Children's Cardiomyopathy Foundation™

Answers from the Experts: CCF Cyberguests

A Compilation of Q & A Pediatric Cardiomyopathy Listserv Sessions 2010–2011

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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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Helping Your Child Cope with Medical Tests or Hospitalization

Toni Millar, M.L.S., C.C.L.S. and Alison Heffer, C.C.L.S. – January 2010

Toni Millar joined Morgan Stanley Children's Hospital of New York (MSCHONY) in January 2008 as the Director of the Child Life department. Previous to MSCHONY, Ms. Millar was the Director of the Child Life and Family Services department at Rainbow Babies and Children's Hospital and has been a Certified Child Life Specialist (CCLS) since 1995. She has held several leadership positions for her national organization, the Child Life Council.

Alison Heffer is a certified child life specialist at MSCHONY. She received her undergraduate degree from Muhlenberg College and her Masters in Special Education with a concentration in child life from Bank Street College. She has been at MSCHONY since 2001.

Question: What is a child life specialist?

Answer: Child life specialists are experts in child development, who promote effective coping through play, preparation, education and self-expression activities. They provide emotional support for families and encourage optimum development of children facing a broad range of challenging experiences, particularly those related to health care and hospitalization. Understanding that a child's well-being depends on the support of the family, child life specialists provide information, support and guidance to parents, siblings and other family members. They also play a vital role in educating caregivers, administrators and the general public about the needs of children under stress. For more information please see the following website: childlife.org.

Question: When should my child see a child life specialist?

Answer: Most major children's hospitals as well as some community hospitals have child life services available. Here again from the child life council are the top ten reasons to utilize a child life specialist:

- Child/ Patient needs preparation for invasive procedure.
- Child/Patient is having difficulty coping with a necessary procedure (i.e. crying, fighting or hiding).
- Child/Patient exhibits oppositional behavior, refusing to cooperate without anger or hostility.
- Child/Patient or siblings express specific fears to staff, needing follow up.
- Child/Patient is perceived by staff as withdrawn.
- Child/Patient is having difficulty taking medications.
- Child/Patient admitted having recently experienced traumatic loss or has a chronic illness (developmental delay).
- Child/Patient is newly diagnosed with chronic illness.

- Child/Patient admitted for injuries resulting from a traumatic accident (i.e., MVA, fire, etc.).
- Child/Patient injuries or diagnosis has resulted from suspected child abuse.

Question: I have a soon-to-be-16-year-old with severe hypertrophic cardiomyopathy. He hates all kinds of medical tests and has outright refused the implantable cardioverter defibrillator (ICD) recommended by several cardiologists. I have had countless sleepless nights over this. He had a bad experience with an MRI and so now refuses that as well. He will not do the Holter monitor tests because "I can't stand the way it feels on my skin." He is very physically sensitive. Doctors have talked to him at length on the need for ICD placement. He feels it will make him look “deformed” and also thinks he will be able to feel it inside him (he has met other teens who have one and this just confirmed his fears). He is also afraid of accidental shocks from the device. What can I do? I know I am legally responsible, but I know I can not force him. He already ran away once when we tried to compel him to have the surgery. The whole family has banded together to support him but he will not budge. He is in counseling, but I'm not sure the therapist knows what to do either--there has been pretty much no movement on this issue.

Answer: Some of the concerns your son has are typical for all adolescents regardless (body image, fear of the unknown etc). It might be helpful to contact your son’s cardiologist to see if there is another patient who has had an ICD and has had some of the experiences your son is afraid of. It might help your son to hear about another patient’s positive experiences with a device. If possible, find out from your son’s cardiologist what hospital he/she is affiliated with. Once you have gathered this information, contact the child life department and they can provide further assistance.

Question: Do you know of any programs that deal specifically with teens living with heart disease? Many of us on this listserv who have teens deal with the fact that adolescence compounds the emotional side of medical issues. I admit I have a particularly stubborn teen; I also think he needs that stubbornness to live with severe HCM. That said, I feel this is an issue that needs serious consideration by doctors, hospitals, child life, etc. My son has met two teens with ICDs; these were good experiences but they did not lead him to change his mind. He sees a counselor, has seen child life staff at two major hospitals, and has talked at length with cardiologists at an HCM center.

Answer: We agree completely that adolescence compounds the emotional side of medical issues for teens. Your thorough approach to helping your son seems to include many of the resources we would suggest.

Question: My daughter is four and is currently on the list for a heart transplant. She has had two major hospitalizations in the last two years and consequent to that, has been fearful of being hospitalized and prodded for tests. Can you suggest ways of "preparing" her for the heart transplant? Thus far, I have briefly told her that she may go into hospital soon, and the doctors will try to fix her heart so that she can be stronger.

Answer: We already see that you are using simple concrete explanations, which is one of the first things we would suggest. Additionally, there is a great book called *The Perfect Gift* by Dianna Veit, which explains heart transplantation on a developmentally appropriate level for your four-year-old. This book can be found either on amazon.com or on kidswithheart.org. Your daughter is at a perfect age to engage in medical play so we recommend contacting the child life specialist at the hospital where you would go for the heart transplant.

Helping Your Child Cope with Feeding Issues

Suzanne Evans Morris, Ph.D. – March 2010

Suzanne Evans Morris, Ph.D. is a speech-language pathologist in private practice near Charlottesville, Virginia. With more than 45 years of clinical experience, she is nationally and internationally known for her work in identifying and treating young children with pre-speech and feeding disorders. She is the director of New Visions (www.new-vis.com), which sponsors innovative workshops for the teaching of feeding-related skills and provides family-oriented clinical services.

Question: When trying to get a child who is having difficulty gaining weight to eat more, how many times a day should a parent offer food?

Answer: Weight gain, as you know, is related to the calories and nutrients that a child eats and to how well the child absorbs these nutrients, but this does not relate directly to the number of times a parent offers food. Some children eat very small amounts at each meal because they simply are not very hungry. Slow or reluctant eating reduces the amount of food that is eaten at one time. Other children are hungry for one or two meals, but not the others. Sometimes when children aren't eating enough, parents will continually offer food all day. To the child it appears that the only way they can please an adult is to eat. They feel they are constantly being pressured to do something that is not comfortable or pleasurable. Out of desperation for the child to gain weight, parents may offer high-calorie cookies, candy bars and other empty calorie foods.

Children eat best when they are considered real partners in the mealtime process. When they know that they will not be pressured by an adult to eat, they are likely to eat more. It is important to offer food at regular intervals that both the child and the child's body can expect. Depending on the child's age, caloric needs, hunger levels, etc. this can be anywhere from three to six meals. This should include three larger meals (breakfast, lunch and dinner). In addition, there should be up to 3 regularly scheduled snacks (morning, afternoon and evening). Meals should consist of healthy, fresh foods whenever possible and should be offered at specific times and in a location that you, as a parent decide. Kids should have the opportunity to share meals with another person or the whole family at a table rather than riding around in the car, grazing all day while watching television, or running through the house.

One of the approaches I have found to be very helpful is the “Division of Responsibility” that is discussed by Ellyn Satter. Ellyn is a dietitian and family therapist from Madison, Wisconsin. Her books describe a structure in which the PARENT is responsible for WHAT foods are served, WHERE they are served, and WHEN they are served. The CHILD is responsible for HOW much of the food is eaten and WHETHER any food is eaten. These guidelines prevent the

battles that often occur around eating for many children and parents. You can find more information on her website at ellynsatter.com/. She has some wonderful books, which include: *Feeding with Love and Good Sense*, *How to Get Your Kid to Eat: But Not Too Much,* and *Secrets of Feeding a Healthy Family.*

Questions: Our son has been fed by a nasogastric and gastric feeding tube since he was four months old. He is now almost two. We have been doing occupational therapy for about a year with very small amounts of progress (he is still 100% tube fed). This past December he had to be on Prednisone for five days for a chest cold, and we saw a VERY dramatic increase in his interest in food--scooping yogurt and putting it to his mouth, grabbing food out of people's hands to put in his own mouth, etc. He was also much more amenable to smelling food when it was offered to him. It made us aware that he has the skills to eat but that his nausea is consistently overwhelming him. We think it is from his medication side effects and poor heart function. We have tried a few things like Reglan, Prilosec and also holding his tube feeds to generate hunger, but we haven't really seen any change. We seem to be doing everything else possible to facilitate eating (having him sit with us with non-smelly food to play with at meals, offering things whenever we're eating, making food kid-appealing) but the nausea is the major roadblock. Is Periactin something that may be helpful for a situation like this? Do you have any other ideas about ways to mitigate nausea?

Answer: Steroids such as Prednisone have a reputation for increasing hunger and food cravings as a side effect. Because of other side effects of steroids, these medications are not appropriate to use long-term to stimulate hunger and appetite. Your short-term experience with Prednisone gave you additional insight into your son's underlying skills and abilities. The nausea that he experiences can be a huge deterrent to wanting to eat. Think about experiences that you have had after the flu or other illnesses when you may have still felt some post-vomiting nausea. When you feel nauseous, you have no feelings of hunger and may even feel strongly antagonistic to wanting any food. This is why a medication such as Periactin would probably have very little effect on his desire to eat.

Nausea, reflux and vomiting can all be increased by stress, especially the stress created by a child's tube feedings. When a child is unwell or growing slowly, professionals place a lot of pressure on parents to give a certain amount of formula through the feeding tube. Even if the child is showing signs of nausea or discomfort with the tube feeding meal, the parent feels that the entire amount of formula must be given. My first guideline in working toward oral feeding is to spend a lot of time observing and working with the child's tube feeding meals. If eating by tube is often accompanied by discomfort, children will associate these feelings with food. They will not choose voluntarily to eat a lot by mouth. I would look for the very first signals of discomfort during the tube feeding. Each child is different so the cues may be restlessness, increased saliva, pulling on an ear or a sour face. At that point I would stop the flow of formula through the tube and we

would "give the tummy a short rest." I work with the child to build awareness of when the tummy feels ready for a bit more. When you keep the level of discomfort low and do not provoke it with more food, the nausea (retching, vomiting, gagging) decreases and the child learns how to recover. This is often the first step toward beginning to self-monitor actual internal feelings of hunger and fullness. I have also had the feeling over the years that when a child has experienced gastrointestinal discomfort at mealtimes, he builds a strong negative association between these sensations and food. When the first sensations of discomfort are experienced, the inner response is to move away from the discomfort and food. Hunger signals in all of us a small amount of discomfort. For us, that discomfort is often followed by sensations of comfort and satisfaction as we are eating our meal. But many children like your son may feel the first discomfort, eat a tiny amount to please a parent or therapist and then strongly refuse to eat more. He is really just taking care of himself based on the meaning that he gives the internal sensation such as "If I eat more, I will feel sick." There are several papers on my New Visions website that discuss these issues in greater depth:

Mealtime Skills-Increasing Gastrointestinal Comfort
new-vis.com/fym/papers/p-feed25.htm

Gastrointestinal Health and the Child with Feeding Problems - Part 1: The Issues
new-vis.com/fym/papers/p-feed16.htm

Gastrointestinal Health and the Child with Feeding Problems - Part 2: Therapy Alternatives
new-vis.com/fym/papers/p-feed17.htm

Stress and Mealtimes
new-vis.com/fym/papers/p-feed26.htm

Question: My son is having some weight gain and feeding issues, which the doctors do not believe is caused by his heart. He is 21 months old and weighs 18.5 pounds. We are beginning the process for a feeding tube but I was able to convince the doctor to try Periactin in the meantime. Oral aversions and sensory issues have been ruled out. It seems the Periactin is working really well! Can you tell me anything about the mechanism by which this drug works? Can you tell me who it seems to work well for and who it does not work for?

Answer: Periactin is actually an antihistamine that has the side effect of increasing hunger. This is a drug that can be effective, as you have seen with your son, when children have the basic desire and skills to eat and drink. It works best when

children do not have gastrointestinal, swallowing or sensory issues that create discomfort associated with food and eating. Parents have found that the effect of Periactin wears off after about two weeks. To counteract this, they often give the medication for two weeks and then stop giving it for a week. After this rest period, they often find that the effect of stimulating hunger and appetite returns. Other parents simply observe their child for any reduced interest in eating. When this occurs, they assume that the medication effect is wearing off and will give them a break.

Some children do not do well on Periactin because, as an antihistamine, it often makes the child very sleepy and irritable. To counteract this, they may give the medication at night before the child goes to bed. Other parents divide the dosage by trial and error and give it throughout the day.

Question: My son sometimes eats four to five bites and then throws the rest of his food on the floor or crumbles it in his hands. He is 21 months old so I know this is somewhat of a normal behavior. Can you tell me the best response to have? The best I can come up with is to hand some back to him from the floor and have him give it back to me or put it on the table. I also try not to give him too much at once but it is still frustrating. He is a smart little guy, and I know he knows by now that it is a "no no" but I do not want to create a negative battle around the food. He will sometimes start eating again after he has thrown or crumbled food, but often he is done at this point.

Answer: I find it most helpful to create a division of responsibility with children in which you are responsible for the food served, the location, and the time for the meal. Your child is responsible for how much he eats or whether he eats. Within these general guidelines, children need and feel most comfortable when adults set limits for what is or is not acceptable during the meal. The four to five bites of a food that your son eats is his decision. However, throwing food on the floor is not an acceptable mealtime behavior, even for a child of 21 months. It may, however, be the only way he has figured out to tell you clearly that he has finished and does not intend to eat any more. Many years ago I developed the concept of an "All-Done Bowl." This is a small bowl that you place next to his food bowl or plate. When he has finished with a food, he can put it or spit it into the “All-Done Bowl,” and you will know that he has finished with any food in this bowl. I use the label "All-Done Bowl" so that he really knows what it is for. If he continues to throw the food on the floor, you gently remind him that you know he has finished the meal but that the "all-done food" goes in the "All-Done Bowl." It is the same concept as not throwing blocks across the room at the cat. You help him learn that his blocks go in the bucket or wherever you designate. Your part of this is that you honor his decision and do not ask him to take anything out of the bowl and eat it. Some kids surprise you and begin to eat whatever is in that bowl when they trust that you are not making this decision for them.

Another idea that I have found helpful is to serve a child's food on a very small plate or in a small bowl. Offer an amount that may seem small to you but that is within the range that he usually eats. I often encourage families to serve the meal family-style with larger serving bowls at the table and the child helping to serve what goes on his plate. In this way, there is always a bit more of the food and the child has the opportunity to ask for more. When children are not gaining weight, our natural tendency is often to serve them very large amounts hoping that they will eat more. The reverse often happens because there is so much food in the bowl that the child never feels a sense of inner satisfaction and accomplishment for the amount he eats. When there are five bites in the bowl and the child eats three or four bites, most of the food has visibly disappeared. Kids feel as though they have really eaten something.

Question: Both of my boys have always been picky eaters. I am a single mom and a pretty diverse eater so I try to feed my children a variety of foods, including kid-friendly foods. At each meal I try to have at least one item that I know they will like. This usually works for my older son and he is semi-willing to try new foods, but my youngest is a really stubborn and challenging one! My younger son is well within the weight/height range for his age. He is about 75% for height and 55% for weight, so he is not underweight although he does appear skinny. He has resolved hypertropic cardiomyopathy and is on Atenolol for his SVT (arrhythmia) issues. He probably has high functioning Autism (Asperger's Syndrome) although I have not sought out a diagnosis.

He has a hard time at all mealtimes. His favorite food is waffles and if I am out of waffles or choose to make another breakfast he will cry and scream. I try to warn him the night before, which sometimes helps. His lunch sometimes consists of pepperoni and a couple of cheese slices. Today he had one single serve applesauce cup, some goldfish and that was it. Dinner is a disaster; he is just as limited there. I have gotten him to eat turkey tacos (in the shape of pizza), sausage, hot dogs, pizza, fish sticks, garlic bread and tater tots. He does not eat a single vegetable. Fruit is touch and go and no pasta or milk. More often than not he goes without dinner.

I do not want mealtime to be a battle. It does not help that at their dad's house, it is nothing but fast food and pizza. By the way, when he eats a hotdog, the skin must be off; he does not eat the roll and does not want ketchup on it. I definitely think these food issues are part of his sensory and auditory processing problems. Any advice?

Answer: Many children are picky eaters. They may have highly preferred foods or categories of food that they eat, and they are very cautious about eating anything unfamiliar. Some of these youngsters, such as your oldest son, are more open to expanding the diet if you continue to offer new foods without the pressure of

making the child eat them. Other children, such as your younger son, are more highly selective in the foods they will eat. They do not like change; things must stay the same. They may pay attention to small differences in a piece of food that you or I would not even notice. If the shape of a piece of pasta is slightly different or the number of spots on a piece of pepperoni are different, the child may consider this a big enough change that he will not eat it. This type of strong food selectivity (more than just being picky) is often part of a sensory processing problem. If your son is seeing an occupational therapist for his sensory and auditory processing problems, you can ask for specific suggestions that might help him become more open and comfortable with changes in the sensations he experiences with food. For example, some children with sensory processing difficulties become more organized and comfortable with swinging or bouncing, jumping, or certain types of music – sensations that can help the body become more organized and deal with new sensations better. Many parents have found that doing activities such as these 15 or 20 minutes before the meal can be very helpful. The types of sensory input that can help a child is very individual, so it is important to talk to your therapist.

You may have more success in increasing your son's openness to new foods if you begin by just making changes in other parts of the meal. Use a different colored plate, different utensils, or something different on the dinner table while giving him familiar foods at the meal. Once he is comfortable with changes in the non-food part of the meal, make very tiny changes in the food he will accept. For example, you mentioned that he loves waffles. You could change the color or taste of the waffle slightly by using a different brand of waffle mix. Or you can make your own waffles from scratch and vary the ingredients slightly. You could add eggs for protein or a tiny amount of a pureed fruit or vegetable. You want to keep these changes very small. You mention that he likes to eat fast food when he is at his dad's house. Find out what types of fast foods he likes. Are there variations that you can make at home that will be similar but healthier? For example, if he likes french fries you could make baked fries. There is a book that many parents have found helpful called *Food Chaining: The Proven 6-Step Plan to Stop Picky Eating, Solve Feeding Problems, and Expand Your Child's Diet* by Cheri Fraker. It breaks things down into these kinds of small steps.

Question: My daughter is six months old and she weighs 13 pounds. She has had dilated cardiomyopathy (DCM) since birth. She takes about 24 ounces of formula, two or three tablespoons of baby rice cereal and two tablespoons of vegetables or fruits everyday. Is this enough for her?

Answer: Have you talked to your doctor or dietitian about the number of ounces of formula that your baby should be taking now? Although there are average or typical amounts of formula at each age, every baby has different needs. The amounts of rice cereal and vegetables or fruits that she is taking are very typical for a six month old baby who is just starting to eat pureed foods. Babies are able to decide

for themselves how much formula to drink. Most babies begin to discover new pureed baby foods by the time they are six months old. Offer a new food every four to five days so that your baby will be happy eating many different types of fruits, vegetables, cereals and meat or other protein. Variety is very important. The most important thing is to keep the mealtimes happy and let your daughter decide how much she will eat.

Question: I am writing because my daughter is not gaining weight or growing as fast as the doctors would like her to. She is 12 months old and only weighs 13 lbs 6 oz. She received a new heart eight months ago. She is very inconsistent with her eating. Some days she eats great; other days she is not interested in eating. She is really active, cruising all around our living room holding on to furniture (hates to sit or lay down as of late). She drinks formula as her main diet, and when we try to mix it any stronger than 24 kcals she refuses to eat it. She also eats baby food and cereal but is inconsistent about that. She seems to like carrots and sweet potatoes, but may refuse those or eat just a tiny bit on some days.

I am concerned because they have referred us to the gastroenterologist and are saying she may have to get a feeding tube placed again. She has been off the feeding tube for six months and has been gaining weight and growing, but very slowly. The tube was a real disaster because she would peel the tape off her face and tear them out quite often requiring me to replace them. I feel like she will be even better at getting them out now that she is older and more active. I am not sure if it is her increased medication that is making her not feel hungry or if she is just going through a phase because of her new-found walking ability. If you have any advice on things to feed her or ideas for helping her to eat better, please let me know! I really would like to avoid the feeding tube if at all possible!

Answer: Your feelings about not going back to a feeding tube are understandable, yet it is an option that sometimes needs to be considered when a child is not growing well. The first question when a child is inconsistent is always "Why?" There is a normal appetite variation in young children, but it sounds as though your daughter goes for days at a time when she is not interested in food or eating at all. One common reason for this is a gastrointestinal problem such as reflux. If it hurts to eat because the esophagus (the body's natural feeding tube) is inflamed from acid, children will choose not to eat. Another condition that can cause similar inflammation is an allergic condition called eosinophilic esophagitis. Since you have been referred to a gastroenterologist, you will have the opportunity to ask the doctor to rule out these two conditions as reasons why your daughter is variable in her desire to eat. The bottom line is that we simply cannot make a child eat. We can offer food, but unless we pry the mouth open and force the food in (never recommended!) we can only offer the food at a time and in an environment that supports any desire the child has to eat. I would offer her a wide variety of foods.

Kids need variety to get the nutrition they need to grow and be healthy. They also get bored when you give them the same foods all the time. If you offer a new food, know that it may take her a while to get used to the new taste.

If your doctor or dietitian does recommend the feeding tube, discuss the option of a tube that goes directly into the stomach (gastrostomy tube). Although the g-tube is placed through surgery, it does not have to be inserted and reinserted through the child's nose and throat. Many parents do not realize that the g-tube is not a permanent tube. It can easily be removed when the child no longer needs it. If parents are feeling a lot of stress about their child's poor growth and nutrition, a feeding tube can be much more supportive in helping the child learn to enjoy food and enjoy eating. Often, the only alternative that parents use is to push food and feed the child for most of the hours during the day. This often results in battles over eating which, in the long run, make it much harder for the child to gain weight and to want to eat more.

You mention that your daughter has been gaining weight and growing slowly since the nasogastric feeding tube (ng-tube) was removed six months ago. Each child has what is called a growth curve. The points on the curve are marked each time the doctor weighs the child. The amount of weight or growth in height is not as important as whether the child remains at the same percentile (i.e. place on the growth chart) each time. For example, if she is following her own growth curve so that she is at the fifth percentile each time, it shows that she is growing at a consistent rate. If, however, she was at the tenth percentile when she was six months and nine months and is now at the fifth percentile, it would mean that she is not following her personal curve and needs to grow at a faster rate.

If at any time you have other questions about feeding and mealtimes, you are welcome to e-mail me at sem@new-vis.com. My website at new-vis.com has “Information Papers” on all aspects of feeding, learning and sensory-motor skills. It also has information about the three-day workshop program, “Becoming a Mealtime Partner,” that I do twice a year for parents and professionals.

New Research on Pediatric Cardiomyopathy

Dr. Seema Mital, M.D., F.A.C.C., F.A.H.A., F.R.C.P.C. – April 2010

Dr. Seema Mital is an Associate Professor of Pediatrics and Staff Cardiologist at the Hospital for Sick Children in Toronto. Her research interests include the genetic basis of heart disease in children and adults, genetic determinants of outcomes in congenital heart disease and cardiomyopathy, stem cell regulation in cardiac malformations, and the molecular basis of right ventricular adaptation and failure. Dr. Mital received a CCF research grant in 2004 for her study "RAAS Gene Polymorphisms Influence Cardiac Remodeling in Children with Hypertrophic Cardiomyopathy."

Question: Pediatric Cardiomyopathy is a rare disease that historically has had a slow rate of progress for research. Has the amount of research into the causes and treatments for pediatric cardiomyopathy increased in recent years?

Answer: I do not have actual numbers to make comparisons about investment in pediatric cardiomyopathy research over time. In general though, investment in this area of research has increased. I think CCF has done a fantastic job of funding research in this area and of aligning with the American Heart Association to support joint funding. Also, the NIH funded Pediatric Heart Network recently completed a study in children with dilated cardiomyopathy, the results of which are being analyzed. There have been some drug trials (e.g. idebenone in Friedreich's Ataxia, enzyme replacement therapy on Pompe's disease) that have focused on or included children. Stem cell research is largely being done in adults, but stem cells are equally important to model disease and to develop new drugs specific to the type of cardiomyopathy. However, there is still a need for greater investment in this area since there are many unanswered questions and a need to find better and more targeted therapies for this.

Questions: Is there any research on stem cells treatment for hypertrophic cardiomyopathy (HCM)?

Answer: There is no research on stem cell therapies in HCM. The utility of stem cells is less clear in HCM since there is less or no loss of heart muscle in HCM compared to other forms of cardiomyopathy where replacement with stem cells may be of benefit. HCM is more complex, and it is not clear that it would benefit from stem cell therapy unless the stem cells were somehow able to correct the gene defect.

Question: Is there any research on stem cell treatment for kids or adults with left ventricular non-compaction cardiomyopathy (LVNC) and dilated cardiomyopathy (DCM)?

Answer: There are no pediatric stem cell therapy trials. There have been multiple, small adult stem cell therapy trials for dilated cardiomyopathy – most of which have shown either no effect or some early improvement in heart function but this is often not sustained. Also, no long term trials have been completed to show if stem cell therapy improves overall survival although these are in the pipeline and results are awaited.

Question: What does the latest research show about the relationship between hypertrophic cardiomyopathy (HCM) gene mutations and the penetration of the disease? Is anything known about the factors that might lead to more severe forms of the disease in certain mutations and in certain individuals?

Answer: HCM genes are in general quite penetrant. There are some predictors of which patients may have more severe forms of HCM. For example:

1. Presentation during infancy: This is largely because the cause of HCM in patients with early presentation is often a metabolic defect that progresses more rapidly than HCM caused by sarcomeric gene mutations.

2. Compound mutations: More than one disease causing mutation in the same patient can also lead to a more severe form of HCM.

3. Modifier genes: These are common variations in genes that can modify the way the heart responds to the gene mutation. As more of these variations are identified with the availability of advanced gene technology, we may be able in the future to predict which gene positive patient is likely to have a more severe manifestation of the disease.

Question: Why do babies with cardiomyopathy have fewer wet diapers? When is it considered a serious problem?

Answer: When the heart gets too weak to pump enough blood to the kidneys, the kidneys do not make enough urine. The number of wet diapers depends on the age of the baby. The child should be seen by a doctor if the number of wet diapers drops a lot or the baby has not had a wet diaper all day or all night.

Question: I would like to know if there is a recommended list of supplements to give a child who has been diagnosed with one of the cardiomyopathies? Have you seen any success in treatment through the use of supplements?

Answer: Supplements are generally recommended only with very specific causes of cardiomyopathies. For example, carnitine supplementation if there is either

carnitine deficiency or some type of fatty acid oxidation defects (where the heart is missing the enzyme required to transport fat into cells for producing energy). Another example is coenzyme Q10 which is also given when there are fatty acid defects. These supplements are only helpful if given for these specific indications in which case they can be quite successful in preserving heart function. They are not indicated in all patients with cardiomyopathies since they do not help in those cases. In some cases, these supplements may even cause harm.

Question: How do you know if there are fatty acid defects? Is this something tested for in cardiomyopathy?

Answer: They can be diagnosed by special tests done on muscle or skin biopsy. This is offered only if the initial metabolic screening of blood or urine suggests a fatty acid defect. In some cases, it may be offered if there is involvement of other organs besides the heart (i.e. muscle, brain or kidneys) that cannot be easily explained.

Question: In relation to cardiomyopathy presenting during infancy, is there any treatment/management of the metabolic defect in order to stop the rapid progress of hypertrophic cardiomyopathy (HCM)? Further, has the type of metabolic defect been identified?

Answer: Several metabolic defects known to cause cardiomyopathy can be detected by blood or urine tests and then confirmed with muscle/skin biopsies. They are a less common cause of cardiomyopathy. Treatment usually involves either providing the missing enzyme (only available in research trials for very few conditions) or the missing protein like carnitine or coenzyme Q10. In addition, the patient needs to avoid long periods of starvation, and diet needs to be modified to a more suitable diet depending on the type of metabolic defect. These interventions often help slow down the disease progression in some of these metabolic defects. For most others, there is still no treatment.

Question: My son is one year old and was diagnosed with dilated cardiomyopathy (DCM) at birth. He has done very well over this year. He is currently on captopril, CoQ10, and levocarnitine. He has never been confirmed to have a mitochondrial disorder. A muscle biopsy came back negative. Should he be on carnitine or coenzyme Q10 if there is no confirmed mitochondrial cause? Our cardiologist's view is there is not enough research to guarantee this cocktail can improve function, but since he is improving it would not hurt to keep him on the two. What is your view?

Answer: In general, I and our metabolic team do not give supplements to patients with cardiomyopathy unless there is a clear indication to do so. Having said that,

it is possible to have a metabolic/mitochondrial defect with a negative muscle and skin biopsy since the disease may predominantly affect the heart in the rare patient. A cardiac biopsy may help to diagnose this but that comes with some risk. Some doctors may choose to give these supplements on the basis of clinical suspicion alone. In general though, we do not advocate routine supplements because of questionable benefit since the majority of isolated cardiomyopathy cases (i.e. those not involving other organ systems) are unlikely to be due to mitochondrial defects.

Question: I was wondering if you could tell me about diastolic dysfunction. What are the problems related to it? Also what are the typical heart pressures to get the diagnosis of restrictive cardiomyopathy (RCM)? Are there any new treatments related to RCM?

Answer: Diastolic dysfunction means that the ventricle does not relax well such that it is stiffer than normal. This interferes with normal filling of the ventricle during diastole (i.e. relaxation phase). If this becomes significant, then blood can back up in the lungs and cause difficulty breathing (i.e. pulmonary edema). If the ventricles get very stiff, they may be unable to push sufficient blood to the body to maintain blood pressure and blood flow to vital organs. There is no absolute cutoff for the diagnosis of RCM. Normal filling pressures in the left ventricle are 5-8 mmHg. In RCM, these pressures are generally higher than 15 mmHg, often much higher. There are no new treatments for RCM. One of the mechanisms for diastolic dysfunction is fibrosis or scarring within the ventricle. New research is therefore trying to understand the process of fibrosis and trying to find ways to prevent or reduce fibrosis. This research however is at a basic research level.

Question: What is the latest news about left ventricular non-compaction cardiomyopathy (LVNC)?

Answer: Regarding LVNC, there are no new therapies. There is more research going on into how to diagnose them better on echocardiography and on genes that cause LVNC. Recent data suggest that some of the same genes that cause hypertrophic cardiomyopathy (HCM) can also cause LVNC so that one should also test for the HCM gene panel in LVNC cases.

There are some interesting data from the Pediatric Cardiomyopathy Registry (PCMR) as well as from a European registry that were presented at the American Heart Association meeting in November 2009. They reported that children with LVNC whose heart function was normal did very well during a five-year follow-up with barely 2% death rate or transplant rate. As patients grow into adulthood and/or heart function decreases, the risk increases.

Question: I would like to know the latest thinking on implantable cardioverter

difibrillators (ICD) and risk factors for children with hypertrophic cardiomyopathy (HCM). For example, is a septum greater than 3.0 considered a huge risk factor for kids if there are no others? Are ICDs recommended for children with one risk factor? It seems like Canada is a bit less enthusiastic about ICD recommendations for kids. Is this true?

Answer: This is an important question but unfortunately there is still no consensus about indications for ICDs in children with HCM. The strongest indications are a strong family history of sudden death or sudden cardiac events. Another strong indication is a previous near death episode, fainting episode or episode of resuscitated arrest or arrhythmia. The other indications are softer indications like brief arrhythmia episodes that are self limited, inability to raise heart rate or blood pressure appropriately during exercise, or a very thick septum. At this time, a very thick septum alone is not considered an absolute indication for ICD by most.

Question: My daughter had to be deprived of food for 2.5 weeks due to complications after a g-tube insertion. She has since shown increasing signs and symptoms of heart failure. She suffers from dilated cardiomyopathy (DCM)/ left ventricular non-compaction cardiomyopathy (LVNC). Do you think that the prolonged length of time without food could have affected her heart? She was being maintained with IV fluids for a week followed by IV nutrition.

Answer: The worsening of heart failure during IV nutrition is likely related to some fluid overload. This sometimes occurs because IV fluids are directly injected into the circulation as opposed to being absorbed from the gut. IV nutrition does provide good calories and nutrients and should not cause nutrient deficiencies or directly affect the heart beyond the secondary effect on fluid status.

Question: What are some things to try with older children who have become intolerant of raw milk products. My son is 13 and the nutritionist has given us Vital Jr. and EleCare. I have not been able to talk him into trying it. Everything that he has tried so far has either caused him to become violently ill or tasted horrible. He had a horrible episode of illness yesterday. He could not hold down water, and phenergan was not working either.

Answer: After transplant, some of the medications in particular, MMF or cellcept, can cause significant gastrointesinal (GI) problems in some patients. One may need to lower medication dose, give antacids, and if still not tolerated, then switch to another medication.

Question: I have heard that the cause of stomach discomfort and pain for children with cardiomyopathy is due to congestion. As the lung becomes congested and

causes tachypnea, the GI tract also becomes congested and causes discomfort creating a sensation of having gastritis. Have you heard of this?

Answer: I have been following the reports of GI symptoms in many children with cardiomyopathy posted today. The explanations as best as we can tell are due to two reasons. First, when the heart is unable to pump sufficient blood to the organs like the gut, this can cause abdominal pain and feeding intolerance due to gut ischemia. The second is as explained in the question such that when the right ventricle is stiff or weak, blood backs up in the veins which can cause the bowel wall to become swollen or edematous and result in GI symptoms. Treatment that can improve heart failure can relieve the symptoms. Other measures like a different formula may help as well.

The Basics on Health Insurance

Lynda Honberg, M.H.S.A. – May 2010

CAPT Lynda Honberg, M.H.S.A. is a Program Director for the Division of Services for Children with Special Health Needs, Maternal and Child Health Bureau Health Resources and Services Administration. CAPT Honberg has over 30 years experience in the private and public health sectors. In her current position, Ms. Honberg oversees several grant programs focused on improving the system of services for children and youth with special health care needs. In the private sector, Ms. Honberg worked for eight years as the Director of Operations for the George Washington University Health Plan of Washington, D.C. where she was responsible for provider relations, member services and health center operations.

Question: My child was denied coverage for physical therapy. What should I do?

Answer: Even with health care reform, a critical element of health insurance is still “what is in the contract.” Most people have never read their benefit contract or summary of benefits and are then surprised to find a benefit is not covered. So I always tell families to be an "educated consumer" and carefully read your benefit summary.

But what happens when it is not clear whether a benefit is covered and the claim is then denied? Health plans typically determine whether the benefit is within the contract or if it is "medically necessary." Never take no for an answer! All health plans have grievance procedures for members to appeal a decision, and often times it takes more than one appeal. Families need to be a bull in the china closet and be persistent in appealing their decision. When that fails, most states have an external review process. Make sure you get all the information about the appeal process, including timelines from your plan. Ask your child's physician(s) for assistance with "medical" documentation, and keep good notes.

Question: Last Friday I presented at an insurance seminar related to children's special health care and Medicaid in Michigan. The audience was mostly professionals and a few families. The seminar covered topics like transition from private to state insurance, tips for families and also learning that “no” doesn't always mean “no” when asking for help. I really support the idea of family centered care not only in the hospitals but also within the insurance world. My question is, do you see any of this going on nation-wide, and what is your opinion of the value of collaboration with families?

Answer: Glad to hear that you were involved in the insurance seminar. The federal Maternal and Child Health Bureau (MHCB) funds the Michigan Family-to-Family Health Information and Education Center which is family run and helps families navigate the system of care.

The answer to your question about family centered care is YES! MCHB has been promoting this concept for years, especially for children and youth with special health care needs. If you go to the Family Voices website (familyvoices.org), there are several resources you can download that explain the principles of family-centered care. It also provides tips on educating families, youth and providers, and gives health plans as well as tools to assess care within a practice.

Question: With the new health insurance regulations, covered children supposedly can not be denied coverage of pre-existing conditions. Is there anything to prevent insurance companies from either denying coverage to these kids altogether or making it so expensive that it's not feasible? In other words, are there cost controls built into the new rules?

Answer: The regulations will be coming out shortly from the Department of Health and Human Services ensuring that children with pre-existing conditions cannot be either denied coverage outright, as well as ensuring that they are covered for costs associated with their pre-existing coverage condition. For children, the ban on these exclusions will start for plan years beginning after September 23, 2010 (six months after the signing of the law).

With regards to whether insurers can raise premiums, there are legal constraints against raising premiums, but these regulations do not go into effect until January, 2014, which is when the Exchanges kick in. Hopefully, people will be keeping an "eye" open for any abuses that could be reported. All federal regulations are open to a public comment period, so you may want to make a comment when the regulations are issued.

Also, while the Exchanges don't go into effect until 2014, states are being required to run high risk pools. They will go into effect July 1, although some states are opting out. There is no age limit but most likely the benefit packages will be very basic. Still, it is a good option for families to consider if they have no other coverage.

There are also new rules that apply to what is called "the Medical Loss Ratio" - the amount of premium dollars that have to be spent on clinical services and quality improvement activities (as opposed to administrative costs). This rule should help keep insurance costs from increasing and they will have to report this ratio every year.

Question: Do you know whether standards are changing as to whether automated external defibrillators will be covered? We were denied coverage by a major carrier, appealed and were denied again.

Answer: I haven't seen anything specific in the health care reform bill. It would have been impossible for the bill to cover all the types of services and equipment

that consumers need. But I would appeal again, and perhaps go to your state insurance commissioner. You may also try to see if you could "borrow" one, perhaps from a local hospital?

Question: Shortly after my daughter's heart transplant someone at the hospital told me she'd be eligible for Medicare coverage, even after the coverage she now has under her father's insurance policy stops. Now we're at a different hospital where I am told that is incorrect; only kidney transplant recipients have lifetime Medicare coverage. Is that true? Also, what happens when she is no longer eligible for his coverage? Or what if he dies, and her coverage ends? What do people with heart transplants do? What happens when our kids are no longer covered under our insurance plans? I know they would attempt to get coverage through their own employment, but I worry that our kids may have a harder time with employment and/or medical benefits as a result of their medical condition. The cost of the medications is incredibly expensive. Any insight would be appreciated!

Answer: These are questions that are difficult to answer because every state and employer health plan is governed by different regulations. I will try to answer some of your questions, but recommend you first follow up with the benefits counselor in your company. Another source of information is the federally funded Family to Family Health Information Center in your state (F2F HICs). F2F HICs are staffed by families of children and youth with special healthcare needs and help families navigate the system. If you go to the website familyvoices.org, you can get the contact person for the F2F HICs in your state.

I am not an expert in Medicare, but I do know that it rarely covers children except for those with end-stage renal disease (permanent kidney failure requiring dialysis or transplant). There is a small provision that sometimes covers dependents of eligibles under 65 in the case of death, but the rules are complex. I would recommend you contact Medicare directly.

The good news is that with the Health Care Reform, children up to age 26 (who are not covered under their own plan) can now stay with their parent(s) insurance. A link with additional information is at kff.org/healthreform/upload/8065.pdf. In addition, the legislation has also banned denial based on pre-existing coverage. In terms of losing your coverage, you are always eligible for COBRA, although you do have to pay for the premiums.

The new health care reform bill is complicated but promises to make some important changes that are long overdue. One website that you may want to go to is healthreform.gov, which is constantly updated as sections of the Affordable Care Act are reviewed and implemented.

Another useful website is Families USA at familiesusa.org. They have a fact sheet on the new options to insure young adults under the age of 26, which is one of the most important features since this age group has the highest rate of not being insured. As I mentioned before, Family Voices is a family run grass roots organization of families with children and youth with special health care needs. They can provide information on health care reform, as well as links to the Family to Family Health Information Centers.

Thanks again for sharing your lives with me. As a mom of young adults with special health care needs, I have also spent years being frustrated when trying to get approval for my daughter's many surgeries. I have boxes filled with appeal letters, and while I wasn't always successful, I did manage to win a few battles!

Ventricular Assist Devices (VADs)

David Rosenthal, M.D. – September 2010

David Rosenthal is a pediatric cardiologist at Lucile Packard Children"s Hospital at Stanford University and director of the Pediatric Advanced Cardiac Treatment (PACT) program for treatment of children with cardiomyopathy and heart failure. Dr. Rosenthal established the PACT program in 2002, which now includes an outpatient and inpatient program as well as a Hypertrophic Cardiomyopathy (HCM) Center. Within the PACT program is the program for Mechanical Circulatory Support for children with advanced heart failure awaiting transplantation. Dr. Rosenthal's research interests are on the investigation of right ventricular disease, and the application and effects of mechanical circulatory support in children.

Question: Can you give a quick review of the different types of ventricular assist devices (VAD)s?

Answer: VADs, or Ventricular Assist Devices, are artificial heart pumps used to support the heart when medications do not suffice. Some are designed for temporary support and are intended to be used for conditions that are expected to be reversible. This might include damage after heart surgery or myocarditis due to a viral inflammatory state. An example of a temporary VAD is the Impella. This device is placed through an artery and can often be deployed without major surgery. More permanent VADs are used to support the failing heart until a transplant donor can be obtained. These devices are usually used only in the setting of a patient who is already listed for heart transplantation. They allow for a longer period of support than the temporary devices, for months rather than days or weeks. The most widely used of these VADs, at least in children, is the Berlin Heart EXCOR.

Each of these VADs support only the heart and cannot be used when lung function is inadequate. In the event of both heart and lung failure, the support technology today would be ECMO, which is short-term support, but it does supply both complete heart and lung function.

Question: I have two young adult children one with congenital heart disease (21 year old male) and one with restrictive cardiomyopathy (19 year old female). My concern is that I have not heard about devices that assist adults with heart failure while waiting for a transplant. When I was in Washington DC at a conference I met a gentleman that informed me that the Berlin Heart is used on children under a certain weight. I also know that ECMO is used on smaller children as well, but can that support an adult? Are there any VADs in place that can be used on adults, and if so what are they?

Answer: There are many more choices of VADs for adults than there are for children, and the results of their use are also better understood in adults than children. These devices include the Heartmate II, Impella, INCOR, Abiomed, Total Artificial Heart and several others. Some of these are already approved for clinical use and others are still undergoing trials. ECMO is occasionally used for adults, but in many places has been replaced by newer forms of VAD support. However, not all forms of heart failure will respond favorably to VAD support. That depends on the severity of illness, the specific cardiac diagnosis and the reason for being ill at that time (infection as compared to worse heart failure).

Question: I have read that children and adults that are diagnosed with restrictive cardiomyopathy (RCM) and waiting for a heart transplant do not respond well to VADs. Do you have percentages of the overall outcome in patients with RCM responding well to VADs and if there is a specific time frame that they should be listed to have a better outcome than others?

Answer: Patients with RCM are not very well understood due to the rarity of the condition. Most cardiologists believe that VADs are less effective in the treatment of RCM than in dilated cardiomyopathy (DCM), but there is not enough experience to provide firm statistics. There is controversy as to when to list patients with RCM for transplantation, particularly in the teens. This question is tough to answer in an abstract fashion, but it is probably safe to say that hospital treatments that are effective with DCM are much less so for RCM, and listing should be considered earlier for RCM patients for this reason.

Parenting Teens with Cardiomyopathy or a Heart Transplant

Miriam Kaufman, M.D. – November 2010

Miriam Kaufman M.D. F.R.C.P.C. is a pediatrician and public educator at the Hospital for Sick Children in Toronto, Canada. Dr. Kaufman's main clinical interests are teens who have a chronic illness or disability. She works with these teens around issues of transition, coping, adherence, sexuality and reproduction, substance use, and body image. Miriam has written books and articles for teens and their parents and speaks at schools and conferences.

Question: Medication compliance is a common issue for teens with cardiomyopathy or a heart transplant. What are some tips for helping teens take a more active and responsible role in their medications?

Answer: The question of medication adherence is a big one in teens. We want teens to learn to be independent in reliably taking their medications, but this does not mean that we are telling parents not to be involved. It can be very hard, as a parent, to find the balance that lies somewhere between being totally in charge of your children's medication (easier in a way, but then they don't learn anything) and letting them take care of it themselves (can lead to disaster).

The first thing to do is to ask your young person what they feel ready to do, such as get their dosette box (pill organizer) ready each week with or without guidance, take their medications without reminders, call in prescriptions to the pharmacy and/or pick them up at the pharmacy. If they are doing or feel they can do some of these things, then you should just keep an eye on them, maybe check the dosette to make sure it has been filled or check it to see if medications have been taken. If they feel they are not ready to do any of this, you can start by having them do the dosette with you or having them name their pills.

Many things can get in the way of taking medication such as times of change: starting a new school, new additions to the family, parental split ups. These situations can all make it difficult for a teen to take their medications. Chaos and adherence don't go well together. Young people need specific daily events to link their medications to, like brushing their teeth or eating breakfast. If there are no daily routines, adherence is very difficult to achieve. Most teens have multiple electronic gadgets that have alarms that can be set to remind them to take their medications. Side effects, of course, have a huge impact also. If you think your teen is skipping medication to avoid side effects, this should be openly addressed with them and with their physician.

Question: Any suggestions on how an 18 year old would explain his lack of work experience on a job application or during a job interview? My son was diagnosed

with dilated cardiomyopathy (DCM) when he turned 15. It took the next two years for him to gain the stamina to get through the school day and graduate from high school on time. Now he is attending college and applying for jobs. My husband and I seem to differ on what to say. He says to say nothing. I feel it is important to let them know why he wasn't working and going to school as a teen. And now that he is showing signs of improvement he is able to handle school and work together.

Answer: The job interview question is an interesting one. One of the reasons that employers want an answer to this question is that they want to know how this person has learned important skills needed for any job such as being organized, arriving on time, communicating well, etc. Therefore, your son needs to address this when asked about work experience. He can say, “For personal and family reasons, I didn’t work during high school. But I still developed important work skills during this time.” He needs to figure out how he got those skills. That might include any significant contributions he made around the house or interactions with elderly relatives. He definitely should not lie about his situation.

There might be an advantage to telling the truth, that he was quite ill in high school and that because of dealing with the medical system intensely, he developed important skills by learning how to be on time for multiple appointments, managing school assignments while missing classes, being responsible for his medications, etc.

In some places there are government programs for youth with disabilities or chronic health issues that subsidize the young person’s salary for several months. Employers get a good deal because they do not have to pay as much themselves.

Obviously, your son should be applying for entry-level jobs that do not require much previous work experience. He might need to volunteer somewhere or do an unpaid internship to get something on his resume and show his capabilities. If he picks his work strategically, he might even end up with a summer job at the end of it. And of course, he should be looking on campus first where there is less commuting time and they preferentially hire students.

Question: I’m wondering how many pre-teen and pre-20s have problems with being angry and getting involved with drugs such as pain killers or street drugs? How do you help the angry child? Our son has been angry since he was told he had hypertrophic cardiomyopathy (HCM) at eight years old and that he was not allowed to play sports anymore. He is now 25 years old and slightly addicted to pain killers. Now the surgeons will not give him any pain medications after surgery for this reason – just Tylenol. How many others out there or in your study have become angry and self-destructive?

Answer: As you have pointed out, it is only a small minority of kids who do not manage to move out of the angry phase, often because of intervening problems, like his surgeries. I am sure that you have already had therapists involved, and I cannot comment on a specific child. I would try therapy, maybe even a group home or school with a therapeutic mandate.

Question: My teen has attention deficit hyperactivity disorder (ADHD). He is able to comprehend what he is supposed to do, but he is forgetful and very quick to do something else. It is frustrating to keep constantly reminding him of these things. I do explain over and over that it is for him and not me that he needs to do this. Any other tips would be appreciated.

Answer: I would try helping him break things down into smaller chunks that can be done at one time (not expecting a long time). I would help him get organized with a big wall calendar and putting in due dates with self-imposed deadlines leading up to the due dates. Also, pick out what is essential and don't remind him about the inessentials. The problem at this age is that if you have not already started these interventions, he is likely to be pretty resistant. Another thing is to make sure that he is exposed to lots of different things. At some point, something will get him motivated and he will realize that he has to get some help around his ADHD.

Question: My daughter was diagnosed with hypertrophic cardiomyopathy (HCM) at 3 1/2 months. Certainly, this is very different from someone being diagnosed later in life. I know that you specialize in teenagers, but I am wondering if you might have some input about an even later diagnosis. My husband, who is 28, was just diagnosed as being gene positive. We have yet to find out if he is exhibiting the phenotype of the disease. I am extremely concerned about his psychological well being. He has never been a person who gets depressed easily but our daughter's illness has thrown him for a loop. I think he is so overwhelmed that his response is to act like nothing is wrong. He continues to lift heavy weights at the gym and refuses to stop. I don't know what to do.

Answer: A lot of men react with denial to something like this, especially when it is not a diagnosis based on how they are feeling but just a lab result. He also might be feeling guilty for passing the gene on to your daughter. I think that at this point, he is dealing with it as best as he can. He knows that you are there to support him. As there is no sign that he has cardiomyopathy, it is probably unrealistic for you to ask him not to stop going to the gym where he feels in control and healthy. On some level, he knows he is taking a risk. If he has a family doctor that he likes, you could encourage him to talk with him about all of this.

Question: Can you tell us generally how having cardiomyopathy and all that might go with it (intensive medical care, many doctor visits, medications, surgeries, implanted devices, transplant, etc.) affects the emotional development of teenagers? Has research been done on this? Lately I've seen in my 16-year-old a lot of emotional maturity and sensitivity, which was preceded by intense grief, anger and depression right after diagnosis (age 11) along with several years of resistance to medical treatment. Age of diagnosis might make a difference in this, but I would like to know what does research have to say about the emotional and psychological development of teens with chronic illness? Also, are there differences in this respect between boys and girls?

Answer: This is pretty much impossible to generalize. It seems to depend on quite complex factors, including how much their life has to change, medication effects, family attitudes about illness, presence or absence of bullying, family breakups, age and function at time of diagnosis, body image issues, predisposition to depression or anxiety, and personality and temperament. Your child's story is one that I hear often, from both boys and girls. Some kids seem to just keep moving along their previous trajectory while others get stuck in the anger and grief part.

Medications and Treatments for Children with Cardiomyopathy and a Heart Transplant

Daphne Hsu, M.D. – December 2010

Dr. Daphne Hsu is the division chief of pediatric cardiology and co-director of the Pediatric Heart Center at the Children's Hospital at Montefiore. Previously she was the director of pediatric heart failure at the Morgan Stanley Children's Hospital of New York. Dr. Hsu has treated more than 600 children with heart failure and more than 250 children undergoing heart transplant. She is also a National Institutes of Health-funded investigator for multi-center clinical trials on pediatric heart disease and is actively involved in the Pediatric Heart Transplant Study Group and the Pediatric Cardiomyopathy Registry.

Question: What should a parent do if a child vomits after taking a medication?

Answer: What to do when a child vomits after taking a medication depends on how soon after taking the medication and whether or not the medication is liquid or pills. In general, if a patient is taking pills and the intact pill is present in the vomit, I would tell a patient to take the dose again. If the pill is not seen, I generally do not repeat the medication. In the case of liquids, it depends on how soon after the medicine the patient vomits. My usual rule is if the child vomits within 15 minutes of taking a liquid medication I have the patient repeat the dose, and if it is within 15 - 30 minutes after the liquid medication I have them repeat 1/2 the dose. After 30 minutes I do not have them repeat the dose. I also advise people to wait about 30 minutes after the vomiting stops before trying to repeat the medicine.

Question: My now 16 year old daughter was born with left ventricular non-compaction. She has had gastrointestinal (GI) problems from birth. Three years ago she had a heart transplant. Her GI problems seem to have worsened. There were 17 hospitalizations, mostly for excruciating abdominal pain, which remained a mystery until a year ago when she had two bowel perforations in three weeks resulting in a colostomy. Pathologists reported that the cause was stercoral perforations. She was the 91st case worldwide and the first child with this condition. Now she gets lots of Miralax everyday. Chronic diarrhea seems to be the only way to prevent blockages and impactions. No one knows why she's chronically constipated. She seemingly has motility problems.

My question is whether you have had any experience with the transplant medications being related to GI problems, such as motility problems, nausea, constipation, reflux or anemia, renal problems or bleeding problems? I should mention that shortly after her transplant she had a bout of rejection, which was treated with massive doses of steroids. I wonder if the steroids play a role in all of this. It became problematic once the doctors began weaning her off of the steroids.

Answer: Many of the heart transplant medications can cause side effects, including GI symptoms (I have usually found them to be diarrhea, not constipation), anemia and renal insufficiency. I have not heard of fecal impaction with the medications. If she has had problems from before the transplant, I'm not sure you could say that the problems she is currently experiencing are from the medications.

Question: My 14 year old has been diagnosed with mild dilated cardiomyopathy since 2007. Last year it had resolved but now it is back. He is also now having coronary spasms. He has been put on vasotec and nitroglycerin for angina (chest pains). He seems to have a small septal defect which I was told harmless. My question is “can this keep changing”? We really thought the DCM had been resolved, and now we are very concerned. He constantly has chest pains, shortness of breath and dizziness. He is also restricted from sports and now full of worries.

Answer: It is unusual for a DCM patient to have angina and need to take nitroglycerin. I would encourage you to take your son back to his pediatric cardiologist and have things checked out. It may be helpful for your son to talk about his concerns with the doctor. You might want to have him write his questions down before the visit and have the doctor answer them during the appointment.

Question: My son was recently transplanted about two years ago when he was seven months old. He is on the usual cocktail of transplant medications. Why do some centers use only one anti-rejection medication and other centers use multiple anti-rejection medications?

Also, my son has been switched from cellcept to imuran and then back to cellcept. From talking to other transplant families, I assume that most everyone is on tacrolimus. What is the difference with anti-rejection drugs like cellcept and imuran. Why would one be preferred to the other?

From researching the long-term effects of these medications and talking with our medical staff, I understand that all of these drugs have to be carefully monitored as they take a toll on the body long-term. If a child is on more than one anti-rejection medication, would it be safe to assume that it is more toxic to the body in the long term? Or do the benefits outweigh the possible future issues.

Answer: You are right to have noticed that there can be variation in the type and amount of immunosuppression given after a heart transplant, and it can be confusing when you are talking to other heart transplant parents. The success of a heart transplant is related to how well the balance is maintained between suppressing the immune system and preventing rejection versus oversuppressing the immune system and having serious infections or cancer develop. The amount

of immunosupppression needed to suppress the immune system “just enough” to prevent rejection and not lead to infection or cancer is different among patients. Some of the things that can influence this balance include: the age of the child, the interaction between the donor and recipient tissue antigens and antibodies, the nutritional status of the child, the strength of the child's immune system, whether or not there are any predisposing factors to infection or cancer, and the overall health of the child.

The immunosuppressive medications that are available to heart transplant patients have overlapping effects on the immune system, similar to the different types of antibiotics one might use to treat an ear infection. The choice of what medication or medications to use in a given child depends on the experience of the transplant cardiologist with particular medications, the response of the child to those medicines (are there side effects or rejection), and what the transplant cardiologist feels is the risk of rejection or infection in a particular patient.

Usually a transplant center has a standard immunosuppression “cocktail” that they like to start patients on and then adjustments are made from that regimen depending on the presence of rejection, infection or side effects. Often there are different combinations of immunosuppressive medications used at the same center based on the risks of infection or rejection in a particular child. For instance, if a child is felt to be at high risk for infection, the immunosuppression may be less intense than the standard and in a case where rejection might be more of a concern, a higher level of immunosuppression would be started.

In response to your question about whether or not cellcept or imuran is preferable, each drug is very effective for preventing rejection and often the choice between them is dictated by the side effects that occur with each one.

You also asked whether or not it is better to be on one medication or multiple immunosuppressive medications. In the early days of transplant, a single medication was used, but to prevent rejection it had to be given at very high doses. This caused a lot of side effects so the idea of combining medications came into being. The different classes of immunosuppressive medications can work together to suppress different parts of the immune system and this allowed a lower dose of each individual medicine to be used, which decreased the side effects that were seen.

Question: Can you talk about the nausea and vomiting that seems to be common in children with cardiomyopathy. My question pertains to gastrointestinal symptoms that are associated with heart failure and possibly from the medications used to treat the disease. Our son, diagnosed with severe dilated cardiomyopathy at four months, is now almost three years old. Prior to his diagnosis and hospitalization

(four months in the ICU due to acute congestive heart failure) he never threw up. Since he started treatment he has been vomiting more often. He now has a g-tube but the vomiting started before that. It got better once we cut out cow's milk but if he has the slightest hint of a cold or congestion, he gets very nauseated and throws up easily. Some days it is for no apparent reason.

We keep trying to figure out what causes it, thinking it might be his medications, the extra work his heart may have to do if he's even a little bit ill, postnasal drip. It is sort of a mystery. I know you aren't a GI specialist, but can you talk about what causes these symptoms for heart failure kids in particular (it didn't start until his treatment for heart failure) and any things you may have seen that help.

Answer: You very nicely outlined the possible causes of nausea and vomiting in a dilated cardiomyopathy (DCM) patient. When I hear about a DCM patient vomiting, I worry that the heart is not pumping enough blood to the GI system. It is important that your cardiologist know that this is happening, so he or she can make sure there are no signs of heart failure. Otherwise, medications can make a child vomit. I am sure that you have figured out a way to combine or separate the medications so that there is less chance that he will throw up. Some children are just kids who vomit when they get a cold; there is not much you can do about it.

Question: We are all concerned about the long-term effects using all the anti-rejection medications. Has any special diet or supplements been shown to reduce potential side effects?

Answer: You are right to be concerned about the long-term effects of the anti-rejection medicines. I don't know of any diet or nutritional supplements that directly counteract the side effects of the medications. The side effects are usually from actions of the medicines at the cellular level, and diet does not change these actions. I would say that it is beneficial for the transplanted children to follow a healthy diet and keep their weight in the normal range by controlling their sugar and fat intake. Although this would not counteract medication side effects, it is important to maintain an ideal body weight and keep the transplanted heart working most efficiently and not create extra demand on the heart from obesity or hypertension.

Feeding Tubes and GI Issues

Elizabeth Gleghorn, M.D. – January 2011

Dr. Elizabeth Gleghorn is the Director of Pediatric Gastroenterology, Hepatology and Nutrition at the Children's Hospital and Research Center in Oakland, CA. She is an expert on ailments such as abdominal pain, liver problems, feeding problems and hepatitis. She is particularly knowledgeable about children who have medical difficulties eating and common pediatric gastrointestinal issues such as reflux, lactose intolerance, inflammatory bowel disease and vomiting.

Question: My son, age 6, has a history of reflux as an infant (on Pepcid, Mylanta) and resolved hypertrophic cardiomyopathy. He has an arrhythmia condition that requires beta-blocker medication. He is also on the autism spectrum with Aspergers.

I have struggled for years with my son's eating/feeding issues. Thankfully he is about 50% for his height/weight, but his diet is severely limited to several foods. He refuses to eat whole groups of foods, from pasta to meat to vegetables. He obviously has food texture issues and is a "stuffer" when he does choose to eat. I am wondering if this is something you see kids outgrow. My current strategy is to use butter, which he loves. He is now willing to take a bite or two of soft veggies such as asparagus and broccoli if it has butter on it.

Answer: I see kids within the autism spectrum sometimes being very picky. They do outgrow it to a certain extent. You can, of course, use any of the bribing strategies that work for him. The trick of using what he likes is perfect, and you are lucky that it does work. Most kids do pretty well on surprisingly limited diets as long as they are not overloaded in salt or fiber. If you are very concerned, keep a record of what he eats and have a pediatric dietitian look at it and brainstorm with you. There is a book you could look for in the library by Ernsperger called *Just Take a Bite*.

Question: I am fairly certain that my son is lactose intolerant. He has refused milk since the age of 2. I know that I am lactose intolerant myself. Again, is this something you see with kids? Any suggestions? My doctor does not seem too concerned since he is "growing," but I am worried about calcium requirements and bone strength.

Answer: If he will not drink milk at all then it is not an issue. You can hide milk powder in many things for a nutrition boost. If you want to know, you can test him; he can have a diagnosis made either by a small bowel biopsy or by a breath test. A big hospital should be able to do either of these. The scope involves anesthesia and half of your day. The breath test involves drinking a clear sweet liquid and

then having breath samples taken from the nose periodically over several hours. You can try to see if he will drink lactaid treated milk, which you can buy or make with OTC enzymes. You can then cook with it, cream soup etc.

Question: My son is four. He is on Enalapril, Lasix (Florosimide), Digoxin and half a baby asprin for his heart. He is also on Pulmicort for his asthma. Do you recommend vitamins for young kids on medication? If not, why not? If yes, what kinds and what brands or types of vitamins do you recommend?

Answer: I think that vitamins cannot hurt and with Lasix you might be driving some things out. Centrum type vitamins are good but any gummy or such vitamin he will take is fine. It is low-level insurance.

Question: Are there particular types of foods which may be good for kids with heart medications to eat? Are there foods I could give my son which would be really helpful to him? I worry about his organs—heart, liver, kidney and bladder —with all the medications he takes.

Answer: I do not think we know the answers to these questions. You can read the interactions and cautions section on the papers that come with your kid's medications. Occasionally they will mention a specific food interaction, which you should heed. If they suggest a possible deficiency, you should ask your doctor because supplements might interfere with other medications. That being said, I think the best you can do is to give your kids fresh, uncomplicated foods. Anything that does not come in a package such as whole fruits and veggies, olive, canola and fish oils, and flax seed oil is good.

Question: My son had a heart transplant at 11 weeks old. He was diagnosed with reflux and asthma as a baby. He is now 13 years old, and in the last three years his health has gone downhill. Three years ago he was diagnosed with eosinophilic esophagitis, eosinophilic gastritis, type 1 diabetes, primary immune deficiency (PID)–hypogammaglobulinemia, kidney disease stage 2, restrictive cardiomyopathy and arthritis. I was told that his immunosuppressant medications caused all the health issues diagnosed during this one-year period.

During the first couple months of last year, he was diagnosed with severe protein losing enteropathy (PLE). He had a severe tricuspid valve leak which was fixed on November 3, 2010. In his abdomen are pleural effusions (excess fluid accumulating in the pleura surrounding the lungs). He has a lot of fluid where it should not be and was in the hospital a long time. He has so much fluid that he looks pregnant but malnourished due to PLE. The heart doctors thought the PID, PLE and fluid problems would get better after surgery but it was made worse.

He has very low albumin and protein levels from PLE, and he is now using some oxygen supplementation during the day due to the volume of fluid pushing up on his diaphragm. He was getting Vital Jr. and Beneprotein to help with malnourishment, but the fluid that was draining into his abdomen and around his lungs is now chyle (lymphatic fluid). He has been on a 50 gram low fat diet since they found the chyle about 6 weeks ago. They have him on Diuril and Lasix. He went into respiratory distress and was flown to the hospital a couple weeks ago due to dehydration from a sudden onset of a stomach virus.

Do you of know anything else that can help get more calories in him? The heart doctors are going to give him a couple more weeks to recover and start increasing his diuretics. The GI doctor that we did have was great, but he has recently moved to another hospital and state. His pulmonologist is trying to get my son in with one who specializes with the malnourishment part.

Answer: This is an enormous and very difficult problem. The protein losing enteropathy and the chyle can be very hard to fix. Until they are fixed you have very few food options. You should see a pediatric cardiology nutrition specialist. MCT oils taste nasty but can add calories. If your child is losing a dangerous amount of weight, your doctor may suggest intravenous nutrition.

Question: My son was diagnosed with hypertrophic cardiomyopathy at 3 months. He had severe congestion and a cough for a lot of his life. We switched him to soy formula because I had been on it as a child and it helped with my congestion. It did seem to help him in a lot of ways. He was put on Propranolol at his diagnosis. The dose was increased in August 2009. He started having a lot of trouble sleeping and would do back bends at night screaming. His cardiac nurse said it was probably reflux. He was put on Prevacid and it helped.

In May we moved and had to switch pediatricians. The new pediatrician suggested taking him off Prevacid. I did and he seemed to do fine. However, he has recently been waking up crying at night and doing the back bending thing again. Could this be reflux? What tests should be run to know if it is reflux? Also, it seems that a lot of heart babies take Prevacid or Pepcid. Is reflux a common problem with heart patients? Could it be caused by the medicines they take for their heart?

Answer: It is hard to diagnose reflux. We can do a pH probe or an impedance study. These involve putting a sensor in his throat (usually a tube in his nose like an NG tube) and leaving it in for a day. With this we can count how much acid comes up and compare to normal. If the amount of time that he is exposed to acid is long, it is possible that he has pain because of it.

We can do an endoscopy, looking down in the throat for damage. This study can be negative in people who still have acid related pain so it is not that good to rule

out acid related pain. However it can rule out another disease that can cause the same symptoms, such as eosinophilic esophagitis (inflammation from allergy to food or inhaled allergens). The upper GI series is not so useful; it only tells you if his anatomy is normal. In most kids the anatomy is normal.

A trial of medication is often used. It is not foolproof; there is placebo effect and there is the child just happening to get better when you started the medication. Sometimes when you take them off the medication they are well for a while and then the acid damage builds up and they experience pain again. You can try the Prevacid again or, if he is old enough, try liquid Maalox or Mylanta to see if it helps. You can also try the less strong drugs like Zantac although the liquid prep is nasty. I do think that a lot of kids with heart disease have gastroesophageal reflux (GER). But as you can tell, being sure is difficult. Nonetheless, I think there is an overall tendency for things to move more slowly in heart kids and that can lead to GER.

Question: I have a question less about disease and more about the bigger picture of treating multiple ailments. What is your opinion on how well different pediatric specialists work together and whether you think we need new approaches to cross-specialty care. As an example, one of my children has type 1 diabetes, celiac disease and has tested positive genetically for familial hypertrophic cardiomyopathy (HCM) which has over a 90% rate of penetrance. All three of these conditions can produce GI symptoms that require different responses and yet are very hard to distinguish. We are always waiting for the HCM to express itself. I know others on this listserv deal with similar issues. I find for the most part that our daughter's various specialists deal with the problem by having us make appointments with each one, who then evaluates the symptoms through his or her particular lens. Sometimes they talk to one another, but there does not seem to be a comprehensive model for thinking about how various systems interact within the whole child. I would love to know your thoughts on how specialists work together, whether it works and whether new approaches are being tried out in some pediatric hospitals.

Answer: What you need is a really good general pediatrician who wants to take a lot of time with you and also a bunch of specialist who want to work cooperatively. If they could all agree to email their separate reports or if they all worked in one place where they have a centralized medical record system that would be ideal. Other ways it can work is to have one specialist service such as endocrinology or cardiology that acts as a clearinghouse for all of the subspecialty services. It is hard and depends on the willingness of specialists to get together. I find shared emails are great to just keep everyone informed.

Question: My son drinks a lot during the day. He drinks a variety of things: milk, juices, water and drinkable yogurts. He drinks much more than other kids his age.

For example, he has finished 24 ounces of liquid in the last 4 hours. Is it normal for heart kids or kids on beta-blockers to drink a lot? He has a lot of wet diapers so I am not concerned that he is retaining anything. Could it be due to the medications or just a symptom of the condition? He is not on diuretics but he is on Propranolol.

Answer: I do not know that there is any reason for this. I am guessing but if his overall blood pressure is low his body might interpret this as dry. However this is really reaching. I would ask your heart doctor for a more experienced view on it.

Maintaining a Committed Relationship

Melissa Groman, L.C.S.W. – May 2011

Melissa Groman is a licensed social worker in private practice in New Jersey. As a marriage therapist, she has been helping couples create satisfying, loving relationships for more than 20 years. Her work has focused on the gentle understanding of human relationships and helping people move forward through difficult times.

Question: Parents may find themselves focusing all their energy on caring for a sick child. What are a few tips to help parents move toward caring for themselves and their committed relationship while still caring for a sick child?

Answer: One of the problems with the idea of taking care of ourselves and our relationships is that, though it certainly makes sense, it can feel like another burden, time commitment or an extra item on the "to do" list. It may feel difficult to connect taking care of yourself, your relationships and your child.

I would be glad to put out a few ideas about how to do it, but I would like to hear from you about what you do to take care of yourselves and your relationships, as well as what your thoughts are about what gets in the way.

Questions: My husband and I did seek counseling ten months ago and went for about six months. It honestly saved our marriage. We still struggle with our differences in "medical views" and opinions, but we are learning to listen to each other even though we may not agree.

Our other saving grace has been Sunday date nights. We were going every week for a long time. Sometimes it was more of a chore, but we went and loved the connection without kids. We left after our child with cardiomyopathy went to bed and were only out for a few hours, so it was very guilt-free for both of us.

Answer: It never ceases to amaze me how important listening is, as you point out. It is not always easy, especially with so many heavy feelings surrounding your child(ren) and their care on top of all the normal relationship issues.

And taking time out, even if it's just a few moments here and there to be with each other, usually goes a long way. It can be reaffirming that there is a connection with each other, even when things are strained, and a connection with ourselves when we take time out alone. I know some folks like to poke fun at "date night" but I am big believer in it when you can swing it.

Question: Getting time alone as a couple is the hardest and most important thing for us. We have managed that by making it a priority to train our loved ones

on how to care for him and let go of the fact that it might not get done perfectly while we are gone.

We have trained about 5 different sets of friends and babysitters on how to do our son's tube feedings, how to give his medicationss (we always draw them and give them to him). We have had 3 overnights away from him. Those overnights have recharged us more than anything. It can sometimes be a logistical nightmare (our last overnight involved coordinating with 10 people) but it is totally worth it for us.

One of the many ways that we are blessed is that my mom comes over every Thursday night to watch our son so we can get out for a weekly date night. Sometimes it is just a movie down the street or we sit in our car and talk after dinner out, but it is like depositing coins in the bank. Before our son started school, my mom stayed over so my partner could get a few hours off in the morning which helped our relationship because she was not completely fried by the weekend and she had something to look forward to.

Figuring out alone, together as a couple, together as a family and one-on-one time with our son is really hard sometimes. I often feel like I don't do any of it very well, but I try to remember that things for me or things that my partner does for herself are like oxygen.

Answer: I love the reference to oxygen that you make! It reminds me of the airplane analogy... that when you fly, the flight attendants make the speech at the beginning of the flight reminding passengers that if they are traveling with small children and the cabin pressure should change, oxygen masks will fall. Also, that it is important for parents to secure their own masks first (to make sure they can breathe) and then put on their children's masks, so that they can continue to be available to help them as needed.

Question: I was wondering how appropriate it is for one parent to be extremely involved in the medical care (doctor's visits, research, feedings, medications, etc.) and for the other not to be. My husband is extremely intelligent and usually does massive amounts of research on illnesses for other family members but for our 2 1/2 year old daughter it is just too tender for him to have to contemplate what could potentially happen. He seems to appreciate that I handle the "heavy" subjects. He loves playing and enjoying her, and he is a great father. I am happy with this arrangement but wonder if it is healthy to have such a stark division between parental responsibilities in a chronic illness case.

Answer: What I think happens when one parent seems to shoulder more or all of the medical responsibilities is that possible resentment can build, or feelings of isolation and emotional disconnect can heighten. This happens in households

where there is not a chronically ill child as well, and it tends to show up around household chores, work, etc. While very important, it certainly does not have the seriousness and implications of having and loving a chronically ill child.

What tends to be a good direction in terms of dealing with things, is to work towards a good and tender discussion of each partner's feelings, needs and emotional barometer, and tolerance for difficult feelings. There is not really a "right" or "wrong" nor does it mean that we are stuck with "what is". Often what is needed is continued emotional connection and mutual support, validation and appreciation, and sometimes a different division of tasks.

Question: One of the hardest things is when two parents have very different ways of dealing with difficult medical realities. It seems as if there's always one parent who is more worried and protective and the other who is more loose or in denial. In my case there are three parents–I'm separated from the child's other parent and they have had a very involved stepmom for the past six years. You would think that the fact that we are all women would make us all anxious, over-involved worriers, but actually we are all quite different and struggle to find ways to accept those differences without being critical or defensive. I am probably the biggest worrier, and I have found it best to seek support from others who understand this—such as on this forum—rather than spilling my worry out onto the rest of the family. This can be hard when I mention fears about some heart-risky activity one of the kids is doing and my partner says, "Really?" as if it never even crossed her mind. On the other hand I think it's great for the kids to have her stability and worry-free attitude.

Answer: I have found, as you mention, that being amongst those or even finding one person who can really understand your feelings and your take on things and offer true support goes a long way! I think that being able to "vent" or talk or just "be" in a place with someone who offers that is one of the best forms of self-care there is.

Acceptance without criticism is quite the challenge, especially when it comes to dealing with everyone's different emotional barometers and all the different feelings that understandably comes up. Things can get tricky when we wish to be on the same page with our partner. That may not come easily sometimes. It's amazing how kids can benefit from the diversity instead of being confused by it. What a strong message you have!

Question: I am a mother of two young adults with heart issues–my son is a congenital heart defect patient and my daughter has restrictive cardiomyopathy. We, as parents, find plenty of time to spend together because our children are older. My question is when is it time to stop being so involved in their medical care.

My son is almost 22 and wants nothing to do with me going to his appointments. Tomorrow is his first time going alone, and I am a wreck. My daughter, however, is 20 and wants me there for everything (even holter tests). My husband and I want to teach them to take care of themselves and maneuver in the medical world, but we are worried that we will miss key information that could change their care or save their lives.

I also worry that we focus so much on the child with a life threatening disease that my other healthy child may feel left out. Although we spend a lot of time with him and focus quite a bit on his sports, I worry that I don't always have enough time for all their needs. How do you balance hospital stays, appointments, day to day, and sports with the healthy child, husband, and still make time for yourself? It is not so much time spent with my husband as it is time spent alone. That rarely happens without feeling guilty.

Answer: I wish there were simple answers to your very important questions. It seems to me that all of you on the listserv are a much better resource for each other than I may be on many of your questions. That being said, it seems that like with all parenting issues, especially with older children, it is usually a good idea to consult with the kids themselves and to learn about, understand and talk with them about what their ideas, wishes and feelings are. When possible, honor them. I always encourage parents to help kids feel that they can talk to them about anything, even things that may be difficult for parents to hear.

Of course, different children will have different needs and wishes, which have to be negotiated individually. It seems to me that there is no general right or wrong. Certainly having less access to the medical information that is crucial is frightening. I tend to think that keeping open communication will go a long way toward finding the right balance between involvement and independence.

As far as guilty feelings go about spending time alone or on yourself, it may help to think of the airplane analogy from one of the earlier posts.

Often parents with chronically ill children underestimate how good their parenting of their children really is and neglect to credit themselves for all the things they are doing right, even the basics. The focus can sometimes be on what does not work or what does not feel good enough, when there is so much good being done and plenty of “good enough” that count as well.

Genetics Related to Cardiomyopathy

Wendy Chung, M.D., Ph.D. – September 2011

Dr. Wendy Chung is Director of Clinical Genetics at Columbia University Medical Center. She has a special interest in congenital heart disease and her laboratory research is focused on identifying new genes for susceptibility to complex genetic traits and developing better diagnostic tools for genetic evaluation of cardiomyopathy.

Question: You last served as a cyberguest on this topic in 2007. How much has the world of genetics related to cardiomyopathy changed in the last few years? Is progress being made?

Answer: The world of genetics has changed remarkably. There is now genetic testing for all the cardiomyopathies, and it is more extensive than was available in 2007. Because of the improved yield of testing, treatment for some forms, and implications for arrhythmias for others, I think all children with dilated, hypertrophic, restrictive, and non-compaction cardiomyopathy should have a genetic test as part of their evaluation.

We know much more about the underlying genetic basis for both isolated, syndrome and metabolic causes of cardiomyopathy. Knowing that information is important to ensure that children get the right care and that families are able to ensure that other family member at risk can be identified and treated early. There is now good treatment available for hypertrophic cardiomyopathy associated with Pompe disease that was not available in 2007.

Questions: Is any thought or research being given to genes being responsible for preventing or blocking arrhythmias?

My father survuved a sudden cardiac arrest and now has an ICD and no evidence of any cardiomyopathy. My son has Wolff-Parkinson-White Syndrome and was diagnosed with mild hypertrophic cardiomyopathy as an infant (now resolved). I am the missing link with no evidence of arrhythmias, but I was born with an atrial septal defect and patent foramen ovale.

It is also concerning to hear of kids with very large hearts being absolutely fine and kids with mild HCM having fatal arrhythmias.

Answer: You raise an interesting point. There are certain genetic forms of cardiomyopathy that are associated with more arrhythmias (LMNA and TNNT2 mutations for instance) even without the hearts being very thick or very large. However even within families with a single genetic form of cardiomyopathy, there can be quite a bit of variability between family members for many aspects of the

disease including sudden cardiac arrest and arrhythmias. This could be due to other interacting genes, gender (males tend to have more problems than females) and other exposures over a lifetime. There is research being done on identifying those other genes and factors that may change the risk of arrhythmia, but the number of patients that will be required to answer that question will be enormous, so the question will not likely be answered quickly.

Question: Can you outline the different genetic tests available to diagnose cardiomyopathy? Also, any thoughts or advice about how to get genetic testing covered by insurance?

Our son's cardiomyopathy is idiopathic at this point after a very basic round of genetic testing while he was in the ICU. We can not decide if we want to do more genetic testing. He is the only one in the family that has cardiomyopathy and we do not plan to have more kids. I wonder if the testing would be worthwhile to possibly identify other family members who may be at risk, if a mutation is found. Do you have thoughts about how to decide whether or not to pursue more testing?

Answer: My personal practice is to genetically evaluate all kids with cardiomyopathy. The specific tests I perform depend on the type of cardiomyopathy (hypertrophic, dilated, restrictive, or non-compaction), whether or not the cardiomyopathy is isolated or not isolated, and what the other features are. In the last two years I evaluated approximately 200 kids with all types of cardiomyopathy combined and identified a genetic cause in 49%.

These days testing is covered by insurance. As long as the child has either private insurance or Medicaid, there is a lab that will do testing. GeneDx has a very good program to limit the out of pocket cost to families to no more than $100 if they have private insurance. Therefore testing is now accessible to the majority of families. It is very important to evaluate kids for treatable metabolic causes of cardiomyopathy, and these are often not recognized or tested by most cardiologists. In the future, treatments are likely to be available for very specific types of cardiomyopathy or particular types of mutations, and I think it is good for patients to be prepared by knowing their child's genetic subtype so they will be prepared when that opportunity arises. Finally, some types of cardiomyopathies run in families and can be associated with a risk of sudden death. Genetic testing provides a valuable means of identifying other family members at risk and treating the condition early to prevent manifestations like sudden death.

Question: Is there value in genetic testing after a child has had a heart transplant? Is the value mostly about understanding implications for their family members as well as their future children? I have always thought we should consider genetic testing, but I also try to be sensitive to the fact that my son has plenty of biopsies,

blood tests, other tests and medical appointments. I would want to be sure there was a purpose for doing it. He had idiopathic dilated cardiomyopathy.

Answer: The value in genetic testing after a heart transplant it to ensure there are no extra-cardiac manifestations, identify other family members who are at risk, and provide information for future children. Blood for the genetic testing can be collected at the time of another blood draw to minimize the burden for your son.

Question: I have a 16 year old son with coarctation of the aorta and had a ventricular septal defect which closed on its own when he was about 5 years old. We had a baby girl who passed away 9 years ago of idiopathic dilated cardiomyopathy (DCM). She was 9 months old. Biopsies were done on her and our doctor reported no genetic concerns for our son. Do you have any recommendations as far as genetic testing?

Answer: If there is no material for genetic testing remaining for your daughter, the genetic testing will be difficult. Your son probably has had and will continue to have echocardiograms that will let you know if there is any evidence of dilation or decreased heart function. You could get genetic testing for DCM for your son, but a negative genetic test will not rule out all risk for DCM in the future for him.

Question: It has been a couple years now since my family has undergone genetic testing and revealed a mutation in the MYBPC3 gene as a cause for my son's hypertrophic cardiomyopathy (HCM). I have two questions. My first question is that sixteen years ago my cousin died while awaiting a heart transplant. They listed dilated cardiomyopathy (DCM) due to a virus on his autopsy report. His heart was 2 1/2 times the normal size, plus he had severe left ventricular hypertrophy and moderate right ventricular hypertrophy. I am gene positive, my only sister is gene positive, my aunt (my cousin's mother) tested positive, my dad was positive, and his mother was positive (who lived to be 101). A cousin of my dad's aunt also tested positive. Is the genetic testing alone confirmation that my cousin had end-stage HCM and not DCM? Or is there a possibility that this gene mutation can cause both HCM and DCM? Either way, we are assuming my cousin's children (who were 5 and 6 months at the time of his death) need to be screened. I have encouraged the genetic testing with no luck.

My second question is that we are military and moved around every few years. We have had the opportunity to see some of the top specialists since my son's diagnosis. Both of our specialists, although glad we have done genetic testing, say that knowing his gene mutation offers us no insight into prognosis or treatment. I am wondering if this is entirely true? It is hard to go on-line and search for articles specifically related to a gene mutation, because it is over most of our heads. Is there a website on genetics that is easy to comprehend and navigate to research

that is open to the general public and not just the medical field?

Answer: For the first part, the MYBPC3 mutation likely started out as HCM in your cousin and with time progressed to a dilated heart. The same gene and same mutation can also cause HCM and DCM (with the DCM progressing through a stage of HCM), and that sounds like it is the case in your family as well. Your cousin's children should be screened as you have tried to suggest.

The actual genetic mutation in your case will not help your cardiologists manage your son going forward. MYBPC3 mutations are not specific in being associated with extra cardiac problems or differential risk of sudden death. There are other genes for which knowing there is a mutation in that gene does help tremendously with management. You do not know before you do testing if you will get a MYBPC3 result or one of the other genes. In families with MYBPC3 mutations, knowing the mutation in the family is most helpful for identifying who in the family is at risk and who is not.

Question: My daughters (ages 7 and 8) and I all have a mutation on the MYH7 gene. My 7 year old daughter was diagnosed with mild dilated cardiomyopathy (DCM) as an infant. Her heart resolved to a normal size by the time she was 1 year old and she has had a normal heart size and function ever since. My 8 year old daughter was diagnosed with severe DCM as an infant and had a transplant in 2010. I have been checked, have a normal heart size and function, and have never had any heart problems, although I have been told that I am at “high risk” for developing DCM sometime during my life.

It appears to me that the technology for testing has improved in that we can identify the causes for some of these cases, but making a prognosis based on this information can be difficult. Any other insight you can provide on the matter would be greatly appreciated.

Answer: Yes, the technology has improved to find the main genetic predisposition to cardiomyopathy in many families, but we still don’t understand yet why some members of families have early and severe disease and others never have any heart problems at all. This is a very important issue. In some cases, there is a mutation in a second gene within the family we can identify that makes the condition worse. There may be differences between males and females in some families. In some individuals there may also be non-genetic factors that contribute that we have not identified yet. The biggest problem is that it will take hundreds of families with each genetic form of cardiomyopathy to answer these questions.

Question: My son became ill suddenly and was diagnosed with dilated cardiomyopathy (DCM). Three months later he had a heart transplant at the age of 13.

The doctors said they did not know if it was genetic but suspected based on some non-compaction in the left ventricle. They did a couple of genetic tests (a carnitine enzyme) but then said it would be costly to do other tests and the yield would be low. My two older children are healthy. They had normal screening echocardiograms. I have mitral valve prolapse (asymptomatic) and a left bundle branch block (LBBB) and so does my husband (LBBB). However we are all healthy and there is no significant cardiac disease in the family. Should we still pursue more testing for the children or their children's benefit?

Answer: It is very possible that your son's left ventricular non-compaction has an underlying genetic basis. Since he has had a heart transplant already and he does not have any non-cardiac problems, the main reason for testing would be to provide reassurance for your older children and to one day provide your younger son with information he needs to make informed decisions about having kids and how to care for his kids. If anyone in the family gets testing, it should be your youngest child first. If and only if a mutation is identified in him, then your other kids can have testing to determine if they are at increased risk for DCM.

Children's Cardiomyopathy Foundation
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