

BHF Cymru response to petition: P-05-870: Heart screening for 10-35 year-olds

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We thank the Petitions Committee for asking us to outline our position on heart screening. Below we have set out our position and the evidence for this, as well as the work of the inherited heart conditions programme underway in Swansea Bay Health Board.

Overview

- We support targeted expert assessment of families with high risk of inherited cardiac disease or where there has been a sudden unexplained death but there is insufficient evidence for the usefulness of universal screening.
- The BHF supports individuals having an increased understanding of their family's medical history. So, if a family member has died suddenly, particularly at a young age, they would be advised to discuss this with their GP, with a view to getting themselves screened and assessed at an inherited heart conditions service, where appropriate.
- The BHF supports cascade testing of individuals who have a first degree relative who has died suddenly and no cause of death has been established (and post mortem reveals a structurally normal heart) or who has been diagnosed with an inherited heart condition.
- While sudden death in the young is particularly tragic, it is also thankfully relatively rare.

Policy Statement: Cardiac Screening for Professional Athletes

Sudden death in young adults is usually caused by ventricular fibrillation (VF), a chaotic heart rhythm disturbance that causes the heart to stop pumping blood around the body and to the brain, also known as cardiac arrest. It is invariably

fatal unless cardiopulmonary resuscitation (CPR) is immediately performed and a defibrillator is used to help shock the heart back into a normal rhythm. There are several conditions that can cause VF in young athletes. Hypertrophic cardiomyopathy (HCM) is the most common, but there are other, rarer conditions that predispose an individual to sudden death due to genetic mutations.

While sudden death in athletes is rare (between 1 in 50,000 and 1 in 100,000 cases annually), it is two to four times more common in athletes than in non-athletes¹. Following the tragic deaths of high profile athletes, screening professional athletes for heart disease has been a significant topic for debate in the UK for the past few years.

BHF Position

The BHF supports targeted expert assessment of families where there is a high risk of an inherited cardiac disease, or where there has been a sudden unexplained death.

However, at present we are not calling for a publicly-funded screening programme for all professional athletes. While a proportion of people may be identified that have an underlying heart condition, screening is not yet at a scientific level of precision whereby all people at risk of sudden cardiac death can be accurately predicted. Misleading screening results can have potentially damaging consequences for healthy athletes and their families.

Many professional sports organisations offer some form of screening for their athletes. Where this occurs, it is essential that the results are interpreted by experts and that the athletes concerned are aware of both the benefits and limitations of such screening.

In the future, we anticipate that the accuracy of screening will improve to a level whereby a combination of ECG, echocardiography and genetic testing could be used to screen all athletes.

Background

Standard cardiovascular risk assessment screening - the type that ideally all adults over the age of 40 should have periodically - would not pick up most of the conditions that cause sudden death in young athletes. Until more evidence is available, physicians have a duty of shared decision-making with athletes when

¹ Ferreira M, Santos-Silva PR, de Abreu LC, Valenti VE, Crispim V, Imaizumi C, Filho CF, Murad N, Meneghini A, Riera AR, de Carvalho TD, Vanderlei LC, Valenti EE, Cisternas JR, Moura Filho OF, Ferreira C. Sudden cardiac death athletes: a systematic review. *Sports Med Arthrosc Rehabil Ther Technol.* 2010 Aug 3;2:19. Available at: <http://www.ncbi.nlm.nih.gov/pubmed/20682064>

discussing the risks and uncertainties of intense exercise in those with underlying cardiovascular conditions².

In contrast, signs of an underlying risk of sudden death can sometimes be picked up by an ECG or an echocardiogram. But these tests are often difficult to interpret, not least because athletes' hearts undergo physiological changes that produce ECG or echocardiographic features that, if found in a non-athlete, would indicate an underlying heart condition, whereas in an athlete may be normal. Multiple gaps in evidence strongly indicate that implementing pre-participation cardiovascular screening of athletes on a large scale would be premature³.

A statement released by the BMJ (2016) states that overall, 25% of people with a condition that may lead to a sudden cardiac death would not be identified, and that up to 5% of healthy people can be suspected of having cardiac disease. The high number of false positives associated with screening programmes leads to over-diagnosis, which for some can cause health-related anxiety and psychological trauma. Athletes may also be restricted or prevented in certain sporting activities unnecessarily and experience issue with employment and insurance throughout their lives⁴.

Most conditions that can cause sudden death in the young are genetically determined. However, understanding of the DNA variations that do and do not lead to an increased risk of sudden death is not yet sufficiently advanced to allow reliable DNA screening for all the mutations that lead to HCM and other potential sudden death syndromes. In the future, it may be possible for a DNA test, possibly combined with an ECG and echocardiogram, to reliably identify most people at risk of developing these conditions.

High profile cases where athletes have suffered cardiac arrests have often led to calls that all athletes should be regularly screened to detect anomalies in the heart that could trigger a cardiac arrest in the future, even though, in some cases, the individuals concerned had undergone screening and been passed as normal.

When screening for genetic or congenital cardiovascular abnormalities, American Heart Association Guidelines recommend that physicians should use the AHA's 14-point screening guidelines combined with a history and physical examination⁵.

² Sharma, S. and D'Silva, A. (2017) *Management of young competitive athletes with cardiovascular conditions*, Available at: <https://heart.bmj.com/content/103/6/463>

³ Sharma, S. (2013) *Cardiac Screening Before Participation in Sports*. *New England Journal of Medicine* 369;21

⁴ British Medical Journal (2016) *Screening young athletes to prevent sudden cardiac arrest not proven to save lives*, Available at: <https://www.bmj.com/company/wp-content/uploads/2016/04/Athlete-screening.pdf>

⁵ American Heart Association (2014) *American Heart Association 14-Element Screening (Maron BJ Circulation 2014)*, Available at: http://med.stanford.edu/content/dam/sm/ppc/documents/HSupervision/AHA_14-point.pdf

The 14-point guideline includes the following assessment: chest pain or pressure related to exertion; unexplained syncope or pre-syncope; dyspnoea, fatigue, or palpitations related to exercise; history of a heart murmur; elevated blood pressure; previous restrictions from sports; previous cardiac testing; family history of premature death; family history of disability from heart disease; family history of hypertrophic or dilated cardiomyopathy, long-QT syndrome, or other ion channelopathies, Marfan syndrome, significant arrhythmias, or specific genetic cardiac conditions; heart murmur on examination; femoral pulses for aortic coarctation; physical examination findings consistent with Marfan syndrome; and brachial artery blood pressure⁶.

The use of 12-lead ECG or echocardiography should not be limited to competitive athletes and may be considered as part of screening in smaller cohorts of young (12 to 25 years of age)

However, universal screening of the general population of young persons with 12-lead ECG is not recommended, regardless of athletic status. When utilized, the limitations of 12-lead ECG as a population screening test (e.g., false-positive or negative results) and cost should be taken into account.

Most data place these cardiovascular sudden deaths in the range of approximately 1 in 80,000 to 1 in 200,000 participants per year.

Evidence

In July 2008, the BMJ carried an editorial which backed a systematic pre-participation screening programme for all competitive athletes. This would use a comprehensive personal and family history questionnaire, physical examination, and screening using an ECG to identify those at risk of sudden cardiac death. This recommendation was based on a study published in the same edition which analysed data from 30,065 Italian athletes who underwent a complete pre-participation cardiovascular evaluation including resting and exercise ECGs⁷. However a subsequent study of British athletes to identify HCM suggests that screening was not cost-effective, as several thousand athletes would have to be screened to identify one with HCM⁸. A screening programme for all athletes may not therefore be appropriate for public funding.

⁶ Barry J., Maron, M.D., Benjamin, D., Levine, MD., Reginald, L., Washington, MD., Aaron, L., Baggish, MD., Kovacs, RJ, Martin, S., (2015) 'Eligibility and Disqualification Recommendations for Competitive Athletes With Cardiovascular Abnormalities: Task Force 2: Preparticipation Screening for Cardiovascular Disease in Competitive Athletes A Scientific Statement From the American Heart Association and American College of Cardiology.', *American Heart Association Journal*, 132(1), pp. 267-272.

⁷ Asif, I., M., Drezner, J., A., and O'Connor, F., G., (2016) 'Cardiovascular Preparticipation Screening in Young Athletes Looking Through One Lens', *Sports Health*, 1(9), pp. 19-21.

⁸ Basavarajaiah S, Wilson M, Whyte G, Shah A, McKenna W, Sharma S. Prevalence of Hypertrophic Cardiomyopathy in Highly Trained Athletes: Relevance to Pre-Participation Screening. *J Am Coll Cardiol*. 2008 Mar 11;51(10):1033-9. Available at: <http://www.ncbi.nlm.nih.gov/pubmed/18325444>

It is also important to note that different screening models (with or without ECG) offer their respective benefits and limitations, but the absence of clear outcomes-based evidence precludes any universal policy for all athletes. It is also clear that some athletic groups are at substantially higher risk of sudden cardiac arrest and death (SCADS) than others, and a “one size fits all” approach is not appropriate. There is limited evidence to support the new guidelines from the American Heart Association.

Malhotra, A. et al. (2018)⁹ investigated outcomes of cardiac screening in adolescent football (soccer) players in the UK. Diseases that are associated with sudden cardiac death were identified in 0.38% of adolescent football players in a cohort that underwent cardiovascular screening. The incidence of sudden cardiac death was 1 per 14,794 person-years, or 6.8 per 100,000 athletes. Most of these deaths were due to cardiomyopathies that had not been detected on screening.

The English Football Association (FA) has run a mandatory cardiac screening program for adolescent athletes in the United Kingdom since 1997 to determine the incidence and causes of sudden cardiac death in this well-defined population of previously screened football players. Each athlete was classified in one of three categories on the basis of the evaluation: normal; further evaluation needed, if an abnormality was detected that required further investigation to confirm or refute the presence of cardiac disease; or cardiac disease detected.

The last category was sub-classified into disorders that are associated with sudden cardiac death or those encompassing congenital septal and valvular conditions that were followed up every 2 years at regional centres.

Following this, decisions to disqualify such athletes were made by the FA cardiology consensus panel after discussions in accordance with current exercise recommendations of the European Society of Cardiology and the American Heart Association.

During the course of these assessments, of the 8 sudden cardiac deaths that occurred, 6 (75%) were in athletes who had normal findings during preliminary screening.

A BMJ review (2016)¹⁰ revealed that while the pre-participation physical evaluation (PPE) may serve many purposes, the ability to identify athletes with cardiac disorders at potential risk of SCADS through a screening history questionnaire and physical examination is significantly limited.

⁹ Malhotra, A. et al (2018) Outcomes of Cardiac Screening in Adolescent Soccer Players. *New England Journal of Medicine* 2018;379:524-34.

¹⁰ <https://www.bmj.com/company/wp-content/uploads/2016/04/Athlete-screening.pdf>

A study by Landry et al (2017)¹¹ supports this further and revealed the incidence of sudden cardiac arrest during participation in competitive sports as 0.76 cases per 100,000 athlete-years. The occurrence of sudden cardiac arrest due to structural heart disease was uncommon during participation in competitive sports. The rarity of sudden cardiac arrest due to structural heart disease raises questions about the potential value of pre-participation screening.

The European Society of Cardiology also recommends that all European countries adopt such screening. This recommendation was based on an Italian study that found a sharp reduction in sudden deaths among athletes once mandatory screening among athletes was introduced (from 3.6 per 100,000 in 1980 to about 0.4 per 100,000 by 2004). However, there are concerns that this study included data only from the two years before the law was implemented when the sudden death rate may have been unusually high¹².

Since 1997, Israel has required ECG screening not only for competitive athletes, but for anyone who wants to join in an 'organised' activity: this includes joining a gym. A 2011 study focused on Israel looked at the decade preceding the screening laws implementation. Researchers compared rates of sudden death from cardiac arrest during the decade before the law with rates in the decade after. Findings indicated that before the law, there were about 2.5 deaths per 100,000 athletes each year; after the law, there were roughly 2.6 deaths per 100,000 each year - the authors therefore concluded that mandatory ECG screening of athletes had no apparent effect on their risk for cardiac death¹³.

A 2010 US study screened 510 Harvard athletes with an ECG, with researchers also using echocardiography. Echocardiography results showed that 11 of the athletes had heart disease. But when limited to screening using medical history and physical examination alone only five of these cases were picked up, with two potentially serious heart abnormalities missed¹⁴. However, there are potential issues concerning false positive results. In the study, one in six athletes who were tested turned out to be a false positive - the authors reported that 'about 20 per cent of ECG screening results could be expected to turn up a potential problem, most of which would be false-positives'¹⁵. A false positive result could have serious

¹¹ Landry et al (2017) Sudden Cardiac Arrest during Participation in Competitive Sports. *New England Journal of Medicine* 377;20

¹² Steinvil A, Chundadze T, Zeltser D, Rogowski O, Halkin A, Galily Y, Perluk H, Viskin S. Mandatory Electrocardiographic Screening of Athletes to Reduce Their Risk for Sudden Death: Proven Fact or Wishful Thinking? *J Am Coll Cardiol*. 2011 Mar 15;57(11):1291-6. Available at: <http://www.ncbi.nlm.nih.gov/pubmed/21392644>

¹³ Ibid

¹⁴ Baggish AL, Hutter AM Jr, Wang F, Yared K, Weiner RB, Kupperman E, Picard MH, Wood MJ. Cardiovascular Screening in College Athletes With and Without Electrocardiography: A Cross-sectional Study. *Ann Intern Med*. 2010 Mar 2;152(5):269-75. Available at: <http://www.ncbi.nlm.nih.gov/pubmed/20194232>

¹⁵ Ibid

ramifications not just for the career of a young athlete, but also their chances to get life or medical insurance in the future, and for their families.

In a British Cardiovascular Society comment piece in 2012, it was highlighted that were results from other programmes replicated in the UK, 40 of the 196 sudden cardiac deaths that are expected each year could be prevented¹⁶. However, for each death prevented, it is estimated that 40,000 individuals would need screening and 791 athletes would be disqualified¹⁷. This net good/harm ratio does not therefore compare favourably with other national screening processes such as for colon or breast cancer, but may still be considered worthwhile in view of the consequences of sudden cardiac death in this young population.

However, the editorial stated that before such a scheme could be introduced in the UK further cost-effectiveness analysis would be necessary, and the infrastructure and funding would need to be put in place to provide trained experts to conduct screening, analyse the ECGs, and deal with the significant number of further specialist consultations that would be generated.

The UK National Screening Committee, who advise Ministers and the NHS in all four UK nations about all aspects of screening policy, considered its policy for HCM and risk of sudden cardiac death screening in 2008 (reviewed March 2015). The policy review concluded that the evidence did not support the introduction of screening, and that pre-participation screening of athletes for HCM or other causes of sudden cardiac death should not be instituted at present¹⁸. However, we are aware that at the time of writing (August 2019) the Committee has recently issued a call for evidence on Sudden Cardiac Death in 12 to 39 year olds.

Recommendations of existing screening programmes

The charity Cardiac Risk in the Young (CRY) runs a subsidised screening programme, offering ECG screening to all young people between the ages of 14 and 35. The charity advocates mandatory screening for all young athletes. CRY currently provides screening services for a number of professional sporting bodies, including the Lawn Tennis Association and a number of FA football teams¹⁹.

The International Olympic Committee (IOC) has recommended, but not mandated, that all countries screen their athletes to minimise the risk of sudden cardiac

¹⁶ Taylor R. Editorial: Pre-participation screening for athletes in the UK. British Cardiovascular Society; 2012. Available at: http://www.bcs.com/pages/news_full.asp?NewsID=19792061

¹⁷ Elston J, Stein K. Public health implications of establishing a national programme to screen young athletes in the UK. British Journal of Sports Medicine, 2011. 45(7): p. 576-582. Available at: <http://www.ncbi.nlm.nih.gov/pubmed/19622527>

¹⁸ The UK National Screening Committee (UK NSC) (2015) *Screening for risk of sudden cardiac death not recommended*, Available at: <https://www.gov.uk/government/news/screening-for-risk-of-sudden-cardiac-death-not-recommended>

¹⁹ <https://www.testmyheart.org.uk/>

death. According to the IOC, if an ECG raises suspicion of an underlying serious problem in an athlete, further tests should be performed²⁰.

British Heart Foundation Activity

- We continue to review any evidence presented that screening techniques have been suitably improved to the point at which they can confidently and consistently detect inherited heart conditions (IHCs) where an athlete is at risk of SCADS.
- We consider research applications that aim to better understand IHCs and SCADS in both athletes and the general public.
- The BHF runs the Genetic Information Service (GIS), which is a unique service where members of the public can speak to a senior cardiac nurse trained in practical genetics at master's level. The service provides information and support about inherited heart conditions to anyone, and our service is confidential. Where indicated, a facilitated referral via the caller's GP can be made to a specialist IHC centre for assessment and consideration for genetic testing.

Inherited Heart Conditions testing in Wales

- The Miles Frost Foundation started after Miles Frost, son of David Frost passed away in 2015 from an undiagnosed inherited heart condition, hypertrophic cardiomyopathy (HCM). The fund has allowed more research to take place into the condition and now supports specialist nurses across the UK to help diagnoses in families who may have inherited heart conditions.
- The BHF award for a nurse specialist post, genetic counsellor and vital admin support is for two years. Louise Norgrove is the specialist nurse funded across ABMU (now Swansea Bay) and Hywel Dda UHBs since November 2018.
- There is considerable evidence of the significant impact this is already having on improving testing, treatment and support for HCM. It is important that this service can be sustained after the first two years and also that it can be rolled out into other areas of Wales, to reach even more families.

²⁰ https://stillmed.olympic.org/media/Document%20Library/OlympicOrg/News/20090716-The-IOC-Consensus-Statement-on-Periodic-Health-Evaluation-of-Elite-Athletes/EN-Health-Evaluation-of-Elite-Athletes-2009-report-1448.pdf#_ga=2.135516209.1617059244.1547820806-189988006.1547820806