To Whom It May Concern:

The Hypertrophic Cardiomyopathy Association is a not for profit organization providing information and support to those affected by HCM, their families, and the medical community. The following is the HCMA’s views on screenings for HCM and basic management of the condition. In the event of the diagnosis of HCM in a family or an HCM related death of a family member, we strongly recommend that all blood relatives be screened. This screening MUST include an EKG/ECG, Echocardiogram, and a cardiac check up by a cardiologist. Ideally the cardiologist should have specialized knowledge of hypertrophic cardiomyopathy. Due to the fact HCM is a rather ‘rare’ condition it may require the patient seek out a specialist in the field of HCM. In many cases this will require travel out of state and in some cases out of the country.

Hypertrophic cardiomyopathy may be suspected because of family history, symptoms, a murmur or an abnormal EKG/ECG. Many symptoms or signs of hypertrophic cardiomyopathy are similar to various other conditions; therefore, it is important to follow doctor’s instructions on complete testing to assure accurate results. An EKG/ECG records the electrical signals of the heart. In hypertrophic cardiomyopathy the EKG/ECG usually shows an abnormal electrical signal due to muscle thickening and disorganization of the muscle structure. In a minority of patients (approximately 10%) the ECG/ECG may be normal or show only minor abnormalities. EKG/ECG abnormalities are also not specific to hypertrophic cardiomyopathy and will be found in many other conditions.

Presently, the diagnosis of hypertrophic cardiomyopathy is made by an ultrasound scan of the heart called “echocardiogram” or “Echo” for short. Like the EKG/ECG, this is an entirely safe test. Excessive thickness of the muscle can be easily measured. Additional equipment called “Doppler” ultrasound can produce a color image of the blood flow within the heart and measure the contractions and filling of the heart. Turbulent flow can be detected using the doppler. Therefore, Echo/Doppler provides a very thorough assessment of hypertrophic cardiomyopathy.

After a diagnosis is made several other tests may be required. Holter monitoring is a continuous recording of the heartbeat over a period of 24 to 48 hours. A holter monitor is a simple and safe test that will detect irregularities of the heart beat (otherwise known as arrhythmia). Stress tests or Stress Echo’s may be used to monitor the heartbeat during exercise as well as blood pressure response and then the echo to see if there have been any structural changes due to the exercise. MRI has been used in more frequent years to gain a more detailed image of the heart. Cardiac catherization may follow but it is not used on all patients with HCM.

The following article was written by Dr. Barry Maron of the Minneapolis Heart Institute Foundation and was in a past HCMA newsletter:

Since HCM is usually a genetic disease, we are often asked, in the context of the family evaluation, how screening for the disease should be carried out. Laboratory genotyping (using 10 cc of blood) would, of course, be the most definitive approach to identifying any individual with the disease (i.e. gene defect) — theoretically. However, such laboratory techniques are expensive, laborious, not routinely available, and do not guarantee a positive answer. Genetic testing is presently confined to a very small number of research laboratories that work on highly selected pedigrees (families) only for investigational purposes. Therefore, such family screening is still best carried out as it has been for many years — with two dimensional echocardiography often performed serially (i.e., more than once) in growing children from families with HCM. The purpose of echocardiography (which is non-invasive, painless and has a "retail cost" in the range of $600.00) is to identify what clinicians refer to as the phenotype or overt expression of HCM — i.e., the thickening of the left ventricular wall, usually of the ventricular septum (the portion of the wall separating the left and right ventricular chambers).
In general terms, the heart wall thickening is not usually evident before the age of 10 and is most likely to be detected after the age of 12. Wall thickness usually increases as the child progresses through puberty with accelerated growth. The change in thickness can be abrupt and striking and therefore the appearance of the heart can change completely between 12 and 14 years of age — often progressing from completely normal to very thick. Experts believe these changes of hypertrophy, while often alarming to the family and even some physicians, represent the normal pattern with which the heart forms in HCM and does not present either deterioration, an alarming clinical sign, or a warning of imminent danger. If the wall thickening becomes evident on the echocardiogram — and is unexplained in other ways, such as by athletic training, other diseases, etc. — then it may be assumed to represent a gene mutation responsible for HCM. Based on the available evidence, we believe that with limited exceptions, if the hypertrophy is not present by the time full growth and maturation is achieved (about age 17-20 years) then it would be less common for it to happen later. Therefore, if an individual is an HCM family is “echo-negative” by the time of adulthood, and then there is a chance of not being affected by the gene. However it is possible to develop HCM at any point in life, therefore the recommendation to continue screening every 5 years throughout life.

But when should echocardiograms be performed in children in families with HCM? Screening echocardiograms before the age of 10 or 12 are optional since these studies are rarely positive at this time, even in the presence of an HCM gene mutation, and recognition of the disease at this age would not necessitate intervention. Exceptions to this would be in selected families with multiple occurrences of premature death due to HCM or if the child is truly a competitive trained athlete. In such instances echocardiographic studies would appear obligatory at young ages, since HCM is the most important cause of sudden death during sports in young people, and a reason for disqualification from training and competition.

Otherwise, we recommend serial echocardiograms about every 18 months or so (every 12 months if a trained athlete) throughout adolescence or until the echocardiogram “converts” from normal to abnormal. Strangely enough, the standard electrocardiogram (EKG) may be abnormal in a genetically affected child long before the echocardiogram changes from normal to abnormal. Recent data does show the possibility of HCM appearing later in life if far more common then previously thought. With this new knowledge it is now suggested that children / young adults continue screening until age 25 on the every 12-18 monthly basis then switch to every 5 years through adulthood.

What to tell your family members about screenings for HCM

This can often be a difficult and stressful topic for discussion. A first great step is to make copies of this letter and give it to family members. What they need to know is rather simple; all first-degree relatives of the affected person should get screened as soon as possible. Example: if your husband is diagnosed with HCM his parents, siblings and children should all be screened. Screenings must be done annually from the onset of puberty (est. age 9-12) through 20, and every 5 years age 25 and over. Screenings must include an ECG (EKG), echocardiogram and a visit to a cardiologist.

In addition to the need for accurate screening is the need for careful case management. It is strongly suggested that once diagnosed with HCM the patient consults with a specialist in the field. There are only a few HCM specialists worldwide: the United States is home to many of them. For more information on these specialists, we suggest you contact the HCMA for names of those doctors closest to your home.

If you are having a difficult time explaining the special concerns of living with HCM to your insurance carrier, send them a copy of this letter and feel free to have them contact the HCMA directly. Most major insurance companies understand the importance of preventative care and careful case management.

If you have any further questions about screening for HCM or the management of HCM, please contact the HCMA office at 973-983-7429.

Sincerely,

Lisa Salberg

Lisa Salberg, President