Children's Cardiomyopathy Foundation™

Answers from the Experts: CCF Cyberguests

A Compilation of Q & A Pediatric Cardiomyopathy Listserv Sessions 2006-2007

Q&A

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A compilation of Q & A listserv sessions on pediatric cardiomyopathy

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

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

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Special Education and Other School Related Issues/Regulations

Dawn McDonald, M.A. — December 2006

Ms. Dawn McDonald is a N.J. certified school psychologist. She has a masters in counseling and is a doctoral candidate at Philadelphia College of Osteopathic Medicine in the school psychology program. She recently completed a one year APA clinical internship in psychology at Jersey Shore University Medical Center.

Her dissertation is on abuse-related traumatic stress on children. She is a school nurse and generally works with kids with learning problems, autism, developmental delay, behavior problems and some with health issues who require special educational programming. Dawn has a son with DCM and is a registered CCF family member.

Question: What is an "IEP" and should my child have one?

Answer: I am glad to be able to share some thoughts with you about special education services for children. It was asked what is an IEP and how does one know if their child needs one? First, an IEP refers to an Individual Education Plan and outlines what specific services, educational modifications and accommodations a child who is eligible for special education must receive. It is a legal document and in NJ, where I practice, the school district is mandated to provide the services outlined in the IEP. The parent, school personnel and any other professionals deemed necessary should be active participants in the creation of the child's educational plan.

To have an IEP, your child must first be eligible for special education and related services. In N.J., this means the child will usually be evaluated by a Child Study Team and the results of the evaluation will determine eligibility. A child can be deemed eligible for services by meeting the criteria of one or more classification categories.

Question: How will a parent know if his/her child needs "special education" or an evaluation?

Answer: A parent has the right to request an evaluation by the Child Study Team of a child who attends a public school. A school may deem an evaluation warranted or not warranted based on the individual circumstances. Many schools require documentation of a variety of interventions attempted prior to evaluation. A parent may wish to consider evaluation if their child lags behind peers in achieving developmental milestones or is having difficulty academically, and this difficulty is not able to be remediated by standard interventions in the classroom. In addition, if a child has serious health concerns, including attention difficulties, that have a significant impact on academic performance as a result of

associated limitations in strength, vitality or alertness, a parent may be warranted in seeking an evaluation. If speech, fine motor skills (including handwriting difficulties) or gross motor skills delays are present, these services may also be available to parents.

If a child requires modification of curriculum or special accommodations to succeed in a particular educational setting, a parent may wish to consider requesting evaluation to determine eligibility for services to be written into an IEP.

Question: What are some of the psychological stresses that parents should be aware of when their children with cardiomyopathy enter school age years?

Answer: As for what psychological stressors that children with CM may face when they become school age, I think that this question is broad and the answer can be as varied and diverse as our children with CM and their individual circumstances and temperaments are. First, children with CM are in many ways like all other children. Our children will likely experience the typical stressors associated with their particular age and developmental levels. The unique issues associated with chronic illness or a history of a traumatic hospitalization or medical intervention may exacerbate and add to some of the typical stressors. For example, our preschool and kindergarten age children, like all children, may experience separation anxiety. It is important that parents and school personnel be aware of and sensitive to youngsters with CM who may have had histories of frightening hospitalizations, etc. This can impact separation fears. Also, younger children who have been ill may or may not have had adequate socialization with peers, and this may be an area that needs to be addressed. Older children with CM may have to deal with the psychological impact of having a chronic illness, disruptions to their schedule, repeated medical procedures, limitations on activities and fears and anxieties about their future. There are a number of issues that could potentially arise. In general, though, I think it is important that children be educated about their condition in an age appropriate way and encouraged to express their feelings and concerns. School personnel should also be educated and encouraged to express concerns and ask questions. In general, if a child with CM, of any age, exhibits any behaviors (excessively withdrawn or sad, acting out, etc) of concern that impact his or her academic or social development, parents should seek out professionals who are able to work with the child on the specific behaviors, feelings or situations that are troublesome to that child. In the school setting, there are counselors and psychologists who may be helpful in addressing these issues on a superficial level. Often though, a therapist outside of the academic setting who is able to collaborate or consult with school personnel may be able to address issues in greater depth.

Question: My son, Evan, is 4.5 years old and has an Individual Education Plan (IEP). Evan is homebound due to his complex medical issues and gets two one-

hour teacher sessions a week. That seems to be a little light on the time spent. Dawn, is there a way to get more hours of homebound services?

Answer: This is an issue that you can and certainly should negotiate with your school district. Your child is entitled to a free and appropriate public education (FAPE), and the district is obligated to meet your child's educational needs, regardless of disability. Since I don't know the particulars of your child's IEP or district's policy on homebound instruction, it will be hard for me to speak about whether or not your child is receiving the minimum services required. I can say, however, that if you feel your child can tolerate longer or more frequent instruction, you should make your concerns known. The first step would be to discuss your concerns with your child's case manager. The IEP usually indicates who this person is and a contact number. You should also request a meeting in writing and copy the supervisor of special services in your district. If you are able to get a meeting, it is important for you to be prepared to discuss what services you would like and your rationale. In addition, be prepared by reviewing the parents' rights in special education. (In N.J., we call it the PRISE and must supply it to all parents whose children receive special services). Be informed, and be persistent. Also, you might want to consider being proactive in setting up a meeting to discuss Evan's reentry into school when you feel he is close to ready, in order to smooth out any potential bumps in the process. It is also a good way to educate the school staff on Evan's condition and needs. In addition, you can begin to discuss a suitable placement and educational plan for Evan for when he is able to return. Keep in mind that the IEP team, at least in N.J., can offer a modified school day for children who cannot tolerate a full day. Good Luck!

Question: We are fortunate in that my son is healthy with his HCM. He has never been hospitalized, and we found out because my ex-husband was diagnosed. My son has an IEP for mild learning differences that he is outgrowing. I have noted his HCM on the IEP, but can I push the issue and get a more stringent IEP regarding his heart? All his teachers are aware, but there is no AED on campus. They don't have a plan if he goes down. The closest fire department knows to bring an AED, but can I do more with an IEP?

Answer: It's great to hear that your son is doing well. In reference to your question, I think it is an important one and should definitely be addressed with the school. Whether or not you would be able to get this written into the IEP seems to me to be a matter of interpretation and how liberally your district is willing to interpret this. The IEP is designed to help children with disabilities gain access to a free appropriate public education. In doing this, children with certain disabilities are entitled to certain services and assistive technology devices that increase, MAINTAIN or improve their functional capacity to participate in their educational program. Whether or not this falls into an "assistive technology" category is, I think, subject to interpretation and really not the standard interpretation of what

assistive technology is. However, public schools also must be appropriately prepared for emergencies and able to show that they act with prudence to ensure the safety of students. I'm not sure if the district would be obligated to provide a device like this on campus, but I certainly would address the issue with both Special Services and the IEP team and the building administrator. Unfortunately, services and devices that are supplied to students vary greatly among districts and may also be influenced by budgeting issues, resources of the district, potential legal liability and persistence and insistence of parents. At the very least, you should initiate a serious discussion about this, including the school nurse and building administrators, and make sure an appropriate emergency plan is in place for your son. It doesn't ever hurt to ask for what you want, and many parents ask more than once.

Question: My son is 2.5, and we are in the daycare/ homecare/ preschool phase of schooling. Are there grounds for a private school or daycare center to refuse to take care of a child for medical reasons? I assume that public schools cannot refuse anyone.

Also, how should we present our children's problems? I am giving the school personnel medical info they have never been exposed to before, I try to keep it very simple and try to be as positive as possible. I say every single good thing that any doctor has ever told us. However, I worry that this is not presenting the situation accurately.

Answer: Children between the ages of birth and three years old who are identified by the school district as being eligible for special education and related services may be eligible for early intervention services. In this case, your school district/case manager would facilitate placement in an appropriate educational program through contractual agreement with an early intervention program provider. Often these are private daycares and preschools. By having a case manager work with you to get an appropriate placement, you can alleviate some of the uncertainty of how to present your child's challenges in order for him to be accepted to a program. They also can help you with any concerns that come up over the course of the year. Private schools considering admission for non classified students aren't supposed to base decisions on disability, race, etc. but it clearly could happen, and it is much easier for the private schools to be selective especially if they don't receive federal funding. Luckily, many do, so there is recourse. Even so, if you aren't already involved in an early intervention program in your district, this may be very helpful for you in navigating these issues. It can be very difficult to discuss your child's challenges with the school, especially if these challenges require more intensive care or present as behavioral challenges that may be disruptive in the setting. I think even with a case manager to guide you, it is important that you as a parent communicate clearly to teachers and caregivers. The best thing to do is to be factual, truthful and informative and

present the information to the school in a neutral manner. If you have effective, appropriate strategies that you use at home to head off problem behavior or specific difficulties, it is always a good idea to share this with school personnel. As parents, we need to educate others about our children's disabilities not apologize for them. It is always best, obviously, to use a collaborative approach, and make sure the school understands that you are both on the same page and what is best for the child is what's most important. While I always recommend parents be firm and informed when advocating for their children, the message must be presented in a way that is conducive to cooperation and collaboration among all involved in the child's care.

Question: How do I get a school district/case manager?

Answer: Your child's school district is determined by where you live, and preschool age children who are identified as having special needs are generally entitled to educational services facilitated by the school district, even if they contract out to a daycare provider for these early intervention services. I don't know exactly how the program is implemented in your area, but a good start would be to call the Board of Education and talk to someone in the Special Education Department regarding early intervention and request that your child be evaluated. A standard evaluation may assess your child's cognitive development and learning ability, speech development, emotional and behavioral needs, social interaction and fine and gross motor skills. Many parents of children with CM and other medical problems may have already had some assessments done through medical providers or as a result of hospital referrals. Often these assessments are good information to share with the team assigned to assess your child for special education services.

So your area's Board of Education should be able to direct you to the appropriate persons who can help you get this started. In N.J., early intervention programs are administered by the Department of Health and Senior Services in collaboration with the Departments of Human Services and Education.

Once an evaluation is underway, you will be assigned a case manager. This is typically some member of the evaluation team. Once your child is determined eligible for services, the case manager should be able to help you navigate placement options, meaning which preschools or daycare centers can provide intervention services for your son. Once eligible, an IEP will be developed for your child and you can have input as to what accommodations and services you would like your child to have.

Question: I have a 2-year-old daughter who was diagnosed with DCM/LVNC a year ago. She's doing quite well clinically and has shown quite a bit of improvement since diagnosis. I work full-time and she's still in a nanny-share type

arrangement. However we are considering putting her in some sort of Montessori daycare as she gets older. I'm uncertain as to whether there are any regulations and whether they can accept/reject her admission based on her heart condition? She does not seem to have any sort of learning difficulties and, if anything, seems quite precocious for her age. The only issue we've had with her in the past and are still having is acceptance of "age-appropriate" foods. She still shies away from table foods and eating on her own. I'm worried about her eating outside of home but also have a feeling she might learn to eat better by watching other kids. My question is regarding how private preschools/home-run preschools accept kids with disabilities. I guess I hesitate to use the word disability since she seems so normal to us!

Answer: As a mom of a now 5-year-old with DCM who was a very picky eater and seemed to catch every cold virus that came around, I can really relate to your concerns about the germs in the daycare setting as well as your concerns about your child eating enough while away from home. I had all of these concerns, and still do, now that my son is in a Montessori kindergarten. In addition, I usually don't consider or refer to my son as having a "disability" because so far he has not demonstrated any developmental lags due to the CM, and therefore he has not been eligible for any special early intervention services. This may be the case with your daughter and therefore a case manager may not be warranted. It is only if the CM is affecting her development in a way that impacts her education that this becomes an issue. Many children with CM, however, may have related syndromes or developmental lags that warrant a need for special education services. Also, some learning weaknesses may not present themselves until later in childhood when the child is working on developing phonemic awareness and pre-reading or reading skills. So this can initially become an issue with older children as well.

I, personally, have not had a problem with a private Montessori school denying my son as a result of a medical issue. Again, private schools obviously should not discriminate based on these issues; however, I am not completely familiar with the legal parameters if a child is not considered "disabled."

When my son needed more intensive care and had a feeding tube, I chose a private home-based babysitter. Childcare and educational decisions are difficult for all parents, but when your child has had a serious illness, it can be very anxiety-provoking transferring their care to someone else and placing them in situations where they may be vulnerable. I think the best we can do as parents is take reasonable precautions, make our child's needs known to the providers and relax and allow our children to engage in as many "normal" activities as possible and as appropriate according to their condition. Often the doctor has good recommendations in this area as well and can be reassuring.

I don't know if discussing my personal experience helps but my son is still quite underweight and gets tired out fast. Sometimes he does not eat as much as I'd like him to while in school, but the cardiologist says his EF is good for now and he is holding up. Like your daughter, he is, in many ways, indistinguishable from the other kids who do not have CM, so it is easy for me to fall into denial about his health. It's a balancing act between keeping them safe and well and in keeping CM and the anxiety it provokes in parents (and often in children) making an able child "disabled."

Disability is a terribly negative term. Unfortunately, it's the legal language that is used to protect children who require these services. We should not, of course, lose sight of the fact that even children "labeled" in this way are "able" in many, many ways.

Medications-Questions and Common Side Effects

Robert Gajarski, M.D. – January 2007

Dr. Robert Gajarski is the director of the heart failure/transplant program at University of Michigan/Mott Children's Hospital, and he also runs the cardiomyopathy and pulmonary hypertension clinics. He has 15 years of experience in the field and has taken care of transplant patients at both Texas Children's Hospital in Houston and University of Michigan. He is also actively involved in research on heart failure and heart transplant patients.

Question: My 2-year-old son has severe hypertrophic cardiomyopathy and also asthma. He takes a nebulizer with xopenex almost daily. We have been continuing with the nebulizer, and the pediatric cardiologist thinks it's probably ok but she seems a little worried. On the web I found some studies linking heart problems with asthma medication. What are your opinions about this?

Answer: In general, beta-agonists like xopenex, are contraindicated in people with hypertrophic cardiomyopathy especially those with the obstructive form since this medication can aggravate the obstruction and increase cardiac symptoms. Having said that, if the only drug that controls the asthma symptoms is a beta-blocker, then xopenex is the best of the group since it theoretically has the fewest cardiac side-effect (although, in my experience I'm not sure it's that much different from albuterol). There are also a host of other medications that may not have negative cardiac effects that can also be used to better control symptoms long term (e.g. inhaled steroids, leukotriene inhibitors). For acute wheezing events, a beta-blocker may still be used for the short term without serious cardiac effects. Also, beta-blockers as a drug choice to treat hypertrophic cardiomyopathy aggravates wheezing in some kids, increasing the need for drugs like xopenex to control wheezing. In those cases, calcium-channel blockers are a nice alternative. Finally, I would encourage you to ask your pulmonologist to work in concert with your cardiologist so the best treatment plan can be devised to treat the wheezing without making the symptoms from hypertrophic cardiomyopathy worse.

Question: Are there any medications that children with cardiomyopathy should definitely NOT take?

Answer: Depending on the medication prescribed, some are contraindicated in kids with cardiomyopathy. In general, most over-the-counter drugs for cold and flu symptoms that have decongestants in them, like ephedrine or pseudoephedrine, should probably be avoided. Other medications may interfere with the medications used for the child's cardiomyopathy. While this cannot be generalized, each family should contact their cardiologist to discuss a potential new

medication to be certain that it will not interact with the new medication or otherwise cause harm.

Question: Are there any antibiotics that should be avoided that can interfere with certain cardiac drugs?

Answer: Once again, this depends on the antibiotic. Some antibiotics can raise and some can lower the amount of effective drug in the system. So, make sure your cardiologist knows if your child is placed on an antibiotic so he/she can verify that it won't dangerously change a drug level in the blood. Since many antibiotics are given for a relatively short time, even if there are transient changes in drug levels, they won't likely be harmful. But check anyway, just to be sure especially with anti-arrhythmics.

Question: Are there any studies showing long-term side effects from taking long-term cardiac medications such as lasix, digoxin or beta-blockers?

Answer: While there aren't a multitude of studies looking at long-term use of these drugs particularly in heart failure, each of these drugs has been used in other areas of congenital heart disease and has been found to be safe without serious long-term adverse effect, provided the drugs are used properly. Any of these drugs when used inappropriately can cause serious harm whether for short or long-term use.

Question: It seems as if so many children with cardiomyopathy have eating difficulties. Is it possible that some of these issues could be related to medication use?

Answer: Yes, while many children with cardiomyopathy may have feeding issues related to their heart failure, many of the drugs we use can have gastrointestinal side effects. In many cases the benefits of the therapy outweigh their adverse effects. There are other ways to treat the feeding issues without needing to sacrifice the potentially life-saving medical therapy.

Question: In regards to long-term risks and adverse effects of beta-blockers, especially using them since infancy, is there a difference between boys and girls? Also, I saw television program on beta-blockers being able to block traumatic memories from "encoding" in the brain. Are more studies planned to see how children's memories are affected?

Answer: In the adult studies of chronic beta-blocker use, most adults report not much in the way of side-effects except transient light-headedness if the medication drops their blood pressure a little too low after taking it. The other effects of mental fogginess, belly discomfort, difficulty concentrating, short-term memory

loss, etc... don't seem to be a big problem or if they are at first, these usually go away over time. I'm not familiar with a big difference between boys and girls. Children, in general, may not experience the same degree of side effects as adults. I didn't see the television program, but feel it's irrelevant because if your child requires this medication as potentially life-saving therapy, he or she needs it. Our job is to balance the benefits with the side-effects, and that's usually done by adjusting doses. It is unlikely that any studies such as that are planned in children, since, at the current time, there is no FDA-mandate to study these drugs in children as the pediatric demand is very small compared with adults.

Question: I know that beta-blockers slow the heart and help prevent an arrhythmia, but have they also been shown to actually decrease the size of the heart? Or is there any decrease from the effect of a slower heart rate?

Answer: In dilated cardiomyopathy, if the beta-blocker works, then yes. As the function gets better, the heart will get a little smaller but may not return to normal size for age (the child may then grow into their heart). The fact that the heart rate is slower will not, per se, make the heart smaller. This would happen only if the drug has some positive effect on function. In hypertrophic cardiomyopathy, beta-blockers will not make the heart smaller in thickness but should improve symptoms if that's why the drug was started in the first place.

Question: What exactly is a z-score and "LV MPI"? What should these numbers be in a heart healthy child?

Answer: A z-score is a way of describing a measurement (in this case, the Left Ventricle (LV) size) which is "normalized" for body size. Remember that the normal size of any heart chamber will necessarily change with age and body growth. Using the "raw" measurement of a heart chamber is not very helpful if the person getting that raw number doesn't even know what the normal range for the measurement is, i.e. if I told you the LV diameter during the relaxation phase of the heart cycle (that's the number we care about) was 50mm, most people would have no idea if that was normal, small or too big. But if I "normalize" that value for body size, it becomes more helpful. Now I can tell you what the normalized value is compared with other children of similar body size. That is the z-score value.

You want your child's LV size to be relatively the same as other normal children his age, so you want the z-score to be around 0 (meaning his LV size does not vary much from the normal population of kids his age and size). The normal range of z-scores is from -2 to +2 z-scores, which you can think of a distant cousin to standard deviation or the normal range of variance in a measurement. We consider an echo measurement like LV size to be within the normal range if it is between -2 and +2 z-scores (0 being the average size). Values smaller than -2

z-scores are considered too small, and values above +2 are considered too big or dilated. For example, many kids with severe dilated cardiomyopathy have LV z-scores +3 or +4 or greater.

LV MPI is an LV myocardial performance index. This is another way of assessing the function. You can report the LV shortening fraction, ejection fraction and some places use the LV MPI (or some combination). Normal LV MPI is around 0.4.

Question: Would a normal z-score for hypertrophic cardiomyopathy still be in the same range?

Answer: A z-score between -2 and +2 would put whatever measurement you're looking at in the normal or expected range. For hypertrophic cardiomyopathy since the LV walls are much thicker, I'd expect the z-score for the walls to be much greater than +2 z-scores (above the mean or average).

Question: How much "growing into a heart" can kids do? My son is 2 1/2 years old and has 17mm hypertrophic cardiomyopathy. It seems that there is not much room for growing into the heart. Do you have experience with that happening?

Answer: The context of growing into a heart was meant to be specific for those children whose cardiomyopathy gets better with treatment (especially those with dilated cardiomyopathy). In those kids, as function improves you'd expect the dilated LV chamber to shrink. It probably won't "shrink" back to normal size but will end up somewhere in-between. If the function stays stable, the LV size will be considered normal as he/she gets bigger and "grows into the heart". This means as the body size increases the Z-score will get closer to 0.

For cases of dilated or hypertrophic cardiomyopathy where the heart is not improving, either the chamber cavity or walls or both can continue to enlarge or grow. In those cases the chest cavity accommodates the growing heart. It can crowd out the lungs (which will still function) or cause one side of chest to appear larger than other (called hemi-hypertrophy of chest wall), but the heart will stay contained within its space in the chest.

Question: As a follow up to my question, genetic testing showed that my son has a fairly standard hypertrophic mutation that he shares with his father's family and also a Noonans variant. In your experience, does Noonans-related hypertrophic cardiomyopathy present differently than standard hypertrophic cardiomyopathy? Do you find this newly expanding field of genetic data to be useful or helpful to your practice?

Answer: We are just beginning to understand more of the genetics of cardiomy-

opathy and how it affects outcomes and treatment choices. Yes, Noonan's related hypertrophic cardiomyopathy appears to be different from other kinds of hypertrophic cardiomyopathy, and its course seems to depend on several factors, which include age of diagnosis and whether congestive heart failure was present when diagnosis was made. We'll know more as we have more children with Noonan's to collect information about and as we understand more of the genetics influencing the disease.

Question: I heard that drugs like Periactin can improve appetite tremendously. Can kids with cardiomyopathy who are on Digoxin, Beta-blockers and ACE inhibitors take it safely?

Answer: I could not find any interaction with the typical heart failure drugs you listed. While it is true that Periactin can be used as an appetite stimulant, one should use caution when prescribing it. It can, in unusual circumstances, cause wheezing, low blood pressure and fast heart rhythms. It is not used in small babies, and in those cases you can usually give extra calories with tube feedings at night. For older kids, you can also give higher calorie shakes in-between meals to give more daily nutrition. It's important to remember that for some kids, growth failure may be a sign of significant heart failure and may indicate a need for transplant. So, be certain the issue is appetite and not worsening heart failure.

Question: I would be very interested to know if you have ever consulted a homeopath or a herbalist for your patients? I live in England and have been told about a plant called Weissdorn (Crataegus Oxyacantha) Hawthorn which is apparently a "heart plant." My aunt takes that together with beta-blockers for a different heart problem. I want to find out more and also ask our cardiologist but just wanted to see if you have come across it in your medical management.

Answer: In general, I do not usually consult a homeopath before treating patients. However, if parents ask about a particular agent, we find information to establish if it may cause harm. With respect to Weissdorn Hawthorn, we did find some interesting information. Hawthorn may act to improve the forcefulness of heart contraction but may also interact with other heart failure drugs like digoxin and beta-blockers. It can cause additive effects for these drugs. While it may help in a small way, you have to be careful when using it with other cardiac medications. In addition, we don't know how to dose this agent in children. So talk to your cardiologist about the pros and cons before starting it to make sure that, in your case, it won't be more of a detriment than a help.

Question: My son who was transplanted at 11 days old and is now almost 6 years old was just taken off his magnesium supplement. They had taken him off it once before and then put him back on it after our pediatrician was concerned about his magnesium levels at his blood draws. Do you have your transplant pa-

tients on magnesium and if so what are the benefits of them being on the magnesium as opposed to not being on it?

Answer: We have several transplant patients on magnesium supplements. Magnesium is needed for several of the cell's functions and also helps regulate the body's calcium. Very low levels can cause the body to be sluggish and not feel well. It so happens that some immunosuppressive drugs can cause magnesium wasting. Therefore, some people require chronic magnesium replacement. We try to replace with the lowest dose possible since oral magnesium can cause stomach upset and diarrhea.

Question: They just started my son, who had a transplant, on aspirin therapy yesterday (his dose is 40 mg once a day). When I went to pick up the aspirin, I got the third degree from the pharmacy tech. She spoke to the pharmacist, and she explained that I need to keep in close contact with his doctor because Reye's Syndrome related to aspirin intake can be serious. Is aspirin therapy something you use with your transplant patients, and how common is the Reye's syndrome with these patients

Answer: I believe many, if not most, transplant cardiologists prescribe aspirin for post-transplant patients since it has been shown to reduce some of the changes associated with the coronary disease post-transplant. Since it keeps platelets from clumping together, this hopefully reduces some of the narrowing that we know comes with this kind of coronary disease. While it has been associated with Reye's syndrome, this is quite rare and most of us feel the potential benefits of this therapy outweigh the very low risk of Reye's syndrome. His dose is perfectly acceptable and what we would use.

Cardiomyopathy and Nutritional/Feeding Issues

Peggy Crum, R.D. – February 2007

Ms. Peggy Crum is a registered dietician and nutritional consultant, and she has been in clinical practice for more than 30 years. Since 1992, she has worked as a pediatric nutritionist at Michigan State University (MSU) in the Department of Pediatrics and Human Development. In her role, she provides nutrition assessment and treatment for children with special needs and serves as the nutritionist for an MSU program aimed at addressing childhood obesity.

Question: Is it recommended that children with heart disease have an evaluation by a nutritionist, or is this something the primary care physician manages?

Answer: I recommend that all children on a tube feed see a pediatric nutritionist or registered dietician for evaluation. Registered dieticians are trained to nutritionally assess a child regardless of the source of the nutrients. A pediatric nutritionist or registered dietician could take a comprehensive look at your child's nutritional intake, adequacy and growth, as well as whether or not the therapy is addressing the child's oral needs. Since a tube feed bypasses the child's internal sense of what and how much to eat, a periodic review with a registered dietician will reassure the child is staying on track.

Question: What are the major nutritional challenges you have seen in your practice facing children with heart disease and other special needs?

Answer: Children with heart disease typically require more energy. This is another reason for the comprehensive evaluation and periodic follow-up — to determine if these increased needs are being met and how best to meet them.

Question: Are there any specific nutritional needs or deficits that parents of children with heart disease should be aware of? For example, what about vitamin depletion resulting from medications?

Answer: For a child who has a tube for feeding, I would suggest approaching feeding in a "normal" way where it's the same food for all family members. You have the ability to feed him adequately with supplemental feedings through the tube so allow him to eat from whatever is offered to siblings at snacks and to the family at meals — on his own plate. In fact, he should be preparing his own plate just like other members of the family. If he wants extra butter or cream, that is fine, but don't force the situation. Congenital heart disease does not make your child more prone to atherosclerotic heart disease.

Question: My son has dilated cardiomyopathy and is on the transplant list. He has almost always gained weight and has eaten well. He has a G-tube that we

use mostly for medications and one night time feeding when he is asleep. His nutritionist wants him to get between 900 and 1000 calories per day. Now that he is nine months he sometimes refuses his food outright and won't eat finger foods yet. He will then want to nurse frequently throughout the day. Our nutritionist helps with the calorie count but does not feel comfortable discussing any sort of "feeding issues" with us. How does a parent balance allowing babies to have some control over their eating but getting all the required calories into them?

Answer: I am so pleased to hear that your son is acting his age! He is determined to eat in an age-appropriate manner. How do you balance your child's need for control with parent's need to get all those calories in? You have a perfect tool in the tube feeding. What you don't want to do is feed too much through the tube so that your son has no motivation (hunger) to eat. At his age (no longer a baby!), he is asserting his independence in something he knows how to do—eat! So let him. We know what normal eating is, and your child should eat normally during his waking hours and you should supplement that during the night to meet his increased needs. I will refer to my most quoted source for explaining normal eating — Ellyn Satter. She has a "division of responsibility" in feeding that all parents find very helpful: the parent is responsible for what, when, and where—you decide what is served, when it is served (6 times per day or about every 2 hours for a toddler) and where it is served (in the high chair). This helps to divide the day so there are times to eat and times to not eat. Your son is responsible for if (whether or not) he eats and how much. See how this approach takes the pressure off both of you? To read more about the division of responsibility and age appropriate feeding, please refer to Ellyn Slatter's book, "How to Get Your Kid to Eat...But Not Too Much."

Question: Will a child lose weight if he has an oppositional day and doesn't eat his three meals?

Answer: Maybe, but the body has an amazing ability to adapt. If your child has a tube for feedings to supplement his daily intake, then you can tube feed more volume on the days when he has not eaten. You may also want to use the tube during naptime if he has not eaten well. But, again, too much use will diminish his drive to eat. Offer food in the high chair 6 times per day.

Question: Will offering too much fruit give a taste for only sweets? Is there a risk of creating lifelong food issues by serving less preferred food masked by preferred ones for infants? For example, covering beans and lentils in pears before spooning them in?

Answer: Human beings are born with a preference for sweet taste. Offer your child the variety of foods that you eat, and he will eat a variety of foods, eventually. Do not play any game with foods. No food is worth risking distrust between

the two of you. Again, put the variety of foods you are serving for the meal (in age-appropriate size and texture) on his high chair tray and let him go at it. If he eats all of the fruit, put more fruit on his tray. He will eat other things at the next snack or meal.

Question: I am the mother of a 3-year-old with hypertrophic cardiomyopathy. My daughter had a modified Konno surgery in June 2005. Since then, the heart situation has been relatively stable and she has been doing okay. She is still fed by using a GT tube. Lately, she has started vomiting a lot. She says she is feeling sick and vomits 2-3 times a day (although not every day, but most days). This has been going on for a month. She does not have the flu. We give the "milk" in very small and regular portions. We have the next cardiologist check up in March. I just wanted to know if you have experienced the same or have any suggestions?

Answer: I am sure there have been many things occurring in your daughter's life since June 2005, but I wonder what the circumstances were around the time that her status changed and she began vomiting. Since this has been going on for a month, it must not be large amounts of food. But I would not wait until March to have this situation addressed. See her primary care physician, and he/she can contact the cardiologist or make an earlier appointment with the cardiologist. A change in status such as this should be addressed.

Question: If a child is nursing three to four times during the day, would you still offer food six times a day? I am offering three times now plus four or three rounds of nursing, and I feel like he spends most of his time eating! Also, if a child does not eat any finger foods yet, do you place the pureed food in front of them and let him "go at it" or is it OK to place food on the spoon and hand that over to them?

Answer: Since your son is 9 months old, if he is nursing 3 or 4 times per day, he won't need to be in the high chair 6 times per day. Perhaps only 3 times is fine. 6 times per day is your goal, and you will take your cues from your son.

Sometimes children want to nurse right after eating as they are thirsty. As he progresses developmentally, starts drinking well from a cup and is less interested in nursing at certain times during the day, offer him another feeding in the high chair. Usually, the last nursing sessions to be given up are sleepy times — before naps and before bed at night. These times will eventually be replaced with snacks in the high chair as well.

Pureed food needs more assistance since toddler age children have difficulty co-ordinating the spoon. If he will let you, spoon feed him. Sometimes two spoons — one for you and one for him — works well. If he wants you to load it and he

wants to put it in his mouth, great. He is asserting his independence. Keep putting out small bits of soft food on his tray so he can practice his hand-eye coordination and his swallowing of foods with increased texture.

Question: What is the difference between a feeding therapist and nutritionist? Which should a family see?

Answer: A feeding therapist may be an occupational therapist (OT) who assesses and makes recommendations about upper body strength and functioning. A feeding therapist may be a speech pathologist who assesses feeding and swallowing — all things to do with the mouth including oral defensiveness. A nutritionist is a registered dietitian (in most states with licensure, these are protected terms) who will nutritionally assess the individual looking at adequacy of the diet, signs of under- or mal-nutrition, and mode of providing nourishment to the person. Parents may want to seek the advice of both and have the feeding therapist (OT or speech) and the registered dietician communicate with each other.

Question: Is there a tool you recommend for keeping track of calories?

Answer: Calorie counting is tough and tedious. I would keep a notebook with details of food consumption at meals/snacks and tube feeding information. Set up a meeting with the registered dietician to go over this. Work out a system that is best for the individual parent or family (estimating by exchange lists, calorie information from labels and then call in or e-mail with registered dietician). It may be hard to keep this up over an extended period and may not serve a purpose every day. I would suggest doing it intermittently.

Question: I was wondering if you have any advice for us parents with very picky eaters. At 18 months old, both my boys went from being excellent eaters to pretty much only eating 4 foods. Weight doesn't seem to be an issue, but I do worry that they are not getting enough protein, vitamins, etc. Do you have any recommendations on how to get them to even try a "new" food?

Answer: I would suggest you take the approach that I always recommend — you are the parent and you have a job to do as the feeder. And that is to serve nutritious food 6 times per day! Make sure your boys come to the table all six times so they can do their job as the eater. The eater gets to choose whether or not to eat and how much to eat of the foods the feeder puts on the table. Ellyn Satter calls this the division of responsibility in feeding, which you can read more about in her books.

Most children become pickier when they are toddlers because there is a dramatic shift in their rate of growth, hence a decrease in caloric need. They just

aren't as interested in food at this age. This allows them to become more selective in what they eat.

When you implement the division of responsibility in feeding, your boys may still eat only 4 foods, but they will be exposed to all of the foods you offer on the table. This creates familiar foods without the food actually going on their plates or entering their mouths. Make meals/snacks successful by offering at least one, maybe two of the foods they have a history of eating at each meal or snack. Most kids will eat bread so I like to see bread served at every meal. Also butter. A complete meal would include a protein, 2 starchy foods or grains, a fruit or vegetable or both, milk and a source of fat. Then sit back, be patient and enjoy your own meal! Remember that children learn most everything by observation of their parents. See how long it takes for them to try foods on their own!

Be consistent. Always serve meals and snacks (comes out to be about every 2 or 3 hours throughout the day) on time, a little earlier if the boys complain of being hungry.

Question: My son is 27 months old and was diagnosed with dilated cardiomyopathy at 10 months old. At the time his weight was around the 3rd percentile. Now it is around the 50th percentile. Nicholas was introduced to solids at 6 months however never really took to them until he was around 13 months. Now he is a good little eater and wants to eat all the time. He is constantly pulling me into the kitchen to get him something to eat. During the day I offer fruit and plain biscuits which he normally starts eating and then I find them lying around the house later. This type of behavior is probably a result of us letting him eat just about anything whenever he wanted at 6-13 months of age. My question is, should I be encouraging this type of eating behavior/habits or should I just refuse and stick to 3 meals and morning and afternoon tea? Is there any link to certain medications making children hungry?

Answer: Your son sounds charming but needs you to set some parameters around eating. He has been used to getting food hand-outs whenever he wanted. So instead, you need to direct him to the high chair when it is time to eat. It won't take long (a few days) for him to figure out that he only gets food when he is in the high chair. So if he is hungry and serious about eating, he will stay there where the food is. It helps children stay on task with eating when they are in an eating environment, that is, in an age-appropriate setting with other people who are also eating and enjoying food. The answer to your question is "yes," stick with 3 meals and morning and afternoon "tea" and also add an evening snack about 2 hours after dinner (a short while before bedtime). Remember the division of responsibility in feeding that I described in previous responses to questions. You, as the mum and primary feeder, are to take care of the what, when and where. Then allow and respect your son to decide whether

or not he will eat and how much. Steroids such as prednisone are notorious for increasing hunger (often referred to as appetite).

Question: When my daughter, who is now 2 years old, was diagnosed with dilated cardiomyopathy she had to be completely tube fed for about three months due to her failure to thrive. She completely refused to eat by mouth while she had the tube. After the feeding tube was removed, she would eat one meal a day, just to the point that she got "full" (10-15 pieces of pasta for example) and refuse after that. This continued for weeks and she lost weight. Eventually, we found a "feeding specialist." Her means and methods of getting kids to eat were unconventional but they worked for us. My concern is that "give them food and let them decide" doesn't work for all kids, and I have a hard time communicating this with most folks, including my pediatrician.

My daughter is now eating well. She's up in the 25-50th percentile back from not being on the charts! The only issue I have is that she's not eating "age-appropriate" foods yet. She does "eat" bits of pasta, rice, chicken nuggets but nothing to sustain her. We still give her Pediasure once a day, and she eats most foods textures but she is not doing so well with table food. I'm concerned from a developmental perspective and also concerned about how she'll do if I put her into preschool. Do you recommend seeing a speech therapist? At this point I'm not concerned about her calories but more so about where her calories come from and that she graduates to self-feeding and eating regular table food.

Answer: I would not be so presumptuous to say, "Put it in front of them and they'll eventually eat it." Some children need a tube for nutritional support — to provide the extra calories needed or to make up the difference between what their hunger/appetite will allow them to eat or for both reasons.

The process I described in the "division of responsibility in feeding" is what I teach parents in all sorts of circumstances. Sometimes called the "trust model," it relies on trust going both ways to make it successful. When properly applied, it respects the developmental stage of the child.

Properly and completely put into practice, if the child is unable to eat enough to grow, a tube feeding is indicated. A tube feeding, for most children, is supplemental to their oral intake. I would suggest you locate a registered dietitian who practices in the area of pediatrics and has experience in the trust model or Ellyn Satter techniques (Ellyn Satter Institute has trained practitioners throughout the US) to guide you. It may take multiple sessions over a period of time.

Cardiomyopathy and Exercise Restrictions

Steve Colan, M.D. – March 2007

Dr. Steve Colan is a pediatric cardiologist at Children's Hospital Boston. The primary focus of his research has been related to the investigation of myocardial function. This includes investigations of basic physiology, development and verification of new indices of ventricular and myocardial performance and clinical studies of ventricular function in children with congenital and acquired heart disease. Dr. Colan also manages the New England Coordinating Center for the National Institutes of Health funded Pediatric Cardiomyopathy Registry.

Question: Our daughter was diagnosed with dilated cardiomyopathy (left ventricular non compaction) at 12 months. Currently she's 27 months, and she's extremely active but I worry that too much activity might not be good for her. How much is too much? When should I limit her activity? She never seems to get tired but I constantly worry when we're out with her that she's "pushing" herself beyond what she can tolerate.

Answer: In general, there is very little reason to suspect that physical activity is in any way harmful to children with dilated cardiomyopathy. There has been some evidence that exercise may be harmful during the acute phase of myocarditis and during the acute phase of rheumatic fever. Otherwise, exercise is not known to be harmful to the heart muscle in dilated cardiomyopathy. There is believed to be some increase in risk of arrhythmias during intense exercise, so most older children are advised to not participate in high-intensity competitive sports. In contrast, there is lots of evidence that exercise is beneficial in terms of overall quality of life and even in terms of cardiac health. As a result, it is important for patients with chronic dilated cardiomyopathy to maintain fitness through regular exercise. This is best accomplished through prolonged, low intensity exercise.

So what about the young children? They do not have the muscle mass to put large burdens on the heart and are not driven by the same sort of competitive urges that drive the teenagers to high intensity exercise. Attempts to restrict activity in this age group are also usually unsuccessful. However, the most important consideration is that there are very real benefits to exercise. This is an important observation that has been made in adults with coronary artery disease. It used to be common practice to exercise-restrict people after a myocardial infarction (heart attack). Then people realized that even though survivors of heart attacks are more likely to have another heart attack during exercise than they are at rest, those who exercise regularly have an overall reduction in risk such that they actually live longer. This has been called the "paradox of exercise" and has important implications. Most importantly it means that it is not sufficient to exclude people from exercise just because they are more likely to have arrhythmias

or even death during exercise because they may still have improved survival if they exercise regularly. The other implication is that it is a bad idea to assume that exercise is dangerous or bad. Unless there is really good evidence to the contrary one should proceed on the assumption that exercise is a good thing, even for patients with heart disease.

Question: My son was diagnosed with a mild form of hypertrophic cardiomyopathy two years ago at age 11. Our pediatric cardiologist has been reluctant to give a list of restricted activities saying he wants folks to think about the activity instead of saying, "It wasn't on the list."

The way I have explained it to the gym teacher is that he can't pit his body against gravity (rope climbing, sit-ups, push-ups), or directly against another kid (wrestling). He can be a kid and play, but shouldn't be in timed races or endurance competitions or do isometric activities. Golf has been encouraged as a sport for a lifetime. Do those general guidelines seem adequate for non-medical folk?

Answer: This one of the hardest aspects of hypertrophic cardiomyopathy. In general, it is believed that the risk relates to the intensity of participation and possibly also to the intensity of the emotional overlay. Prolonged, low-intensity exercise appears to provide the greatest degree of benefit while minimizing the risk. I am asked all the time for specifics about particular sports, and there have been lists developed based on the response of the heart to various forms of exercise and on the reported risk associated with various sports. Gym class is tricky because many of the activities do not really fall into the usual team sports activities and the level of intensity is often much lower.

I have to say that your rule of thumb is not bad, and it does reflect that you are trying to follow your cardiologist's advice. I am an advocate of normalizing the lives of these children to the maximum degree that can be achieved, and avoiding social stigmatization by forced exclusion from activities is an important aspect.

Question: My daughter is 3.5 years old and has hypertrophic obstructive cardiomyopathy (HOCM). After her septal myectomy surgery, she had boundless amounts of energy and was doing well keeping up with the other kids. However, the obstruction in her heart is increasing again, and she seems to tire more quickly now and can't keep up with the other kids when running. I am trying to figure out if I should be limiting her in some way. Most of the time I don't, and she will say, "Mommy, I'm really tired," when she just can't keep up anymore. I'm worried that at that point she may have already pushed herself too far.
What type of physical limitations, if any, do you recommend for someone this age, with HOCM? Is there anything I should be doing or watching for when she

is out running around to help reduce her chances of having a life-threatening event? Does having the "obstructive" type of hypertrophic cardiomyopathy increase her risk?

Answer: This raises a host of issues, most of which are pretty hard to answer. Most children this age self restrict (they don't push themselves beyond tolerance), in contrast to older children. Although there is a known risk with exercise in adolescents and older patients, such a risk has not been demonstrated in preadolescents. Self reporting of fatigue or shortness of breath is not a very reliable method for determining level of exertion. Imposing limits at this age is very difficult because it is so dependent on the child recognizing his or her own tolerance, something they are usually not able to do. Attempts to impose limits in this age group can be more stressful to the child (and parent!) than the exercise itself and can create an undesirable level of conflict between the child and parent. Overall, I generally do not advise attempts at limiting activity at this age. However, it is also important to not place the child in situations where he or she feels obligated to exercise beyond perceived tolerance. This is very unusual in pre-schoolers, but in a few years she may be faced with friends or gym teachers pushing her beyond her comfort level, which is not a good idea.

The other question you posed is whether having outflow tract obstruction represents a risk. In hypertrophic cardiomyopathy, one of the common associations is narrowing of the left ventricular outflow tract. This relates to having muscle that protrudes into the ventricle and forces the blood to squeeze between this muscle and the mitral valve. This narrowing increases the chances of symptoms such as tiring with exercise, particularly when there is also leakage of the mitral valve (mitral regurgitation). Elimination of this obstruction usually improves or relieves these symptoms. In older patients it is very rare for the obstruction to reappear after this sort of surgery.

Unfortunately, this does happen more frequently in younger children, and it is disappointing to hear that it seems to be happening with your daughter.

The much harder question is whether obstruction increases the risk of sudden death. This has been looked at pretty carefully and despite many studies there is no evidence that it does. There have also been many studies looking at whether relief of obstruction reduces the risk of sudden death, and it does not seem to do so.

There are also some children who have obstruction on both ventricles, and this is a very different situation. Having both severe right and left ventricular outflow tract obstruction is associated with a substantially increased risk of both symptoms and sudden death. When both ventricles are severely obstructed, intervention becomes a priority, regardless of symptoms.

Question: I have been reading the questions and answers about what level of physical activity is acceptable for children afflicted with hypertrophic cardiomyopathy. I am compelled to tell the story of my son, 13-years-old, diagnosed with hypertrophic cardiomyopathy.

My son was diagnosed at 2 months. He never exhibited any symptoms — dizziness, chest pain, shortness of breath, fainting, etc. He limited his activities, primarily choosing not to participate in sports like soccer, basketball, etc. However, we let him play "normally" with kids in the yard, offered guidance to the gym teacher and even allowed him to play Little League baseball. While playing "normally" with kids in our yard while we were having a BBQ, he suffered cardiac arrest -- later described as sudden ventricular fibrillation.

When my son was transported to the hospital, the first thing his doctor said to me was, "He was supposed to give us some warning." My son spent 2 months in the hospital, the first 10 days to address immediate medical problems and implant an implantable cardioverter defibrillator (ICD). The rest of the time was spent with inpatient rehabilitation where he had to relearn how to walk, talk, eat, go to the bathroom, etc. The 25 minutes without a heart beat had caused anoxic encephalopathy which led to profuse brain damage. He was eventually discharged from the hospital without any need for special accommodations at home. He learned (again) his ABCs, to count, to write, to read and eventually returned to his 5th grade classroom. Today, he is in 7th grade with minimal accommodations, reading at a 6th grade level, interacting socially as any "normal" 13-year-old would. He is playing the piano again, has taken up golf, and has a "normal" 13-year-old attitude.

I'm not convinced that anyone really knows what a safe level of activity is. I have since learned that it is becoming more common to implant a defibrillator in kids afflicted with hypertrophic cardiomyopathy. Would this have saved Kyle from the recovery process that he experienced?

Answer: First, with regard to the use of ICDs in cardiomyopathy, this is a rapidly changing field. The ICDs have become smaller and last longer than before, making their use in children much more feasible. However, they are not yet a cure-all. They also carry risks, including false discharges that can at times be fatal. The decision about using these devices relates balancing the risks against the benefits, where one thinks the risk of an adverse event related to the HCM is greater than the risks related to the ICD. This equation is different in children than it is in adults, because there are excellent data that show that the risk associated with these devices is higher in children than it is in adults. With the continued improvements in these devices I would not be at all surprised if someday everyone with HCM gets a device, but we are not there just yet. In the mean time, we try to assess the level of risk based on certain predictors of outcome,

such as family history, presence of abnormal heart rhythms, a history of passing out, the response to exercise, and some of the findings on echocardiography or MRI. If we find anything that suggests that someone is at higher risk, then an ICD is recommended. Otherwise, the risk associated with the ICD is likely to outweigh the risk of the disease, so an ICD is not recommended.

Secondly, I would like to make a comment about what we can and can't learn from events such as this. Whenever things like this happen, we (both the physicians and the parents) do a lot of soul searching to try and figure out if things should have been done differently. There are two issues that are raised here with regard to management: whether activity should have been more restricted and whether an ICD should have been placed earlier, and I hear people asking whether this should have been handled differently. This is completely natural and appropriate, but I want to emphasize that just because something bad happens, it does not mean that the wrong decisions were made. I do have enough experience with this situation to know that lots of people will decide that since he had this arrest we can conclude that he should not have been exercising and/or that he should have had an ICD. I am in the fortunate position of not knowing enough about your son's situation to be able to make any comments about his management one way or the other, so I will keep my comments completely abstract. I want to make a plea that people resist the urge to conclude that since a bad event happened, the decisions that were made were the wrong decisions. People will often conclude that because of a bad outcome in an individual, the decision about the course of action was incorrect. Although we do this all the time, it is actually an incorrect conclusion. No one can predict the future.

All decisions are correct or incorrect based on the information that is available at the time the decision is made, not based on the outcome. If the decision is based on the correct probabilities, it is a correct decision. For example, if I have an accident while driving to work tomorrow, it does not mean that I made the wrong decision when I decided to drive to work. If the road is generally safe, and I will derive benefit from going to work, then I made the decision that gave me the highest probability of a good outcome, and that decision remains the correct decision regardless of whether I have an accident or not. If the overall incidence of accidents goes up and the probability of an accident increases, then the equation changes, and I may have to make different decisions in the future, but today it is still the right decision.

The decision about activity participation and ICD use is similar. Rather than discussing your son's situation, which is very personal and emotional and therefore it is difficult to take an objective view, consider my father with coronary artery disease. We know that some patients with coronary artery disease will have heart attacks while they are exercising. We also know that their probability of survival is better, and their probability of a heart attack is lower if they exercise regularly,

so I tell my father every time I speak with him to exercise because it increases his chances of a good outcome. It is absolutely true that there is appropriate and inappropriate types of exercise for him, and the recommendations have to be tailored to his situation. However, just like driving to work, if he has a heart attack during exercise, it does not mean that he made the wrong decision when he decided to exercise, because he decided to do what had the highest probability of a good outcome. I guess my primary message here is that it is a big mistake to conclude too much from what happens in an individual case. The decisions about exercise and ICD use are very complex; for people to decide based on any individual case that the management of their child should be changed would be a very bad idea.

It is always fair to ask questions, and I think parents have to constantly challenge their physicians to justify their decisions and support them with the best available data, but it would be a bad idea if they changed management based on individual cases. I would hate to tell you the number of bad medical decisions that have been made when physicians have decided to make their decisions based on their last case rather than on the overall statistics!

No one really knows what a safe level of activity is, if what is meant by “safe” is activity that is free of all risk. However, this does not mean that the solution is for everyone to be excluded from activity because some risk is beneficial. I know this sounds paradoxical and it is particularly hard to accept this with regard to our children when every instinct we have is to protect them from risks, but stated otherwise we have to recognize that there is a risk associated with exclusions from exercise, and the challenge here is to find the right balance.

As to the conclusion that an ICD would have prevented this event, I really have to disagree with this statement. It is fair to say that an ICD might have prevented this event, but it could also have failed to prevent this event (ICDs are not 100% successful) and it could even have caused this type of event (the ICDs have sometimes caused arrests by inappropriate discharges). The decision to put in an ICD after an arrest is easy because one of the best predictors of sudden death in hypertrophic cardiomyopathy is a history of resuscitated sudden death, so we know the risk/benefit ratio is in favor of the ICD. However, it is impossible to conclude that things would have been better if the ICD had been placed previously.

Although we (physicians) often give advice as if it is all based on hard science, many of the recommendations that we make come down to balancing quality of life versus risk, which are issues that are fundamentally based on personal philosophy rather than on science. For example, we all know that even completely healthy children are at risk for injury or death during sports. Most parents decide that the quality of life benefits for the child are worth this risk, although at times

we all have more than a little hesitation. For patients with heart disease, this balance gets altered and there is no "scientific" way to decide when the "risk" exceeds the "benefit" since this judgment is based on personal philosophy rather than science. Parents need to ask the basis for any recommendations to make sure their philosophy is aligned with their physician's.

In case it is not completely obvious, I should disclose that I am heavily biased towards the quality of life end of the spectrum. I strive to "normalize" the lives of my patients to the greatest extent possible, attempting to avoid the social isolation and stigmatization that a chronic disease can impose. This often implies more risk than would be associated with greater restrictions on patients. I strive to have frank discussions with my families so they know that this is where I am coming from. In parallel with this, I also am biased towards taking short-term risks for long-term benefits. When we send one of our small patients off to open heart surgery we take a large short-term risk, knowing that surgery increases the chances of death in the short term with the anticipated benefit of a diminished chance of death and disability and an improved quality of life if the surgery is successful. This is clearly a balancing act, and there are physicians who lean more in one direction or the other about undertaking high risk surgery. Although I definitely lean towards accepting the higher risks if it maximizes the potential for longer and better quality of life, I feel it is my obligation to explain to families that this influences my recommendations. Not all of my patients agree with my philosophy, by the way, and I respect their opinion. There is no right or wrong answer to this balance, only opinions.

Question: I was wondering if there are any particular signs that you might look for that would let you know that your child might be pushing activity too far. I ask about signs because my almost 11 year old son has dilated cardiomyopathy as part of a mitochondrial disorder. His muscles are globally impacted but he can walk well, although he fatigues easily. Any higher strength activities such as walking up inclines and climbing stairs take a lot of effort and can have him bent over catching his breath.

Although he has always worked hard to keep up with his peers, it is more recently that he seems to be really pushing himself at school to keep up. We don't worry about him at home because he seems comfortable with a slower pace there. We have always let him make these decisions, although we have been pressured by school at times to insist he take the elevator. We feel it is so important for his independence to participate as fully as he wants to. Is there any danger for him if he pushes until he has to catch his breath and needs to squat on the ground to recover?

Answer: The mitochondrial defects are a particularly complex scenario to deal with. In addition to the heart problem, they may also have skeletal muscle prob-

lems. This makes it much harder to tell which is causing the symptoms. If the heart is affected, it has a limited capacity to increase output in response to exercise. If the skeletal muscle is affected the mitochondrial defect creates an inability to utilize oxygen which means two things. First, the cardiac output will go up even more than usual for the level of exercise as the body responds to the signals the muscles are sending out that they are not getting enough blood. Second, the muscles convert very early in exercise to anaerobic metabolism, which is basically energy production that does not require oxygen. This allows exercise to go on but results in acid buildup in the blood, and the body responds to this by rapid respiration. The physiology is complex but the bottom line is that the patients feel really short of breath, but it may be due to the skeletal muscle response rather than the heart response. There are some relatively easy tests that can be done to separate which of these is the bigger problem.

Whatever the explanation is, you want to know what the level of risk is. These are very rare diseases, and there is very little information available about the risk associated with exercise in this specific disorder, so we are stuck with having to base recommendations on the experience in more common disorders. In general, risk is related to intensity of participation, and this is graded on the basis of how the body responds rather than the type of exercise. If he is really short of breath and if the recovery time is long (more than a few minutes) then he is probably pushing himself too far.

One of the unusual features of the mitochondrial myopathies is that their switch to anaerobic metabolism is caused by the mitochondrial defect rather than because of inadequate blood flow. If you or I exercise to the point of anaerobic metabolism, we get lots of pain in the exercising muscle because of the buildup of acid in the muscles. Therefore, when we switch to anaerobic metabolism our muscles hurt, and we slow down or stop because of the pain. However, the patients with mitochondrial problems have plenty of blood flow which washes the acid out of the muscle, so even though they switch to anaerobic metabolism they don't get pain, so they can keep exercising until their total body acid levels are extremely high, and they are really, really short of breath! Please note that their shortness of breath is because of the high acid, not because they are not getting enough oxygen. The bottom line is that whereas most of us have a warning system in the form of muscle pain that tells us when we are overdoing it, patients with mitochondrial defects don't get this warning and can push themselves to very high acid levels. The only solution is for your son to learn to respond to shortness of breath rather than muscle pain. The other trick he can use is to test himself: If one set of stairs makes him require more that 1-2 minutes to recover, he either needs to stop halfway to recover before completing the climb or else take the elevator. If he can handle one set of stairs but not two, he can decide on that basis. His tolerance should be pretty reproducible. If he is still significantly short of breath for more than 2 minutes after stopping exercise, it is probably excessive for him.

Once again, exercise is beneficial and so it is important for him to not reduce his activities too much. In addition to all the usual benefits of exercise, in the particular situation of the mitochondrial defects there are some data indicating that regular exercise can increase the number of mitochondria in the muscle which can overcome in part that abnormal function of the mitochondria. So, once again the message is to get lots of exercise, but keep the exercise low to moderate in intensity.

Question: How does heat and cold affect children on beta-blockers and children with heart disease? I ask because my son seems so affected by the elements, and I was curious if this is related to his condition in any way. He absolutely hates the cold, and he seems to get very cold quickly. The heat makes him want to lie down and rest.

Answer: Patients with significant heart disease may be intolerant to temperature extremes. The body regulates temperature loss primarily through the control of blood flow to the skin. In high ambient temperatures, the body shunts larger amounts of blood flow to the skin to get rid of excess heat and maintain a normal core temperature. Patients with a limited capacity to increase cardiac output (the total amount of flow from the heart) may be excessively burdened with fatigue during high temperatures. Patients with diminished cardiac capacity may be excessively sensitive to the cold because their skin blood flow may be already diminished. However, there is marked variation among people anyway, and it is often hard to say what part of this is constitutional and what part is related to the cardiac problems or medications. Although there are mechanisms by which the beta-blockers can alter the control of body temperature, most people do not experience significant change when they start these medications, so I would say that this is a possible but unlikely cause of intolerance to swings in temperature.

Question: What is the most rewarding thing for you about working with families that struggle on a regular basis with childhood disease?

Answer: I have a teenage patient who was just admitted to the hospital on Friday and is quite ill in the intensive care unit. I first started taking care of him when he was two hours old, and he had his first catheterization within hours, aborting an otherwise certain death. I have put him through 4 catheterizations and 3 open heart surgeries since then and shared the angst and anxieties with his parents on each occasion.

He is now a robust, well-adjusted teenager and a star on the school tennis team. Now there's a reward for you! His admission on Friday left me in tears, and I spent quite some time thinking why it affected me so deeply. When you share this sort of thing with people it becomes much more than a service provided — it becomes personal. I know the tragedies are not my own, since even though I

share them with my families, when I go home I have two healthy children to greet me. Nevertheless, it is impossible to not get invested in these situations. Fortunately, my patient's current illness has nothing to do with his heart disease and he has now turned the corner with expectations of full and rapid recovery. We are able to help a lot of children and a lot of parents. What more could I ask for?

Marriage/Relationship and Family Issues with a Chronically Ill Child

Maryann Rosenthal, Ph.D. – April 2007

Dr. Maryann Rosenthal is a clinical psychologist and a highly regarded author and international speaker on family dynamics and life achievement issues. Appointed by the governor of California, she served on the California Developmental Disabilities Board and is president of Global Leadership Enterprises. Among her publications is "Be a Parent, Not a Pushover," which was selected by USA Book News as "Best Book" in their family/parenting category. An expert on human behavior, she has been selected by Yahoo! Health to be a featured relationship expert. Dr. Rosenthal received her M.A. in Psychology from United States International University (USIU) and her Ph.D. in Psychology from California Institute for Human Science. In addition, Dr. Rosenthal also has a grandson with dilated cardiomyopathy.

Question: What do you see as the greatest struggles within the marriage or significant other relationship in parents raising children with cardiomyopathy?

Answer: Professionally, families and relationships are my passion. Personally, I am connected to all of you because of my grandson's diagnosis of dilated cardiomyopathy at 3 months of age. From that moment on, I knew that none of us would walk on this planet the way that we ever thought that we would. From that moment on, we would know something about life that other people would never know. We were going to know and embrace what living life to the fullest would mean for ourselves, our children and our grandchildren.

I often tell my clients that the male/female relationship is a "cosmic joke" and we might as well get used to it. All relationships, even without a special needs child, are a balancing act within the family system, and all families develop their own unique ways of interacting. In a marriage that is unstable, the stress of dealing with your child's cardiomyopathy can cause the family system to collapse. On the other hand, in a relationship that is strong, your child's illness may develop increased closeness and strength in the marriage.

Parents must learn to cope with the fact that their child will never fulfill the life that they had hoped and dreamed of for them. Coping is a process that evolves over time and each parent will develop their own style to deal with the stress and strain of the disease. Each parent may exhibit a different behavior in response to illness related events, and there will often be changes in couple's role definitions. The wife's perception of support from her spouse may be related to the husband's involvement in the care of the child, and the husband's perception of support may become related to the wife's availability to him and the rest of the family. So strengthen your family resilience by taking care of each other and your rela-

tionship. Build and focus on what you have. Share your feelings with one another, so that the other partner doesn't have to guess what you are feeling. Holding in your emotions and feelings protects no one and may only make your partner feel even more isolated

Question: Are there specific things you can recommend to parents to help explain to other family members what it is like to raise a child with chronic illness? How can parents help extended family to understand what they go through with their child?

Answer: No one can possibly know what it is like to raise a child with a chronic illness unless they know through experience. It is simply not possible, but in response to helping other family members try to "get it", communication is the key. In the beginning, many family members may be overwhelmed by the diagnosis and be in denial. They simply can't accept the fact that this child is not " normal and is chronically ill." A suggestion might be to ask family members to write down all of their questions and once you have the answers call a family meeting to share the information. Encourage family members to educate themselves. Remember, it is important to recognize that they are also juggling feelings of anger, grief, protectiveness and love all jumbled together. Often, they will take their cues from you and truly want to be part of your support system given the chance.

Question: In your last answer you talk about how parents need to learn to cope with the fact that their child will never fulfill the life they had hoped and dreamed of for them. Maybe that sentence should read "may never" instead of "will never." I know it sounds silly but something so small can really affect the hopes of some parents who may be new to this disease. Our daughter is the perfect example of a child who was in the late stages when diagnosed and is now doing great and leads a perfectly normal life.

Answer: Thank you for your feedback, and I understand and appreciate your comments. It is wonderful that your daughter is doing so well. I know that "never" sounds so final, but in my own experience with my grandson, I needed to face the reality of his illness knowing that he would go on to do wonderful and magical things with the life that he was given; it would just be different. All parents with special needs children must adapt to a different reality, and one of the "faces" of hope is the courage to accept this reality. When I think of "special" needs, I don't think about the physical challenge, but about how truly special these children are in their own unique ways with their God given gifts and talents.

Question: I know I don't always like to admit it but my wife and I don't always agree on things regarding our son's care. Just the other day she was frantically screaming, "I am going to call 911." I was yelling,"You better not." You see, my

son had lost his airway by his trach tube coming out and we couldn't get it back in. If the 911 respondents would show up they would have no clue about a child with a trach. I have never called them yet. We finally agreed to take him to the emergency room where there are more hands and they typically don't panic. After we left the emergency room, my wife and I both giggled a bit and said, "That was an eventful day." The big problem was that our 8-year-old son was with us all the time, and it scared him quite a bit. He didn't want to go to the hospital with us because he thought his younger brother was going to die. We had already had a panic situation when my wife took my son in for an urgent chest x-ray earlier in the week and they told her he was in acute heart failure and they wanted to admit him to the PICU. I asked her to stay at the hospital until I got there so we could assess the situation together, but she and the nurse were determined to let him go home. You are probably getting the picture — my wife and I are the experts on our son's condition, and it is a nightmare trying to keep everyone on the same page. Any suggestions will be appreciated.

Answer: All parents have a certain style of parenting that is based on personality, experience and their own upbringing. That style, in my opinion, becomes even more apparent during a crisis. In fact, it is rare that the styles will be compatible because we always fall back on what feels comfortable to us in times of stress. Think of your style of parenting as the filter through which passes all of your interactions with your children, and it might be quite different than your spouse or significant other's filter. As needs increase or a crisis occurs, the emotions experienced will be the strongest that they will ever be. We put on our "game face" and we act with a single-minded focus...coping with the challenge.

And, of course, men and women simply do not think the same. It is the unique combination of characteristics and strengths that each partner has that creates the resilience and protective factor, defending your child from the disruptions and crises of his illness. As with all parents of kids with a chronic illness, you are the experts and if you put two experts in the same room together, they will not always agree, and that's okay. Agree to disagree. Give up trying to get everyone on the same page, but be very clear with each other about what you will not compromise on. That might take some negotiation with each other during the "quiet" times—never during the crisis. Try to find a balance (even if you don't want to) because this is truly a shared experience.

Because emotions are a constant juggling act, here are a couple of tips to help create the "safety net:"

> Keep your perspective by keeping your sense of humor (I love that you and your wife were able to giggle about it afterwards.)
>
> Communicate.

Find a physician that you can really trust...not just one who has all of the proper credentials. It really involves trust, and that physician will become part of your heart and soul, whether he likes it or not. That is the power that you have together, and he can be a very leveling force in your lives.

Question: My husband and I have three children ages 6, 4 and 22 months. Our middle child Felicia was diagnosed with dilated cardiomyopathy on January 2006. Prior to this she was diagnosed with Autism Spectrum Disorder in May 2005. To this day I have not cried over my daughter having a heart condition. I have been experiencing depression, which of course affects my marriage and relationships with friends. Is it normal to feel afraid to be happy again? It seems feeling down has become a normal part of daily life. How can a family get past this? Will there come a time when we won't be afraid to be happy and not worry so much about the unknowns?

Another major concern is trying to balance all three children. How do people do it? I find it so difficult. Your opinion and advice would be greatly appreciated. I'm sure I'm not the only who has feelings about this.

Answer: There are two issues that I want to address. The first is family resilience, and the second is the very serious concern of depression.

All parents of kids with chronic illness experience depression and anxiety in different ways and at different times. In addition to the stress of the illness, the financial drain, quality of care, anger, grief, protectiveness, isolation all create a cumulative layer of stress that puts you into overload. All of your feelings are valid and normal. They may vary slightly, but you express the feelings of many others. There are no pat answers, but the only way to survive is to make you and your family as resilient as possible. Dr. Sol Gordon states, "It is not the child's disability that handicaps and disintegrates families; it is the way they react to it and to each other." Recognize your personal feelings and emotions, try to find a balance and never lose hope.

Having more than one child is always a balancing act, whether one is special needs or not. Having three children will always be difficult, at times. I say that because sometimes it is important to "normalize" some of your feelings so that not every single emotion feels catastrophic. It helps to keep a perspective.

I often ask myself, "How many times can these parents be expected to bounce back?" Well, the answer is forever. Because of that "forever" your main source of strength will come from fostering family resilience. Here are some tips:

Open up lines of communication (a constant theme of mine.)

Make time for each other...difficult, but not impossible.

Develop a support network that you truly trust...not always family.

Embrace either a formal or non-formal respite...so important for busy moms.

Seek professional help.

When emotions become so overwhelming that there is no ability to "bounce back," it may be a depression that needs treatment. Depression and anxiety are treatable, and help is out there. Counseling can give you the opportunity to redirect some of your emotions into something positive for your family unit. It can be a very empowering experience and get you "back on track." It is never a sign of weakness.

Question: We recently had to deal with my daughter, who is 11 years old, having a second open heart surgery. She knows she has health problems. In her language (she is somewhat delayed) she says, "I have a puzzle piece missing and a horsy in my heart." She had a pulmonary valve put in and an Atrial Septal Defect closed and had nightmares about the procedure.

After her cardiologist offered some reassurance and explanation of the procedure, she now will sleep in her room again, and she hasn't expressed any more fears. Unfortunately, we do discuss health problems in front of her.

We know she is going to have more surgery in the future, but she hasn't grasped it yet. I am thinking of using mental health professionals to help her understand what is going on but haven't done anything about it. What are your suggestions?

Answer: Your daughter is an adolescent, which is a time of upheaval in any child's life and a challenge for parents. There are so many changes happening both physically and mentally. Seeking professional help can offer a source of comfort as she explores her emotions, attitudes and feelings. Even with reassuring parents, a counselor can provide another source of support for all family members. All modern day parents are so busy, and parents of kids with special needs have even less time. Because of their changing brain, adolescents process information differently than adults. It is important to remember that she might be hearing things quite differently, so I encourage parents to remember this when discussing health information in front of your child. Some of the information that she hears can be a source of anxiety. Mental health professionals are trained to be an added support and can be helpful to both parents and children.

Question: Our daughter, who has just turned one, was diagnosed at 4 weeks old

with a familial type of dilated cardiomyopathy, called non-compaction (or LVNC). We also lost a daughter nearly 5 years ago, as an infant, to the exact same diagnosis. We thought and were told at that time that it was an unlucky genetic error, and it was unlikely to happen again. After our loss we went on to have a healthy son, who is now 3 years old.

We obviously made the choice to have more children, knowing the risks, however small we thought them to be at the time. I worry that our affected daughter will blame us for making a decision that has so greatly impacted her lifespan and her quality of life. I have two other children entering their teenage years. I know what they are like, and parents get blamed for a lot of decisions, which doesn't ordinarily worry me in the slightest. My youngest daughter is a different case. Our decision has affected her entire well-being, has affected all our other children and our extended family and friends. I wouldn't say I feel guilty about having her, and I certainly don't obsess about it, but how do we approach the subject with her when it arises? I would like to hear your views on this subject.

Answer: You bring up a great question about how we tell our kids that they have a chronic and life-threatening illness. If it becomes too painful and frightening for your child, he/she could go into denial that an illness exists. It is such a personal call because every family situation is different, and every child is different. There is no set rule because the age of the child, the severity of the disease and the family dynamic all affect the course of action. I can, however, make a few suggestions. The most important thing is to be honest without overwhelming them with emotion. Age appropriate information geared toward your child's learning ability and emotional ability will vary from child to child. A 3 year old probably does not understand the concept of being sick, while a 6 year old will understand. Imagination is often worse than reality, but only give them facts that they can understand based on their intellectual capacity.

The mother asking the question mentioned that teenagers blame parents for a lot of decisions. She is so right. We have to remember that an adolescent's job is to drive their parents crazy. Teens want information about their lives, but a rule of thumb is not to go overboard with details unless they ask. Take your cues from them, answer questions straightforwardly, and then wait until you get asked another question. This is a time when kids often go into denial regarding their illness. They do not want "this disease" to take over their life any more. I hate to keep repeating myself but foster their resilience at an early age through communication, and you will limit the level of potential conflict in the teenage years. Buffer the feelings that your child will experience with as much normalcy as possible. The positive message is to carry on as normal as possible knowing they have a life threatening illness, but at the same time they are not going to let it control their lives. That message is very reassuring to children.

Question: Our son (15 months old) was diagnosed with dilated cardiomyopathy and arrhythmia at 7 months. Since that time I have tried to source as much information as possible about the condition and the medications being used to manage his dilated cardiomyopathy. I have found this listserve to be a great source of both information and support. My husband, on the other hand, does not want to read any information or listserve comments. He believes that I am becoming immersed in "doom and gloom" stories and that the doctors have told us all the information we need to know right now. I have articulated to him that I believe knowledge is power, and this knowledge will enable us to manage our son's condition better. I truly believe he is in denial, as Max appears to be a healthy looking little boy who is bright and energetic. How do I deal with my husband's unwillingness to search for answers and information? He maintains that he is simply being extremely positive about Max's condition and his future.

Answer: Your question addresses a common theme that prevails among couples with children who have a life-threatening illness: NOT BEING IN THE SAME PLACE! What I know from my own family experience and counseling is that the process of coming together evolves over time. For you and your husband, the experience is very new and each partner needs time to "absorb" the shock. It is second nature for moms to be caregivers, no matter what the circumstances, while dads usually need more time to adjust to any new baby in the house.

When a couple has a baby, there is always such joy at the life they are bringing into the world. That dream becomes shattered when their child is diagnosed. A grieving process begins, and parents often feel a deep sense of guilt as if they have caused the disability. A long process begins of coming to terms with the disability, their own struggle with emotions and those of all others on their child's life. What may seem endless for mom may just be "sinking in" for dad. The information to help is always available but only when the person is ready.

During a calm moment, explain to your partner that you each have different ways of coping and acknowledge his/her feelings. Agree to stay away from any name calling. Come to an understanding that you will not make assumptions or guess as to what the other partner is feeling. In time, you will create a safe place for feelings to be shared. Denial is a way to cope and survive and pace our feelings of grief. It is a way of only allowing what we can handle at the time. It can serve to make us stronger over time. Be patient.

As much as it is a shared experience, you each will deal with it differently. For the mother asking the question and her husband, there already is a dynamic evolving where they are seeking a balance, which can prove to be quite helpful as their journey progresses. She is intently focused on her son's care and her husband is trying to picture a bright future. With the right mindset, it does not have to be adversarial, but a system of checks and balances and mutual respect for where you are in the process.

Echo Reports and Lab Reports

Daphne Hsu, M.D. – May 2007

Dr. Daphne Hsu is professor of Clinical Pediatrics at the Columbia University Medical Center and director of Pediatric Heart Failure at Morgan Stanley Children's Hospital of New York. She has been caring for children with cardiomyopathy since 1984. She is a National Institutes of Health-funded investigator for multicenter clinical trials in pediatric heart disease. Her special interests are in heart failure, cardiomyopathy, and heart transplantation and is actively involved with the Pediatric Heart Transplant Study Group and the Pediatric Cardiomyopathy Registry. Dr. Hsu is also on CCF's medical board of advisors.

Question: How would you suggest that parents best digest and understand the echocardiogram (ECHO) report since many of the abbreviations can be overwhelming?

Answer: I would suggest that parents speak with their cardiologist in order to understand the degree of abnormality that is present on the echocardiogram and whether it has changed from the previous studies. The abbreviations and other detailed measurements are not as important as the overall assessment of the results.

Question: What is the main difference between shortening fraction (SF) and ejection fraction (EF)? When is each used?

Answer: Shortening fraction (SF) and ejection fraction (EF) are both measurements that reflect the function of the left ventricle. The calculations for each are different, so the important issue is whether or not the measurements fall in the normal range for the child's size and age. The decision to use the SF or EF is based on the preference of the laboratory, as each is a valid measurement of how well the heart is squeezing.

Question: Is it best to have the same echo lab or blood lab do the test each time since there can be operator variation? Or can this be accounted for?

Answer: In most cases, the SF or EF is a fairly standard measurement that all echo labs can perform. However there can be local variations in the standard views obtained in order to calculate the measurements between labs. I would recommend that echocardiograms be performed in the same lab if possible, especially because since it is important for the patient and family to have a relationship with a single institution and/or physician so that the evaluation of the child, including the echo, can be provided in a consistent manner. There is no need to have the same echo technician perform the study though.

There are very strict standards of practice when performing clinical laboratory tests, and commercial laboratories must go through an CLIA approval process before they can perform testing. This means that results from different laboratories are comparable. In special cases, such as with genetic testing, there are limited places that perform the testing. Or, the testing is performed as part of a research study, and those studies need to be performed in the same laboratory.

Question: Our son was diagnosed with dilated cardiomyopathy as an infant, and at age three it was determined that his dilated cardiomyopathy is part of a mitochondrial disorder. He is now 11 years old. He gets very regular echocardiograms so we have become relatively comfortable with what we are seeing. Dilation and systolic dysfunction are always noted, as would be expected. But more recently the echocardiogram reports are noting diastolic dysfunction as well. How do you determine diastolic dysfunction from an echo? What is being looked at to diagnose this? What does this mean for overall heart function? Can shortness of breath be a symptom of diastolic dysfunction even without fluid in the lungs? Our son takes a hefty dose of diuretics and has not had any fluid build-up in his lungs, but shortness of breath (can be intermittent) is a more prominent part of the picture.

Answer: Systole is the part of the heart beat when the ventricle is squeezing or contracting. Systolic function is measured by the shortening fraction or ejection fraction. The ejection fraction or shortening fraction is a good measure of how well the heart is contracting and has been shown to be associated with the condition of the patient. Diastole is the part of the heart beat when the ventricle is relaxing and filling with blood. There is no proven way to measure diastolic function by echocardiogram. One of the new echo techniques that has been proposed is called tissue doppler. Tissue doppler measures how fast the heart muscle contracts and relaxes. Many echo laboratories are doing these measurements, but no one is sure what they mean. No studies have convincingly shown that the tissue doppler measurements are an accurate reflection of the diastolic function of the heart or predict how the patients will do in the future. Although our echo lab does do the measurements at the present time, I am skeptical, as I have seen patients with very abnormal hearts who have normal tissue doppler measurements and others who have normal hearts with abnormal numbers.

Systolic and diastolic dysfunction both can result in fluid build-up in the lungs and shortness of breath with exertion or exercise. In both cases the lungs should have increased fluid on chest x-ray.

Question: Have you ever heard of a connection between Isolated non-compaction of the left ventricle and the production of little or no human growth hormone (HGH)? My 13-year-old daughter, diagnosed with dilated cardiomyopathy

at 5 months, was found a year ago to produce no HGH. She now gives herself daily injections. Do you know of any connections to the disease?

Answer: There have been some reports of growth hormone deficiency and dilated cardiomyopathy, although the connection is not well characterized and it is not clear if the cardiomyopathy improves with growth hormone replacement.

Question: I was wondering if there is a "normal" fractional shortening. I remember when my son was first diagnosed at 3 months old they told me that "normal" was 30+. Now that he is 2, I'm just curious at this age, what is considered "normal." What does fractional shortening really mean?

Answer: Normal fractional shortening is different by age and by the laboratory performing the examination. In our laboratory, 28-40% is considered normal in your son's age, but you would need to check with your cardiologist as to what is normal in his/her laboratory.

Fractional shortening is a calculation based on the diameter of the heart in systole (smallest at maximal contraction) and diastole (largest at maximal relaxation). The fractional shortening is calculated by the formula below and turned into a percentile by multiplying by 100. In essence we are comparing the biggest to the smallest diameter of the heart. If the heart is not working well, there is less difference between the biggest and smallest diameter and the fractional shortening is lower. It is a measure of how well the heart contracts, which then tells us how strong the heart muscle is.

Diameter (in diastole) - Diameter (in systole)/Diameter (in diastole)

Question: My daughter has dilated cardiomyopathy, and we have not seen much of a difference in her echocardiograms over the past year since she started taking medications. I was told her case is somewhat rare. The doctors think she had an aneurysm to the septum while I was pregnant with her. They told me it's a large ventricular septal aneurysm affecting two-thirds of her interventricular septum (the lower two-thirds towards the apex.) There is thinning of the septum and paradoxical motion.

I am wondering how accurate her echocardiograms are considering these facts. Based on this, should her echocardiograms be done by the same person at each appointment? Can we really get accurate numbers? Is it possible some echo machines are better than others? We live in eastern Canada, and this is the first time the doctors here have seen a case like hers where she has a large ventricular septal aneurysm. Because her condition is rare, do I need to get echocardiograms done at another hospital that has more experience with this?

I was also told that my daughter is an excellent candidate for a biventricular pacemaker. Does a doctor base his decision on whether or not a child gets a biventricular pacemaker with what the echocardiogram shows and the numbers the echocardiogram gives?

Answer: It sounds like the doctors are able to get good pictures of your daughter's heart by your description. The ventricular septal aneurysm is unusual, but it does happen. I do not think the echocardiogram needs to be performed by the same person or on the same machine, but the studies should be compared to see if there has been any change over time.

Biventricular pacing has been used in adult patients, but the data in children is sparse and the decision to place a biventricular pacemaker should be made by a team of pediatric cardiologists who have expertise in cardiomyopathy, heart failure and electrophysiology.

Question: I was told that my son's septum is thickened to about 5 mm. Is that considered a lot? I've heard of kids that have a septum of 10 mm so I have trouble determining what is significant. He is otherwise asymptomatic.

Answer: When we decide if a heart is "thicker than normal" we are comparing the measurements to what would be expected based on the size of the child. Your doctor should be able to tell you if the measurement of 5 mm is in the normal range for your son, or how much thicker the septum is compared to the expected range for a child your son's age.

Question: My 17-year-old son has been on the cardiac transplant waiting list for nearly 18 months. We have recently entered into a new phase of routine hospitalizations for Dobutamine drips to assist his lung/internal organ function due to heart failure. Does anyone know about using the BNP blood test as a marker for heart failure?

Answer: BNP stands for b-type natriuretic peptide (formerly know as brain natriuretic peptide) and is a blood test that measures a hormone that rises when there is clinical heart failure. BNP is used in adult patients who have trouble breathing to help decide if the trouble is from a lung or a heart problem. More recently studies in adult patients are suggesting that the BNP level is an indicator of how severe the heart failure is in a patient. One of the problems with BNP levels it that they range from 0-10,000 and can change within a day in an individual patient. There is not much information about normal BNP levels in children, although more studies are being reported. At the moment, pediatric cardiologists are considering BNP as one of the indicators of heart failure, but it is not an exact

test and we still don't know whether the BNP level will be able to predict how the patient will do in the future or whether it gives us any more information than the clinical picture of the child.

Genetics and Pediatric Cardiomyopathy

Wendy Chung, M.D., Ph.D. – June 2007

Dr. Wendy Chung, M.D., Ph.D., is a clinical and molecular geneticist who directs the genetics program at Columbia University Medical Center. She has served as the clinical geneticist with the pediatric cardiomyopathy program at Children's Hospital of New York for the past 7 years. She performs laboratory research to identify new genes for susceptibility to complex genetic traits and develop better diagnostic tools for genetic evaluation of cardiomyopathy.

Question: Do you recommend that all families with a new diagnosis of cardiomyopathy see a pediatric geneticist?

Answer: Ideally, all newly diagnosed children with cardiomyopathy should have a genetic evaluation. Depending on the level of experience of your cardiologist, this may be accomplished by having him/her perform the evaluation including a full evaluation of your family history or by referring your child to a clinical geneticist familiar with pediatric cardiomyopathies. For young children diagnosed with cardiomyopathies, children with cardiomyopathy that have additional medical problems/developmental delay/growth problems, and for any child with hypertrophic cardiomyopathy, it is especially important to be genetically evaluated since there is a greater likelihood that the cause is genetic. A genetic evaluation will assist with determining the prognosis, whether there is a specific treatment for the condition and risk for other family members/future children.

Question: How can you assess risk for family planning with cardiomyopathy? Does this vary from family to family? If so, what is the variation based on?

Answer: The risk definitely varies by family. Cardiomyopathies can be caused by viruses or toxins (i.e. not genetic.) The risk of recurrence in these cases would be essentially zero.

In other cases, it may be autosomal dominantly inherited with a 50% chance of having a predisposed child if a parent carries the genetic mutation. This is most commonly seen with Noonan syndrome and many of the isolated hypertrophic cardiomyopathies that start in teenagers.

In some cases, it may be X-linked with a 50% of recurrence in boys predominantly. Examples include Barth syndrome, Fabry disease and Duchenne muscular dystrophy. In other cases it may be autosomal recessive with a 25% risk of recurrence in girls or boys. Many of these cases are due to metabolic problems or inborn errors of metabolism in which the body cannot appropriately break down food substances and extract energy. Some of these have treatments available.

In rare cases, it is encoded by mitochondrial genes coming from the mother only, and the risk of recurrence is variable. Determining which of the above fits with your family often requires determining your child's underlying diagnosis and/or analyzing the pattern of inheritance in your family.

Question: The jargon of genetics can be overwhelming to families. Is there a website or tool you recommend to help families understand the complex world of genetics and inheritance?

Answer: This website is a good start: http://ghr.nlm.nih.gov/

Question: Both of my children and I were diagnosed with hypertrophic cardiomyopathy. We were all asymptomatic and have no known family history. Since the diagnosis, both kids' now present with hypertrophic cardiomyopathy (HCM), left ventricular non-compaction (LVNC) and possible dilated cardiomyopathy (DCM).

I have questioned whether we need to have genetic testing performed. Our pediatric cardiologist says that it is "obviously" familial so why bother with genetic tests? She also believes that it would be nearly impossible to identify the marker and would probably be a waste of time and resources. If both kids have the physical presentation of LVNC, wouldn't it be reasonable to assume that it came from somewhere and could be tracked genetically? Could I possibly carry the mutation and not the physical symptoms? My doctor tells me not to worry; as long as the physical symptoms aren't there it doesn't matter if the mutation is there or not!

One last note, my father passed away at 49 due to a cardiac issue of some sort. I have received his medical records from 1998 and there are several mentions of left ventricle hypertrophy, IHSS and septal hypertrophy. This leads me to believe that our HCM/LVNC possibly came from him. What are your thoughts on this? Am I wasting energy wondering if genetic testing could shed some light on the situation?

Answer: In general I find genetic testing to be most helpful in the following situations:

1. Evaluate an inborn error of metabolism for which there might be a treatment/cure.
2. Determine risk of recurrence and for family planning.
3. Determine who else in the family is at risk (which can also many times be done by echo.)
4. Help with prognosis, especially when this is not an isolated cardiomyopathy, and to answer the lingering question of what caused the cardiomyopathy.

5. Some day perhaps identify individuals at risk to allow for prevention of the cardiomyopathy. This is not currently possible but is the hope for the future.

In your case, the disease appears clearly to be familial with a 50% risk to the children of anyone who has cardiomyopathy. It does look like it came down from your father. The cardiomyopathy can have a different course and manifestations in different members of the family. This someday will provide important clues about how to prevent/treat cardiomyopathy by understanding why some people genetically at risk (like yourself) are asymptomatic.

Whether to test and when to test? Practically, in your family it probably won't make a huge difference in the near term for you or your children if this is an isolated cardiomyopathy since it is not likely to be an inborn error of metabolism. Genetic testing can be costly and may not be something all families can afford. Genetic testing might help with prognosis. If you have brothers or sister, they should have screening echocardiograms. As we learn more about the genetic basis of cardiomyopathy, we will be able to provide more/better prognostic information for the future for our children and grandchildren.

In part we can only learn these things as people get tested, so it's a bit of a CATCH-22. Therefore, although you might decide not to test now, you may want to test in the future or potentially join a genetic research study that will help to provide answers for the future.

Finally, your children may be concerned about passing this onto their children some day. Multiple options are available to ensure their children won't carry this if that is of concern to them, but these options can only be exercised if the genetic basis is known. It may take some time to figure that out, so give yourself plenty of time when you decide it's important.

Question: When our 6-month-old son was first diagnosed with hypertrophic cardiomyopathy (HCM) and Wolff-Parkinson-White Syndrome (WPW) we were told that genetic testing might someday help us learn why he had the disease (no family history that we can tell of), and whether our other son and we ourselves were at risk. They said this was about 10 years down the road.

Now that our son has had several "clean" echocardiograms, with no sign of hypertrophic cardiomyopathy, I think it is even more important to get genetic testing done to know what risk lies ahead. Obviously there is so much we don't know about this disease and we were warned it could come back at anytime — probably during the teenage years, if it does

Do you think it would be appropriate to have the testing done on someone with a

history of the disease in infancy but is now resolved? When do you imagine the genetic screening will be "good enough" to make it worthwhile to have the test on a child with no family history? Is there a better age to do this at?

Answer: If your son has/had absolutely no other medical problems besides the HCM and WPW and a normal family history documented by screening echocardiograms, the likelihood of finding a genetic cause given your current testing is probably 20% but not zero. There are specific genetic types of cardiomyopathy associated with WPW. Normal genetic test results would not rule out the possibility of an underlying genetic cause since our testing panels are not currently comprehensive.

The testing itself is only a blood test; however, the cost of testing is not inexpensive and you may have some out of pocket expenses even if insurance covers the testing. Therefore, your decision of if and when to pursue testing depends in part on how much you want to know this information. As a general rule, genetic testing improves with time; it gets faster, better, more comprehensive and less expensive. Good testing is available today, but better testing will always be available tomorrow.

The age at which testing is performed does not matter in the sense that the genes do not change over time. With time, your son could demonstrate additional clinical features that might suggest one diagnosis or another that would narrow the number of genetic conditions to consider.

Question: Can you say something about the relationship between a positive genetic test for hypertrophic cardiomyopathy (or perhaps any cardiomyopathy with a genetic cause) and the severity of the disease itself? In other words, to what extent does the particular mutation determine how the disease will actually develop in an individual, and what is the range of variation in that development?

Answer: If there is information known about the particular mutation for hypertrophic cardiomyopathy in a family, your doctor may be able to make some predictions about the clinical course. In addition, looking carefully at your own family and seeing who in the family carries the mutation and how they have done is useful. However, even with the same mutation and even within a family there can be variation in age of onset and severity that may be the result of other interacting genes, gender, treatment, physical activity and other factors we don't know. Thus, we can at best predict a range of possibilities. If, as is often the case, your family's specific mutation has never been observed before, careful studies of other members of your family are the most helpful means of clarifying prognosis.

Question: My son was diagnosed at 15 months with restrictive cardiomyopathy and transplanted at 18 months old. He had a genetic test done on him to check

for genes that are known to cause hypertrophic cardiomyopathy. He tested positive for a mutation in the Beta Myosin Heavy Chain. My husband and I were tested also, and my husband had the same results as our son. However, my husband had an echocardiogram last year, and it was found to be normal. Do you know of the relationship between the gene mutations my son and husband have and restrictive cardiomyopathy? Do you know of any genetic link between different types of cardiomyopathies?

My husband's only sister just found out she has the same gene mutation, and she is 27 weeks pregnant with a son with severe heart problems and hydrops fetalis. She also has never had any health problems. We can't understand why my husband and his sister are fine but our children are not.

Answer: It is unusual to have multiple asymptomatic adults in a family and a child who was symptomatic so young. It would be helpful to check other members of your husband's family if that's possible. It makes me wonder if the genetic variant reported in the beta myosin heavy chain genes was really a disease associated mutation or simply a benign normal genetic variation. Sometimes non-genetic doctors will misinterpret complicated genetic test reports and not realize the subtle distinction. It is also possible that your son (and perhaps your sister- in-law's fetus) have something in addition to the beta myosin heavy chain variant that was reported that has not yet been identified in another gene. Although not common, I have several families in whom there is more than one genetic factor contributing to a child's cardiomyopathy. In general, there is some overlap in genes for restrictive cardiomyopathy and hypertrophic cardiomyopathy and with dilated cardiomyopathy as well.

Question: My son, who is 2 years old, has mild hypertrophic cardiomyopathy (HCM) and Noonan syndrome. It has never been said that his HCM could be related to Noonan syndrome. I've read that a small number of Noonan syndrome children can suffer from HCM in addition to other specific heart defects, so although the numbers are on the lower side, there has been a link. At no stage has anyone mentioned whether or not it would be worthwhile having myself, my partner or my other 3 older children checked for HCM. It appears that since HCM can be a part of Noonan syndrome and since neither my son's father nor I carry the genetic defect for Noonan syndrome, that there is no point pursing this further. My eldest daughter also has a small problem with her pulmonary valve, but there has been no mention of any other problems or HCM anywhere with her.

Do you think it worthwhile for us all to be screened for HCM and heart defects in general considering that there is no genetic link between my son and the rest of our family but there are similar heart problems between the two siblings?

Answer: If I understand correctly your son has a proven molecular genetic diag-

nosis of Noonan syndrome due to a spontaneous mutation carried by neither you nor his father. Your son's mild HCM is almost assuredly caused by his Noonan syndrome and that alone. Given that no one else in the family carries the mutation, the likelihood there is a second cause for the HCM (given that it is mild) is remote.

However, I do have one patient with genetically proven Noonan syndrome with HCM who also has a mutation in another gene causing HCM that comes from his father. This is EXTREMELY rare, but it can happen. Therefore, to be safe I would screen the children, you and your son's father by echocardiogram but would anticipate that it is unlikely anyone else will have HCM.

Question: My daughter, age 5, was diagnosed with dilated cardiomyopathy and Autism Spectrum Disorder. We were told her case is rare because she also has a large ventricular septal aneurysm affecting two-thirds of her interventricular septum towards the apex. There is thinning of the septum and paradoxical motion. Some genetic testing was done. Her karyotype was normal; 46,XX and her FISH 22q11 deletion test was normal in that no deletion was detected. Her urine organic acid levels were normal. Another test was done called Microarray Comparative Genomic Hybridization, which also came back normal.

Would you recommend any other genetic testing that could be performed on my daughter? Have you seen any cases like hers where the cause was genetic? I have two other children that had echocardiograms done in addition to my husband and me. We are all fine. My husband has two other children from a previous marriage. Do you feel it would be important for his other children to be screened with echocardiograms?

Answer: It is difficult to assess the specifics of your daughter's situation without doing a comprehensive assessment. Based upon what you have said, her heart condition is quite unusual, and it is hard for me to know whether or not it is a true dilated cardiomyopathy. I would have her half siblings screened by echocardiograms. It appears that your daughter has had a good genetic assessment. There are additional tests for inborn errors of metabolism in addition to the urine organic acids that were performed. These are blood tests such as amino acids and acylcarnitine profile if your geneticist feels this is warranted based upon his/her assessment.

Question: Do you envision a time when there will be 100% conclusive tests for hypertrophic cardiomyopathy and dilated cardiomyopathy and other genetic diseases? Or, because of the multiple numbers of genes involved, will this be something not done in our lifetime?

Answer: Cardiomyopathy genetic testing for children is complicated because of

the number of genes that can cause cardiomyopathy in children, many of which we don't yet know. In addition, there may be interactions of multiple genes and environmental aspects as well. My hunch is that for genetic cardiomyopathies in young children, the genetics will be due to single genes. However, in older children or adults it could be more complicated. The real problem is that not enough research is being done on pediatric cardiomyopathy, so we know the cause of cardiomyopathy in only a small fraction of children. Hopefully, this will change with time which will allow us to develop genetic tests that are more useful for patients and families. It is rare that any genetic test is 100% conclusive, but that's our goal.

Question: My youngest son was diagnosed with neonatal viral myocarditis at 8 days old. He had all the classic symptoms. I ran a fever the day he was born so they assume he caught the virus from me. They cultured the coxsackie virus, I believe, from his nasopharynx. They did not do a heart biopsy because he was too critical to survive it. Our local pediatric cardiologist always told us that my son's dilated cardiomyopathy was caused by the virus and that the rest of the family was not risk. However, three years ago when we took him to another doctor for a second opinion, he told us that we could not be 100% sure that his dilated cardiomyopathy was caused by a virus since they did not perform a heart biopsy. He recommended echocardiograms for me, my husband and our 3 older boys (ages 14, 12 and 10.) We did this, and they were all fine. Do you think we need to have regular echocardiograms for the rest of us or can we assume that we are okay?

Answer: Unlike hypertrophic cardiomyopathy, dilated cardiomyopathy certainly is more likely to have a viral etiology. Given the age of onset of your youngest son and the normal echocardiograms in your older boys, you and your husband, I think you can be fairly confident this will be isolated to your youngest.

Question: My son was diagnosed with dilated cardiomyopathy at 3 months old. Prior to this diagnosis I had not heard of dilated cardiomyopathy (DCM), but my husband has Endocardiofobrielastosis (EFE). Is EFE the same as DCM?
We have gone through one round of genetic testing, of which the results came back "normal," and our geneticist says that there are no other tests to pursue. Do you know of certain hospitals/studies that have promising genetic testing for familial DCM? I have been told that no genes have yet been identified for DCM. Is that true, or has a gene been identified but it is hard to identify? My husband is adopted, so our family history ends there. My son's pediatric cardiologist has classified his condition as familial (with no medical proof), and is leaning towards the idea that it's carried in the male gene in our family since we have a daughter that is unaffected.

What kind of screening is recommended for siblings in a situation like ours? If

my son's condition was viral or idiopathic I would think one good echocardiogram from siblings would be enough, but since we feel sure that his condition is familial, we are not sure what kind of screening and how often for our other children.

Answer: Without knowing all the details of your case, if what you say is true, the cardiomyopathy in your husband and son is very likely to be genetic and I would guess autosomal dominantly inherited with a 50% chance for each of your children to have the condition. Screening should be done by echocardiogram. Whether or not a single echocardiogram or with what frequency this needs to be done depends on the particular presentation in your son and husband. If the heart condition is obvious early in life, your daughter may be out of the woods if her echocardiogram was normal.

There are several genes known for DCM, some of which may have been tested. EFE is not exactly the same as DCM, and there are some genes specifically for EFE such as Barth syndrome. My laboratory is doing research on familial forms of pediatric cardiomyopathy and would be glad to enroll your family if this is of interest.

Question: We have had two daughters with left ventricular non-compaction cardiomyopathy (LVNC). One of them died at 10 days old. Our second daughter was diagnosed at 4 weeks. Our son appears not to have LVNC. I have two daughters from a previous marriage who have also been echoed and do not appear to be affected.

Our second daughter is now 15 months old and has grown normally; her weight and height is in the 90th percentile. Her development (walking, talking etc.) has been age-appropriate, and she has no other health issues at this point. She is on medication, including beta-blockers, digoxin and ace inhibitors which appear to have had a positive effect on her well being and heart function. My first daughter who had LVNC and died was extensively examined from a metabolic standpoint, and nothing was revealed. She was also tested for mitochondrial disorders and that was negative. As a result, our second affected daughter has not had any of these investigations. They assume that her case would be the same as her sister's, although the severity appears to be different.

What is the likelihood that my son and my daughters (half sisters) would pass LVNC on to children of their own? From your knowledge of the genetics of LVNC, would it be possible that the LVNC would manifest itself later in life in the children that appear to be unaffected. How could my husband and I pass this to our children if we do not appear to have it yet?

Answer: My best guess is that within your family the LVNC is probably autosomal recessive (i.e. both you and your husband are carriers but are and will con-

tinue to be unaffected.) I would predict that the risk of recurrence would be 25% for you and your husband. I assume the LVNC will manifest during infancy in your family and that your other children are therefore likely off the hook and probably won't pass this down to their children. LVNC is, however, a rare form of cardiomyopathy so these are only my best guesses based on a few families I have seen similar to yours for whom we are now trying to find a genetic basis in our research lab.

Question: My son was diagnosed with spongiform cardiomyopathy (LVNC) on day one of life. He is currently 11 months old and shows no symptoms and takes no medication. When I delivered him he was born with a low-grade fever and was brought to the NICU for a spinal tap for meningitis and testing for infection. All results were negative. Both my husband and I had echocardiograms which showed no issues and no LVNC. Neither my husband nor I has heart condition issues in our family. With that in mind, my question is related to my husband and I wanting to have another child. Since my son's case seems to be an isolated issue, should we have genetic testing performed in order to plan for our second child? Will it really make a difference? Should my son have genetic testing done now to know what the future would be like for his children, or should we wait until he is older? I ask because I wonder if things do improve with his heart that it will be "too late" to know what the original problem was.

Answer: Given that you are seeking information to plan for a second child, it would be important for your son to have a genetic evaluation and try to determine a genetic diagnosis. If no genetic diagnosis is identified, I would recommend considering a series of fetal echocardiograms — although these may not pick up a heart problem until late in the third trimester or possibly not until after birth.

If you are only concerned about your son's future children, that can wait until he is older. His genes will not change over his lifetime so whether he is tested now or when he is an adult, his genetic information will be the same.

Question: Both of my children have been diagnosed with left ventricular noncompaction (LVNC) in addition to hypertrophic cardiomyopathy (HCM). The HCM was the original diagnosis, and both were screened several times before the LVNC presented. They have different fathers so I am the common link. I have no symptoms of LVNC but do have HCM. It has been suggested that I obviously carry some sort of LVNC genetic issue. What are your thoughts?

Answer: I agree that you are the common link. Within your family it appears there is an autosomal dominant gene that predisposes to cardiomyopathy. The manifestations appear to differ a bit by individual and perhaps age, but I presume it is the same genetic basis in all three of you.

The Spectrum of Grief as it Relates to Cardiomyopathy

Judith E. Brady, Ph.D. – July 2007

Dr. Judith E. Brady, Ph.D., completed her graduate work at the University of Minnesota Institute of Child Development in 1983 and is currently a pediatric psychologist in the Department of Pediatrics and Human Development at Michigan State University. Dr. Brady's area of clinical and research interest is in how children, adolescents and their families cope with the challenges of acute and chronic medical conditions.

Question: My husband and I have three children ages 6, 5 and 2. Our middle child was diagnosed with autism in May 2005 and then dilated cardiomyopathy. Our third child was born just one month after our daughter was diagnosed with Autism. Since all of this has happened, it's almost like we are afraid to have fun and enjoy life. I'm not sure if other parents feel this way. I've lost friends over all of this because they just don't understand about the autism and the heart condition. My daughter with cardiomyopathy looks "normal," not sick at all or autistic. People just don't get it. I've also battled depression and am currently taking antidepressants. My husband and I don't fight but we have gone to counseling. He has his own business and works long hours. He depends on me to manage everything when it comes to my daughter. I feel so alone because I have all of this on my shoulders, and nobody understands. Is there a way I can get past this or is this just part of having a sick and special needs child? There are so many times I go into her room in the morning afraid to find her not breathing. Does that feeling ever go away? Any ideas or suggestions would help.

Answer: Do you have opportunities to interact with other mothers of children with autism or children with autism along with other medical conditions? From my experience the feelings that you describe — the wondering if it is okay to experience joy, whether things will be normal again — may be a part of having a special needs child but need not be the whole story of having a special needs child. The story is very different for each mother, child and family. For some, that dread when going into the child's room never completely goes away, but other feelings are experienced such as the excitement of whatever is planned for the day or the smile at seeing your child sleeping in a funny position. These other feelings can be acknowledged and can co-exist alongside the dread. Joy can reenter. Some mothers talk about finding a new "normal." One can't really go back and recreate circumstances that preceded any event (be it sad or happy) but we can have a say in creating a new "normal" that may permit experiencing a broader range of feelings.

Depression, too, can put a grey cloud over life and significantly interfere with one's ability to think creatively, to be flexible and to experience a range of emo-

tions. Recognizing that and taking care of that for yourself is so important for yourself, your children and your marriage and something many mothers do not give themselves permission to do.

Question: My daughter is 17 months old. She has a familial form of cardiomyopathy, which means that my husband and I have passed the disease on to her. We lost a daughter (at 10 days old) 5 years ago from the same disease. My daughter is not showing any outward symptoms of cardiomyopathy but is on medication. She has a fairly unusual form of cardiomyopathy; therefore the outlook for her is unknown. Our specialist will not speculate, and we do not know what will happen to our daughter as she grows older. I struggle with this since I am a very factual person and like to know what is happening next. From day to day, I do not worry obsessively about her, as I rationalize it in this way — something may happen to any child at any time and there is no telling when or how it will happen. However, there is a deep dread in me, as I know from experience how unspeakably horrible it is to lose your child. I do not want my family to go through that again nor do I want my daughter to have a life which is anything less than what she could have. As a result, I feel that I am either one of two things, in denial or existing on hope. Do you have any suggestions for helping me come to terms with our life, which like all parents in this situation, is lived in limbo not knowing what will happen.

Answer: I think that living in that place between alternate denial and hope is a very understandable experience to any parent who has lost or who must live with the possibility of the loss of a child. I am not certain what for you will constitute "coming to terms" with your life. That is likely different for all of us. Do you have a sense of what your life would look like were that to be the case?

Many parents I have met speak of coming to terms as if that were a point in time past which all would somehow be different. Yet I am not sure that such a moment in time happens for most. Coming to terms is a process that one can feel some relative peace one day only to have something come along the next day to challenge it. On the other hand, one can be truly struggling and then experience an event, a developmental milestone or life event, and there can come joy and comfort.

From my experience both the denial and hope have bases in the reality of one's situation and have live sustaining functions. That is keeping us vigilant to the needs of our children and allowing us to cherish them and share with them life as they discover it.

I suspect I am telling you nothing that you haven't already told yourself. Having some insight into what you are experiencing, accepting those feelings as normal and understandable and having someone or someplace to share is a core part of

the coming to terms process. One mother described the fine line she walked on as this — she could spend each day dreading something terrible only for nothing to happen and to feel that she had not enjoyed the time she had. Or, she could recognize the uncertainties and also enjoy each day so even if something dreadful occurred she would be able to look back at the joy she had experienced along the way.

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