Children's Cardiomyopathy Foundation™

Answers from the Experts: CCF Cyberguests

A Compilation of Q & A Pediatric Cardiomyopathy Listserv Sessions
January 2014 - December 2014

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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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Psychosocial Adjustment to Medical Illness

Anne Farrar-Anton, M.D. – January 2014

Dr. Anne Farrar-Anton is a licensed psychologist at Joseph M. Sanzari Children's Hospital and Hackensack University Medical Center in New Jersey, as well as the Psychological Supervisor at Tomorrows Children's Institute. She is a previous executive board member of the New Jersey Psychological Association and the Bergen County Psychological Association.

Question: I have been thinking a lot lately about our 2 year old daughter and the ramifications later in life of going through so much poking, prodding and numerous doctor visits, especially when it comes to self-confidence about herself and her illness, and medical compliance as an adolescent.

I was wondering whether or not there have been any studies on appropriate ways to involve children with chronic illness in their medical care, at different stages of their lives, and whether this kind of involvement and self ownership leads to better self image and higher compliance as a teenager. We feel it is something that can only help, but we struggle to figure out what are appropriate ways at her age to make her feel involved in her care and to give her the choice to refuse some things. Obviously nothing critical, but enough so that she feels like she's in control of herself and has some bodily autonomy.

Answer: I truly appreciate the concept of allowing a child/adolescent involvement and some control. Yes, there is plenty of research that talks about children/adolescents being involved in their care and having positive outcomes. My philosophy when working with parents is how to set up guidelines that are consistent yet appropriately flexible to allow children some control. All children like to make decisions, but they need parameters. For example, do not provide open-ended questions such as "What would you like for dinner?" What if they respond "Morton's?" Is that a realistic option? I always suggest providing them with two or three options and then let them choose. You can use the same question of "What would you like for dinner – Wendy's or pizza?" This way you are allowing them involvement but you are setting the guidelines. Transfer that approach to medical issues such as taking a pill or swallowing, and you can say "You can take the pill now or in 5 minutes, but there will be no television until it happens."

One of the most common problems I see with children who struggle with medical issues is that parents feel guilty that their child is sick and then let them get away with things that they would not allow with a "healthy" child. I am not saying all families do this, but many do. The problem with letting them get what they want when they want is that this becomes their norm and then it continues way past the active illness.

My suggestion is to always include a child in an age-appropriate description of their illness and to have open communication about what this means. Work together with them on how to best handle what needs to occur. Allow them to be a part of the team. Also, child-life specialists who are typically employed in pediatric centers can be called on to teach the child/adolescent about upcoming procedures and help demystify the experience via play and vocabulary that makes sense. We often keep the truth away from children because we are scared of how they will handle it. We find that when kids know that they are not getting the whole story they will fill in the gaps with what they feel is going on and this can often be worse than reality.

The long and short of it is that I would always recommend involving children and providing options but remain consistent in expectations and clear on rewards and consequences.

Question: How can you tell the difference between normal stresses versus an adjustment disorder to an illness?

Answer: When it comes to an adjustment disorder, there are criteria that must be met. The criteria are:

1) The development of emotional or behavioral symptoms is in response to an identifiable stressor occurring within three months of the onset of the stressor(s).

2) These symptoms or behaviors are clinically significant as evidenced by either of the following: 1) marked distress that is in excess of what would be expected from exposure to the stressor and 2) significant impairment in social or occupational/academic functioning.

3) The stress-related disturbance does not meet the criteria for another specific disorder.

4) The symptoms do not represent bereavement.

5) Once the stressor or its consequences have terminated, the symptoms do not persist for more than an additional six months.

What does all this clinical jargon mean? It basically means that in order to have an adjustment disorder, there needs to be an identifiable stressor and then there needs to be significant changes to emotions and/or behavior that impairs functioning at work, school or in social performance. Once the stressor/consequence has finished, that behavior returns to a normal state.

I would be less concerned about whether or not it is a disorder and focus on whether or not you feel that you need help for yourself or for your child. It is interesting that we all go to school for many years to do lots of things in life but not on how to parent. I find that we parent one of two ways in general. One is we love something that our parents did and we do the same thing or we hate something that our parents did and we do the exact opposite. This would potentially work if you are raising a little you and there are also different life circumstances that emerge. This is where I think that groups such as CCF for support and guidance are so important.

I hope this helps to clarify and focus on the social, emotional and behavioral impact and how to remedy the concerns that are emerging over the diagnostic criteria. The reason that diagnosis is important is really for communication among practitioners and it does help define some treatments. However, you always need to take into acount the individual concerns and situations.

Question: My son is not quite 4 yet, but I want to make sure we are taking appropriate steps now to help him as we go down this road. Could you talk about things from the sibling stand point. My son is a middle child. He was only 4 months old when diagnosed and has been seeing doctors and taking daily medicine for nearly his whole life. His almost 6-year-old sister is showing signs of understanding that he is different. She wants to take medicine like him, see doctors like him and is always caring for a sick toy (probably fairly common for kids her age). I don't know how to differentiate between normal and reactive in our case. I want to make sure his brother and sister are appropriately supported through all of this, as well as our affected son, but I am unsure of what to do.

Answer: I appreciate your questions about how to help siblings. I think that they are often the "forgotten" part of medical illness. I remember when I was first starting out in medical psychology and I had a six year old tell me that she wanted to have leukemia like her brother. It took me a second to catch my open mouth but when I sat back I realized what she was saying was she didn't want leukemia, but her brother was getting all the attention. She wanted to feel important; she wanted to be noticed; and she wanted the family to focus on her.

My suggestion on your daughter is to open the lines of communication with her and see what she would like from you. My guess is that she is looking for "special time" – time where it is just about her. She does not understand the real reasons why her brother gets so much attention. In her mind, she wants to be important like him. I find that scheduling "special time," which could be 15 minutes, will produce a positive change. In those 15 minutes, she can choose the activity. I think this is a small but important step in the process of helping siblings.

Question: My daughter, age 11, was diagnosed with mild restrictive cardiomyopathy (RCM) in September 2011. The only symptoms she is starting to exhibit are getting super pale and tired very suddenly and easily. Her care providers have tried to involve her in decision making as much as possible. Her father, my ex-husband, thinks she should not have any say in her care as in who her doctors are, where her cardiac catheterizations are performed) etc. She is a very mature young woman and is handling all of the restrictions as well as possible. In your opinion, at what age should a child be told about their disease and treatment paths and be given a say in things?

Answer: I do think that this can vary depending on the child. At our hospital, which is pretty consistent across medical centers, nine is considered the age at which children should be "assenting" to treatment. This is different from "consent" which involves a parent's decision, but this gives the child a voice and a chance to review their medical plan. You will often hear the words "patient-centered care" and in pediatrics we talk about "family-centered care." In this model, the patient and family are part of the treatment team and should all be involved in medical care decisions.

Children need to be told the truth about their medical condition in age-appropriate terms and to have a plan on how to include their voice in their treatment plan. While I agree that nine should be the age of assent, I work with children as early as 4 years old to help them to understand their body and ask questions about what is happening. For example, they can take their medication before dinner or after dinner but there is no choice about taking the medication. The choice is about "when."

I serve as a donor advocate for our bone marrow transplant team for siblings who have been asked to donate their bone marrow. In my opinion, it is important that children know that they have a voice. It is their body and while they do not necessarily have the executive functioning skills of planning and understanding consequences to make these decisions alone, we should include them in the conversations in an age appropriate way, hear their questions and concerns, and consider their input.

Question: I have a 30 month old. When she was 7 months old, she was diagnosed with dilated cardiomyopathy and received a heart transplant at 8.5 months old. She has done well in many areas post-transplant. She had a strong oral aversion, but has been eating orally exclusively for four months now. It was extremely slow progress, but then one day she started eating normally. She used to be terrified of doctors and would start crying the second she saw the clinic. Now she is fine and does not even cry for blood draws.

The two issues we have now are separation anxiety and sleep. We tried to send her to a preschool playgroup this fall, but she was asked to leave because she

would get so upset when I left that she would upset the other children. The teacher has been doing this for 12 years and had never seen a kid get so upset! There has been progress though. Since November she is fine with babysitters she knows well. Six months ago, she had to be in the same room as me at all times. We are trying to send her to preschool next fall.

The real issue is she wakes up 4-12 times a night and has done this her entire life. When she was diagnosed, she could not breathe well when placed in a vertical position. She wakes up screaming or crying and takes 30-60 minutes to get back to sleep. Sometimes she is barely awake and goes back to sleep quickly. We have read all the sleep books, but it seems like her sleep has not developed normally. My daughter spends at least 50% of her playtime playing doctor or "sad baby." I think this has really helped her with her fear of doctors and separation anxiety, but not sure it can help her with her sleep.

Answer: In terms of preschool, can she talk to you about her fears when you leave? Does she truly understand that it is "normal" for her to be at school without you and that you are always coming back? Sometimes kids do not logically understand this and may get emotional. I think it would be important for her to start preschool in the Fall. Her progress is "slow and steady," as six months ago she would not let you out of her sight. I think we need to start off with where she is at and then use the next eight months to build up to longer separations and then to "reward" her for her ability to be without you in very slow steps. If she allows you to be one room away, build up to two rooms away and increase from there. I would also talk about it with her. When children get stressed their "thinking" brain tends to stop working and their emotions take over. When we start to think about things in a rational way we typically can overcome our fears.

With regard to sleep, I am not sure that I have any additional information. I agree that the early years of waking up due to her breathing has probably patterned her sleep. However, that does not mean it will be that way forever. My initial thought is to talk with her about what she would like you to do when she wakes up. This way she is part of the planning. You would want to help soothe her but not necessarily talk to her. This may help her return to sleep faster. The first thing I would work on is getting her to routinely fall asleep with less disruption and then her body will get used to being asleep for longer periods of time. The more they are "awake" during the night the harder it is to fall asleep. I would talk to her about this because if you are going to stop talking to her during the night she understands why and will try to focus on non-verbal ways of soothing.

I believe that medical play is a great way for children to work through their own inner emotions and thoughts. Putting it on a doll helps them to master it.

Question: If a family has several family members with heart defects, surgeries or

have experienced life-threatening situations, how long does it take to adjust? Can children adjust or will they be so traumatized as to get posttraumatic stress disorder (PTSD)? I myself have gone through two open-heart surgeries with two sons who we almost lost a few times. There have been three pacemaker installments in three family members, and I am diagnosed with left ventricular non-compaction. We are getting our third son evaluated as he's starting to show symptoms as well. What are the long-term prospects for these patients, families and individuals for PTSD and other conditions with respect to trauma and stress related illness?

Answer: I don't believe there is a time frame of adjustment for families. This varies based on family, medical situations, social context, and individual differences. I think most people do adjust and some do have aspects of PTSD but that can also improve.

Words that I have learned along the way that I use in talking to families is being "flexibly structured." Trauma and stress as it relates to medical illness is something most experience but the question is how do you choose to live with it. If you are always looking over your shoulder and worried about what bad thing is coming next, then there is never a break. I am not saying that bad things will not happen; one needs to be cognizant of symptoms but we also need to try and live in the present. Many people focus on the stress in the past and not the here and now. They are worried about the future, which I understand, but as a result there is no focus on the now. This is where "flexibly structured" comes in. There needs to be day-to-day expectations of all family members. Everyone should have a schedule of typical daily responsibilities and should be held accountable to that. This gets a family out of living in "sick mode" and getting everyone to participate. Obviously there may be days in which someone is not able to accomplish daily expectations and that is fine when it is not the norm. If it is the norm, then we need to assess what they can actually do and then build up slowly from there.

Question: My daughter, who has developmental disabilities and is unable to speak, was diagnosed with hypertrophic cardiomyopathy (HCM) in November of this year. Being a health care provider, I know a little too much about this and seem to focus on the "what ifs." They want me to keep her heart rate down, but she is hyperactive and mentally delayed so she does not understand what the risks are. I sometimes imagine scenarios about what if she died, and how would my day go without her and I begin to cry. Do these feelings fade? How can I stay vigilant without getting anxious?

Answer: Being a health care provider can make it worse in some ways because of having too much information. In regards to imagining your life without your daughter, I don't know if those feelings fade but I think that they can become less of a focus. That is when we put more emphasis on living in the now. Staying vigilant without getting anxious is a talent and I wish I had an easy answer. Keep doing

what you are doing which is thinking about it and seeking answers but not letting it consume you. This is definitely easier said than done, but I think the doctors and family can help reinforce in a developmentally appropriate way any restrictions on her activities and help her to follow them to the best extent possible. I know that you mentioned she is developmentally disabled and is hyperactive but I am guessing she knows how to pull herself together when she wants something. I think you can help her to meet her physical restrictions by rewarding her with the things that she enjoys as opposed to her just always having access to them.

In terms of your thoughts, I refer you back to self-care. This is so important and is often the last thing that parents do even though it is necessary. If you are so focused and anxious about bad things happening, then she is going to pick up on your anxiety. She may not understand why and misunderstand what it is about. When we see others anxious we think there is something that we do not know and often put the story together in a fragmented and untrue way.

Question: Our son will be 6 years old in April. He is doing great right now (normal heart function, still enlarged heart but stable) but it's been a hell of a road to get here.

When he was 4 months old, we took him to the pediatrician for respiratory distress. She suspected pneumonia and sent us to ER for a chest x-ray. He ended up on life support by the next morning due to acute congestive heart failure because of undiagnosed dilated cardiomyopathy/left ventricular non-compaction.

We spent 4 months in the ICU with lots of ups and downs. He has been through a lot of trauma. It took them seven hours to get an IV in him the first morning and he ended up with a line in his neck after they stuck him about 30 times, got septic 2 times, had a gastrointestinal hemorrhage due to a reaction between Coumadin and antibiotics, and had a spinal tap. He has had three surgeries (broviac placement, G-tube placement and ear tubes). We also were at a point where we consulted with a transplant surgeon because he was deteriorating. We decided not to list him and thought we were basically going to have to let him die. He started a new medicine and began improving, but we thought we had signed a death sentence for him which obviously had an impact on us, and in some way on him since he saw us so grief-stricken.

We just had a really hard six months with moving in and out of our house due to construction, doing a very intensive three week inpatient feeding therapy program and switching schools because he was so overwhelmed by the number of kids. He had a very hard time with all these transitions and started showing a lot of anger (hitting kids and teachers), experiencing very fast and hard mood swings and showing intense stubbornness (even teacher/feeding team noted this).

Question 1: All of these behaviors are completely understandable given his medical history of having so many things done to him and not having control, but it is making life pretty unpleasant for us and impacting his time at school and his

social peer interactions. I am hoping it will get better as the dust settles, but I am afraid of this pattern getting set. Do you have thoughts or ideas about how to help him figure out more constructive ways to work his feelings out?

Question 2: My wife and I totally have post-traumatic stress disorder (PTSD) from all this. Just last night we spun out because he woke up super sweaty and wanted us to come be with him. My wife thought she felt his heart beating erratically and freaked out thinking that just as things were getting back to normal, his heart was now in trouble. It doesn't take much for us to go from fine to sheer terror. What are your thoughts about how to help parents work through PTSD when the reality is that cardiomyopathy is unpredictable and bad things are more likely to happen to us than for parents of healthy kids?

Answer: Thank you for your sharing your story. It sounds like you have all endured a very challenging medical crisis and are here to tell the story but not without the emotional wounds.

With regard to question 1, I always tell everyone that I work with that there is nothing wrong with anger and all the various feelings we experience, but it is not okay to express it inappropriately. There is a time and place where we can let it out and as long as it doesn't work against anyone or anything then it is good. I suggest physical activity that is medically approved as well as anger management strategies, such as hitting a bedroom pillow, blow up or boxing bag when stressed, ripping up "read" newspaper, bubble blowing as well as listening to calming sounds (ocean music). Having a time where one can let out our anger in an appropriate place is basically what we are looking for.

With regard to Question 2, I agree that the unpredictability of health issues is very complicated because you do not know when things might change. I can appreciate the panic and fear at something that might be very small but that it might be life-threatening. The best advice is self-care. Making sure that you are both taking time to care for yourselves and find time for things that you enjoy, whether it be reading a book, taking a walk, taking a bath or watching a good movies. In order for you to be present in the moment you need to take care of yourself first. This may sound strange because parents feel that they need to care for their child first. I want you to think about the airlines and what they tell you to do first. They tell you to put your oxygen mask on yourself first and then help those around you. This is true for parenting. If you are not taking care of yourself, it is hard to be fully there for your child. I suggest time alone and also time for you both as a couple. This will help strengthen your emotional energy.

Question: My husband and I have three children, age 7, 6 and our angel who passed away from dilated cardiomyopathy (DCM) 4 years ago at age 1. Currently we have no cardiomyopathy related issues. Our son had a LMNA

gene mutation come back on his genetic test. My husband and my younger daughter also had these same test results. My oldest daughter and myself do not have the genetic mutation. We do still have both girls monitored for cardiomyopathy with our son's cardiologist. It was never determined what the trigger for his DCM was. My first concern is constant worry over whether or not someone else will get sick with DCM. When you have already lost one child, you tend to be hypervigilant. Constant worry accentuates my anxiety disorder. Second concern is I worry about how the girls are handling their brother's death. Are we focusing too much or too little on him. There is no roadmap for how to handle these things. Third concern is since my youngest daughter has the same genetic result, I am worried about the appropriate age to tell her about it. Just because she has the genetic mutation does not mean she will develop DCM. Our son was born with it and his case was particularly severe.

Answer: Constant worry can and does lead to anxiety and the hard part is how do we find a way to not live in a constant state of worry and anxiety. This is the challenge. As I have mentioned previously, worry and anxiety keep us in a state of stress and prevent us from living in the moment. It is important to have self-care and work on keeping the worry in your mind but not allow it to take over. I would recommend self-care activities such as taking a walk, taking a bath and engaging in an activity that you find relaxing. When you are able to be in the moment, the stress around you becomes less powerful.

I agree that there is no roadmap on how to handle the death of a child/sibling. We all expect that the typical path is that parents will die when older and their children will outlive them. Since, this is not the case with your family, you and your family need to figure out what works best for you as a unit. I think communication is the key. It is important to tell your children that you want to hear their thoughts and feelings about their brother and that they can let you know if they want to talk about him less or more. As I am sure you know already, children experience grief in random periods. They can be upset one minute and playing the next. This is normal and does not indicate that they are not suffering. It means that they are trying to incorporate their feelings but also do developmentally appropriate things as well.

With regard to your daughter, I think talking in developmentally appropriate language is all you can do. I would focus more on what she has and what she will need to do to follow up with medical recommendations instead of focusing on your son. If she asks if this is what he had then you can mention exactly what you told me about his rareness/severity and that it is similar yet different.

I hope this information is useful for you and thank you again for sharing. With focus on today and living in the now, the power of the trauma gets diminished. I am not saying it is easy but it is a goal.

Familial Forms of Cardiomyopathy

J. Carter Ralphe, M.D. – February 2014

Dr. J. Carter Ralphe is chief of pediatric cardiology at the University of Wisconsin School of Medicine and Public Health and co-director of University of Wisconsin Health's Pediatric Heart Program. Dr. Ralphe's clinical interests include diagnosis and treatment of congenital heart defects, acquired and familial forms of cardiomyopathy and pediatric heart failure.

Question: Our daughter was recently diagnosed with hypertrophic cardiomyopathy (HCM). She is 8 years old and we discovered it when she developed pneumonia and a heart murmur was heard. Our daughter also has developmental delays. She exhibits patterns seen in cerebral palsy, autism, as well as Angelman's, but is none of these in its true form.

No one else in our family (8 children) or close or distant family members have HCM that we are aware of. It does not appear to be genetic. Do you know of any syndromes associated with HCM that we may zoom in on to try to understand her overall condition better?

Answer: While hypertrophic cardiomyopathy seems to occur most frequently in isolation, it does also occur as part of recognized genetic syndromes. I agree with your assessment that your family does not appear to carry a genetic mutation commonly associated with familial forms of HCM, but there is the possibility that your daughter carries one of the known mutations that has not occurred in your family before. This is called a spontaneous or de novo mutation.

I suspect that the other issues your daughter is dealing with are unrelated. However, genetic syndromes often present with variable features, each with varying degrees of severity that makes syndrome identification and diagnosis difficult. Noonan syndrome is one that comes to mind. In its fully manifested form it is easily identified, but full manifestation is not present, leaving physicians to connect the dots among subtle findings. It is associated with HCM in approximately 20% of children and can have a wide range of other manifestations that can be quite pronounced or quite subtle.

In situations like yours I always enlist the help of a geneticist. These physicians have training in identifying unusual features and tying them together in a global diagnosis. Based on their evaluation it might then be advised to pursue a specific genetic confirmation.

Questions: When is one considered to have hypertrophic cardiomyopathy (HCM)? Is one considered to have HCM when identified as a carrier of a familial gene with

what appears to be a healthy heart? Or is one considered to have the condition when symptoms actually arise?

Answer: I understand your confusion with this, and I am not so sure the medical community would provide a consistent answer to you. I personally consider making the diagnosis of HCM when a patient is identified as having an abnormally thickened heart muscle. It gets complicated when we realize that some of the known HCM mutations can present in people as a rhythm problem before any evidence of hypertrophy. This is where I think genetic testing and identification of a mutation in a family can really help. For those carrying the mutation with what appears to be a normal heart, I do not make the diagnosis of HCM but refer to them as carriers. As a carrier these individuals get regular screening evaluations, which can include EKG, holters, echocardograms, or exercise tests to look for any indication of progression from the carrier status to disease status.

Question: I am very confused by this gene that some of our family members carry. Is it the same as many genetic predispositions? In other words, is this a gene that guarantees the eventual diagnosis of HCM in one form or another or is it a gene that indicates that one could someday develop the symptoms of mild to severe HCM?

Answer: The answer depends a bit on the actual mutation and what the pattern is in your family. The presentation of HCM has a wide range. Some people may carry the mutation for life with minimal to no issues while others with the same mutation present with severe HCM early in life, while still others present with arrhythmias. Adding to the complexity is that the range of presentation can be found within the same extended family. We can only assume that HCM represents a combination of factors beginning with the primary genetic mutation in one of the genes known to cause HCM and combined with the genetic background of the individual and the additional influence of environment (high blood pressure or intensity of athletic participation). In short, there is probably no guarantee of eventual diagnosis and even less ability to predict severity. This gets back to the need for regular surveillance screening by a cardiologist.

Question: I understand that a genetic carrier has a 50/50 chance of passing this gene forward to future generations. My question is whether or not there is any data on the percentage of familial genetic carriers who might actually develop symptoms of mild to severe HCM? To date, only one of our many family genetic carriers has been diagnosed with HCM.

Answer: I think the previous answer covers this question. The fact that your family has only one disease-manifesting individual among many carriers might be hopeful but does not negate the recommendation for regular surveillance.

Question: Is there any research or documented concern that viruses, bacteria and/or exposure to environmental factors such as certain medications (post natal or pre-natal) could trigger a person with the gene to develop the symptoms of HCM? I ask this because our one diagnosed person out of several carriers ranging over several generations was ill prior to diagnosis. Or, is the situation kind of like the chicken and the egg? Who knows which actually came first?

Answer: We do not have a clear, fact-based answer for this yet. Anecdotally there seems to be a triggering stressor in some patients. Many of us believe that there is an environmental component and by this I mean any of the things you mentioned above that contributes to the severity of the disease. I am not aware of any strong data to back this up yet.

Question: My daughter is 12 and HCM positive gene ACTC1 at age 9. She has low blood pressure and low heart rate with activity. She has prolonged QT, postural orthostatic tachycardia syndrome (POTS) and has had an implantable cardioverter defibrillator (ICD) placed in Oct 2013. Her left ventricular ejection fraction (LVEF) is 50%, but her right ventricular ejection fraction (RVEF) is 39%. She has been on Florinef since August, her BNP levels are around 4500 on our local hospital scale and over 1200 at our cardiologist's hospital. She complains of sweating a lot and being restless. I am confused as to why this is not partial heart failure. Any advice or suggestions? She does not handle beta blockers; she only takes 10mg nightly.

Answer: This is a difficult situation to provide any concrete advice. The combination of things you describe here would make her management complicated and delicate. I would suggest that you return to your primary cardiologist to discuss your concerns. Depending on their background and area of expertise, you might also consult with a pediatric cardiologist who specializes in heart failure if you have not done so already.

As your daughter's primary advocate, you are empowered to ask questions until your concerns are either addressed or you understand and are comfortable with the explanations provided by the doctor.

Question: How often is the Gene Nkx2-5 associated with left ventricular non-compaction cardiomyopathy (LVNC)?

Answer: To my knowledge, we do not have a number to put on this association. Since Nkx2-5 is an early developmental regulatory gene that affects many aspects of cardiogenesis, it is not surprising that it has been associated with LVNC. Since LVNC itself is such a variable condition with probable multiple starting points that end up with the manifestation of non-compaction, it is not surprising that Nkx2-5 would be implicated. So, where does that leave us? It is associated, but it remains to be determined if mutations in Nkx2-5 are in and of themselves enough to result in a form of LVNC.

Question: Our 16 year old son was recently diagnosed with left ventricular non-compaction cardiomyopathy (LVNC) after experiencing tachycardia during a soccer game. He has never had any symptoms before or since. After he produced 125% of normal on a cardio pulmonary exercise test (CPET) with left ventricular dilation just barely outside normal limits, they assessed him as likely having "athlete's heart" and cleared him to resume competitive soccer with annual echo follow ups. Our 13-year-old daughter and myself have just had echos that indicate no LVNC; my husband has yet to have an echo. Can I be a carrier and never have a LVNC diagnosis and can our daughter?

Answer: Yes, you can. There are growing lists of genes that can contain mutations associated with the condition of left ventricular non-compaction. The occurrence of the disease and the severity of manifestation, even when a person is known to carry one of the mutations, can be highly variable. There are likely individuals who carry specific mutations and do not show any changes in their hearts. The fact that your son's severity is mild might provide some reassurance. Obtaining a genetic diagnosis however would give your family the opportunity to identify if a specific mutation is present, and if so, who else in the family might be carrying it. This knowledge could result in peace of mind or loss of peace of mind depending on the result and how the individual responds. My bias is to want to know if I am a carrier so that I would be able to choose regular screening or not, but this is my bias. Unfortunately, what we know about LVNC is just beginning to accumulate, and there are likely causes that are not yet included in the genetic tests.

Question: Our son had a two vessel umbilical cord, which I understand can be associated with heart defects. Do you know of any correlation between two vessel cords and left ventricular non-compaction (LVNC) specifically?

Answer: No, none that I am aware of. Two vessel cords are certainly associated with other forms of congenital heart disease, but not to my knowledge with LVNC or other forms of dilated cardiomyopathy or hypertrophic cardiomyopathy.

Question: Do you know of any recent research focused on teen athletes, left ventricular non-compaction (LVNC) and athlete's heart?

Answer: None that I am aware of focusing on this particular scenario. The way that I think about athlete's heart is that it is a benign condition of physiologic hypertrophy of the heart muscle in response to athletic training and the increased workload placed on the heart. That being said, it is critical to exclude other possibilities such as HCM or other diseased processes like unrecognized hypertension. I make the diagnosis of athlete's heart in very specific settings; the non-hypertensive athlete with a negative medical history, negative family history and marginally increased concentric left ventricle wall thicknesses. When I reviewed the current literature I was surprised to see how many people are writing papers on this topic, but we do not seem any closer to a better understanding of the phenomenon.

Question: I have two daughters affected by dilated cardiomyopathy (DCM). The oldest was diagnosed at 11 days of age in 2003, received a heart transplant at the age of six in 2010 and is now ten and in 5th grade. The youngest was diagnosed at three weeks of age in 2004, was medicated as a baby, but her heart normalized and she came off of medications by the age of one year. She is now nine and in third grade and continues to have an echocardiogram bi-annually to be sure that there is not a recurrence.

In 2010, we underwent genetic testing. It was found that both my daughters, and I (their mother) have a mutation on the MYH7 gene, which is attributed to cardiomyopathies. Both of my parents were tested as well and neither of them presented with the gene, so my understanding is that the mutation developed somehow within me. The cause is unknown; it could be because of some exposure or it could have happened during fetal development.

Since 2010, a cardiologist has followed me regularly and my function is in the low-normal range (ejection fracture of 55-60%), but I am considered high risk for developing cardiomyopathy at some point. I am now 39 years old, have never had heart problems and do not take any heart medications.

Is there any research or statistic that shows the likelihood of recurrence in familial cardiomyopathies, such as in my youngest daughter's case? I understand that the disease is highly variable and that it can present at different times for different family members. As a medical layperson I find it odd that both of my daughters were diagnosed as infants and that I might present at sometime in my adulthood. Do you have any thoughts or information about the ages that familial cardiomyopathies present?

Of course, we will all continue to be seen by our cardiologists as they have recommended. I am just wondering if you have any other information in this area or if there is any new research or statistics.

Answer: I presume by what you wrote that your daughter(s) have manifested with DCM associated with the MYH7 mutation they carry? And you have not been diagnosed? There unfortunately is no reliable way to predict who, among carriers, will develop signs, the timing of development or the severity. Some mutations seem to be more powerful than others in that more or all of the identified carriers are affected; other mutations are more variable in presentation. As someone conducting research on cardiomyopathy, this is one of the most difficult questions to answer. My own theory is that the context of the mutation is important in determining how and when the disease presents. By context, I mean a combination of the rest of the genes of any given individual and/or the environmental exposures accumulated during normal life. We know that every individual harbors countless silent mutations in genes that alone do not cause disease (often recognized as benign polymorphisms), but when

combined with other mutated genes can lead to disease. For HCM we are actively trying to identify other potential genetic modifiers. This might explain why you and your daughters are following such a dramatically different course even though you have the same mutation. Unfortunately, this is the answer I have that does not answer your question. Your situation underscores why we need to continue to solve this puzzle.

Question: My son's heart triggered concern while in utero, and he was diagnosed with left ventricular non-compaction cardiomyopathy (LVNC) after an echocardiogram following his birth. Knowing this, my husband and I both had echocardiograms and they diagnosed LVNC for me. My two brothers, mother, and father all have had echocardiograms and LVNC was only cited in one of my brothers and I. It is my understanding one of my parents may be a carrier but not exhibit the phenotypical traits/characteristics and that some chemical change likely triggered the phenotypic traits for my brother and I to exhibit this. Could you please provide any detail or explanation for what may have triggered LVNC or how exactly this works and if there is a general time frame such would be triggered by. I have a little girl, as does my brother, who does not exhibit LVNC and my other brother who is 22. I am wondering if there is a time they will be out of the woods. In 2010 during our diagnosis we had genetic testing done but at the time they found nothing. However I understand testing has likely advanced since then.

Answer: LVNC understanding is evolving at a very rapid rate, but a lot of questions remain. I still regard LVNC as largely a developmental issue with the left ventricle that occurs early during fetal development. This implies that if there is no evidence of LVNC on an individual's echocardiogram, it is not likely to develop in the future. Unfortunately this view is being challenged by cases in which LVNC appears years after a normal echo. The theory of failure of normal compaction of the left ventricle during heart development is failing to explain all of the cases. I do believe that a normal echocardiogram with no signs of LVNC is very reassuring. That being said, I would also continue to screen individuals in the family every five years.

We have added quite a few more candidate mutations to the screening list since 2010. It might be worth repeating screening of one affected family member to see if something comes up positive. This way you could rationally approach other members of your family and look for that specific mutation. I would certainly consult with a genetic counselor before embarking down this road.

Pediatric Cardiovascular Genetics and Pediatric Genetic Testing Related to Cardiomyopathy

Amy Roberts, M.D. – April 2014

Dr. Amy Roberts is the Director of the Cardiovascular Genetics Research Program at Boston Children's Hospital and Assistant Professor of Pediatrics at Harvard Medical School. Dr. Roberts has a certificate in pediatric medical genetics and specializes in cardiovascular genetics, clinical genetics and Noonan Syndrome.

Question: Is there any progress on the reimbursement of genetic testing for cardiomyopathy through insurance?

Answer: This differs by each insurance company, but it is worth inquiring about prior approval and soliciting a letter of medical necessity from your physician if requested.

Question: Has genetic testing for cardiomyopathy changed or improved over the years? Is it detecting more genetic mutations and more accurate in finding causes in children with cardiomyopathy?

Answer: Yes! It is not that the testing is more or less accurate, it is that our understanding of specific mutations has increased as more families have been tested and the number of genes tested has also increased. Thus, there is a higher likelihood now of finding a mutation than there was on previous smaller gene testing panels.

Question: How likely is it that there may be more than one genetic mutation in affected families? Would genetic testing be able to detect multiple mutations?

Answer: In general, cardiomyopathy gene testing is done as a "panel" meaning the most important genes are tested all at the same time. Therefore, if more than one mutation is present it will be detected. In one study of genetic testing in pediatric hypertrophic cardiomyopathy, 12 percent had more than one sarcomere gene mutation (Morita et al, *New England Journal of Medicine,* 2008).

Question: If a family had genetic testing done in the past and no mutation was found, when should they retest due to advancements in the field and new genes/panels available for testing?

Answer: The field is rapidly evolving so I would recommend checking in with your geneticist every two to three years for updates on newly available tests.

Question: We have had two children born with severe hypertrophic cardiomyopathy. The first child passed away at 11.5 weeks and the second child received a heart

transplant at 5.5 weeks. We have three other children who present normally with no known extended family history of childhood cardiomyopathy. No genes have been identified in the testing that was done pre-transplant, and we have not yet had any of the more extensive testing done.

My question is about having that "more extensive" genetic testing done. We need to weigh the risk/benefit/cost of additional testing. The only advantages that have been explained to us are that if a gene is identified, our three healthy children will be able to forgo their yearly echoes, and for family planning purposes. Are there other benefits to having this testing done? Do you foresee any possibilities of our healthy children being discriminated against in any way in regards to future health insurance if it were discovered that they were found to be carrying an identified gene?

Answer: Without knowing what testing has already been done, I can not comment on the additional testing being proposed. The utility of genetic testing would potentially be to both your affected children and at risk family members. If the exact genetic cause can be determined in your affected child, then you would know if the genetic change causes isolated cardiomyopathy or if there are other medical complications one should be on the lookout for. For example, boys with cardiomyopathy due to a mutation in the TAZ gene have Barth syndrome and in addition to cardiomyopathy can have other non-cardiac medical problems. Knowing the exact genetic diagnosis would be helpful in the future if your affected child decides to have children. He/she and his/her partner would be eligible for pre-implantation genetic diagnosis (PGD), which would allow pregnancy with embryos that did not inherit the gene change. More detailed information about PGD can be found at: http://www.geneticalliance.org.uk/aboutpgd.htm. As you said, if the genetic cause is found then you and your unaffected children can be screened, and only those that inherited the gene change would need to be followed by an echocardiogram.

There is protection from the Genetic Information Nondiscrimination Act (GINA), which makes it illegal for an insurance company to deny coverage based upon a genetic diagnosis. Please note, though, that GINA does not provide protection on life insurance coverage. More detailed information about GINA can be found at http://ghr.nlm.nih.gov/spotlight=thegeneticinformationnondiscriminationactgina.

Question: Our son has hypertrophic cardiomyopathy (HCM) and has genetically tested positive to have LEOPARD Syndrome. Our first child was born at 32 weeks 3 days, and spent 50 days in the NICU. Does HCM or LEOPARD relate to premature birth? Also, with HCM due to LEOPARD, would it be beneficial to stay on a beta-blocker for life? Our son was removed from his beta-blocker in December 2013 at 15 months, as he has had no arrhythmias in a year. However, now that there are no preventative measures it makes us worry. We have another echocardiogram on June 2 and will most likely ask that he be put back on it as

his diagnosis was caught early and therefore his heart is working that much harder all the time.

Answer: I do not know of an association between LEOPARD syndrome (now also called Noonan Syndrome with Multiple Lentigines or NSML) and prematurity nor HCM related to prematurity. I will defer to your cardiologist with regard to the choice to be on beta-blockers.

Question: My daughter acquired idiopathic dilated cardiomyopathy (DCM) at 2 months old and had a transplant at 2 1/2 years. She just turned 5 a couple days ago and has been fantastic except for lingering neutropenia that no one can figure it out. In an effort to see if she was even making white blood cells and if she could have some type of Noonan Syndrome, we decided to do a bone marrow biopsy during her most recent heart biopsy. They were surprised to find Mosaic Turner Syndrome. She was missing the second X chromosome in 4 per twenty. We finally met with a genetic specialist two months ago who checked her blood and did not find any evidence of Turner syndrome. They felt that the Turner is just in her bone marrow and not in her blood. She basically does not have it and we do not need any follow up.

Have you had any experience with a chromosome issues in bone marrow and not blood? If so, were there really no symptoms? Do you think this is something she could carry on to a daughter, if she is able to have children someday?

Answer: If your daughter has no signs of Turner syndrome on physical exam (short stature, eyelids that don't open all of the way, short/wide neck, widely spaced nipples, swelling in her feet) or by her medical history (aortic coarctation, hypoplastic left heart, kidney malformation) then it is most likely that the Turner cells (the cells missing one copy of the X chromosome) are just in the bone marrow. DCM is not a feature of Turner syndrome. When and if your daughter has children, it would be prudent to at least mention this finding though it is unlikely to be present in her egg cells if the rest of her body is unaffected.

Question: We have a 15-year-old son who was born with hypertrophic cardiomyopathy (HCM). He needed to have a myectomy at 6 years old. Currently he is doing well on a beta-blocker and has had no real issues in the last several years.

We also have been through genetic testing in the years 2007 and 2008. They took a significant amount of tissue from his heart during his myectomy, and as I understand that tissue is on file at the Laboratory for Molecular Medicine, Center for Genetics and Genomics, Harvard Medical School.

Question 1: Could they still use that tissue specimen to test for other HCM genes? Our genetic testing revealed a heterozygous 3330+2T>G mutation (IVS30+2T>G)

in intron 30 of the MYBPC3. I do have the mutation and I passed it onto my first-born son. The laboratory indicated that at the time we had our testing done there was no known reported literature on our mutation and its clinical significance was not known. However, I am aware of an Amish study done on this gene mutation and the clinical course did not have a good outcome.

Question 2: Is there any current relevant information on our genetic mutation now?

Question 3: Is this mutation associated with a higher incidence of sudden cardiac arrest or does it have more of a benign clinical course?

We also have a second son who tested negative for this gene mutation. However because our first son presented before the age of one, and has a massive septal measurement, they are hesitant to totally clear our second son of HCM. They continue to feel that our first son may harbor a secondary genetic mutation that caused his HCM to present so young and so severe. Our second son is still seen every two years by a cardiologist for echocardiogram/EKG. He continues to show no signs of HCM.

Question 4: Would it behoove us to undergo a second genetic test to try and uncover a secondary mutation that may or may not exist?

Question 5: Do you know of any other families that have our same genetic mutation that have a secondary mutation that they are speaking of?

Answer: Question 1: Could they still use that tissue specimen to test for other HCM genes? The panel of genes available for genetic testing for HCM has expanded since 2007/2008 and the Laboratory for Molecular Medicine likely still has DNA saved for further testing. Whoever ordered your first son's testing in the first place can easily reorder the gene panel. In addition, the understanding of the significance of a given gene mutation changes with time as more and more families are tested. I would also encourage you to ask the physician who ordered the test to call the lab to ask if anything new about this variant has been determined since your son's report was written.

Question 2 and 3: Is there any current relevant information on our genetic mutation now? Is this mutation associated with a higher incidence of sudden cardiac arrest or does it have more of a benign clinical course? The reported cases of this mutation are in infants with a homozygous mutation, meaning a mutation in both copies of their MYBPC3 gene, which your son does not have. One weakness of the Amish study is that they did not echocardiogram the parents who, like your son, have a heterozygous mutation. The authors note that the parents were almost all asymptomatic in their twenties. One of the parents who did present with cardiac abnormalities had a mild form of asymmetric septal hypertrophy with a mild degree

of left ventricular outflow tract obstruction. We believe that for people with only one MYBPC3 mutation there may be a small risk and, if HCM develops, it develops later. However, because it is not truly known, anyone with this gene change with a normal echo (like you and your second son) should probably have regular echocardiograms.

Question 4: Would it behoove us to undergo a second genetic test to try and uncover a secondary mutation that may or may not exist? Based upon what we know about your first son's MYBPC3 variant, it seems likely that there is a different explanation for his early onset HCM, and additional genetic testing might uncover that mutation. It is possible that a second gene change, in and of itself, caused your first son's early HCM presentation, or it is possible that the combination of the MYBPC3 mutation and a second mutation together explains his early onset.

Question 5: Do you know of any other families that have our same genetic mutation that have a secondary mutation that they are speaking of? I don't personally know of a family with this particular MYBPC3 gene mutation and a second mutation but it is not uncommon to find more than one HCM gene mutation in children who have HCM. There is one family published in the literature with this mutation and a second MYBPC3 mutation.

Question: My family and I have had genetic tests done for the NXKY2.5 mutation. Two out of three people in our family are showing up with the mutation. The doctors can't explain why my youngest son had one atrial fibrillation incident and 21 tachycardia spells. Although my youngest son is not presenting with left ventricular non-compaction like myself, he is showing tachycardia like I just went through. Can NXKY2.5 cause rhythm issues not associated with any underlining heart condition? What can NXKY2.5 cause? What chromosome is it associated with and how can I find out more about this mutation?

Answer: I assume you are referring to the NKX2-5 gene? Mutations in this gene can cause atrial septal defect, ventricular septal defect, tetralogy of fallot, arrhythmia with or without a heart malformation, and hypothyroidism. The NKX2-5 gene is on chromosome 5. You can read more about it here: http://www.omim.org/entry/600584.

Question: Our daughter, now eight, has some sort of syndrome but unsure of what. We live in a remote place, (American Samoa) and do not have access to genetic testing. She may have low functioning autism, Cerebral Palsy or a similar syndrome. No one else in the family has hypertrophic cardiomyopathy (HCM). We have 7 biological children and one adopted. She is the last child. I am 53 and will not be having more children so genetic counseling is not needed in regards to pregnancy counseling.

We would love to know if the HCM is part of her undiagnosed syndrome or simply a random mutation, but to do genetic testing would be a huge expense. The blood cannot be shipped in time to any genetic center from our location, we do not have insurance in American Samoa, and then there is airfare. We have been continually encouraged to get genetic testing done, but my question is would it really be necessary other than to satisfy our curiosity about her diagnosis?

Answer: If your daughter has a syndromic cause of her HCM then the benefit of a diagnosis would be in knowing if there are any other medical or developmental problems that are associated with that syndrome for which she should be assessed or followed for over time. One of the critical components of making the diagnosis of a syndrome is a physical exam, and often a diagnosis can be made based upon physical exam and medical history without having to do genetic testing. I am sorry but I have no knowledge of the medical system in American Samoa so I do not know if there is ever a geneticist who visits to provide evaluations? Another option is that medical centers in remote locations like yours often have a Telemedicine program, which allows doctors in different locations to “examine” a patient by seeing them on a video conference. Perhaps you could ask your doctor about that possibility?

Exercise Rehabilitation and Nutrition

Gabriel Somarriba, D.P.T. – May 2014

Dr. Somarriba is an assistant professor at University of Miami and Exercise Physiologist and Senior Research Associate at Holtz Children's Hospital. Dr. Somarriba has authored several publications on nutrition and exercise rehabilitation on pediatric cardiomyopathy.

Question: Is it a general rule that hypertrophic cardiomyopathy (HCM) patients cannot play any competitive sports due to the risks associated with the disease, or is it a case-by-case decision? I have a teen that wants to participate in sports. She has never had an incident and has an implantable cardioverter defibrillator (ICD). With the exception of the possibility of hitting the ICD, would volleyball be an acceptable sport for her? She wants to do basketball, soccer or softball but none have been permitted. What about volleyball?

Question: My daughter is 13 and has dilated cardiomyopathy (DCM). She is asymptomatic but on some medications. Exercising and incorporating better nutrition have always been a problem. It is not because she cannot tolerate it; she just does not like to exercise and eat healthy foods. Over the years we were able to lessen her portions, which has helped with how much she is eating, but how do you get a teenager to begin eating more vegetables and fruits? How do you make healthy eating fun? She is not permitted to join sports, but can walk and jog. How can we make exercise fun for her? We explain to her the importance of both exercise and eating right, especially for someone with her heart condition. We don't want to repeat ourselves to the point of scaring her.

Answer: For children and adolescents with cardiomyopathy, participation in physical activity is a component of their lives that should be encouraged and monitored. As medical professionals continue to improve the lives of all your children, the roles that we take on at home become equally, if not more, important. Physical activity and nutrition are modifiable health behaviors that we should all pay attention to.

When it comes to the types, amounts, frequency, and intensity of physical activity for children and adolescents with cardiomyopathy, activities are on a case-by-case issue. Certainly, your cardiologist must be involved to help determine guidelines. Some patients are allowed to participate within the entire spectrum of physical activity, while some need to keep exercise at a low intensity. There are many factors that go into this decision and your cardiologist will help guide you.

When it comes to competitive sports, a few additional concerns are raised. Being educated is critical. We need to monitor kids in non-competitive sports

so they learn their bodies. What makes competitive sports more challenging is the difficulty in monitoring the intensity of the event. In competitive sports, the intensity is variable and the event may make it more difficult to keep intensity below the prescribed levels. This is where education is critical; the recognition of symptoms and the patients' understanding of when they should take a step back.

I am an advocate for physical activity for all, but in these special circumstances working closely with your cardiologist is very important. A stress test is an excellent test that helps set ranges of intensity for activity levels. I always begin physical activity at low levels and learn the kid's response. Building slowly will help recognize differences between what may be fatigue and what may be a symptom. Beginning with activities that are very controlled, such as walking, bike riding, kayaking, and other similar activities is a simple way to monitor children. From here, the activities may be increased in duration and intensity.

Questions: My daughters were followed by a children's hospital throughout childhood because my husband has hypertrophic cardiomyopathy (HCM). Electrocardiograms (EKGs) and echocardiograms (echos) were negative until the ages of 12 and 16 when they were both diagnosed with HCM. Before they were diagnosed, they participated in gymnastics, dance, karate, basketball, field hockey, and soccer. After a four hour training session in 90-degree weather, my 16-year-old old complained of shortness of breath and decreased endurance. It has been upsetting for them to give up competitive sports. They are now 17 and 21. They try to stay fit by going to the gym, but were told no isometric exercises. What exactly does that include? Are planks & sit-ups/crunches in this category? Is it safe to use the weight machines?

Answer: Isometric stands for "same length", iso = same and metric = length, so exercises that are considered isometric are exercises like planks, wall sits, or any exercise where the goal is to sustain a certain position with or without resistance. The main concern is related to breath holding. Isometric exercises are commonly associated with breath holding and when we think of ourselves doing any of these exercises we are more likely to hold our breath as we begin fatiguing. This can lead to dizziness and shortness of breath. Crunches or sit-ups are not isometric exercises and neither are weight machines, but you can imagine if the resistance is high on weight machines, chances of breath holding increases. I would highly recommend instructing them on the importance of breathing through each repetition.

Question: Are there any specific nutritional needs or deficits that parents of children with cardiomyopathy should be aware of? For example, what about vitamin depletion resulting from medications and the benefits of supplements? How can parents address these needs and deficits safely?

Question: We have been discovering a spectrum of side effects from medications, specifically decreased bone density due to a combination of metabolic acidosis, anemia, and low magnesium levels mostly due to Prograff and stress on the kidneys. We have been advised to just strive for a great diet and lots of exercise, which we always have. Are there supplements or other things that we should be doing? What else should we be looking out for?

Answer: Whenever we begin to discuss any chronic disease including cardiomyopathy, the initial concern as it relates to nutrition is primarily malnutrition. A discussion about nutrition is very in depth and important but requires a lot of back and forth as it is very individualized.

When seeing patients with cardiomyopathy, the question about nutrition usually comes up. The approach we have initially is general. I am interested in looking specifically at what one's intake is and analyzing it. With cardiomyopathy there may be an increased metabolic demand because of the disease, but even with that being said we need to determine what the current dietary intake is. A lot of information can be derived from a 24-hour recall assuming it's a typical intake. My recommendation is to begin with an analysis of what is being consumed and from there determine if there are any macro (protein, carbohydrate, fat) or micro (vitamin, minerals) deficiencies. It would be advisable to make sure the diet is adequate prior to taking a look at additional factors.

Supplements have a place but I do not believe that they should be the initial approach. Supplements may be used to help fill in some gaps in the intake, but first we should make sure the intake from foods is adequate.

I am also attaching a manuscript I co-authored on nutrition for cardiomyopathy, where you can find more specific recommendations:
http://www.ncbi.nlm.nih.gov/pmc/articles/PMC2151740/pdf/nihms33842.pdf

Medication Management in Pediatric Cardiomyopathy Patients

Teresa Lee, M.D. – July 2014

Dr. Teresa Lee is Assistant Professor of Pediatrics at Columbia University Medical Center and a pediatric cardiologist at Morgan Stanley Children's Hospital in New York City. Her specialties are cardiomyopathy, congenital heart disease, heart failure, heart transplantation, and cardiogenetics. She has received several academic awards and published in several scientific journals.

Question: Why is it important to make sure your pediatrician and cardiologist communicate what medications are being prescribed at any time?

Answer: It is important for all members of your child's medical team to be in good communication. Depending on how complicated your child's medical needs are, multiple specialists may follow your child. Since your pediatrician is often the one coordinating much of the care, it is vital that they be kept in the loop. Many medications can interact with other medications, so it is important you bring a full list of all the medications your child is taking to every doctor's appointment. Also, certain medications may pose additional risks in children with cardiomyopathy, so it is important that other doctors know about your child's condition, and if there is ever a question they should contact your cardiologist.

Question: I have a 3-year-old daughter, who was diagnosed with hypertrophic cardiomyopathy (HCM). So far she has had no health problems at all and is asymptomatic. HCM was discovered because of a murmur that was not very expressive at all. We live and receive treatment in Slovenia, where there is a lack of experience with HCM, especially in infants. Doctors prescribed metoprolol therapy twice a day for my daughter. When is medication therapy necessary?

Answer: Any given child's response to medical treatment can be variable so treatment has to be tailored to each individual. Metoprolol is a beta-adrenergic receptor blocking agent that works by slowing down the heart rate to help with ventricular relaxation and filling. Other common formulations include atenolol, nadolol and propranolol. Medical management is generally initiated when children are symptomatic. Medication may also be used in certain asymptomatic cases but there is a lack of consensus to the utility in these cases.

Question: Is there anything preventative we can do for "asymptomatic" Hypertrophic Obstructive Cardiomyopathy (HOCM) patients except "wait for it to rear its ugly head again?"

Answer: At this time, there is no published human data that adequately addresses the effectiveness of prophylactic treatment. We hope that as science advances

things may improve. This is one of the reasons we always recommend continued follow-up with a pediatric cardiologist.

Question: My son is now 10 months old with dilated cardiomyopathy (DCM) and is taking accupril, 2 diuretics (furosemide and spirolonactone), aspirin, digoxin (low dose), carvedilol, and vitamin C and pedia multivitamin supplements as well. He has had two blood transfusions recently because his hemoglobin keeps dipping below the norm. We are now consulting a pediatric hematologist in addition to his cardiologist and general pediatrician, but need to wait a few months for him to be able to do an osmotic fragility test on his blood to determine what type of anemia he has. So now he is also on folic acid and iron drops. We have also tried to increase his solids and introduced iron-rich foods. Is there a possibility that his anemia is caused by some of his medications? Have you seen cases where the anemia is related to or caused by the heart condition? Or should we be looking at other factors for this new development on his blood? Are there complications that we need to be aware of that could impact his DCM and left bundle branch block (LBBB) because of his anemia?

Answer: While rare, certain cardiac medications can be associated with anemia. In particular, aspirin is an anti-platelet medication, and it can sometimes lead to bleeding in the gut that may not be obvious. Make sure all of your child's doctors know every medication they are on, even if it is a medication that they only take once in a while or as needed. Anemia can sometimes be observed in patients with heart failure, especially those with severe disease, but these patients often have other medical problems as well. However, if you are anemic, your heart does need to work harder to get oxygen to all the vital organs. In cardiac patients, we do not want to put added strain on their hearts so we are often more aggressive about treating anemia.

Question: If a child with DCM has shown improved heart functioning and appears to be in the 'resolved' category following long-term use of medications and is gradually weaning off of all medications except carvedilol, is it recommended to take the child off the remaining beta-blocker medication? Is there any research to support remaining on carvedilol (or other beta blockers) or alternatively, taking the patient off medications, when heart functioning has improved and remains stable?

Answer: It is common practice at our center to slowly take children off their cardiac medications if their heart function and heart size normalize. Carvedilol is a unique beta-blocker in that it is also an alpha1-blocker. In studies, it has extended the favorable effects of selective beta-blocking agents and helped to reduce the mortality from heart failure. It may also play a role in cardiac remodeling. We mostly hear about cardiac remodeling in the context of adult studies on another type of medication called angiotensin-converting enzyme (ACE) inhibitors but some believe that carvedilol could have a similar effect. However, there is very little

evidence, especially in pediatrics, so it is best to discuss with your doctor the reasoning behind why they may be choosing to continue or discontinue a medication. Practices can vary and often times we have to see what works best for a child.

Question: What are your suggestions for helping teens to be more compliant with their medications? What tips can be suggested for helping them to take ownership of their medication management as they transition into adulthood?

Answer: We (parents and medical teams alike) all struggle with our teens. We love that they want to assert their independence, and yet we know that they still sometimes need our guidance. Many programs offer some sort of a transition program. It starts by having the child and then teen be more and more involved in their health care as they are able and at their own pace. Some of our 8-year-olds know all their medications while some 18-year-olds may still struggle. We will often conduct the visit specifically to the teen and ask him or her, not the parent, to name all of their medications and dosages. We emphasize the reason why they need to take their medications. In particular, when teens feel well they do not want to have to take daily medications or anything else that might make them different from their peers. Parents can play an active role by letting their teen be responsible for their medications and even letting them call for appointments or questions (of course, with parent supervision). Now that most teens have some sort of smartphone or device, those can be great tools with alarm reminders and the like. We find that the more active of a role teens play and the more that they understand about their health the more compliant they tend to be.

Question: Our son was diagnosed with dilated cardiomyopathy (DCM) when he was 4 months old and is now 6 years old. He is only on enalapril and carvedilol (Coreg) and CoQ10 supplement in terms of medications. He was on Coumidin, Lasix, aldactone, when he was sicker. We have had to use Miralax daily for years because of chronic constipation. We are not sure why he experiences constipation, whether it is the medication side effect or part of his autism spectrum diagnosis perhaps. We have recently been hearing about a possible link between chronic use of Miralax and autism. He was diagnosed on the spectrum last year. Have you heard of this and/or have any thoughts about it?

Answer: I do not know of a peer-reviewed human study that links Miralax (or its generic) to autism. While this has not been formally studied, I think that we can all anecdotally agree that there seems to an increased incidence of constipation in children with cardiomyopathy, as is the case with almost every chronic pediatric disorder. Many of our children are on multiple medications that can cause or exacerbate constipation. In children with additional medical issues there may be more than one reason for them to be constipated. It could be related to the typical picky toddler/child diet and the fact that about one-third of all children have

constipation. Since children with cardiomyopathy also see more doctors and have more medical appointments, it is also likely that they get treated with medication more than the average child whose parents may just ride it out.

Our kids still need to be doing the normal things like eating a healthy, fiber-rich diet. If, despite these maneuvers, they are still very constipated then I do not see the harm in something like Miralax. Many cardiac programs are closely affiliated with a gastrointestinal/nutrition program. Even one or two visits may be helpful if you are really struggling, so I would encourage you to ask your cardiologist if they think a referral would be appropriate.

Ventricular Assist Devices in Children with Cardiomyopathy

Scott Auerbach, M.D. – August 2014

Dr. Auerbach is the medical director of the ventricular assist device (VAD) program and assistant professor of pediatrics at the Children's Hospital of Colorado. Dr. Auerbach specializes in pediatric cardiomyopathy, heart failure, mechanical circulatory support, and heart transplantation. His research interests include biomarkers, diastolic function and predictors of outcomes in pediatric heart failure and transplantation.

Question: When would a ventricular assist device (VAD) be recommended for a child in heart failure?

Answer: In general, a VAD is recommended for a child in heart failure when standard medical management has failed and the child will not survive to transplant without improvement in cardiac output. When considering VAD therapy, it is important to have a clear goal for treatment. The vast majority of pediatric VAD patients need a VAD in order to survive to heart transplantation. If the child has been in heart failure for a long period of time and the heart failure has become progressive, the best option for long-term survival is heart transplantation. If heart failure is so severe that liver, kidney, lung, or intestinal function is compromised, then a VAD can restore blood flow to vital organs and improve organ function. Normal function in the lung, liver, kidneys, and intestine are critical to recovery after heart transplant surgery.

An international database called the Interagency Registry for Mechanically Assisted Circulatory Support (INTERMACS) has created different profiles to help us decide when a VAD is indicated. The pediatric arm of the registry is called PediMACS. A VAD is not indicated for INTERMACS profile 1, which consists of patients in shock due to severe heart failure. INTERMACS profile 1 patients usually have failure of other vital organs and do poorly after VAD placement. These patients often will require Extracorporeal Membrane Oxygenation (ECMO), which takes over the function of both the heart and the lungs to stabilize the patient and allow for recovery of lung, liver, kidney, and intestinal function. Once organ function has improved and the child has recovered from shock, then a VAD can be placed. INTERMACS profile 2 patients are slowly worsening on inotropic medications (IV medications that help the heart squeeze and include milrinone, dopamine, or dobutamine) with signs that other vital organs are being affected. INTERMACS profile 2 patients benefit most from VAD support. INTERMACS profile 3 patients have stable cardiac output and vital organ function on inotropic medications but cannot be weaned off these medications. This group can be considered for VAD placement. Practice varies from program to program with this group of patients as they may remain stable on inotropic medications until transplantation. This group requires close monitoring of vital organ function if a VAD is not placed. Patients

with INTERMACS profile 4 and above have heart failure that can be managed with oral medications and typically do not require VAD support.

Rare indications for VAD placement include: 1) a “bridge to recovery” if it is thought that the heart can recover function with VAD support and 2) a “bridge to decision” when more information is needed to determine if a patient is a transplant candidate. This strategy is used on a case-by-case basis and many factors will need to be weighed prior to VAD placement. In adults, patients can be considered for what is called destination therapy. Destination therapy means that a patient is not considered a transplant candidate and will live for the rest of his or her life with the VAD. There are currently no implantable devices in the US approved for destination therapy in patients under the age of 18.

Question: Is a VAD a helpful bridge until transplant if you have transplant coronary artery disease?

Answer: If a patient has transplant coronary artery disease, it is best to list for retransplantation prior to the need for VAD support. Most programs will recommend listing for retransplantation once an angiogram has shown that coronary artery disease is severe. However, there are rare situations in which coronary disease progresses much more quickly than usual, leading to progressive heart failure. Such patients can be considered for VAD support, but the need to continue immunosuppression even when a VAD is in place makes standard VAD placement complicated. If immunosuppression is continued, VAD patients are at higher risk of infection of the device. If immunosuppression is stopped, then the body will reject the heart and make the management of the VAD much more difficult. Due to the risk of rejection, transplant patients typically require biventricular support where a pump takes over the function of both the right and left ventricles. Furthermore, the surgical procedure to place the VAD may stimulate the body’s immune system and cause rejection even if immunosuppression is continued.

The total artificial heart is becoming more widely used as a bridge to retransplantation since it allows for complete removal of the heart and withdrawal of immunosuppression. This makes the total artificial heart a more preferable way to restore cardiac output and recover vital organ function while the patient awaits a donor heart. The problem is that the currently approved device can only be placed if the distance between the spine and chest wall is 10cm or more, which limits the device to teenage patients. Fortunately, there is a smaller total artificial heart that is undergoing study and may be available for use in the future.

Question: If a child is on beta adrenergic blockers, will they need to continue that medication while on the VAD?

Answer: Beta blockers should not be used in the immediate post-operative period following VAD placement, especially if only a left sided VAD (LVAD) is placed. The right ventricle will need to function well enough to supply blood flow to the LVAD. In the early post-operative period, the function of the right ventricle is usually decreased and needs medications like epinephrine or dopamine to improve the amount of blood it is pumping. Beta blockers directly counter these medications and can make it more difficult for the right ventricle to do its job. Beta blockers can be used once the patient has recovered from surgery, but they are no longer needed for the same reasons they were used prior to VAD placement since cardiac output will have been restored. Beta blockers may be used after recovery from VAD placement to treat abnormal heart rhythms such as atrial or ventricular tachycardia (fast heart rates originating abnormally from the upper or lower chambers of the heart, respectively). These abnormal heart rhythms can decrease the amount of blood pumped from the right ventricle to the LVAD. Beta blockers can also be used to treat high blood pressure. Another situation in which a beta blocker may be beneficial is in patients that have a VAD for recovering heart muscle function as in "bridge to recovery." Beta blockers can help improve heart function, size and shape while on a VAD. However, a VAD indication of "bridge to recovery" is a rarely used in children with cardiomyopathies. The overall experience with VAD removal in pediatric patients with cardiomyopathies is very limited. Beta blockers are generally not necessary after VAD placement if arrhythmias, high blood pressure or potential for recovery are not present.

Question: I am a mom to a 7 month old diagnosed two months ago. Are all VADs a bridge to transplant or are there any that can help a heart to work without a transplant? Is there any VAD in development to avoid a transplant and have a normal/semi normal heart function?

Answer: The vast majority of children that have a VAD placed for heart failure should expect that they will be listed for a heart transplant. A VAD for the purpose of "bridge to recovery" so that heart function can return to "normal/semi normal" is a rarely used in children with cardiomyopathies. The overall experience with VAD removal because of recovery of heart function in children with cardiomyopathies is very limited. We are still trying to understand which children have potential for recovery of heart function after a VAD is placed. This is an area of ongoing research. The potential for recovery of heart function needs to be assessed on a case by case basis.

Question: Is a VAD ever used for people with left ventricular non-compaction cardiomyopathy (LVNC)? Is it used on adults as well as in children? How bad does the heart need to be to have a VAD placed?

Answer: A VAD can be used in left ventricular non-compaction (LVNC) in both children and in adults. LVNC can make it more difficult to place a VAD due to many

muscle bundles within the left ventricle. The decision of whether a patient with LVNC can receive a VAD must be made on a case by case basis in discussion with the VAD surgeon.

In general, a VAD is recommended for a patient in heart failure when standard medical management has failed and survival to transplant is unlikely without improvement in cardiac output. If heart failure is so severe that liver, kidney, lung, or intestinal function is compromised, then a VAD can restore blood flow to vital organs and improve organ function. Normal function of the lung, liver, kidneys, and intestine are critical to recovery after heart transplant surgery.

Pediatric Cardiovascular Anesthesiology in Pediatric Cardiomyopathy Patients

Courtney Hardy, M.D. – November 2014

Dr. Courtney Hardy is the director of pediatric cardiac anesthesia at Ann & Robert H. Lurie Children's Hospital and associate professor of anesthesiology at Northwestern University Feinberg College of Medicine in Chicago. Dr. Hardy specializes in pediatric cardiovascular anesthesiology and her main area of research interest revolves around safety in pediatric cardiac anesthesia and simulation based training to improve clinical care in the operating room.

Question: Why is it important to let the anesthesiologist know of any recent changes in medications and/or dosages before a procedure?

Answer: It is important to let your anesthesiologist know of any drug changes or dosages as many cardiac medications do have an impact on which anesthetic we choose for your child. In addition, changes in dosages and drugs may indicate a change in your child's cardiac status, which would be very important for your anesthesiologist to know.

Question: For a child who is not exhibiting symptoms, should there be any concerns about anesthetics used in dental work?

Answer: Yes, any child with cardiomyopathy even in the absence of symptoms does pose specific challenges for the anesthesiologist, even for procedures such as dental work. Your anesthesiologist would want a very detailed history of your child and would want to see a recent evaluation from your child's cardiologist prior to planning a safe anesthetic plan for your child.

Question: Is there something better to use besides chloral hydrate for sedated echocardiograms, or is this the best option?

Answer: Sedated echocardiograms are done differently depending on the institution, age of the child and maturity of the child. Chloral hydrate is used safely at many institutions for sedated echos.

Question: In dental work and anesthesia, is there anything else you need to let the technician know besides their medications (if any) and the condition in which they have?

Answer: When having dental work in an office without anesthesia they should know the medications and condition, and a recent clinic visit report from your cardiologist would be helpful information as well.

Question: Our 8 year old daughter was diagnosed with restrictive cardiomyopathy (RCM), and she declined very rapidly once diagnosed. What kind of effects can procedures requiring general anesthesia such as cardiac catheterization or implantation of an implantable cardioverter defibrillator (ICD) have on the "stability" of a patient with RCM? We know that within cardiomyopathies, RCM is the rarest and least known about form, but we continue to wonder what role general anesthesia played in our daughter's steep decline following her surgical procedures. Have any studies been done that explore the trajectories of these diseases with repeated instances of general anesthesia?

Answer: Clearly, anesthesia in these type of patients does have risks, but I do not know of any evidence to indicate that the actual anesthesia or procedure itself would be the cause of the rapid decline. Anesthesia alone for patients with RCM can involve significant risks, however cardiac catheterizations and ICDs are necessary treatments for this condition and without anesthesia these treatments would not be an option.

Question: Is there a certain age recommendation for children with cardiomyopathy in which it is safe to undergo different types of anesthesia?

Answer: All cardiomyopathy children pose additional risk compared to children without a diagnosis of cardiomyopathy, regardless of age. The specific timing of the surgery should be discussed in great detail with your cardiologist.

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