Dear Family Member,

If you are reading this letter, it is because I care about you and want you to have important information about my health, and maybe your health too. I have been diagnosed with hypertrophic cardiomyopathy (HCM). This letter was created by the Hypertrophic Cardiomyopathy Association, which is a non-profit organization (501 C3-41-1855605) that provides information, support, and advocacy services to those affected by HCM, their families, and the medical community. The purpose of this letter is to help me explain what HCM is, why you should be screened, and what you can do to take care of your heart. The following information has been compiled from scientific journals and expert input on who, when, and how people should be screened for HCM.

What is HCM?

HCM is a genetic cardiac disorder that causes the heart to become thick, and may cause structural changes and electrical disturbances in the heart. HCM can lead to sudden cardiac arrest and heart failure if not managed properly. ***If one member of the family has HCM, it is highly likely that others do as well.*** This is an autosomal dominant genetic disorder meaning you only need one copy of the gene to have HCM transmitted to you. HCM is actually very common. Over 1 in 400 people (that is nearly a million people in the USA and tens of millions worldwide) have the disease or carry the gene for it.

Why do you need to be screened?

You should be screened because I have a diagnosis of HCM. It is strongly recommended that, once there has been either a diagnosis of HCM or an HCM-related death in a family, ***all blood relatives in previous generations (parents), your generation (siblings), and the next generation (children) should be screened.***

What does HCM screening include?

Screening for HCM includes a cardiac check up by a cardiologist, who, ideally, has specialized knowledge of HCM. You will also need an electrocardiogram and an echocardiogram; these tests will examine the electrical function and anatomy of your heart. The cardiologist will also review your family history and symptoms, and may require other testing to verify or rule out a diagnosis of HCM.

HCM 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 your doctor's instructions on complete testing to assure accurate results. HCM is often misdiagnosed as asthma, an innocent heart murmur, mitral valve prolapse (MVP), or panic/anxiety attacks.

Further tests may be ordered for a more detailed look at your heart and how it reacts to exercise. The following tests, however, should be performed on all first degree and likely second degree relatives of those with HCM:

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- **EKG/ECG**-Records the electrical signals of the heart.
  - In HCM, the EKG/ECG usually shows and abnormal electrical signal due to muscle thickening and disorganization of the muscle structure.
  - In a minority of patients (approximately 5-10%), the EKG/ECG may be normal or show only minor abnormalities.
  - EKG/ECG abnormalities are not specific to HCM and will be found in many other conditions.
- **Echocardiogram (Echo**)-An ultrasound scan of the heart
  - This is an entirely safe and noninvasive test. Excessive thickness of the muscle can be easily measured with an echo, in most cases.
  - 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.
  - Echo/Doppler provides a very thorough assessment of hypertrophic cardiomyopathy.
- **Cardiac MRI (CMR)**- Cardiovascular Magnetic Resonance Imaging
  - Non-invasive
  - In some cases this is used to gain a better view of the heart
  - It is important that the cardiac MRI be done by a center with a protocol specifically for the HCM heart to ensure proper views.

Where should HCM screening should be performed?

A cardiologist's office or medical center can conduct a complete screening for HCM. However, because HCM is a rather poorly understood condition, you may need to seek out a specialist in the field of HCM for accurate screening. The HCMA maintains a list of HCMA Recognized Centers of Excellence Programs on their website, www.4hcm.org.

What are doctors looking for when they perform these tests?

In general, the heart wall thickening is not usually evident before the age of 10 (but can be), 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. 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, while often alarming to the family, and even some physicians, represents the normal pattern with which the heart forms in HCM; it does not indicate deterioration, an alarming clinical sign, or a warning of imminent danger. Based on the available evidence, it is believed that with limited exceptions, if the hypertrophy is not present by the time full growth and maturation is achieved (about 22 years of age), then it would be less common for it to happen later, but it does. It is, however, possible to develop HCM at any point in life, so the recommendation is to continue screening every 3-5 years throughout life.

Is there a genetic test available to see if you are affected?

In some families, genetic testing on the ***index person***, or the first person with a clinical diagnosis, can be performed to identify the mutation responsible for HCM. If a family member has had genetic testing, and a mutation was found, you might benefit from also have genetic testing performed.

- ☐ YES-I have had genetic testing- Contact me if you would like to discuss the results
- ☐ YES- I have had genetic testing, but they were unable to find the mutation responsible
- ☐ NO- I have not had genetic testing

What do you do if you find out you have HCM?

In addition to the need for accurate screening, there is a 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 specials worldwide; the United States is home to many of them. For more information on these specialists, we suggest you contact the HCMA or visit www.4hcm.org for the names of those doctors closest to your home.

If you have any questions about HCM, please contact the HCMA at support@4hcm.org or call them directly at 973-983-7429. They will be happy to help you!

Sincerely,

Your big-hearted family member.