyond stabilize and have a prolonged “honeymoon period” where their heart function is very stable. Children with Barth have more difficulty with their immune system and with growth. Routine illnesses can affect the heart function more severely than one would predict and children with Barth syndrome need to be monitored closely.

Question: My 2 year old son was diagnosed with LVNC at one month old. At birth it was thought that he had Ebstein's Anomaly, which he does not. Subsequent genetic testing revealed an MYH7 mutation on Exon 16. He did have systolic dysfunction and a dilated left ventricular, but medication stabilized that. My question is could his LVNC be a benign form since his function is now within normal ranges even though there is a genetic link? I asked his doctor but she was unsure if it might in fact be benign.

Answer: MYH7 mutations are a recently recognized cause of Ebstein's and LVNC. Here is the citation for the medical literature if your doctor wants to review some of the information on patients who have this:

> "Mutations in the Sarcomere Gene *MYH7* in Ebstein Anomaly"
> Alex V. Postma, PhD; Klaartje van Engelen, MD et al.
> *Circulation Cardiovascular Genetics* 2011;4:43-50.

In this study, which was comprised mostly of adult patients, the majority of patients with LVNC had evidence of cardiac dysfunction by echocardiogram.

Question: My 12 year old daughter has Noonan syndrome, though her genetic tests did not confirm this. Are there any new advances in Noonan research in the past five years?

Answer: There have been many new developments in Noonan syndrome research. Eight different genes are now known to cause Noonan syndrome. There is increasing recognition of specific signs and symptoms associated with particular genetic changes. Management guidelines have been standardized and published. The pathways that cause Noonan syndrome are now understood in much better detail and researchers are studying how these pathways cause particular features of Noonan syndrome. There is some promise that new pharmacologic therapy will be developed that targets these pathways.

Question: I would like to know what you recommend for supplements for the heart kids that have genetic disorders. I know about the "Mito" cocktail and my son takes coenzyme q10, levocarnitine, and creatine. I would like your input on the effect and benefits of supplements. My son was born with an atrial septal defect, ventricular septal defect, and dilated cardiomyopathy with LVNC. He has been diagnosed with Complex IV Deficiency (the cause for his DCM supposedly) and he has MELAS in his blood/urine but no symptoms.

How do we know what and how much to give? I know this is newer science and geneticists do not always agree to treatment approaches, so I worry that his kidneys or liver will be affected if we are too aggressive on the supplements. He also takes enalapril and carvedilol.

Answer: As you probably know, patients with mitochondrial disorders typically have difficulties in generating energy efficiently, since one of the major jobs of mitochondria within the cell is to produce energy. Mitochondria perform a number of other important functions for the body as well, such as helping with fat metabolism, detoxifying and preventing free radical damage, playing a role in iron metabolism and regulating ion levels. There is no cure for mitochondrial disorders. The mito cocktails that you refer

to are essentially providing additional cofactors or vitamins to help the mitochondrial do their job more efficiently. Typically these medicines do not cause harm or have side effects. In patients missing a specific cofactor, they can have a great impact. In patients with a more global impact, they can be beneficial but in some patients they do not have a therapeutic effect.

You are correct that many physicians use slightly different cocktails and there is not a standard approach or consensus. Sometimes the cocktail is modified depending on which aspect of mitochondrial function is most significantly impaired. There is very little evidence based medicine for or against the use of these medicines, but, as stated previously, they are considered quite safe and have potential benefit. For coenzyme q10 and carnitine, the target range is to get to two times the typical level (levels can be tested with blood tests).

Question: I would like to know the same thing about supplements, but for hypertrophic obstructive cardiomyopathy kids with really fat septums!

Answer: The mitochondrial cocktail does not vary depending on the type of mitochondrial cardiomyopathy. However, the cardiac medications will change.

Question: My daughter was born six months ago with a ventricular septal defect and hypertrophic cardiomyopathy (HCM). It took five months to get a diagnosis. We saw a geneticist who thinks she may have Noonan syndrome. The geneticist is only testing for Noonan and said there is no need to test for HCM because she already has it. Lastly, how does one determine if LVNC is present?

Answer: A diagnosis of Noonan syndrome can be made one of two ways. One is through genetic testing which shows a mutation. Not everyone with Noonan syndrome will have positive genetic testing (about 70% will). The second way is by having characteristic features and being diagnosed by a physician, frequently a geneticist. Testing for HCM is also very high yield but not perfect. Testing should be a consideration if it is unclear whether the individual has Noonan or isolated HCM and the Noonan testing is normal. One reason for considering this additional genetic testing, even though it is clear that the patient has HCM, is due to the implications for other family members who are at a 50% risk. A diagnosis of LVNC is made by echocardiogram or MRI.

Question: My daughter has dilated cardiomyopathy (DCM) and she was diagnosed at 8 months. At first, they thought a virus caused it, but then we discovered my maternal grandmother had DCM. She was diagnosed in her 30s and died of ventricular tachycardia in her 60s.

We were told that genetic testing would only have a 30% chance of finding the gene.

I plan on having an echocardiogram, but am very concerned about what this could mean for my other child and the third child I would still like to have. Is there any better way for me to find answers?

Answer: DCM is common enough that your maternal grandmother's history could be related or unrelated to your daughter's diagnosis since she is several generations removed. I agree that you should have an echocardiogram.

Unfortunately, DCM can have many different genetic causes and not all of them are known at this time. It is correct that about 30% of children with DCM will have a positive genetic test identifying the genetic basis of their DCM. A positive test is very helpful for at-risk family members so that they can know whether they inherited a genetic predisposition. In the absence of a positive genetic test, all first-degree relatives (siblings and parents) should have routine cardiac surveillance by imaging. These are the current guidelines for management of DCM in families. For genetic testing, we always recommend testing an affected individual. So, if your echocardiogram were normal, we would not recommend genetic testing for you. The most appropriate person in the family to test at that point would be your daughter.

Question: My youngest daughter was diagnosed at 2 months old with left ventricular non-compaction/hypertrophic cardiomyopathy (LVNC/HCM). When my older daughters had their echocardiograms they were noted to have non-compaction in the left ventricle also, but not officially diagnosed with LVNC. The recommendation at that time was yearly echocardiograms for both and maybe at some point every two years. Both of their heart functions were normal.

What kind of questions should I be asking the cardiologist when we take the older girls in for their echocardiograms? If they have normal heart function why would they need to be monitored? Should I expect them to be followed throughout their entire life or just until they hit puberty? What are the chances that it can turn into an official diagnosis of LVNC?

At my youngest daughter's genetics appointment the doctor looked up one of my older daughter's echocardiograms and asked me what medication she was on. She was not on any medications for LVNC so that puzzled me.

Answer: LVNC can be a difficult diagnosis to make and may be isolated or may be accompanied by hypertrophy/HCM as in your youngest daughter or dilation/DCM. Because both HCM and DCM can develop at any age, ongoing cardiac screening is recommended for first degree relatives of someone with LVNC/HCM. If the genetic cause is identified in an affected individual (your youngest daughter) then other family members could also be tested for that genetic cause and this would allow more precise recommendations to be made about ongoing cardiac surveillance. You should

ask your cardiologist about the function of the heart and whether there is any evidence of hypertrophy or dilation. You can also ask for updates on genetic testing.

Question: What is new in the research world of hypertrophic obstructive cardiomyopathy (HOCM)? What are now considered the best predictors or risk factors for sudden cardiac arrest in kids? What happens when they hit puberty?

Answer: There is new research into causes and risk factors for the progression of hypertrophic cardiomyopathy. There is also research being done to try to improve the sensitivity of detection and better understand when to start medical management. Risk factors include the thickness of the heart and whether there is obstruction, history of heart rhythm problems of passing out, the degree of scarring (fibrosis) of the heart and a family history of sudden death. With regard to the question of puberty, it is not completely clear why there is frequently progression during puberty, although it is often attributed to the hormonal changes that occur.

Cardiomyopathy Evaluation and Diagnostic Screening

Irene Lytrivi, M.D. – August 2013

Dr. Irene Lytrivi is an Assistant Professor of Pediatric Cardiology and Medical Director of the Pediatric Heart Failure and Heart Transplant Service at Mount Sinai Hospital. After completing her residency in General Pediatrics at Monmouth Medical Center in New Jersey and her fellowship at Mount Sinai Medical Center where she trained in non-invasive imaging, Dr. Lytrivi returned to her native Greece where she worked in the largest pediatric cardiologist center in Athens for two years. She returned to the U.S. for advanced training in heart failure/heart transplant at Boston Children's Hospital before joining the faculty at Mount Sinai.

Question: When my child goes for their annual evaluation, what are some questions I should be asking about his or her EKG (electrocardiogram) and echocardiogram test results?

Answer: On clinically stable children, the frequency of the evaluation varies between 6 months to a year. The issues to discuss with the cardiology team are mainly related to the echo results: is the function stable, is the ventricle more dilated and whether any arrhythmia is detected on the EKG. Other tests that the team may order (depending on the exam, results, and age of the child) are exercise stress tests, Holter monitors and also MRI. These can all help quantify the risk of each child for adverse events and may push the team to recommend listing for transplant earlier.

Question: I have a daughter diagnosed with dilated cardiomyopathy (DCM) with an ejection fraction (EF) of 31%. Our daughter's situation is unique in the fact that she has another disease called Sanfilippo syndrome type B. Life expectancy for Sanfilippo B is early teens. Our eldest daughter passed away at 13 years of age with Sanfilippo B syndrome. We also have a son with the syndrome at age 4. Our daughter, who I initially mentioned with DCM and Sanfilippo, is 5 years of age. I was wondering if there is any correlation between the syndrome and DCM. Our geneticist assures me there is no link. Does anyone else have a child with an underlying shortened life expectant disease independent of DCM.

Answer: To my knowledge DCM is not linked to Sanfilippo syndrome. Children with the syndrome may develop leaking heart valves and heart failure symptoms if the leakage is severe but not DCM.

Question: In response to the 5 year old child with Sanfilippo and DCM with a EF of 31%, how long before we should expect our child to develop symptoms? I only ask this in terms of planning. I recently signed a DNR order, which the palliative team suggested. I would like to fly to Disney World with her but unsure if it would be responsible to fly with her.

Answer: I wish I had a good answer for you but there is a lot of uncertainty regarding the development of symptoms or the risk of a sudden arrhythmia. You may want to ask her cardiologist to communicate with a team in Florida in case she gets sick when you are there.

Question: Is there any connection between high cerebral spinal fluid pressure and cardiomyopathy? My 14-year-old son has a pressure of 50 and hypertrophic cardiomyopathy (HCM). His myectomy was performed 2 years ago.

Answer: I am sorry for the delayed response but I was not aware of any association and I tried to research this. I found a couple of reports on rare associations of high CSF (cerebral spinal fluid) pressure and HCM, mainly syndromes such as Costello and Dandy-Walker malformation with HCM. Has your son being diagnosed with a syndrome? Did they have to treat the high CSF pressure?

Question: My 6 year old son has dilated cardiomyopathy (DCM) due to viral myocarditis as an infant. He has a patch of scar tissue in his left ventricle (LV) that is thinning and dyskinetic. His cardiologist refers to it as a "potential aneurysm." It has not changed since it was discovered a year and a half ago. His cardiologist would like to wait until he is older before doing an MRI, which would give us more info. My question is have you heard of any cases of children having a potential aneurysm like this, and do you know how rapidly they developed it and if surgery was an option? Our doctor said most of the information about this problem is in senior cardiac patients, so it's unknown how this will progress for a child. He also said given its location in the LV, removal through surgery might not be an option.

Answer: I would agree with your cardiologist; there is little information about the evolution of these abnormal wall segments into a true aneurysm. He will probably be cooperative for an MRI in a couple of years or so and an MRI would definitely characterize the area better. How is the rest of his heart functioning? Is he taking any medicines? I assume his EKG shows no abnormal rhythms.

Question: My son is 14, 6'1" and weighs 250. He has hypertrophic cardiomyopathy (HCM), metabolic syndrome and Acanthrosis Nigricans. He has had 2 lumbar punctures for high CSF (pressures 50 and 29). He is on Diamox and topirimate, atenolol, metformin and lovaza. He continues to have bad headaches, nosebleeds and chest pain. He is in a wheelchair as he cannot walk more than 10 feet without symptoms such as shortness of breath, headaches, chest pain, blurry vision, tinnitus, muscle weakness in legs and hyperventilating.

Answer: I did find some reports linking acanthosis nigricans with increased intracranial pressure so probably this is part of the reason your son has these symptoms. I really believe the HCM is unrelated.

Question: How much variation is typically seen in an echocardiogram before it is considered a significant change? My oldest daughter is 16; she was diagnosed with dilated cardiomyopathy (DCM) at age 12 and an automatic implantable cardioverter defibrillator was placed within 4 months of diagnosis. Initially her EF was 20%. Now it is up to 36%. Last year we were told it was "about the same" at every appointment that we went to until last August when they recommended a transplant because of her "steady decline" in ejection fraction (EF). At that time, it had gone from 36% to 34% to 32% but they never told us it was declining until they recommended a transplant. Since that time it has come back up and we are holding off on a heart transplant. When I asked our doctors what number they were looking for on an EF, they were vague and would not give us a straight answer. I realize it is a complicated issue and not only dependent on EF, but are there general standard values that point to transplant?

Answer: The truth is there is no absolute number to go by. Other things to take into consideration are activity level, irregular rhythms that you may be picked up by a Holter and symptoms of heart failure such as shortness of breath. Some kids self-limit their activities, so you have to be very persistent in asking questions about their activity level. An exercise test may also help with that. EF in the mid-30's is quite low so if they see other indicators as well, this may be the reason they are recommending a transplant.

Question: Now that we know my kids have hypertrophic cardiomyopathy (HCM), is there anyway to stop it from progressing to where their father's condition was in years to come?

Answer: At this point there is no clear evidence that any medication will prevent progression of the hypertrophy. However, in some recent publications, they have tried different medications (statins and diltiazem, a calcium channel blocker) in animal models and have reported slowing of the progression. This is obviously very preliminary and needs systematic study before it is applied in humans, but it offers hope that in the near future we will be able to alter the course of this disease. For the time being, medications are only used in patients who have symptoms, and more invasive treatments such as an automatic implantable cardioverter defibrillator are used when the patient is considered to be at high risk for sudden events.

Question: Are there any standardized guidelines on the frequency of screening a sibling of a diagnosed child?

Answer: In HCM, the recommendation is part of the published guidelines and it calls for EKG and echo evaluations every 12-18 months during childhood and adolescence and every 5 years in adulthood. In dilated cardiomyopathy, every 3-5 years is advisable.

For the less common cardiomyopathies such as restrictive and non-compaction, the recommendation is less clear but it is probably prudent to follow the same guideline as for dilated cardiomyopathy.

Question: Are there any new developments in how cardiomyopathy is evaluated and diagnosed in the future? Measurement and interpretation variability is still an issue that needs to be addressed.

Answer: As for new developments, genetic diagnosis is advancing greatly and gene panels for cardiomyopathy are expanding fast, increasing the chance of identifying a mutation in the patient and then screen the family members. MRI and newer echo techniques (strain, torsion, etc.) are also used more frequently to identify scar tissue in the walls of the heart and potentially risk stratify patients.

The other aim for the future would be to become more precise when we assess the risk of death in these kids and thus make the decision to urgently list them for transplant. In adult literature, there are different risk calculation models that use different variables (exercise endurance, kidney function, and many more) to assess what would be the survival advantage if a heart failure patient is offered a heart transplant. So far, and mostly due to the small numbers and vast heterogeneity of the diagnoses that lead to end-stage heart failure in children, we do not have these tools but hope to develop it in the near future.

Question: Can you explain how to look at ejection fraction and how to compare it to what is the norm for a child's age or weight?

Answer: Ejection fraction is a measurement of the pumping ability of the left ventricle. It measures the volume of blood that is pumped out with every beat. For example, when the heart is relaxed (in diastole) the left ventricle contains 62 cc of blood. If when it squeezes (systole), it can pump out 40 cc (so 22 remain in the ventricle), the ejection fraction is calculated as the percent change: 62-22/62 so 64%. 60-65% is the average ejection fraction that most kids have. The lower limit of normal is 50-55%. This is true for all ages, including adults.

Question: What would you consider a normal ejection fraction for an eight-year-old boy?

Answer: The lower limit of normal would be 53-55%; on average the ejection fraction should be in the low 60's. This is true across age groups pretty much.

Question: When is a MRI of a heart used for diagnosis? Are there different types of echocardiograms and when are they used for diagnosis?

Answer: At this point MRI is not routinely necessary for most forms of cardiomyopathy provided that the echocardiogram images are satisfactory. One exception is hypertrophic cardiomyopathy where the MRI can look at fibrosis of the walls, (fibrosis has started to emerge as a risk factor for adverse events). It can also be helpful in the diagnosis of more rare forms of cardiomyopathy such as arrhythmogenic right ventricular dysplasia (ARVD) and may help with more accurate estimation of function in non-compaction cardiomyopathy. In adults it is also used for diagnosis of secondary restrictive cardiomyopathy (RCM), which is not as helpful in children since in the overwhelming majority of RCM in kids is idiopathic.

As for the echocardiograms, we have started doing more stress echos. This is an echo immediately after an exercise test to look at dysfunction that is not present at rest but appears when the heart is challenged with exercise. This helps us in estimating individual risk with intense activity.

Question: My grandson has an ejection fraction of 48%. He also has a bicuspid aortic valve. His left ventricle is slightly enlarged. He takes enalapril, spironolactone, and coreg twice daily. His mother and aunt (my daughters) had cardiac transplants in 2001 and 2006 respectively. Both had dilated cardiomyopathy. His mother was twelve years old at transplant. His aunt was 20 years old at transplant. My grandson's pediatric cardiologist is now referring us to a transplant center. I am unsure if he is just being cautious or if this means that my grandson is definitely going to need a transplant. He is asymptomatic right now. I am wondering what new information will be gained by him going through a cardiac workup at a transplant center (catheterization, etc.) and how that will effect his future treatment?

Answer: I do not think it is a bad idea to speak to the transplant team early on, even if listing is not imminent. Based on the fact that he is asymptomatic and his function is only mildly depressed, I suspect they will decide to follow him closely after having collected all the information they need from their work-up. In cases of cardiomyopathy that affect multiple family members, the risk of another family member having it is quite high, but the way it will manifest, the timing of severe cardiac dysfunction and the need for transplant will vary, even within the same family.

Question: How do they determine if a child should undergo a 24-hour or 48-hour Holter test? Under what circumstances should they do a stress test?

Answer: It is really individualized. When symptoms occur infrequently, leaving a Holter on for 48 hours increases the chance to catch irregular heart beats. This is the reason we sometimes put event monitors that can stay on for a month.

As for the stress test, it is used differently in different cardiomyopathies. For example,

in hypertrophic cardiomyopathy the response of the blood pressure to maximum exercise and whether or not the patient develops arrhythmias can put the patient at a different risk category. In severe dilated cardiomyopathy, on someone with few symptoms at rest, the exercise endurance can help with the decision to list for transplant or not.

Question: I have a daughter age 9, diagnosed with dilated cardiomyopathy (DCM) at 11 days old. She has had some scary times due to her DCM. She had a mitral valve repair done when she was 3 due to further damage done to her through infective endocarditis. Her heart was too damaged to hold a replacement valve. She also had double pneumonia and spent months in the hospital. We were advised three years ago that our daughter would need at transplant further down the line perhaps in her teens as there is nothing more that can be done with her heart.

On her last cardiology appointment she showed no signs of heart failure although she still has a dilated left ventricle. We have been told that puberty will be a testing time for her heart even though she is not showing signs of heart failure at this time. Could you please explain why that would be. I myself was recently diagnosed at 43 with non-ischemic DCM and my son, age 11, was also noted to have an enlarged left ventricle in October. We are waiting to see a geneticist in November.

Answer: Based on the fact that both you and your children have DCM, there is a higher probability that genetic testing will be positive so I hope you will get some answers. The truth is, as you have experienced first-hand, that even in the same family the disease can present at any age. Sometimes the onset is gradual; sometimes it is not. Puberty is a time of rapid growth and many hormonal changes. We do see patients who present very ill in infancy and then start deteriorating again in their early adolescence with different types of cardiomyopathy. That is not an absolute, though.

Cardiac Electrophysiology

Robert H. Pass, M.D. – August 2013

Dr. Pass is the Pediatric Electrophysiology Director and Director of Pediatric Interventional Cardiology at The Children's Hospital at Montefiore. Dr. Pass holds the distinction of being New York tri-state area's most experienced pediatric electrophysiologist, having performed over 1,000 electrophysiology studies and over 500 arrhythmia ablation procedures. Previously, Dr. Pass was the Director of Pediatric Electrophysiology at the Children's Hospital of New York Presbyterian. He received his medical training at New York Hospital- Cornell Medical Center and Children's Hospital of Boston.

Question: What types of questions should parents ask when their child is being evaluated for an implantable cardioverter defibrillator (ICD)? How does the criteria for a child differ from an adult getting an ICD? Are there different considerations for infants versus older children?

Answer: In regards to your questions regarding ICDs, the important questions usually revolve around what the indication for the device is. For certain forms of cardiomyopathy, such as hypertrophic cardiomyopathy, the indications are fairly clear and have been elucidated in multiple papers with the majority written by Dr. Barry Maron. For other forms of cardiomyopathy, the indications are less well established and the decision is more commonly based upon specifics of the particular patient and their history.

For certain, the biggest issue in pediatrics relates to the size limitations to place ICDs. We have placed ICDs in rare situations in very small infants. However, such an implantation is a tour de force that is not without risk. Generally, for smaller patients, epicardial systems (on the surface of the heart) are more common. These are generally reliable but often break and have to be revised. As patients get older, a more typical “adult” transvenous system with the main ICD lead being in the vein and heart are more common. The size and weight of a patient loom large in the decision making process.

Question: I am wondering if you have any experience with children with resolved hypertrophic cardiomyopathy (HCM) and Wolff-Parkinson-White Syndrome (WPW) and what the general trend is as these children head into adolescence and early adulthood.

My 9 year old son was diagnosed with mild HCM and WPW at age 6 months and started beta-blocker therapy. By 18 months of age the HCM had resolved and his heart measurements were at the high end of normal where they remain today. He experienced breakthroughs of supraventricular tachycardia (SVT) or rapid heart

rhythm requiring medical intervention to about age 5. We have been able to manage his arrhythmia successfully and while he has had episodes where his heart rate has increased and we thought he was going into SVT, he has not required medical intervention. He had a treadmill stress test last year and his heart was found to convert to a normal rhythm at high levels of exertion which as I understand it puts him at low risk for a cardiac event. While I know that no one can predict the future, my concern and question is how likely is it that the HCM will return.

My son was quite resistant to medication as an infant/toddler and we struggled to keep him SVT-free and to convert him when he did go into SVT. Can repeated runs of SVT make a heart hypertrophic or dilated/etc.? We would like to have him ablated at some point so that he does not have to endure an adolescence of SVT and ER visits.

To complicate matters my father (son's maternal grandfather) is a sudden cardiac arrest survivor and has an implantable cardioverter defibrillator. They assume it was an arrhythmia as no other cause was found for his cardiac event. Heart attack was ruled out as well as all arrhythmic conditions.

Answer: In reference to your question, WPW does not cause HCM though the two have been described to be associated with one another. Most patients with WPW do not have a cardiomyopathy but a small percentage of HCM patients do have WPW. Multiple runs of SVT do not cause the heart to become hypertrophic. If one were in SVT for a very prolonged period of time, the heart might dilate and function may be affected but with the resumption of normal rhythm. Both the dilation and function might resolve in short order.

Question: I have a 5 year old diagnosed with Wolff-Parkinson-White Syndrome (WPW) at 18 days old when he was in supraventricular tachycardia (SVT). He had a few more episodes until he was about 5 weeks old and has not since. He is now off medication but still presents with WPW.

I have a 2-year-old boy that doesn't present with anything. I also have a baby girl that was diagnosed with WPW at birth and left ventricular non-compaction (LVNC) at 17 days old. She is only 6 weeks old now and so far no SVT and therefore is not medicated. If WPW cannot cause cardiomyopathy, how does it complicate the LVNC? Is LVNC cardiomyopathy? I am confused and feel like the information is conflicting. Her heart function is not concerning right now but we have an appointment with our cardiologist Friday.

I am in the beginning stages of trying to figure out exactly what is going on. We are seeing a geneticist in October. My husband and I are going to the cardiologist in September as well. No one in either of our families has had cardiomyopathy, but my mom’s family has a lot of cardiac related deaths over 30 years ago. But they all smoked as well.

My oldest has attention and impulsivity issues and had irritability and digestive issues as a baby. My baby girl has the same digestive issues and irritability. My middle child, who does not present with any congenital heart defect, never had any developmental or digestive issues and still does not. Are there any genetic syndromes or disorders that I should be thinking about going into the genetic testing? Can my middle child present with something later in life, and should I be taking him to see the cardiologist every year? My oldest goes every six months and has a Holter twice a year. The baby will go every month for now.

Answer: I am not, unfortunately, much of an expert on LVNC. However, in regards to your questions on how it may complicate things, if a patient has any form of cardiomyopathy it is likely that any episode of tachycardia will possibly be less well tolerated than in someone who does not have this condition. Thus, as a general rule, we are usually more aggressive in offering ablation to our WPW cardiomyopathy patients in order to take that risk "off the table." The natural history for WPW patients is that most do not have problems with SVT until they reach their teen years. At your child's age, I would say that any further episodes of SVT would likely be reasonable rationale to consider ablation. I have mentioned previously that the devil is in the details and more about your son would be needed to make a formal recommendation.

Question: I was wondering if there is an optimal age to perform an ablation. My nephew is 6 years old with supraventricular tachycardia (SVT) and has been told to wait a few years, if possible. I was wondering if you agree.

Answer: As a general rule, children who are 6 years of age can undergo ablation. The reason that we, as electrophysiologists, sometimes would prefer to wait is that when a child does not have WPW, it is not possible to know precisely where the SVT is arising from within the heart. If the SVT is coming from an area close to the normal conduction system ("the AV node") then, in a small child, there is a slight increased risk of having to "back off" for fear of injuring the normal conduction system in any attempt to fix the arrhythmia. Thus, in order to increase the odds of a single procedure being definitively successful, many electrophysiologists prefer pediatric patients to be somewhat older for ablation in order to improve the odds that the procedure will work and a second repeat attempt will not be needed. The general success rates for ablation of SVT in 6 year olds is still high, perhaps in the high 80% range. In older children, that percentage of success may rise a few points, and some parents and doctors would rather wait in order to "stack the odds" in favor of the procedure working on the first try.

Question: What are the side effects or potential complications of having an implantable cardioverter defibrillator (ICD) in a child? Is there any research on the quality of life for children that have ICDs?

Answer: ICDs in children, though potentially life-saving, are not without their "issues." The most important one is that the lead or wire placed in the vein in the heart tends to be fairly large relative to the vein size in children. Therefore, if an ICD lead is placed in a small child there is a chance over time that the vein may become occluded making it more challenging to replace the lead if it is too short for a growing patient or if it is simply broken (as can sometimes occur). Children also have an increased risk of what we refer to as "inappropriate shocks." This means that the device believes the child is in an arrhythmia requiring therapy but they are not. Inappropriate shocks are typically very painful and unpleasant. Despite these limitations, for the correct patient, ICDs are lifesaving therapy and can be critical for survival.

In reference to the second question regarding quality of life in patients with ICDs, recent studies suggest that pediatric patients with ICDs perceive a lower quality of life. There are many different possible reasons for this from fear of shocks to body image issues. Though not yet proven, it is increasingly believed that psychological therapy may be useful to help the child with an ICD who is not feeling well or who may be frightened by their device. I believe it is always important to remind children that it is not unusual to feel uncomfortable or sad about having an ICD as it helps to know that they are not alone. I also believe that forums such as this where many different people with the same problems and worries can speak with one another is a good way to help allay fears. After all, we are all in this together!

Question: How are decisions made for getting an implantable cardioverter defibrillator (ICD)? For example, if a child is diagnosed with hypertrophic cardiomyopathy (HCM), has no current arrhythmia issues but has a family history of the disease, what course of treatment is advisable?

Answer: As a general rule, the decision to place an ICD is based upon an estimate made by the doctors caring for a cardiomyopathy patient on their risk of sudden cardiac death from serious ventricular arrhythmia or abnormal heart rhythm. Depending upon the diagnosis, there are multiple factors that have been studied to predict a possible serious life-threatening arrhythmia.

Hypertrophic cardiomyopathy (HCM) is a good example of a disorder that has been very aggressively studied in multiple studies to evaluate just this issue. The following factors have been demonstrated to be associated with an increased risk for serious life-threatening arrhythmias in HCM patients:

1) Prior personal history of averted sudden cardiac death with serious life-threatening arrhythmia

2) Sustained ventricular tachycardia (multiple beats in a row of serious arrhythmia from the lower chamber of the heart)

3) Family History

4) Unexplained syncope (fainting)

5) Episodes of “nonsustained ventricular tachycardia” (short episodes of multiple beats in a row of arrhythmia from the lower heart chamber)

6) Left ventricular thickening (hypertrophy)

7) Abnormal blood pressure response to exercise

All of the above have been shown to increase risk of serious arrhythmias in adults with HCM, and studies in children, though fewer than in adults, have shown that some of these risk factors are also important prognostic signs in children as well. For sure risk factors 1, 2, 3 and 6 are particularly worrisome in HCM patients. Recently, I participated in a multicenter study with Dr. Barry Maron, and we showed that left ventricular hypertrophy of severe degree was highly associated with serious life threatening arrhythmias.

Thus, this is just an example of one cardiomyopathy where risk factors have been identified for possible need for an ICD. There are similar lists for other types, and in some, we are not yet as knowledgeable. Electrophysiologists and cardiomyopathy experts need to work together to try to predict risk in order to make good decisions regarding the placement of an ICD in a small patient. The decision is often a difficult one, and there are many factors that enter into this challenging decision.

Question: My daughter was diagnosed with hypertrophic and restrictive cardiomyopathy after going into sudden cardiac arrest while attending a tennis camp at the age of 9. Fortunately for us they had a defibrillator and were able to revive her. She had an implantable cardioverter defibrillator (ICD) put in which has appropriately fired during 4 separate events. The last time it needed to fire 5 times to get her heart out of ventricular fibrillation. At that time, she was put on 200 mg of Amiodarone and is monitored for liver and thyroid problems. She was placed on the heart transplant list 2.5 years ago as a status 2 and has been on Amiodarone for almost 2 years with no arrhythmias. She does have one of the rare genetic mutations for HCM.

You mentioned for those who have not had any arrhythmias during a 1-2 year period, a trial off Amiodarone may be considered. Does the risk of side effects increase with time on this medication? How long can someone stay on it? Is there a timeframe beyond which you should be taken off it? If side effects do happen, what other alternatives are available?

Answer: I am sorry to hear of your daughter's problems but happy to hear that she has been doing much better in recent years. In reference to your questions about Amiodarone, the risks or side effects related to its use are generally greater the longer one is taking the medicine. Some can occur fairly early such as thyroid issues. However, others, such as the effects on lung function, are usually seen after many years (>3-5 years) of therapy.

The good news in regards to Amiodarone is that most of the effects are reversible meaning that upon stopping the medicine the effects will abate. The one which is most likely to be "permanent" is the effect seen on lung function which is the one most commonly seen in long-term therapy with the agent. If your daughter is on a transplant list, it seems unlikely she will be on this medicine long enough to encounter these side effects (e.g. lung fibrosis). Annual pulmonary function studies are typically performed to monitor for these side effects.

In general, Amiodarone is a wonderful life saving therapy that has helped prolong the lives of many patients with various different sorts of heart problems. There are at least two features that make it particularly useful and well tolerated. First, it has virtually no negative effects on the function of a patient's heart. Almost all of the arrhythmias medicines we have can negatively affect heart function with the exception of Amiodarone. This is clearly a major benefit for cardiomyopathy patients who already may have issues with heart function. The other major benefit of this agent "pro-arrhythmic." Pro-arrhythmic agents are those that can actually cause arrhythmias. Though most arrhythmias medicines are used to prevent arrhythmias, some can actually cause arrhythmias and these effects are called "pro-arrhythmia." Despite its very powerful positive anti-arrhythmia effects, Amiodarone rarely causes arrhythmias and if carefully monitored, it is a very safe agent to use for many different sorts of arrhythmias in many different kinds of patients.

I am glad to hear that your child is benefitting from the use of this outstanding medicine. Though it has a number of potential associated side effects, in general, when carefully monitored, this medicine can be life saving.

Question: My 10 year old daughter was diagnosed with hypertrophic cardiomyopathy (HCM) in 2011 following a sudden cardiac arrest at school. What we have learned in the past two years is that her hypertrophy is moderate yet her arrhythmias are quite serious and incongruous with what one would expect to see with her HCM. She has a Medtronic pacer/implantable cardioverter defibrillator (ICD), which was placed September 7, 2011. Atenolol kept her steady until April of 2013, when her defibrillator fired 11 times on four different events. She paces about 93% of the time now and is on Amioderone 200mg daily, and 50 mg atenolol twice a day. Dr. Towbin at Cincinnati Children's Hospital is part of our team and with him we have initiated genetic testing. Our electrophysiologist (EP), Dr. Chris

Johnsrude, is in Louisville. What are your thoughts, as an EP, as to what our options are moving forward? Do electrical systems ever correct?

Answer: I am very sorry to hear of the arrhythmias that your daughter has been experiencing. We have seen a number of HCM patients who have had similar histories and it is always very challenging to manage patients like your daughter as they do appear to be particularly arrhythmogenic or susceptible to arrhythmias. Dr. Johnsrude, a very competent and excellent electrophysiologist, seems to be managing things most appropriately. Unfortunately, Amiodarone can sometimes be the only treatment for certain HCM patients. Though very effective, it can carry a number of side effects. However, most can be managed in the short to mid term.

Unfortunately, every case is different from the others. I would say that for now, continuing in the manner prescribed makes good sense. If your daughter goes a long while (1-2 years) without arrhythmias, consideration of a trial off the Amiodarone might make sense but as in all medical things, close consultation with Dr. Johnsrude will be of obvious importance. Having the ICD in place makes any such trial significantly safer than in patients who do not have an ICD. Please rest assured that you are in good hands with Dr. Johnsrude.

Heart Transplant: Before, During and After

Yuk M. Law, M.D. – November 2013

Dr. Yuk Law is the Director of Cardiac Transplant and Heart Failure Services at Seattle Children's Hospital, as well as, a Professor in the Department of Pediatrics at the University of Washington School of Medicine. Dr. Law's academic interest and clinical expertise is in cardiopulmonary failure and heart transplant and is a founding member of the Pediatric Heart Failure Group, a national professional interest group. He received Seattle Magazine's *Top Doctor award in 2011 and 2012.*

Question: What are the criteria used to determine if a child is eligible to receive a transplant? Under what circumstances are children not able to be listed?

Answer: In the evaluation for pediatric heart transplantation – likely to be no different in concept than other solid organs – it comes down to the general principle of how much the transplant state will benefit the quality of life of the recipient. What this translates to given the dire state of the cardiac condition is, will the replaced heart allow that individual to live very well. Because these can be relative and sometimes subjective assessments; it really does have to be taken on a case-by-case basis and the evaluation reviewed by a multidisciplinary group. It typically takes 2-3 full days to conduct a proper heart transplant evaluation. Some "more" straight forward examples of why a patient would not be a good candidate would be if the individual has a malignancy that is not eradicated by the oncologist and the transplant immunosuppression might cause the malignancy to spread. Or, there is significant multi-organ failure to the point that these organ systems will not recover function even after transplant. Hence the quality of life, if not the success of the transplant, will be significantly affected.

Question: My child has Sanfilippo Syndrome and was diagnosed with dilated cardiomyopathy (DCM) with an ejection fraction of 31% last year. Her life expectancy with Sanfilippo is preteen. In your opinion, would a child with a shortened life expectancy be eligible for transplant?

Answer: Since a transplant patient without any other medical problems other than those that can occur from transplant itself has a shortened life span, a shorter life span from a non-transplant condition should not be viewed as a contraindication. One has to be fair to the child and the family and determine if the expected survival from Sanfilippo will be further affected by the transplant state. Also, will the quality of life be significantly better after transplant and then compare survival duration with transplant versus transplant/Sanfilippo. The median survival (50% alive) at large (entire group of transplant recipients) is about 15 years. Like everything in medicine, the final decision is a bit science, a bit mathematics (statistics) and a lot of judgment.

Question: Do you have any thoughts or ideas on whether or not Sanfilippo B combined with dilated cardiomyopathy would have any impact on my daughter's already existing shortened life expectancy. I was told that they are two separate systems. Sanfilippo is primarily affecting the central nervous system.

Answer: I would say that systemic diseases such as Sanfilippo and other metabolic diseases can affect the heart muscle once there is heart disease even if cardiomyopathy is not part of the syndrome to begin with because the cardiovascular system is closely connected with the rest of the body. No organ system truly functions in isolation.

Question: After a transplant, (1) what steps should be taken to lower the risk of rejection and infection and (2) what are potential warning signs of rejection that parents should be aware of?

Answer: On question 1 – We have immunosuppression and antibiotic prophylaxis strategies worked out over the years to minimize rejection. Rejection and infection are somewhat linked or tied together. If too immunosuppressed, then risk of infection increases, and if too under suppressed, infection risk decreases and rejection risk increases. To find that delicate balance, we deliver more immunosuppression early on when rejection risk itself is higher and lower it over time so that the cumulative risk of infection lessens as well over time. We also use antimicrobials to minimize certain infections (early on only). Obviously, there is not a drug that prevents all infections, so we only do this early after transplant. Lastly, we take great care in monitoring for rejection with diagnostic tests and minimize risk of infection by good wound care, avoidance of contagions (people/animals/foods within reason), continue to do seasonal vaccinations as well as complete vaccination pre-transplant.

On question 2 – Warning signs are general lack of well being, as well as early signs of heart failure which many families are already familiar with because heart failure is a common indication for transplant. We also ask families to monitor heart rate at home and keep a log. An increase in heart rate can be very helpful in the triage for something going on including rejection.

Question: What does a child look like who is having a bout of rejection? All we have been told is that often you don't see it until it shows up on a biopsy or can look like another infection such as flu. This seems vague. Anything else?

Answer: The majority of rejection events are asymptomatic (no signs and symptoms). That is what is referred to as you will not see it except on biopsy. Of course, there are different grades or severity of rejection. If it is very bad, a patient will be symptomatic. Typically with symptomatic rejection, the patient presents with signs and symptoms of heart failure like during pre-transplant. In those with symptomatic

rejection, there is typically echocardiogram and even laboratory markers of change. What is more controversial is if we can pick up rejection before this stage without a biopsy. I think we can some of the time with the use of monitoring heart rate changes, labs, EKG (electrocardiogram) and a more comprehensive echocardiographic study. So for example, at Seattle Children's, we do very few biopsies because we rely on echo, etc. But the echo we do is very detailed and performed by only a few sonographers so we can more reliably pick up small changes within an individual as well as to compare differences with the transplant population at large. If we see these changes then we would do a biopsy as opposed to doing scheduled surveillance biopsies regardless of the pre-test (pre-biopsy) probability of rejection.

Question: Our son has restrictive cardiomyopathy (RCM), and he just turned ten. We are waiting for him to be put on the transplant list once his lungs begin to have increased pressures. Is it your opinion that it is better to place a child on the transplant list while he/she is still strong and active, since the surgery and recovery are difficult? Or is it better to wait until the lungs are beginning to be compromised?

Answer: It is amazing how families recognize the same dilemma that we heart failure and transplant subspecialists also ponder over. In general, RCM does not have as good a prognosis as dilated or hypertrophic cardiomyopathy, but that is at the "group" not "individual" level so we always have to keep that in mind when we apply population or group data to our patients. Part of the reason for the difference in prognosis is perhaps there is no therapy that slows progression for RCM. At the same time, transplant is a one-way street. So the two main drivers in determining timing of transplant are active heart failure symptoms and pulmonary artery pressure (lung pressure). Since your son is already diagnosed, these should not sneak up on him and if both are not present, it is usually ok to wait. For example, if he was diagnosed at 1 year of age he would have used up 9 years of his transplant time. I hope these generalities in how I would approach RCM are helpful to you.

Question: My daughter is currently on the transplant list as a status 2. Over the past couple of years, I have learned that each transplant center appears to follow different protocols for medication regiments post transplant. For example, I have heard of some centers that may not use steroids post transplant. Why are there different protocols? Why has there not been a best practice protocol established across all centers? Also, what is your center's opinion on Allomap versus heart biopsy for measuring rejection? Finally, what can be done to keep the kidneys from failing from all medications and what can be done to keep coronary artery disease (CAD) from occurring post transplant?

Answer: I will point out the basic challenges and general trends:

1) Different protocols are used because there has not been any large multicenter randomized clinical trials to compare protocols. So what we end up using is from

experience and adult trials extracted to children. Amazingly, for the most part, what we do works, so this further hampers the demand for trials.

2) Typically, steroids are used less and weaned off earlier than in the past. Most centers will still use but not as high doses as before.

3) Common protocols are an induction phase with steroid and Thymoglobulin followed by maintenance drugs of calcineurin inhibitor (tacrolimus or cyclosporine) and mycophenolic mofetil.

4) My feel on Allomap is that it is not applicable enough to pediatric patients and utility (cost-benefit and how it is incorporated into modern transplant programs) is not strong enough to be routine. Not all will agree with this sentiment though.

5) Sparing kidneys: keep calcineurin levels at a minimum to maintain rejection free state. Minimize any renal toxic drug exposure.

6) CAD: diagnose early use a statin plus sirolimus. The timing of introducing each medication is a point of controversy currently in pediatric heart transplant.

Question: Have there been any quality of life studies on children that have received a heart transplant?

Answer: There are studies. There can be more and better conducted to involve a larger number of patients. In general, the findings are consistent with personal observations. Quality of life is quite good, particularly in terms of physical and functional capacity. The emotional adjustment, which is also a quality measure, can be better based on my anecdotal observation. This is particularly true in the older teenagers-young adults.

Question: What are the different wait list statuses for heart transplantation?

Answer: 1A (which is the highest priority), followed by 1B, followed by status 2. Status 1A is for patients who are on a large amount of critical care type of support. Status 2 is for patients waiting at home without inotropes. Status 1B is in between in terms of acuity and amount of support. There is also a 1A and 1B by exception and that is for very unusual situations where a patient deserves 1A or 1B (not 2) but does not fulfill the criteria automatically. For example, if a patient has significant arrhythmias and cannot go on inotropes.

Question: How are donor hearts matched?

Answer: Donors are matched to recipients by blood type, weight, and distance,

among other factors. But these are the big three.

Question: Are there any donor-related factors that could affect the success of the transplant surgery or health following surgery?

Answer: Infections or so-called donor transmitted diseases, how well the donor heart functions at time of procurement, age, presence of coronary artery disease, and distance from donor hospital to recipient hospital are some of the major issues to be considered in the risk-benefit ratio.

Question: Would this child be a suitable candidate for transplant and would/shoud they be placed on a list? If not, why? The patient: a 16 year old male who presents with dilated cardiomyopathy, Long QT Syndrome (LQTS), persistent patent ductus arteriosus (PDA) with failed closure, chronic idiopathic pericardial effusion with 2 pericardiocentisis, an ejection fracture of 48% and on enalapril.

Answer: This is a very pertinent question and a good example where one really has to know all the facts about this patient including actually seeing the patient. That is why we perform a thorough transplant evaluation even if the patient is one of our own patient. I can still try to answer based on what you provided below. I do not see any contraindications to transplant. The bigger issue is readiness. If this child is highly symptomatic, has no good option with medical or surgical treatment and the disease has been progressive, then there is an indication for transplant. DCM, LQTS, PDA (patent ductus arteriosus), pericardial effusion even when recurrent, per se are not reasons for a transplant.

Gastrointestinal Issues Pertaining to Cardiomyopathy

Philip G. Kazlow, M.D., December 2013

Dr. Philip Kazlow is a Clinical Professor of Pediatrics at the Division of Gastroenterology, Hepatology and Nutrition at Columbia University Medical Center and Director of Clinical Gastroenterology at Children's Hospital of New York. With 29 years of experience, he has published many articles on pediatric gastroenterology in peer-reviewed journals and is rated one of America's Top Doctors in the Metro Area by Castle Connolly and Best Doctor by New York Magazine*.*

Question: Are there any ways to minimize negative side effects of magnesium supplementation in pediatric heart transplant recipients or increase/maximize uptake without increasing supplementation? Also, do you find that it is common for these kids to have increased incidents of diarrheal illnesses and what can be done to shorten duration and increase resistance? Is probiotic supplementation safe for these kids?

Answer: Magnesium salts, by their very nature, are cathartic. For example, Milk of Magnesia is chiefly made up of magnesium salts. Some of the side effects may be slightly minimized if they are taken together with food. In transplant patients, any diarrheal illness should be taken seriously. Stool should be evaluated for possible pathogens, such as clostridia difficile toxin, bacteria and parasites. As a rule, probiotics are thought to be not harmful. However, there is some emergent literature that suggests that they may be problematic in patients who are heavily immunosuppressed.

Question: What are common gastrointestinal (GI) side effects to cardiomyopathy medications and what are ways to treat these side effects?

Answer: Common GI side effects include nausea, vomiting, diarrhea and loss of appetite. There is no perfect way to treat these symptoms. Each case must be dealt with on an individual basis. However, some general guidelines would include trying to space the administration of the medications as far apart as possible. When not contraindicated, taking them with meals may help. In addition the use of certain over the counter medications such as Mylanta, or Pepto Bismol may help relieve GI discomfort.

Question: What about supplements such as Co-Q10, levocarnitine and creatine for kids with cardiomyopathy?

Answer: Some of these supplements, especially carnitine, can cause diarrhea. Patients must follow the instructions of their cardiologist. There is ample medical evidence that these supplements may be beneficial to patients with cardiomyopathy. As mentioned previously, taking them with food and using pre-medication may help.

Question: My son was diagnosed with acid reflux a few years ago (he is now 9). We did Prilosec, 20 mg in the morning and Zantac, 150 mg in the evening. We have done this on and off throughout the years whenever his tummy issues flared up. In August of this year, he was diagnosed with hypertrophic cardiomyopathy (HCM) and had a myectomy in October. Before surgery, he was put on propranolol and verapamil. There were no side effects with the propranolol, but the verapamil made him feel like he had the flu for three weeks. In the process of taking him back to the doctor, we found out verapamil was not working anyway and scheduled surgery. He was kept on the verapamil but on a lesser dose. In the hospital, they switched him to Cardizem CD 120 mg because of the horrible side effects of the verapamil. He does not feel as bad as he did, but I have noticed that, ever since we have been home from the hospital, his stomach issues have come back. Sometimes they are so bad he cannot eat. He throws up after he eats; he bends over from the pain, etc. He says it feels like sharp knives in his stomach. I asked the pediatric cardiologist about it at our last visit, and they suggested seeing a pediatric gastroenterologist. Did the surgery itself somehow increase his stomach pain issues or is it the medication?

Answer: This child would surely benefit from a gastroenterology (GI) consultation. As a general rule severe stress, including metabolic stress such as what was described, can lead to increased acid output and gastrointestinal upset. The child needs to be carefully evaluated and may need an upper endoscopy. Until that happens, he would probably be able to take an acid reducer such as Zantac, which he had been on previously, if this does not interact adversely with his other medication.

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