Short QT Syndrome Associated with Noncompacted Myocardium

Cardiomyopathies are a heterogeneous group of myocardial diseases associated with mechanical or electrical dysfunction that usually (but not invariably) exhibit inappropriate ventricular hypertrophy or dilatation and are due to a variety of frequently genetic causes. Cardiomyopathies are confined to the heart or are part of generalized systemic disorders, which may lead to cardiovascular death or progressive heart failure-related disability. (1)

Noncompacted myocardium is a primary genetic cardiomyopathy first described in 1984 by Engberding, characterized by myocardial hypertrabeculation with sinusoids in communication with the ventricular cavity, resulting from a sudden arrest in the normal embryogenesis prior to compaction, and involving predominantly the distal portion of the left ventricle. (2)

Short QT syndrome (SQTS), introduced as an association with sudden death (SD) in 1993 by Algra, but first described by Ghusack in 2000, who reported 6 unrelated family cases with atrial fibrillation as the form of presentation, is a primary cardiomyopathy described within the channelopathies generating electrical dysfunction. (3) Diagnosing SQTS is not easy, since in a first instance, numerous studies associated corrected QT interval (QTc) <300 ms with SD, but then, the limit point proposed was 2 standard deviations from the normal QTc (350 ms), so the cut-off point to consider a short QT interval would be 320 ms. In 2011, Gollob et al. proposed a diagnostic score based on QTc, the JT interval, the genotype, and the patient’s medical record and his/her family history, assigning points to each category. An overall score of 4 points or greater indicates a high-probability of SQTS, 3 points an intermediate probability and 2 points or less a low probability of SQTS. (4) In 2015, the European Society of Cardiology (ESC) standardized criteria and determined that the presence of QTc <340 ms (Class I, level of evidence C), or a QTc <360 ms with at least one associated genetic mutation, family history of SQTS, family history of SD before the age of 40, or survival from an episode of ventricular fibrillation (VF) or documented ventricular tachycardia (VT) in the absence of structural heart disease (Class IIa C) constitute diagnosis of SQTS.

A 19-year-old male patient, without known personal pathological history, with a history of a 4th degree relative (cousin) who died of SD at the age of 1 year, and a 2nd degree relative (maternal uncle) who also died of SD at the age of 15 months, consulted for paroxysmal palpitations and dyspnea in FC II-III associated with an episode of chest pain. Sinus bradycardia at 40 bpm was detected, with adequate chronotropic response.

The ECG showed absolute QTc of 320 ms, and corrected by Bazett’s formula of 292 ms (Figure 1). The echocardiogram revealed myocardial hypertrabeculation in the mid-apical region of the low and lateral walls, suggestive of noncompacted myocardium (Figure 2). The MRI reported preserved left ventricular size and function and normal myocardial wall thickness. Within the intraventricular cavity, abundant trabeculae in the lateral-apical region and apical segment were observed (Figure 1), with a relation between noncompacted and compacted areas of 2:4 (noncompacted area 16.5 mm, and compacted area 6.8 mm in a short axis view), meeting the diagnostic criteria for noncompacted myocardium (Figure 2).

The 24-hour Holter evidenced average HR of 45 bpm, minimum 33 bpm, maximum 94 bpm, without ventricular arrhythmia, maximum and minimum QTc of 375 ms and 276 ms respectively, and minimum absolute QT of 360 ms, with scarce daily variability despite high HR variability.

No familial phenotypes have been found in the screening for familial SQTS.

With the diagnosis of SQTS and the patient’s consent, a cardioverter defibrillator (ICD) was implanted.

So far, SQTS associated with noncompacted myocardium has not been described.

Short QT syndrome was diagnosed according to the 2016 ESC guidelines for ventricular arrhythmias and prevention of SD due to QTc <340 ms (class IC). Based on the Gollob score, we have a patient with high probability of SQTS (4 points QTc <330 ms, and second-degree family history of SD) without performing genetic testing. The yield of genetic testing for SQTS is low (14%). (4)

So far, noncompacted myocardium has been described in association with other conditions, such as long QT syndrome; recently, a novel entity has been introduced, which identified the mutation in the HCN4 gene that is a phenotypic manifestation of sinus node dysfunction, noncompacted myocardium, mitral valve prolapse, and aortic dilation. Of all the family cases analyzed, no patients with short QT have been reported. (5)

Moreover, SQTS has been associated with Brugada syndrome and early repolarization syndrome, with sig-
We describe for the first time this new entity that associates a structural cardiomyopathy (SCM) with a channelopathy (SQTS). It will be difficult to discriminate whether it is merely an incidental association or it is due to a genetic mutation and constitute its phenotypic manifestation, together with persistent sinus bradycardia. Follow-up and description of further cases, as well as family genetic testing of this novel syndrome will be central to answer this question.

Conflicts of interest
None declared.

Ethical considerations
Not applicable

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Abdominal Aortic Coarctation in Noonan Syndrome

Abdominal aortic coarctation or hypoplasia is a rare condition. Its incidence, evaluated in a serial autopsy study, was 1/62,500. (1) Magnoli et al. (2) reported 20 cases of aortic coarctation among 1,500 patients treated for aortoiliac obstruction. Three of these patients also had aneurysmal aortic dilatation. Coarctation of the abdominal aorta is more common in women than in men. In a series of 18 cases presented by Delaurentis et al., (3) there was only one man. This condition was first described

Fig. 1. Patient ECG showing heart rate of 50 bpm, absolute QT of 320 ms and a corrected QT by Bazett’s formula of 292 ms.

Fig. 2. Cardiac MRI. Top left: Four-chamber view; on the right: Two-chamber view of the left ventricle; both images show myocardial hypertrabeculation predominantly in the apical region. Bottom: Short axis view with hypertrabeculation, typical of noncompacted myocardium. Noncompacted/compacted myocardium ratio > 2.4