Letters to the Editor

Emergency Physicians Also Knocking on the Door of Echocardiography

Los médicos de urgencias también llaman a la puerta de la ecocardiografía

To the Editor,

We thank Garcia Fernandez for his excellent editorial on echocardiography performed by noncardiology specialists.[1] We would like to add that emergency physicians are also a group of professionals with a strong interest in improving the quality of our care through the application of noninvasive techniques such as basic clinical echocardiography, particularly for assessing patients with acute heart failure. In 45% of admissions, no prior echocardiographic information is available. Although this lack of information is not associated with a worse prognosis in the emergency room, we believe that it may lead to underuse of basic treatments with a recognized prognostic benefit.[2] It would therefore seem to be common sense to introduce basic bedside echocardiography in the assessment of acute heart failure for certain selected patients.[3] Thus, in our tertiary university hospital, which serves 1.5 million inhabitants, we have set up a training program in basic echocardiography with the following aims: detection of pericardial fluid, subjective estimation of ventricular function, assessment of presence of segmentary wall movement disorders, and measurement of chamber size. The training comprised a part dedicated to theory (8 hours covering the principles of ultrasound scans and basic echocardiography) and a part to practice (25 to 30 echocardiographic studies under the supervision of cardiologists who work in the emergency room). The degree of satisfaction and acceptance of the technique was excellent; the extent to which the technique was applied in clinical practice was high. The main objective of echocardiography was subjective estimation of left ventricular function in 49%, detection of pericardial fluid in 33%, and assessment of shock and intravascular volume in 18% of cases. We also found that the learning curve was very steep, with appropriate choice of axis and identification of pericardial fluid in the first 5 echocardiographic studies. In addition, even in this short training period, participants were able to make a subjective assessment of cardiac contractility, segmental contraction defects, and dilation and size of atria and ventricles. All this information may help decide whether a patient with acute heart failure and previously undetected impaired ventricular contraction may benefit from a full work-up in the cardiology department. We should, however, be aware that our basic echocardiographic studies are in no way a substitute for those performed by specialists. We cannot stem the tide.

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Emergency Physicians Also Knocking on the Door of Echocardiography. Response

Los médicos de urgencias también llaman a la puerta de la ecocardiografía. Respuesta

To the Editor,

I appreciate the response by Jacob et al to my editorial published recently in *Revista Española de Cardiología*[1] on echocardiographic training for noncardiologists. Their letter is a clear demonstration of the need to regulate the practice of echocardiography by nonspecalists, as well as the training these noncardiologists should receive and the scope of their echocardiographic studies. This needs to be done as soon as possible to ensure that the training for each particular need does not descend into chaos and that the physician acquires the necessary knowledge to attain competence in an orderly fashion.

As emergency physicians, the authors have identified a problem (the need for echocardiographic training) and have attempted to solve it as best they could with an empirical approach. In the absence of current guidelines, they decided, in agreement with cardiologist colleagues, on the number of hours of training, the scope of training, and the specific skills needed to record an echocardiogram in their specific area. And they did this because there were no regulations or established levels of competence required for their daily practice.

Without doubt, things should be different, and they should not need to take the initiative themselves. Rather, our Society, through its Imaging Working Group and based on experience, should provide training guidance according to the different needs, as I proposed in my editorial. I recommend the autors carefully read an attractive, and in my opinion, fantastic proposal from the influential Italian Society of Echocardiography,[2] which reclassifies the definition of echocardiography according to different levels of training, competency, and use. The society defines 4 types of use of

echocardiography, which require completely different training, competencies, and application. The first is the basic level (ultrasound for initial diagnosis, determining the extent of disease, and examination). The second is general echocardiography (transthoracic echocardiography, transesophageal echocardiography, stress tests, and 3-dimensional echocardiography). The third type is application of echocardiography in specific situations (intraoperative, catheterization laboratory, coronary artery disease unit, critical care unit). Finally, there is the application in the emergency room, with echocardiography performed on admission of the patient to the emergency room or out-of-hospital in an emergency situation. Obviously, each level is regulated with homogeneous training to obtain appropriate competency in each case.

To ensure a rational use of echocardiography and so to provide the best care we can for our patients, I suggest that the Spanish Society of Cardiology take up the challenge and establish training criteria according to the needs of each specific area of use, with particular attention to uses outside the cardiology department.

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Usefulness of Genetic Diagnosis in a Woman With Hypertrophic Cardiomyopathy and the Desire for Motherhood: Information Is Key

Utilidad del diagnóstico genético en la miocardiopatía hipertrófica de una mujer que desea ser madre: la información es clave

To the Editor,

We would like to congratulate Villacorta et al[1] for their letter recently published in this journal, but we think it is appropriate to add some caveats.

The authors report the case of a patient with hypertrophic cardiomyopathy (HCM) who wanted to become a mother. To avoid transmitting the disease to her offspring, she requested a preimplantation diagnostic test. The genetic study detected 2 mutations in the *MYBPC3* gene: a previously unreported truncating mutation (Asn1023Lysfs*28) and a previously published missense mutation (Gly5Arg). Of the large family shown in the family tree, data are only presented for her parents and a brother. Each parent is a carrier of 1 of the transmitted mutations and her brother is a carrier of the truncating mutation. Given that the patient is a carrier of 2 mutations, each inherited from a different parent, the probability of transmitting at least 1 mutation would be 100%. The authors considered both mutations to be pathogenic and therefore recommended not to proceed with the preimplantation diangosis.[1]

We agree that the Asn1023Lysfs*28 mutation can be considered pathogenic. Several mutations have been reported in the same functional region of the *MYBPC3* gene with a similar mechanism, and all were associated with HCM. We have identified this mutation in a female patient with HCM.

However, we are more hesitant to consider the Gly5Arg mutation as pathogenic. This mutation has been reported in at least 7 publications on 7 carriers from 5 different families. The index cases were 3 patients with HCM, 1 with dilated cardiomyopathy and 1 with noncompaction cardiomyopathy. However, the publications do not present a detailed description of the patients and their families. Thus, for example, one of the patients with HCM was a carrier of another *MYBPC3* mutation (Arg502Trp, a known pathogenic mutation),[2] the family members of the patient with dilated cardiomyopathy were not genotyped (and therefore we cannot know whether the mutation cosegregates in the family),[3] another patient with HCM had right ventricular hypertrophy (very infrequent in sarcomeric HCM), and there is no information on a family study.[4] In all patients the genetic studies were incomplete (few genes were studied, and therefore mutations in other genes may have been present).[2–5] We have detected a heterozygous Gly5Arg mutation in a newborn child with severe hypertrophy who died at the age of 1 month. In addition, this patient had a mutation in the *GAA* gene, causing Pompe disease. When we studied the family, we found that Gly5Arg did not cosegregate with the disease.

We searched for information on the Gly5Arg variant in public databases such as the Exome Sequencing Project,[6] which contains information on genetic studies in the healthy population (without cardiomyopathy), and found that it has been identified in 7 out of 4159 Caucasian Americans (0.16%). If the prevalence of HCM in the general population is 1 in 500 (0.2%), Gly5Arg alone would have a prevalence close to that reported for the entire disease. Therefore, we believe that Gly5Arg is an uncommon polymorphism in the general population and that its pathogenicity should be placed in doubt. This polymorphism may have a modifying effect in the presence of another mutation, but it is unlikely that it is pathogenic by itself.

This case requires a reflection on the interpretation of genetic studies. Their usefulness is clearly demonstrated and supported by current clinical guidelines. However, we should treat the results critically and not consider a genetic variant as a pathogenic mutation merely because it has been published previously. We should also take into account the number of publications, the number of genes studied, the number of carriers (symptomatic and healthy), the presence of complete family studies, and whether additional clinical information, functional studies, etc, are available. The public databases (Single Nucleotide Polymorphism Database, Exome Sequencing Project, etc) are very useful, as they provide information on the presence of these variants in thousands of controls.

Finally, we agree with the authors that there is a need for cooperation among different scientific societies to reach a consensus on which types of disease can be screened in a preimplantation diagnosis.

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