The Genetics and Semantics of Hypertrophic Cardiomyopathy

To the Editor:

We read with interest the article by Maron et al.1 We would like to commend them for utilizing the term, "hypertrophic cardiomyopathy," in this report, instead of the term, "asymmetric septal hypertrophy," which has now proven rather nonspecific. We were also pleased to note that Maron et al1 now recognize that asymmetric septal hypertrophy is not pathognomonic of hypertrophic cardiomyopathy.

Nevertheless, we are concerned about the use of the term, "disproportionate ventricular septal thickening," which seems to further confuse the semantics. It would appear that there may be some difference between disproportionate ventricular septal thickening and asymmetric septal hypertrophy, particularly in view of the following opening statement in a previous report: "Asymmetric septal hypertrophy (ASH) or hypertrophic cardiomyopathy, is a genetically transmitted disease of cardiac muscle that is characterized by disproportionate thickening of the septum.1"(2,9,20) This statement was supported by several references from the same group,2-5 yet the difference is not clear, as both terms are defined as a septal-free wall ratio of 1.3 or more.1,3

The finding in this article that disproportionate ventricular septal thickening or asymmetric septal hypertrophy occurs in 6 percent (2/33) of the patients with systemic hypertension is considered by Maron et al2 to reflect a relatively low incidence; however, since hypertension is found in 15 to 30 percent of the US population,6 the number of patients with disproportionate ventricular septal thickening secondary to high blood pressure may exceed the number of patients with asymmetric septal hypertrophy associated with hypertrophic cardiomyopathy. The relative prevalence of "genetic asymmetric septal hypertrophy" would be further diminished if we consider the patients with nongenetic asymmetric septal hypertrophy and coronary arterial disease6 or congenital heart disease.7

Since the initial genetic studies4 were performed at a time when asymmetric septal hypertrophy was believed to be specific for hypertrophic cardiomyopathy and when the occurrence of this entity with symmetric hypertrophy8,9 was not yet recognized, one wonders whether the opening statement that "hypertrophic cardiomyopathy is a genetically transmitted disease of cardiac muscle that is characterized by asymmetric septal hypertrophy1"(2,4,6,8) is still appropriate? A clarification by Maron et al9 of their present understanding of the genetics and semantics of hypertrophic cardiomyopathy in the light of their recent data would therefore be very helpful.

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REFERENCES

To the Editor:

We are in agreement with Chahine and associates that one of the barriers in communicating information regarding hypertrophic cardiomyopathy has been the confusion resulting from the multiplicity of terms used to describe this disease. Furthermore, we regret it if we have contributed to this problem by introducing yet another descriptive term, "disproportionate septal thickening (DST),"7 in several publications,1,5-8 including one that recently appeared in Chest.5

Perhaps a historical perspective would be helpful in explaining the factors that led us to introduce yet another term to an area of investigation already overburdened in this respect. The application of echocardiography to cardiac diagnosis in the early 1970s demonstrated that a hypertrophied ventricular septum which was at least 1.3 times the thickness of the left ventricular free wall was present in virtually all patients with hypertrophic cardiomyopathy. This observation led to introduction of the term, "asymmetric septal hypertrophy" (and the acronym, "ASH"), to describe the disease, hypertrophic cardiomyopathy.6

Nevertheless, as Chahine and associates point out and as our echocardiographic and postmortem data in over 900 hearts, substantiate,1-8 ASH is not pathognomonic of hypertrophic cardiomyopathy. Specifically, we have found that a septal-free wall thickness ratio of 1.3 or greater is present in about 10 percent of the patients with a variety of congenital or acquired heart diseases. When such an abnormal septal-free wall ratio occurs in these patients, the septal thickening usu-
ally is found to be secondary to the patient's underlying lesion and not a manifestation of a coexistent primary cardiomyopathy.

Since the term, "asymmetric septal hypertrophy (ASH)," had already been appropriated for the name of the genetically transmitted primary cardiomyopathy, these findings led to a dilemma in nomenclature, ie, how to refer to the type of disproportionate septal thickening that occurred secondary to other types of heart disease. Hence, the descriptive term, "disproportionate septal thickening (DST)," was introduced1-3 to describe the secondary form of "asymmetric septal hypertrophy." While ASH and DST were thus used to describe the same gross anatomic abnormality (a hypertrophied ventricular septum that was at least 1.3 times the thickness of the left ventricular free wall), each term had a very different meaning; ASH implied the existence of a primary cardiomyopathy, while DST implied a secondary form of septal thickening.

While we now believe that the most appropriate name for the primary cardiomyopathic disease is hypertrophic cardiomyopathy, we continue to use the acronym, "ASH," to specifically describe the asymmetrically thickened ventricular septum in patients with hypertrophic cardiomyopathy and to use the term, "disproportionate septal thickening (DST)," to refer to the same gross anatomic abnormality present in patients with other forms of heart disease. Put another way, "DST" is characterized by (1) ventricular septal to left ventricular free wall thickness ratio of 1.3 or more, (2) the presence of congenital or acquired heart disease (ie, valvular, coronary, or hypertensive), and (3) no evidence of genetic transmission.1-9

We therefore agree with Chahine and associates that the sentence, "asymmetric septal hypertrophy (ASH) or hypertrophic cardiomyopathy, is a genetically transmitted disease of cardiac muscle that is characterized by disproportionate thickening of the ventricular septum,"2(1925) is potentially confusing; however, we hope that we have put the sentence into historical perspective and also hope that this letter serves to clarify ambiguities in nomenclature.

Finally, we concur with the point made by Chahine et al that in the US population the number of patients with disproportionate septal thickening secondary to systemic hypertension may exceed the number of patients with hypertrophic cardiomyopathy. We believe that this possibility emphasizes the importance of our observations regarding disproportionate septal thickening in hypertension (as well as in a variety of other relatively common congenital or acquired heart diseases).

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9 Maron BJ, Epstein SE, Roberts WC: Cardiac muscle cell disorganization in the ventricular septum: Evidence from quantitative histology that it is a highly sensitive marker of hypertrophic cardiomyopathy (abstract). Am J Cardiol 41:435, 1978

Correction
To the Editor:

Flipping through the pages of the newly delivered July 1978 issue of Chest, I was pleasantly surprised to note the heading, "Illustrative Electrocardiogram," on page 98; however, my interest, aroused by my deep involvement in electrocardiography, was short-lived when, on reading the article, I realized that what appeared to be the beginning of a new series on electrocardiology was actually an old series on echocardiography. There was some consolation in noting that the table of contents on page A-9 did not lie. Although I may have previously pointed out typographic errors in your journal (Chest 72:129, 1977), my communications to you are not aimed at acquiring the label of a typographic critic. I enjoy reading your publication very much and treasure each issue for future reference in my office.

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Who Should Be Trained To Do Fiberoptic Bronchoscopy?

To the Editor:

In his article entitled "Bronchoscopy Training" in the May 1978 supplement (Chest 73:776-778, 1978), Faber has left out the anesthetist as a potential candidate for learning and performing endoscopic procedures. Performing tracheal intubation in daily anesthetic practice and respiratory care is usually a simple procedure. When anatomic alterations make visualization of the larynx an exceedingly difficult task, the options and safety provided by the fiberoptic bronchoscope make it superior to other techniques advocated for difficult intubation.1,2

When faced with a difficult intubation during induction of anesthesia for emergency surgery, familiarity of the anesthesiologist with the use of the fiberoptic bronchoscope would prevent a disastrous outcome. To perform in stressful situations, he should receive supervised training and practice on..."