Inherited arrhythmia syndromes (IAS) are a heterogeneous group of genetic heart diseases predisposing to sudden cardiac death. Patients with IAS and their families receive diagnostic and therapeutic management, which is heterogeneous across centers and suboptimal with regard to adherence to the current guidelines.

General practitioners (GPs) can be crucial to the identification of subjects with family history of and suspected IAS, for onward referral to specialists for appropriate diagnostic and therapeutic management, and for screening of family members. Nevertheless, awareness and knowledge of genetic diseases among GPs and their confidence in managing these patients need to be improved in different countries.

The aim of this statewide survey was to obtain real-life data from GPs and private practice cardiologists (PPCs) and to assess their role in ensuring further investigations for subjects with IAS or family history of sudden cardiac death.

After ethics approval (Swiss Ethics 2019-00754), a self-administered questionnaire, consisting of 15 questions, was sent by regular mail in October 2019 to 306 medically qualified GPs and 21 PPCs active in Canton Ticino, Switzerland. National sources with doctors’ addresses and specialties were used. One reminder letter was sent 1 month later and nonrespondents were contacted by telephone. Physicians were asked to report data on patients with IAS followed-up in their practice, and to provide information on the management of subjects with family history of sudden cardiac death in the young (SCDY, <40 years of age). SCDY was defined as a fatal event at age <40 years in the presence of known cardiac disease, cardiac or vascular anomaly at autopsy, or when a cardiac arrhythmia is the most probable cause.

Information on the perceived indication for further diagnostic examinations (ie, 12-lead ECG, pharmacological challenges, and genetic testing) and family screening was obtained. The data that support the findings of this study are available upon reasonable request.

A total of 106 GPs and 13 PPCs completed the questionnaire (response rate: 35% and 62%, respectively). Results are depicted in Table. While all PPCs reported having seen patients with family history of SCDY, only 40% of GPs declared encountering this condition in their practice. Similarly, out of 106 GPs, 64 (60%) reported no patient with IAS in their practice.

In the presence of family history of SCDY or IAS suspicion, 63 GPs (59%) and 9 PPCs (69%) would indicate further investigations and specific consultation in a dedicated center to management of IAS. Specifically, 12-lead ECG and pharmacological challenges would be considered by 54% and 20% of GPs, respectively. Genetic testing is considered valuable by 21% of GPs and 46% of cardiologists. There were 80 patients with IAS reported by 42 GPs. Among IAS, Brugada syndrome was the most commonly managed disease (33 patients, 41.5%), followed by early repolarization syndrome (22, 27.5%), long-QT syndrome (14, 17.5%), idiopathic ventricular fibrillation (10, 12.5%), and catecholaminergic polymorphic tachycardia (1, 1%). Among GPs and PPCs managing these cases (42 GPs and 13 PPCs), only 21% and 46% reported patients diagnosed with IAS as a result of family screening, respectively.

To the best of our knowledge, this is the first study assessing GPs' attitudes in the management of subjects...
with family history of SCDY and patients with IAS. Taking an adequate family history, ideally involving a 3-generation pedigree, is essential for the appropriate management of a genetic disease. In this study, a low proportion of GPs (40%) reported to have seen subjects with a family history of SCDY or suspected IAS in their current practice. Potentially this is due to the lack of awareness of the importance of a family history of SCDY as a red flag leading to the suspicion of an undetected genetic disease in a given family. SCDY is a rare event with a reported incidence of 2 cases per 100,000 persons every year. According to the demographic characteristics of the studied region (350,000 inhabitants), up to 7 sudden cardiac death young victims per year can be expected and 7 families may require a specific diagnostic assessment. The referral to a specialist center in genetic cardiac diseases is of paramount importance for proper patients’ diagnostic and therapeutic management. The need for further evaluation by a specialist and diagnostic investigations was reported by a similar but suboptimal rate of GPs (59%) and PPCs (69%). Moreover, new IAS diagnoses by family screening were reported only by a minority of GPs (21%) and PPCs (46%), suggesting the lack of systematic diagnostic strategies in the management of first-degree relatives.

Brugada syndrome, together with LOTS, is known to be one of the most common IAS; therefore, it is not surprising that GPs reported Brugada syndrome as the most common IAS seen during their practice. As far as early repolarization syndrome is concerned, the over-reported rate is possibly due to an inappropriate interpretation of ECG in patients with benign early repolarization.

Although the response rate was relatively low, this rate is comparable with similar postal surveys and all appropriate methods were used to increase the response rate. Up to 40% of GPs and 30% of PPCs do not consider further investigations in the presence of family history of SCDY, indicating the need for more awareness activities in the primary care medical community. Moreover, the referral to dedicated centers is suboptimal and should be implemented to ensure a proper management of IAS patients and their families.

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