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Review article

Familial hypercholesterolaemia patient support groups and advocacy: A multinational perspective

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HIGHLIGHTS

- Familial hypercholesterolaemia (FH) is mostly an unidentified disease that does not discriminate.
- More awareness is required to increase the number of FH patients identified.
- Patient empowerment and the patient voice is paramount in raising awareness and getting system change to identify and optimally treat all FH cases.
- Patient organisations and healthcare professionals work together to find all FH patients worldwide, the earlier the better.
- Networking by and between FH patient organisations is important to share information and learning.

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ABSTRACT

Familial hypercholesterolaemia (FH) is an autosomal-dominant disorder associated with high low-density lipoprotein cholesterol (LDL-C). Left untreated, 50% of men with FH will develop coronary heart disease by the age of 50 and 30% of women by the age 60 [1,2]. It is estimated that the prevalence may be as high as one in 250 people, with most undiagnosed.

This article explores the development of advocacy in FH patient organisations, citing examples from Canada, the Netherlands, Spain, the US and the UK as well as the pan-European patient organisation, FH Europe. The article demonstrates that for patient advocacy, the link with medical and scientific expertise is essential to ensure that advocacy for familial hypercholesterolaemia is well-founded and credible and that patient associations are prepared to take a long-term view on achieving improvements in identification and treatment.

1. Introduction

Familial hypercholesterolaemia (FH) is an autosomal-dominant disorder associated with high low-density lipoprotein cholesterol (LDL-C). Left untreated, 50% of men with FH will develop coronoary heart disease by the age of 50 and 30% of women by the age of 60 [1,2]. It is estimated that the prevalence may be as high as one in 250 people, with most undiagnosed. Early diagnosis, family history, lipid profile and genetic testing should help doctors to identify members of the whole family as there are available treatments to avoid early premature cardiovascular disease. As the disease is inherited, once patients are diagnosed, other family members normally become involved as measures are taken to test for further family members who may have this disorder.

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This article is based on the work of five patient associations and seeks to illustrate the development of patient groups from an initial support function, focusing on education about the disease, and sharing lifestyle and treatment advice and the gradual evolution towards awareness-raising and subsequently seeking to influence the decisions that lead to early identification and improved treatment. As national influence grows, the effectiveness of coming together as a community where small and larger organisations can benefit from shared experiences becomes apparent, as does interaction with specialists in the medical and research fields. Where a history of patient action exists, a model exists for newer associations to start from a well-informed base.

Despite different health care systems which operate in differing ways, the common goal is to increase awareness of FH and work together with a wide range of health care professionals and other providers to deliver improvements to patient care.

2. How patient associations become involved in advocacy

2.1. The story of patient support groups for familial hypercholesterolaemia

The general story of most FH patient associations is the creation of small voluntary groups of patients and family members, for whom education regarding this genetic disorder is a key focus. Many patients will be anxious, fearful and often angry upon diagnosis, especially as it has implications for families and children. Initial awareness of the disease and a gradual increase in scientific knowledge can and do lead patients to desire to achieve more to ensure early identification, better and more affordable treatment with equality of access.

The shared bond between patients in national associations can take many forms - learning more about the disease and treatments helps ensure informed choices. For others, lifestyle advice on day-to-day living with their condition plays an important social and lifestyle role - advice on the importance of diet and exercise in supporting treatment, often via websites and dedicated Facebook pages, or even cookery groups.

At national level, fundraising events by volunteers can emphasise the benefits of healthy lifestyle. Both Ireland's Croí Heart & Stroke Charity and the UK organise participation in the Virgin London Marathon, whilst countries such as Switzerland set targeted distances for running teams in Swiss marathons, allowing a range of volunteers to play their part The FH Foundation® organises a “Race for FH” in conjunction with their annual FH awareness campaign that includes a broad public relations campaign and a fundraising component to support the mission and work.

As groups gain in knowledge and confidence, they often seek to improve identification and improvements in availability and cost of treatment. This wider perspective requires obtaining support from decision-making authorities, be they health commissioning bodies or government officials and representatives.

The FH Foundation is a research and advocacy organisation based in the United States that serves as a global resource for healthcare providers and individuals impacted by FH. The FH Foundation started FH Awareness Day on 24 September in 2012. FH Awareness Day is now recognised on a global scale and generates publicity for FH for this highly under-recognised condition. Raising consciousness of the condition helps receptiveness in later discussions with a wider public and decision-makers. The FH Foundation is an integrated partnership of healthcare providers and individuals born with FH.

2.2. What does advocacy mean for familial hypercholesterolaemia patients?

Governments and other decision-makers (at national, EU and supranational level) can affect the lives of FH patients – actually or potentially, positively or negatively. To improve the diagnosis and treatment of FH, members of patient associations need to express in their own words, how the decisions that are made have an impact on the lives of FH patients.

Advocacy involves raising awareness of FH and prioritising the challenges faced within and across national boundaries. Advocacy is about interaction with decision-makers, as they adopt policies and laws relevant to patient groups. These may include government ministers, politicians or government officials in national, regional or local government.

There are various building blocks required to support successful advocacy. These include a foundation of science, accuracy and reliable evidence to engender confidence that existing and novel treatments have been fully validated.

While the internet remains a major source of information for both the public and health care professionals, it can often provide conflicting, misleading, inaccurate and often dangerous information. In order to rebut inaccurate information and ‘fake news’, in particular regarding cholesterol and the use of statins, patient associations are seeking to develop expertise to provide consistently high-quality information in a way that encourages public trust and supports the work of health care professionals.

The development of scientific material to support early identification of FH includes cascade screening and registries, the latter providing health care professionals and researchers with data for clinical, scientific and policy purposes. From the patient perspective, their role in improving and monitoring the outcome and quality of treatment and in reviewing best practice, becomes a tool for patient advocacy to ensure continued development and improvements are embedded in healthcare policies.

2.3. New media as a tool in patient advocacy

To maximise their reach across boundaries – both national and generational, all FH patient associations use e-media to inform and advise.

In the United Kingdom, HEART UK's website [3] has more than two million visitors a year and also provides a regular e-news both to supporters and to health care professionals.

The FH Europe website is a hub to provide general and easily understood information on the condition for patients - their families, the wider public and decision-makers - and to promote contact with members. Its private member area offers a substantial resource for reference and training material. FH Europe's e-Flash newsletter is issued on a regular basis to provide an overview of FH-related activity to interested parties in patient associations, healthcare professionals and others. Facebook and Twitter also play a part, but it is the personal contact between members that provides the motivation for network activity and can ensure that volunteers are energised.

2.4. Screening - the role of patient organisations in the development of a FH cascade screening programme

2.4.1. The example of fundación hipercolesterolemia familiar of Spain

The Spanish patient association notes that since Familial Hypercholesterolaemia (FH) is the most frequent genetic disorder, which affects 50% of the family members, besides clinical criteria, its final diagnosis should be genetic. It fulfils the WHO criteria for genetic screening: FH is a silent disease, invisible for many people. Its physical signs are not always present (xanthomas 15–20%) especially in the young, and it has serious cardiovascular consequences for young and middle-aged people.

The aim of the Spanish association's contribution to data generation is both to help improve understanding of the FH risk and to support decision-makers at the Spanish Central and Regional level, the latter being one of the purposes of the SAFEHEART nationwide Registry, which is the Spanish FH cohort follow-up study, created and conducted by the Foundation with the collaboration of 28 lipid clinics.

2.4.1.1. The voluntary contribution made by patient associations and the experience acquired is exemplified in Spain's 'Weekend days' work' FH screening programme. The SAFEHEART Registry started in 2004; growth rate was slow as specialists did not have sufficient time to conduct family cascade screening. The Spanish patient association sought a novel solution and in 2009, the FHF team started delivering family cascade screening themselves.

In Spain, specialists at hospital lipid clinics (the case for most diagnoses), and also doctors in primary care identify Index Cases (IC) by clinical criteria confirmed by DNA test. Once the genetic result is positive, doctors inform the IC and obtain consent for the FHF to trace the family members. Trained personnel contact relatives to explain the importance of early diagnosis since FH is a silent disease with a family history. That is an important information process. Once the specific 'Weekend Days' work' is fixed, the association organises the family tree with the IC: this is followed by an information process to the family members, firstly by means of a phone call, followed by sending written information regarding the whole process.

Fundación Hipercolesterolemia Familiar (FHF) stresses the importance of listening to the patient, as this initial contact is not an easy task. Confidence and trust are key elements that FHF volunteers require, not only from the IC, but also from all the family members whose traits may be quite different. Listening to them is the best way to ensure that the messages have been taken on board. FHF believes that "empathy and complicity are also our best allies to accomplish our task."

2.4.1.2. How FHF puts screening procedures into practice in 'Weekend days' work'. On a specific weekend, the FHF team, the IC and family members meet at the hospital where the index cases were identified. Work is carried out on Saturday and Sunday from 8.30am to 2.00pm. In the morning, 4 or 5 doctors, 2 nurses, 3 laboratory technicians, 1 dietitian, and a cascade coordinator will enrol 120 people on average. FHF provides a physical examination, Clinical and Dietitian questionnaires, as well as blood samples, following SAFEHEART requirements, as they will be part of the follow-up Cohort Study.

2.4.2. Government-led screening programmes – Netherlands case study

Until 2013, the Netherlands had a national screening programme for Familial Hypercholesterolaemia. The programme included visiting family members at home. Largely as a result of this programme, 42% of the people with FH were identified [4,5]. However, funding was stopped and the activity ceased, and thus many unidentified FH patients remain.

Harteraad is collaborating with LEEFH [6] (the 'Landelijk Expertisecentrum Erfelijksheidsonderzoek Familiaire Hart-en Vaatziekten' foundation), internists, cardiologists and the Dutch Heart Foundation, to lobby for the reintroduction of family screening for FH [7].

All people with FH and their relatives can ask for, or are being offered, DNA research by their general practitioner. Harteraad has participated in the development of 'consultkaart FH'. With this information chart it helps people to make a well-balanced decision with their doctor to start DNA research on FH in their families, 'yes' or 'no'? This 'consultkaart' is based on the international option grids model [8].

2.5. Registries - development of a predicting risk tool of cardiovascular events on FH patients

HEART UK promotes the use of a single FH Registry for the UK, to capture all the data for FH families being identified. This system is a pedigree cascade system, which enables information to be shared across hospitals with clear benefits for testing of family members across the country. HEART UK operates a FH Registry Advisory Group, which includes membership from a range of lipidologists, paediatricians and FH nurses. This Registry Advisory Group oversees the governance arrangements for the registry and management of its development, including the relationship with other registers established by cardiovascular specialist societies.

The foundation of science, accuracy and reliable evidence enables HEART UK to provide solid support for the public and patients, enabling it to play a pivotal role in important key decision-making bodies at a government level, such as NHS England's FH Steering Group chaired by the National Clinical Director for Heart Disease, helping to drive forward FH services and the CVD Collaboration Group and the National Cardiovascular Intelligence Network.

Spain has used Cohort Study data from its SAFEHEART (The SpAnish Familial HypErcHolEsterolaemiA CohoRt STUDy) Registry to develop an equation to estimate the risk of cardiovascular events in FH patients at 5 and 10 years. Although risk factors for atherosclerosis cardiovascular disease (ASCVD) in FH have been described, models for predicting incident ASCVD have been not reported. The Cohort Study is a prospective, open-label, multicentre, long-term follow-up study of a cohort of people with a genetic diagnosis of FH in Spain by the Foundation, conducted with the collaboration of 28 lipid clinics. Grounding details of this equation development procedure were published in 2017 in the prestigious medical journal *Circulation* [9].

The use of this innovative application, or App, is intended to help both patients and physician in charge of the care of adult heterozygous FH. The tool is available in its website version, as well as through App ("Fundación Hipercolesterolemia Familiar" and "Safeheart risk equation) for Android and Apple.

FH patient associations have welcomed such developments. To obtain worldwide intelligence on familial hypercholesterolaemia will require a global database, with a framework capable of adapting and harmonising measurements made in different countries and health systems. It is understood that research is ongoing by EAS-FH Studies Collaboration and the US FH Foundation.

The FH Foundation established and operates the CASCADE FH® Registry. Over 5000 patients are currently being followed longitudinally at 37 lipid and cardiovascular academic centres. Within this database, the majority of patients already have established cardiovascular disease with an average FH diagnosis age of 47. The CASCADE FH Registry is a hybrid design, which also includes an Internet based portal that is currently available in Japanese, Spanish, Portuguese, and English. This patient-centric tool allows FH individuals to track key medical metrics longitudinally and provides insights into qualitative issues such as understanding of FH, perceived burden of disease, and extent of family screening. All data is given back to patients in a visual dashboard format to help them track the trajectory of their care. Importantly, a sharable pedigree tool is built into the patient portal to allow individuals to track their own family history and encourage family screening.

2.6. Monitoring developments in research

Equally important to patient associations is being kept abreast of developments which may help identify those at risk and of developments in treatment for existing patients.

2.6.1. The development of new therapies for FH patients

The most significant recent development in treatment of FH is PCSK9 inhibitor therapy. Patient and healthcare professionals alike have been monitoring the outcome of clinical trials and development.

Partnership working with pharmaceutical companies has proven to be a successful model to ensure that the appropriate treatment is given to FH and other lipid patients. However, patient organisations have had to undertake advocacy and engagement to increase the number of patients having access to appropriate treatment.

For example, during the consultation on access to PCSK9 inhibitor therapy, the Canadian Heart Patient Alliance submitted evidence and responded to encourage access to PCSK9 inhibitor therapy to be included in the public drug plan. The Canadian Agency for Drugs and Technology finally approved the medication and the Canadian Heart

Patient Alliance continues to engage with private and public plans to ensure the drug remains available.

In the UK, HEART UK had a pivotal role in arguing for access to PCSK9 inhibitors, after initial reluctance for its approval. Following a comprehensive consultation involving patients and healthcare professionals through its 'say yes to PCSK9' campaign, HEART UK's submission and evidence of the need for innovative medicines helped overturn a decision by NICE (National Institute for Health and Care Excellence), the public body whose published guidelines include the use of health technologies within the NHS and clinical practice.

In the United States, individuals with FH lobbied at Federal Drug Administration hearings for the approval of PCSK9 inhibitors. Using an in-house database that includes laboratory results, medical and procedure claims from over 200 million Americans, the FH Foundation has published a manuscript in *Circulation* detailing potential barriers to access to PCSK9 inhibitors in FH patients. The FH Foundation has also undertaken an educational campaign with major pharmacy benefit managers and insurance companies to improve the process for approval of novel therapies for FH patients.

On 29 June 2018, Lomitapide ('Lojuxta' in the EU) was approved by NHS England's Specialised Commissioning Group for use in England for homozygous FH patients. HEART UK's December 2016 statement on the management of homozygous familial hypercholesterolaemia in the United Kingdom [10] was instrumental in this development for patients.

2.6.2. Developing testing for paediatric cases – the example of the UK

HEART UK has been championing adopting the child-parent approach presented in Professor David Wald's 2016 paper. This detailed the outcome of research at Queen Mary University of London: testing for cholesterol and FH genetic mutations was carried out on 10,059 children while they were undergoing routine vaccinations in infancy at 92 general practices in England [11]. Forty of the children were found to be FH positive.

HEART UK has contributed to the UK National Screening Committee on this matter. At present all FH cases in the UK are identified starting with an adult, usually aged around mid-50s. Introducing the additional approach of child-parent screening would introduce a constant source of index cases and would also address the inequalities that exist around who and where the cases are identified. HEART UK believes delivered paediatric testing in addition to the adult service will help prevent early heart attacks and enable FH patients to have a normal life expectancy.

2.7. Health advocacy at supra-national level and current constraints

International bodies are aware of the constraints on national health budgets. A 2011 World Health Organisation (WHO) Bulletin [12] cited Romanian doctors who declared 'Prevention is better than cure' and recounted the struggles of their under funded health system which focused on treatment rather than prevention.

Under the principle of subsidiarity, European Union member states decide how to deliver and organise their health services: the European Union, however, has certain public health powers under Article 168 of the Treaty for European Union (TFEU) [13].

The European Commission's Public Health and Food Safety Directorate General has stated that its policies on non-communicable diseases will align with those of the World Health Organisation. It collaborates with the WHO on monitoring of chronic disease risk factors and on its Non-communicable disease (NCD) strategy introduced in 2015. It also collaborates with the Paris-based Organisation for Economic Cooperation and Development (OECD) with regard to the economics of preventions, country profiles and knowledge.

Part of the Commission's strategy is to create a new health policy platform and invite health NGOs to share expertise and country knowledge. FH Europe is currently exploring how to achieve this to promote best practice in paediatric testing. Professor Albert Wiegman of the Netherlands visited Brussels with two FH Europe Trustees in spring 2017. This direct contact opened the door to dialogue with the European institutions and made it possible to present the current results arising from 15 years of studies. These will be published in 2018 and work that is ongoing in the Netherlands, Spain, the Czech Republic and elsewhere points to the benefits of early testing in avoiding premature cardiac events. Cascade screening can then identify family members at risk and FH member associations intend to gather information on the extent to which cascade testing is used in their respective countries.

2.8. FH Europe – an umbrella organisation that acts on behalf of national groups and gives patient organisations the tools for advocacy

FH Europe is an active network of 23 organisations spanning 22 European countries, having been established as recently as 2015 and achieving charitable status in late 2016. It brought together long-standing FH patient organisations, such as HEART UK and new organisations such as the Swiss patient association, founded in 2015.

2.8.1. Development of support group collaboration

On 20 October 2015, representatives of the fledgling FH Europe patient network attended a meeting in the European Parliament in Brussels to launch a Call to Action on FH. A number of elected representatives, members of the MEP Heart Group, signed their support. A number of FH patient associations met in Gothenburg, Sweden in November to discuss and plan creating a formal FH patient network for Europe.

From the outset, FH Europe set itself some ambitious goals as a focus for member involvement, namely:

- Early identification of FH patients
- Raising awareness of FH amongst health care professionals, the public, decision-makers in policy and clinical commissioning fields
- Sharing best practice, information and success stories
- Supporting the creation and development of new/smaller patient groups
- Campaigning at EU/European level to ensure that there is a Europe-wide approach

In order to fulfil these aims, more needed to be known about FH treatment in member countries. A brochure published in 2016 set out member associations' aims and objectives. In early 2017 FH Europe produced a 'heat map'; this illustrated the response from member associations to three simple questions regarding the national presence of adult registries, paediatric registries and whether cascade testing was available on a national basis. The results, published on the FH Europe website [14], reflect the disparate status of treatment across the continent (see Supplementary Figure).

The mix of activities in national associations helps create a cohesive group that can act together to promote awareness of the disease and engage with decision-makers at regional and national level to help improve access to healthcare and reimbursement of treatments.

2.9. Case study - the Spanish FH National detection plan

The Spanish FH National Detection Plan is an ambitious target (pending approval by the health Ministry in 2018).

2.9.1. Why is it so important to gain approval?

Most patients (80%) with FH are still undiagnosed and untreated. This poses a public health problem and therefore a public health challenge. Treatment is often started in the late stages of the disease, which means that most of the patients are underdiagnosed, and attended in Primary care, where GPs' information should be a priority for the health care system. However, neither policies nor support are available since national health care systems are not sufficiently aware

of the problem.

A National Detection Plan for FH with the participation of Specialist and GPs may be the best way for FH patients in Spain to be diagnosed, breaking the main barrier that prevents them from getting the right cardiovascular care. The FHF has been instrumental in bringing a proposal for a National FH Detection Plan not only to the Spanish Health Ministry, but also at regional level.

A National Detection Plan was approved by the Parliament in 2010 and is now a reality in one of the seventeen autonomous regions that comprise Spanish health care since 2008. There has also been a pilot study in Madrid since 2006. However, approval from the Spanish Health Ministry is a mandatory requirement for implementation to take place throughout Spain. Despite Parliament's support for government implementation, the Detection Plan is still pending Health Ministry approval.

On 5th July 2017, FHF was instrumental in bringing the proposal, once again, to the attention of the Health Ministry and also the different regional level authorities in the Spanish Health Ministry. The discussions emphasised the benefits to patient associations of evidence-based arguments, including in this case, two publications which confirm that a Detection FH Plan is always cost-effective [15,16]: The meeting between health care professionals and economic experts, to discuss how Spain can improve outcomes for FH patients resulted in a proposed plan:

- Horizon 10 years (2018–2027)
- Detection of 9000 FH cases per year (2250 IC and 6750 relatives):1 Index Case/3 relatives.
- Total detection could be 90,000 cases – at present only 20,000 FH patients have been detected in Spain.

After the presentation, each decision-making body, the Minister, and the majority of high public authorities accepted a compromise to go further with the FH Detection Plan. The date for granting approval is eagerly awaited by FHF.

2.10. Case study – the advocacy examples from the UK

Advocacy with decision makers can take the form of joint meetings between patient representatives, specialists and decision-makers or responses to government and health funding programmes. Patient associations may also set their own targets for the long or short-term priorities and extend influence by involving members of parliaments who are not normally involved in health committees or legislation.

2.10.1. Championing familial hypercholesterolaemia

HEART UK lobbies to keep cardiovascular disease and cholesterol as a top health priority for the British Government and within that, FH is a key condition. The charity was instrumental in getting FH included within the Government Cardiovascular Outcomes Strategy in 2013 and has built on this success to keep the pressure on to establish a national FH cascade testing service with one place to capture the data. Although progress has been observed, it is not as yet a national service. As with the Spanish Detection plan, persistence is required and so the charity is maintaining pressure.

2.10.2. Specific targets – a long-term example

A recent HEART UK campaign theme includes a sensitive area around Death Certificates. HEART UK firmly believes that families should be made aware of an inherited condition so that family members have the option to be tested and life-saving treatment offered. The Royal College of Pathologists has been very supportive of enhancing the training of medical examiners and others so that there is greater awareness of FH. HEART UK is working very closely with them to provide a perspective on FH.

2.10.3. Case study – establishment of an international classification of disease, tenth edition (ICD-10) code for familia hypercholesterolemia

Central to the mission of the FH Foundation is taking a systematic approach to addressing the underlying and complex issues that contribute to the lack of diagnosis of FH. This genetic condition puts a significant subset of the population around the world at risk of developing aggressive heart disease or dying during their prime of their lives. The FH population has remained largely invisible as a cause of intergenerational heart disease. In January 2014, the FH Foundation submitted an application and attended a hearing at the Coordination and Maintenance Committee for Medicare and Mediaid Services (CMS) and the Centres for Disease Control and Prevention (CDC). The result was the creation of a distinct ICD-10 code for FH and also one for a family history of FH. In October 2016, E.78.01 was established as the diagnostic code for FH and Z83.42 as the code for Family History of FH.

2.10.4. The patients' role in influencing

HEART UK's patient and supporter network invites patients to share their knowledge and learn more about treatment options, food and lifestyle from clinicians and dietitians. More active patients may become HEART UK ambassadors and are campaigners, awareness raisers and influencers particularly on HEART UK's Patient and Supporter Committee.

During National Cholesterol Month in October 2017, HEART UK held an FH Day of Action [17] and invited those living with or affected by FH to meet their MPs in the Houses of Parliament to share their experience of their initial diagnosis, accessing services and treatment. Thirty-one MPs attended the event and held discussions with FH patients and families, a number of questions were asked in Parliament, letters and statements asked of the Department of Health and copies of correspondence on FH service development between the Prime Minister and the Secretary of State for Health were sent to HEART UK. One patient ambassador was sufficiently empowered to persuade their local healthcare funding commissioning group to start an FH service.

3. Conclusion

This article illustrates that approaches to familial hypercholesterolaemia patient advocacy vary from country to country and are adapted to respective national healthcare regimes. No one model suits all, as healthcare, governmental and legal systems vary greatly. However, associations benefit from sharing experience and best practice; combining forces within a wider network enables a wide-ranging perspective on the challenges faced at national and pan-European, or international, level. Broader networks represent a harmonised view of patient association aims and are able to offer support to nascent patient support groups across the globe. The patient association can give voice to concerns regarding identification of treatment, treatments and funding and its collaboration with specialist medical and research activity is crucial in ensuring that advocacy is well-founded and grounded within a reliable scientific context. Other disciplines may also be involved, such as in evaluation of socio-economic benefits of patients receiving appropriate treatment at an early age so that they are able to lead active and productive lives.

The benefits of a collaborative approach to advocacy are essential in bringing about the necessary changes to care for and support the FH population around the world. As has been demonstrated above, shared experiences and knowledge can achieve great gains at regional, national and international level.

Conflicts of interest

The authors declared they do not have anything to disclose regarding conflict of interest with respect to this manuscript.

Appendix A. Supplementary data

Supplementary data related to this article can be found at https://doi.org/10.1016/j.atherosclerosis.2018.08.020.

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