UK National Screening Committee

14/365

UK National Screening Committee

Screening for Sudden Cardiac Death (SCD)

19 March 2015

Aim

1. This document provides background on the item addressing screening to reduce Sudden Cardiac Death in people aged 12 to 39.

Current policy

2. In 2008, a review was conducted on the leading cause of SCD in young people: hypertrophic cardiomyopathy. The recommendation was that systematic screening should not be introduced.

Current review

3. The scope of this review focuses on screening people between the age of 12 and 39, and explores a broad range of conditions associated with SCD, expanding on the previous review of hypertrophic cardiomyopathy.

4. Interest to explore the broader range of conditions was stimulated by a number of meetings between UKNSC representatives, CRY and Ministers. . The UK NSC are aware of the established screening programme for SCD in Italy however the impact of the programme to prevent SCD has not been reported.

5 Dr Phil Wiffen (Cochrane Pain, Palliative and Supportive Care Group) and Dr Mike Clarke (University of Belfast) were asked to review the evidence published since 2008 and the resulting document is attached. This reported that:

- While SCD is an important health problem, there is little peer reviewed evidence to enable an accurate assessment of the number of people suffering from SCD.

- The conditions that lead to sudden cardiac death are poorly understood and there is no evidence to guide clinicians regarding treatment or lifestyle advice when such a problem is found in a family member or when detected at a screening examination. Guidelines for the management of patients identified as being at risk are consensus based due to a lack of high quality evidence.

- No studies reporting on test performance (sensitivity or specificity) were identified by the literature search so it is not possible to recommend its use in a national programme.

- The literature largely addresses screening in young people participating in sporting activity, is predominantly not peer reviewed and the published outcomes have been questioned in peer reviewed literature.

- No direct evidence, for example, in a US population, was identified to conclude that an ECG or any other cardiovascular screening programme will reduce the incidence of SCD in any of the patient populations thought to be at increased risk.

Consultation

7. A three month consultation was hosted on the UK NSC website, and the following organizations were contacted directly: British Cardiovascular Society, British Congenital Cardiac Association, British Heart Foundation, Cardiac Risk in The Young, Cardio & Vascular Coalition, The Cardiomyopathy Association, Children's Heart Federation, Circulation Foundation, Faculty of Public Health, HEART UK, Institute of Child Health, The Oliver King Foundation, Royal College of Physicians, Royal College of GPs, and Royal College of Paediatrics and Child Health.

8. Three responses were received from members the public whose families were affected by SCD, and the following organisations:

    a. British Cardiovascular Society

Affirmed the overall conduct and main conclusions of NSC review and highlighted the limitations of the evidence and the potential harms of screening.

b. NHS England

A personal submission from the National Clinical Director for Heart Disease focused on ongoing work to ensure prompt management of affected individuals and follow up of family members.

c. Royal College of Paediatrics and Child Health

Responded to with no comments on the document or its conclusions.

d. Cardiac Risk in the Young (CRY)

Acknowledged many of the concerns identified by the review, in particular; the limitations of much of the data on incidence in the UK and internationally, the lack of high quality evidence relating to the effectiveness of screening in preventing SCD, the limited evidence base underpinning management of some risk factors for SCD and the ongoing uncertainty relating to the optimal screening strategy.

However CRY also suggested that the review:

- had conflated SADS (Sudden Arrhythmic death syndrome) and SCD which underestimated the incidence of SCD and had missed important published UK data on incidence published in a paper which had been submitted
- omitted the substantial evidence base underpinning the management of most risk factors for SCD which provided a rationale for the implementation of cascade testing and management services,
- did not take into account that ECG appeared to be the more favourable strategy when compared to other approaches, and that data on key test performance outcomes had been reported in papers which had been submitted
- in its current form, was likely to detract from work being undertaken to prevent and manage risk of SCD and to contradict national policy established by the National Service Framework and NICE guidance on the use of ECG in the management of transient loss of consciousness.

9. Discussion with the reviewers has taken place and this suggests that the submitted comments and papers would not lead to a change in the review's conclusions. A closer reading of the submitted papers is ongoing and the results of this will be reported to the UKNSC meeting.

Recommendation

10. The Committee is asked to consider and approve the following recommendations:

"The UK NSC does not recommend a systematic population screening programme for Sudden Cardiac Death.

There are serious limitations in the literature on fundamental issues relating to the condition, test, intervention and cost-effectiveness of a screening programme."

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Screening to reduce sudden cardiac death in people aged 12-39 years: an appraisal against UKNSC criteria

Response by Dr Ed Duncan on behalf of the British Cardiac Society

Sudden cardiac death in young people is devastating but rare. The role of screening young people to identify those at risk continues to be debated around the world. Currently practices differ widely and various models of screening program exist in some countries e.g. Italy and Israel. The BCS acknowledges that there are strong supporters of screening for SCD within the UK. However on balance, the BCS supports the NSC review document and it's major findings. The document is well researched. Concerns regarding the introduction of national screening to reduce sudden cardiac death in young people include:

1. Uncertainty regarding the actual incidence of sudden cardiac death in this population – the incidence is likely to be low.
2. Uncertainty about the best way to screen this population
3. Lack of evidence demonstrating that screening reduces the incidence of sudden cardiac death.
4. The high number needed to screen to identify those at risk of sudden cardiac death
5. The high false positive rate during screening
6. The lack of data demonstrating the cost effectiveness of screening

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Thanks Adrian

The Cardiovascular Disease Outcomes Strategy published by the Department of Health in 2013 recognised the potential for lives to be saved from out of hospital cardiac arrest, by increasing awareness of cardiopulmonary resuscitation (CPR) skills, access to public access defibrillators (PAD) and identifying family members of those who suffer cardiac arrest when young (<40 years). NHS England's National Clinical Director for Cardiac Care is working with the Chief Coroner, Resuscitation Council UK, the Ambulance Services, the British Heart Foundation and other charities to help pursue this goal.

Best wishes

Huon

Professor Huon H Gray MD FRCP FESC FACC
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XXXX XXXX
XXXX XXXX

UK National Screening Committee

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Dear Adrian

Thank you for inviting the Royal College of Paediatrics and Child Health to comment on the Screening to prevent Sudden Cardiac Death Policy Review. We have not received any responses for this consultation.

I would be grateful if you could please acknowledge receipt.

Kindest regards,
Sara

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Leading the Way in Children's Health

Response relating to the latest evidence summary produced by the National Screening Committee on: Screening to reduce sudden cardiac death in people aged 12-39 years: an appraisal against UKNSC criteria

We read with interest the report commissioned by the UK National Screening Committee (NSC) that essentially summarised part of the existing literature on screening to reduce sudden cardiac death (SCD) in young individuals. Although, there is little doubt, based on current evidence, that screening of young individuals for conditions predisposing to SCD remains a challenge, the review commissioned does very little to advance the field. The false premise and misleading conclusions of the report are a cause for concern as it depicts the field of inherited cardiac diseases and sports cardiology void of any evidence to guide investigation and management of individuals suspected or diagnosed with conditions associated with SCD. Of most concern, however, is the fact that this document will be potentially read by the thousands of families devastated by SCD and will falsely lead them to believe that there is very little anyone can do to prevent further tragedies in their family. In addition, this review is likely to cause confusion and undo all the hard work that the charitable organization Cardiac Risk in the Young (CRY) has done, as its conclusions are in direct opposition to the recommendations of the Department of Health and the National Institute of Health and Care Excellence (NICE).

Although we concede that high quality evidence relating to the effectiveness of screening in preventing SCD is lacking, it is imperative that major parts of this report are revised to ensure the NSC provides the public and in particular health system stakeholders, policy and decision makers with accurate conclusions. Below we provide a list of key points that should be reviewed. It is worth noting that most of these points were highlighted to the NSC during the consultation process. Unfortunately, it appears that the views and information provided by experts in the field were not conveyed to the authors of the document and as such the literature review forming the basis of this summary missed important evidence. The involvement of individuals with expertise in the field under investigation is of paramount importance in order to reach accurate and clinically useful conclusions that will benefit the population's health.

1. **Incidence data in Europe and the rest of the World:** We disagree with the authors that there is little peer-reviewed evidence relating to the incidence of SCD in the young. There are numerous studies in multiple countries in the literature that report on the incidence of SCD in the young, many quoted by the authors of the NSC review. Although there is variability relating to the quality of the evidence and definitions of the terms "young" and "athlete", it is the responsibility of the reviewers to assess the quality of the data. For example data relating to the studies by Maron et al. are predominantly based on a large dataset that Dr Maron has collected over the years. Although the effort is admirable, that dataset is largely dependent on media reports, which is hardly a sound scientific methodology upon which to base policy. It is very hard to believe that those data are

reliable as they attempt to collect deaths from the whole of the United States (more than 300 million). As such they can be considered, at best, the absolute minimum rather than an accurate estimate of the incidence of SCD in the US. The same limitations apply to the Israeli data by Steinvil et al. where the authors are basing their whole research and conclusions on a "systematic review" of two main newspapers.

Additional studies across Europe and the US that the authors should consider including in the review are studies in Ireland, Sweden and studies in young military recruits. For the benefit of the committee we provide a table summary with respective references.

Table 1: Summary of studies reporting the incidence of SCD and SADS in different young populations.

Country (Ref) (Region)	Studied population	Nature of study	Time period	Age in years	Incidence of SCD (SADS) per 100,000
USA[1] (Olmsted county)	General	Prospective	1960-1989	20-40	3.6
USA[2]	Military recruits	Retrospective	1977-2001	18-35	11.2
USA[3]	Military personnel	Retrospective	1998-2008	18-35	3.5
Sweden[4]	General	Retrospective	1992-1999	15-35	0.93
Denmark[5]	General	Retrospective	2000-2006	1-35	2.8
Ireland[6]	General	Retrospective	2005-2007	14-35	2.9
USA[7] (Minnesota)	High school athletes	Retrospective	1985-1997	15-17	0.46
USA[8]	High school athletes	Retrospective	2006-2007	14-17	4.4
Italy[9]	Athletes	Prospective	1979-2004	12-35	1.9

(Veneto)					

2. **Incidence data in the UK:** The section relating to the incidence of SCD in the UK is unfortunately inaccurate and incomplete. 1. The authors reference the database from the UK National Audit of Sudden Arrhythmic Death Syndrome and give the impression that within a period of four and a half years there were only 165 young deaths. The reader could justifiably infer that those numbers equate to less than 40 young deaths within the UK per annum, which is clearly not the case. Sudden Arrhythmic Death Syndrome (SADS) is only one aspect of young SCD. The term SADS is reserved for SCDs where the post-mortem fails to identify a cause of death and the heart appears structurally normal. Research has shown that a significant proportion of such deaths is due to inherited primary arrhythmia syndromes such as Long QT syndrome and Brugada Syndrome amongst others. So the terms SADS and SCD should not be used interchangeably. In addition, the SADS audit although initially an admirable initiative, it unfortunately failed to serve its purpose due to a number of reasons that I am sure the NSC committee identified during the initial consultation. As such the reported numbers are likely to be an underestimate of the true impact of SADS. 2. The authors refer to the study by Elston et al. It is important to highlight that the NSC commissioned this study and it was a “simulation” of the Italian data as published by Corrado et al. in the UK population. As such although the study may offer some insights to the potential impact of screening of young athletes in the UK, it should definitely not be used to quote incidence rates. 3. There is no reference to the most important UK data published to date (Papadakis M, et al. The magnitude of sudden cardiac death in the young: a death certificate-based review in England and Wales. Europace. 2009;11:1353-1358). It is

hard to understand how this could be the case as this study refers to the issue under investigation by the authors and in the relevant population cohort. In that particular study we reported an incidence of SCD of 1.8 per 100,000 per year for England and Wales, alone, with SADS accounting only for 57 of the 419 (13.6%) SCDs annually. 4. Finally, in the same section the research paper by Wilson et al. is quoted. This paper reported on diagnostic rates after cardiac screening of conditions predisposing to SCD, not incidence rates of young SCD and does not belong in this section.

3. **Investigation and management of individuals identified through screening:** This section lacks scientific rigor and the conclusions are false and misleading. The authors state in their key messages that "The conditions that lead to sudden cardiac death are poorly understood and there is no evidence to guide clinicians regarding treatment or lifestyle advice when such a problem is found in a family member or when detected at a screening examination." The authors go on to quote the consensus statements form the American Heart Association, the American College of Cardiology and the European Society of Cardiology relating to the management of patients with ventricular arrhythmias and participation of athletes with cardiac disease in competitive sports. It is beyond the scope of our response to outline the relevant literature relating to the investigation and management of conditions implicated in young SCD. However, below are a few pertinent points: 1. Screening to reduce SCD in the young is indeed challenging as we are looking for more than one condition, some of which are fairly novel and lack evidence based management. 2. However, for most conditions there is reasonable evidence in the literature. Such conditions include Hypertrophic

cardiomyopathy, Arrhythmogenic right ventricular cardiomyopathy, Dilated cardiomyopathy, Long QT syndrome, Cathecholaminergic polymorphic ventricular tachycardia, Brugada Syndrome and Accessory pathways (WPW), amongst others. 3. The fact that there are guidelines/recommendations for some of the conditions indicates that there is guidance for physicians who identify individuals with heart disease, irrespective of the level of evidence such recommendations are base on. 4. The conclusions of the review contradict the UK government. If indeed there is no established treatment then why did the Department of Health commission the 8th chapter of the National Service Framework for coronary heart disease, aimed at facilitating early identification of individuals at risk of SCD. That particular chapter states that “Quality requirement three: Sudden Cardiac Death. When sudden cardiac death occurs, NHS services have systems in place to identify family members at risk and provide personally tailored, sensitive and expert support, diagnosis, treatment, information and advice to close relatives.” It also states “Sudden cardiac death in younger people is often indicative of inherited cardiac disease. There is real potential to prevent further tragedies by the appropriate care of family members in these cases.” In line with these recommendations a number of specialists units, such as the one we run at St George’s Healthcare Trust have been created, in order to accommodate for the family needs, provide appropriate investigations, risk stratify individuals and prevent further SCDs.

Consistency or more explicit justification of recommendation differences between the NSC, Department of Health and NICE is imperative to avoid confusion of the public in general and health system stakeholders, policy and decision makers in particular.

4. **The different approaches to screening:** In their conclusion the authors state that until further evidence is available the dichotomy between the American Heart Association and the Italian approach to screening is likely to remain unresolved. This statement is correct but it is important to highlight a few key points: 1. The American Heart Association approach to screening is not supported by any evidence whatsoever, while ECG based screening is. On the contrary, there are studies, even from Dr Maron, the chair of the recommendations committee, which suggest that most individuals are unlikely to benefit from the American Heart Association approach to screening. So the question based on the available scientific data is either no screening or screening utilizing the 12-lead ECG. 2. There are a number of studies, most included in your references, comparing the two models, which unanimously show that ECG based screening is the most cost-effective option of the two, identifying more individuals at risk at the expense of more people being subjected to investigations (higher false positive rate). 3. The AHA views do not represent the views of the whole US scientific community and this is reflected in their screening practice. Most of the professional sporting organisations and colleges follow the Italian protocol utilizing the 12-lead ECG. There are even charitable organisations, similar to Cardiac Risk in the Young, that perform ECG screening of young, physically active individuals in a large scale (Young Hearts for Life, http://www.yh4l.org/, Marek et al. Feasibility and findings of large-scale electrocardiographic screening in young adults: Data from 32,561 subjects. Heart Rhythm. 2011; 8:1555–1559).

5. **The test:** 1. Although there is no doubt that the ECG, similarly to other screening tests, has its limitations, it is a simple, safe and validated test as required by the WHO and UKNSC criteria. Moreover, the ECG has been studied extensively as a screening tool, particularly in the UK, where our group amongst others has tested the ECG in young individuals of different gender, ethnicity, level of exercise and sporting discipline. Our group, through its extensive screening programme is leading the field, Internationally, and there are two clear recommendation documents from the ESC and the Seattle group that not only provide the clinician with clear recommendations of how to interpret an athlete's ECG but are also accompanied by on-line training modules. For the benefit of the committee I have appended some of our seminal publications on ECG, all conducted in a British population (References 10-16, up till Jan 2014). 2. There are a number of studies referring to the sensitivity and specificity of the 12-lead ECG as a screening tool which are not referenced in the review. (References 17-23, up till Jan 2014). 3. The conclusions of the authors appear to be directly at odds with the NICE support for commissioning for transient loss of consciousness **(**http://www.nice.org.uk/guidance/sfcgs71/chapter/the-quality-statements-and-their-commissioning-and-resource-implications), which states that "A 12-lead ECG is an important initial diagnostic test for identifying the likely cause of transient loss of consciousness in some people, and especially in predicting adverse events (for example, ECG abnormalities that are 'red flag' signs or symptoms may suggest structural heart disease or potential for arrhythmic syncope)."

6. **There should be evidence from high quality Randomised Controlled Trials that the screening programme is effective in reducing mortality or morbidity:** We agree in principle with the authors that this is the ideal standard we should all aspire to. However, that standard is not always achievable in many aspects of science and life, in general. Sudden cardiac death in the young is thankfully not a common occurrence and the conditions that cause it are relatively rare. As such a large population would be required over decades to prove benefit. This is something we aspire to do through our CRY screening programme. However, it would be technically and ethically challenging to perform a large scale randomized controlled study, particularly in the light of the results of the 2006 Corrado et al. study and studies relating to the effective management of individuals identified with an inherited cardiac disease. In addition, in the UK, as is the case with most European countries and North America, there has been increasing demand for screening of young individuals. As such substantial parts of the population are being screened throughout the country. It would therefore be impossible to identify a large enough population, which is not being screened and will not be screened over the next decade.

This particular point has been discussed extensively on previous occasions with the NSC, as we feel that is unacceptable to fall back on the lack of randomized controlled trials as the basis for insufficient evidence. We would expect the NSC to be more explicit on what they would require from research to influence policy. If a randomized controlled trial is the standard they require, then commissioning regular reviews is a thankless task.

REFERENCES

1. Shen WK, Edwards WD, Hammill SC, Bailey KR, Ballard DJ, Gersh BJ. Sudden unexpected nontraumatic death in 54 young adults: a 30-year population-based study. *Am J Cardiol*. 1995;76:148–152.

2. Eckart RE, Scoville SL, Campbell CL, Shry EA, Stajduhar KC, Potter RN, Pearse LA, Virmani R. Sudden death in young adults: a 25-year review of autopsies in military recruits. *Ann Intern Med*. 2004;141:829–834.

3. Eckart RE, Shry EA, Burke AP, McNear JA, Appel DA, Castillo-Rojas LM, Avedissian L, Pearse LA, Potter RN, Tremaine L, Gentlesk PJ, Huffer L, Reich SS, Stevenson WG, Department of Defense Cardiovascular Death Registry Group. Sudden death in young adults: an autopsy-based series of a population undergoing active surveillance. *J Am Coll Cardiol*. 2011;58:1254–1261.

4. Wisten A, Forsberg H, Krantz P, Messner T. Sudden cardiac death in 15-35-year olds in Sweden during 1992-99. *J Intern Med*. 2002;252:529–536.

5. Winkel BG, Holst AG, Theilade J, Kristensen IB, Thomsen JL, Ottesen GL, Bundgaard H, Svendsen JH, Haunsø S, Tfelt-Hansen J. Nationwide study of sudden cardiac death in persons aged 1-35 years. *Eur Heart J*. 2011;32:983–990.

6. Margey R, Roy A, Tobin S, O'Keane CJ, McGorrian C, Morris V, Jennings S, Galvin J. Sudden cardiac death in 14- to 35-year olds in Ireland from 2005 to 2007: a retrospective registry. *Europace*. 2011;13:1411–1418.

7. Maron BJ, Gohman TE, Aeppli D. Prevalence of sudden cardiac death during competitive sports activities in Minnesota high school athletes. *J Am Coll Cardiol*. 1998;32:1881–1884.

8. Drezner JA, Rao AL, Heistand J, Bloomingdale MK, Harmon KG. Effectiveness of emergency response planning for sudden cardiac arrest in United States high schools with automated external defibrillators. *Circulation*. 2009;120:518–525.

9. Corrado D, Basso C, Pavei A, Michieli P, Schiavon M, Thiene G. Trends in sudden cardiovascular death in young competitive athletes after implementation of a preparticipation screening program. *JAMA*. 2006;296:1593–1601.

10. Basavarajaiah S, Wilson M, Whyte G, Shah A, Behr E, Sharma S. Prevalence and significance of an isolated long QT interval in elite athletes. Eur Heart J 2007;28:2944-2959.

11. Papadakis M, Basavarajaiah S, Rawlins J, Edwards C, Makan J, Firoozi S, Carby L, Sharma S.. Prevalence and significance of T-wave inversions in predominantly

Caucasian adolescent athletes. Eur Heart J 2009;30:1728-1735.

12. Rawlins J, Carre F, Kervio G, Papadakis M, Chandra N, Edwards C, Whyte GP, Sharma S. Ethnic differences in physiological adaptation to intense physical exercise in highly trained female athletes. Circulation 2010;121:1078-1085.

13. Papadakis M, Carre F, Kervio G, Rawlins J, Panoulas VF, Chandra N, Basavarajaiah S, Carby L, Fonseca T, Sharma S. The prevalence, distribution and clinical outcomes of electrocardiographic repolarisation patterns in male athletes of African/Afro-Caribbean origin. Eur Heart J 2011;32:2304-2313.

14. Sheikh N, Papadakis M, Carré F, Kervio G, Panoulas VF, Ghani S, Zaidi A, Gati S, Rawlins J, Wilson MG, Sharma S. Cardiac adaptation to exercise in adolescent athletes of African ethnicity: an emergent elite athletic population. Br J Sports Med 2013;47:585-592.

15. Zaidi A, Ghani S, Sheikh N, Gati S, Bastiaenen R, Madden B, Papadakis M, Raju H, Reed M, Sharma R, Behr ER, Sharma S. Clinical significance of electrocardiographic right ventricular hypertrophy in athletes: Comparison with arrhythmogenic right ventricular cardiomyopathy and pulmonary hypertension. Eur Heart J 2013;34:3649-3656.

16. Gati S, Sheikh N, Ghani S, Zaidi S, Wilson M, Raju H, Cox A, Reed M, Papadakis M, Sharma S. Should axis deviation or atrial enlargement be categorised as abnormal in young athletes? The athlete's electrocardiogram: time for re-appraisal of markers of

pathology. Eur Heart J 2013, 34: 3641-3648.

17. Wilson MG, Basavarajaiah S, Whyte GP, Cox S, Loosemore M, Sharma S. Efficacy of personal symptom and family history questionnaires when screening for inherited cardiac pathologies: the role of electrocardiography. Br J Sports Med 2008;42:207–211.

18. Bessem B, Groot FP, Nieuwland W. The Lausanne recommendations: a Dutch experience. Br J Sports Med 2009;43:708–715.

19. Hevia AC, Fernández MM, Palacio JM, Martín EH, Castro MG, Reguero JJ. ECG as a part of the preparticipation screening programme: an old and still present international dilemma. Br J Sports Med 2011;45:776–779.

20. Vetter VL, Dugan N, Guo R, Mercer-Rosa L, Gleason M, Cohen M, Vogel RL, Iyer R. A pilot study of the feasibility of heart screening for sudden cardiac arrest in healthy children. Am Heart J. 2011 May;161(5):1000-1006.

21. Drezner JA1, Asif IM, Owens DS, Prutkin JM, Salerno JC, Fean R, Rao AL, Stout K, Harmon KG. Accuracy of ECG interpretation in competitive athletes: the impact of using standised ECG criteria. Br J Sports Med. 2012;46:335-40.

22. Koch S, Cassel M, Linné K, Mayer F, Scharhag J. ECG and echocardiographic findings in 10-15-year-old elite athletes. Eur J Prev Cardiol. 2012:24;21:774-781.

23. Weiner RB, Hutter AM, Wang F, Kim JH, Wood MJ, Wang TJ, Picard MH, Baggish AL. Performance of the 2010 European Society of Cardiology criteria for ECG interpretation in athletes. Heart. 2011;97:1573-1577.

UK National Screening Committee
Screening for Sudden Cardiac Death- an evidence review

Consultation comments pro-forma

Name:	**Mr XXXX XXXX**	**Email address:**	**XXXX XXXX**
Organisation (if appropriate):	**Family of XXXX XXXX XXXX XXXX XXXX XXXX XXXX XXXX**		
Role:	Father of **XXXX XXXX**		

Do you consent to your name being published on the UK NSC website alongside your response?

Yes ☒ **No** ☐

Section and / or page number	Text or issue to which comments relate	Comment
21	SCD Poorly Understood	To my knowledge there is a clear understanding of the causes of SCD, in particular SADS, and Arrhythmia based inherited heart conditions and how to detect them. An article published in the New Scientist on Feb 1st 2015 would seem to support this, and the need to screen. I believe this report should be taken into account. http://www.newscientist.com/article/dn26890-virtual-hearts-get-to-the-crux-of-sudden-cardiac-death.html#.VM3s0jUeLMI Content follows:

Virtual hearts get to the crux of sudden cardiac death

- 00:01 01 February 2015 by **Michael Slezak**
- For similar stories, visit the **Genetics** Topic Guide

Virtual human hearts beating on supercomputers are helping get to the bottom of the most mysterious of heart diseases – sudden arrhythmic death syndrome.

When someone dies suddenly and unexpectedly, there is often an underlying cardiac problem. If a post-mortem doesn't find one, sudden arrhythmic death syndrome (SADS) is recorded as the cause. SADS can result from a number of genetic conditions that affect the way electrical signals pass through the cardiac muscle making the heart beat. One day – often during physical exertion – the person's heart may begin to beat in a fast, uncontrolled way. This can kill them if their heart doesn't right itself quickly enough. Around 1.3 deaths in every 100,000 can probably be attributed to SADS, and the same genetic problems may also play a role in sudden infant deaths.

If someone has the genetic mutations, they can be treated with drugs or have a defibrillator implanted in their chest. But how do you work out who is at risk? Genetic tests can help but not everyone with the altered genes seems to have the syndrome. Electrocardiograms or ECGs can measure the heart's electrical activity, but exactly how features on the ECG relate to risk is not fully understood.

All in the t-wave

Enter the virtual heart. By running hundreds of genetically customised hearts on a supercomputer, each for many thousands of beats, Adam Hill and his colleagues from the Victor Change Cardiac Research Institute in Sydney, Australia, have cracked some of the secrets of SADS.

		One sign that someone has the genetic condition that most commonly leads to SADS, known as long QT syndrome, is a distinctive bump or notched t-wave in their ECG readout. "For the past 30 years, that notched t-wave has been in the diagnostic criteria but nobody's known what's caused it," says Hill. "We show what causes it." With the wealth of virtual data created by running the simulations, they were able to establish that the more extreme the bump in the ECG is, the higher a person's risk of dying. What's more, they found the main genes thought to cause the problem can be either amplified or compensated for by complex combinations of other genes. **Better diagnosis** "We show that the degree of t-wave notching is correlated with how much risk they are at," says team member Arash Sadrieh. "So person A can have the mutation [but his ECG shows] he's absolutely normal, so you don't need to do the complex surgery to prevent sudden cardiac death. And if his sister has a more notched t-wave, then she is at more risk." It would have been impractical to use real hearts for this research as you'd need huge numbers of people with specific genetic combinations, all with their full genome sequenced, hooked up to an ECG for days. Hill says the team has taken the virtual trial data, applied it to patient records of ECGs and found the finer grained analysis of the ECG led to more accurate diagnoses. They're also making progress using the simulations to distinguish between different types of long QT syndrome. "The work is quite a milestone in terms of how thoroughly they've investigated this issue of the notched t-wave…and how you interpret it," says Peter Hunter from the University of Auckland in New Zealand, one of the world's leading cardiac modelling experts. "This has pushed it to a new level."

		Journal Reference: *Nature Communications*, DOI: 10.1038/ncomms6069 Much data has been gathered by the Charity CRY (Cardiac Risk in the Young) from their excellent and forward thinking screening program that actually detects I understand between 2 and 3% of those screened require further Cardiac Investigations. Their screening program is PROACTIVE and saves lives. NOT REACTIVE as per the current NSF 8.0 Directive that so few GP practices seem to be aware of.
22	SCD Definition	Recent evidence suggests that SCD or SADS can ALSO happen without physical activity having taken place. In fact I understand many young people never wake from their sleep. There is also I believed a suspected linkage to Sudden Cot Death. In the case of our daughter, she was in a Jacuzzi following exercise, and it is suspected that the increase in temperature due to time in this unit (which was at a correct use temperature) caused her to die from SADS (Brugada Syndrome?). We are 99.99% sure it was Brugada Syndrome as **XXXX XXXX** has been diagnosed (and treated with an ICD) as a result of subsequent screening to have Brugada Syndrome that has been passed down via **XXXX XXXX** blood line! **XXXX XXXX** also have Brugada, diagnosed through screening. I accept that further gentic work is required to identify the gene that is defective, but the current treatment is effective (ICD). If our daughter had been screened PROACTIVELY she would be alive today!
23	SADS Screening	Italy has been screening active young people I understand for over 25 Years, and have reduced the mortality rate due to SADS by 90%. This seems to have been ignored in the report. SADS is in many cases caused by genetically inherited defects, not by lifestyle. If it is triggered by lifestyle, and you do not know you have one of the genetic defects or SADS conditions through PROACTIVE screening, then how can you be expected to alter your lifestyle? If our daughter had known she had Brugada syndrome should would NOT have entered into the Jacuzzi, and also would probably have had an ICD fitted!
24	SCD Deaths	I understand that AT LEAST 12 young people die each WEEK in the UK from SADS, with the number probably closer to 20 per week or more. Currently not all Coroner reports conclude SADS as the cause of death, but still put down the death as natural

		causes. This I presume is due to lack of awareness and education. It is NOT I understand just prevalent in Athletes, but also happens when resting or without physical activity! Detecting a heart defect via voluntary screening, which MIGHT be a problem is a POSITIVE, PROACTIVE and sensible approach. After all it is the person's life! Not that of the establishment! Waiting until a person dies who does not know they have a problem, which could have been detected by screening is UNFORGIVEABLE nor is it ethically acceptable. The impact to the family is immense and unrecoverable. And the loss to the country should also be considered.
25	Screening	It would appear Italy & Israel seem to have good screening programmes in place for ACTIVE young people, Also the sports bodies in the UK. The UK NSF 8.0 would appear to be very outdated and inadequate with respect to PROCATIVE SCREENING, and has a poor awareness of its existence by GP's (from family experience) in the UK.
Conclusion	General	It would appear the report and its findings is written in such way as to influence the reasons NOT to introduce PROACTIVE Screening, as opposed to benefits to the community as a whole of the benefits of PROACTIVE screening and the phased introduction. It does not appear to offer any clear recommendations on the way forward.

UK National Screening Committee

14/365

Response to the appraisal of screening for Sudden Cardiac Death by Wiffen and Clarke dated June 2014 commissioned by UKNSC

We are writing in response to the UKNSC appraisal for Sudden Cardiac Death carried out by Wiffen and Clarke and welcome the opportunity to respond.

We assume that the National Screening Committee will only base their findings on available published data but currently this will not provide a complete picture and reflect what is actually happening across the UK. We accept firm data is hard to come by. Indeed, we were told at a meeting at The Department of Health in July 2013 by Dr Anne Mackie that based on available documented evidence there were probably only 60 deaths a year from sudden cardiac death which after a challenge from ourselves was increased in November 2013 to an estimate of 400 deaths a year by The National Service Framework on Coronary Heart Disease; rather a large difference.
This number is without doubt on the conservative side.

The report dated June 2014 by Wiffen and Clarke correlates data available up to 31st March 2013 and therefore, by the time the report is finalised the data will be two years out of date. In the authors disclaimer they state that they "do not warrant the accuracy of this document" and "do not warrant that the information contained in these pages is current, accurate or complete". We wonder how the UKNSC can base any decisions at all on this report.

We are neither medics nor statisticians and therefore do not have any relevant statistics but it is important that you have input from personal experience to balance that obtained from published scientific data.

It is recognised that until recently not all sudden cardiac deaths have been recorded in Coroners Inquest verdicts and of course this discrepancy will not show up in your current statistics.
Many deaths have also been incorrectly recorded. We know this from personal experience.

Our own daughter, our only child, died suddenly in February 2012 from a rare cardiac related condition which combined with horrendous failings by **XXXX XXXX** hospital, caused her to have a cardiac arrest with tragic consequences.
She was a fit and healthy 21 years old.

Since her death we have been acutely aware of the alarming number of seemingly healthy young people who have died from sudden cardiac death.

Annually, there are hundreds of bereaved families who contact charities such as CRY (Cardiac Risk in the Young) following the sudden death of a young family member from

cardiac related conditions. It should be noted that not all families bereaved in this way make contact with any charity.
We assume you have all the data from CRY.

It is imperative that you consider the wider picture as behind every statistic there is a human story and the effects of a sudden death in a young person has far reaching effects.

The immense impact that the grief and shock caused by the sudden death of a young person will have on family, friends and the wider community must never be underestimated. The consequences of bereavement can have huge effects mentally, emotionally and consequently physically on all those concerned.
This in itself results in hidden costs to the NHS.

We doubt you will have any relevant statistics for this.

Screening may not have helped our daughter but there are many deaths in the
12 - 39 age group that could be avoided by simple electrocardiology (ECG) and Trans-thoracic Echocardiography (TTE) testing.
The Wiffen and Clarke report highlights the point that many young people are engaged in sport often at a highly competitive level and it is this group that appears to be the most vulnerable from undetected cardiac conditions. However, this group are not routinely tested in the UK unless funded privately by professional bodies such as the FA and LTA.

This does not mean that young people who do not play sport are any less at risk.
Their conditions may not manifest themselves till later in life and of course these deaths in the over 39 age group will not appear in your statistics.

Currently a young person in the 12 to 39 age group, whether they engage in competitive sport or not, cannot request an ECG test through their GP. The GP will only agree to this test if symptoms of cardiac related problems are evident or suspected or they have a family history of cardiac issues. The only option available is to attend screening sessions funded by charities such as CRY usually paid for by funds raised by bereaved families or by paying for an ECG at privately funded hospitals.

This situation is either ideal or fair.

Where can these young people go to be tested? There is no easily available option.

Screening tests may not be 100% accurate and will show false positive and negative results such as the PSA test for prostate cancer. However, these false positives/negatives are a small price to pay.

Cardiac screening too may show false positive/negative results or abnormalities following an ECG test.
This will naturally cause a high degree of anxiety especially in a young person.
However, if a TTE test is done immediately after an abnormal ECG to establish whether there is a major issue then much of this anxiety can be eliminated.
Currently there is often an unnecessarily long delay between an abnormal ECG and a TTE being carried out.

It has been accepted by the Department of Health that young people with family histories of cardiac problems should be identified and offered screening. It was only in January 2014 that Professor Huon H Gray, the National Clinical Director for Cardiac Care at NHS England following a meeting with the Chief Coroner in October 2013, agreed a letter with NHS England that went out to all Coroners encouraging them to identify cases of sudden cardiac death in those under 40 years of age so that family members can contact their local specialist Inherited Cardiac Conditions service. This is a help but perhaps of greater benefit to the family member who died from sudden cardiac death would have been knowing that they had a potentially life threatening condition by a simple cardiac screening test which could have prevented their death.

Is it acceptable for one young family member to die before any testing of siblings takes place?

In our opinion, and in an ideal world, all parents of children aged 14 and above should have the opportunity for their children to undergo an ECG test and if it shows irregularities then an immediate TTE test should be available. Adults between 18 and 39 years old should also have this option and this screening should be available whether or not they participate in sport.

If it is not feasible or realistic to organise screening at schools or GP practices then it would seem logical to have regional centres where this is possible and if not then mobile screening units could be an option.
Doing nothing should not be the default position.

Screening may not save everyone at risk but the absence of a screening programme will not save anyone.

In the Wiffen and Clarke conclusion the authors acknowledged that “it is always regrettable that young people die whatever the circumstances” but “until a reliable evidence base is gathered … the issue of cost and benefit remain unresolved”

The only conclusion that can be derived from those statements is that the hundreds of families a year (check the CRY and the National Service Framework on Coronary Heart Disease data) which are affected by a sudden cardiac death with all the devastation,

heartache, despair and pain that death inevitably brings not just to close family members but peer groups and friends are an acceptable consequence of the inability of the NHS to implement a cardiac screening programme because the cost/benefit issue cannot be resolved.

What if this was your child who had died?

A recent report has stated that more women die from cardiac related problems than from breast cancer. Whilst lifestyle choices may have an impact on these figures how many of these deaths in the over 39 age group could be saved by early screening? Of course there are no statistics or data available to answer that question.

As already mentioned there are also no statistics available for the huge number of bereaved families who through the intense emotional stress caused by the death of a child have to resort to the NHS for treatment of one kind or another.

What is the resultant cost to the NHS?

What is the resultant cost to the NHS for hugely expensive cardiac procedures on the over 39 age group whose symptoms could have been identified and treated at an earlier age and at less cost?

What cost does the NHS put on the lives of those 400 young people at least who die from sudden cardiac death every year?

In conclusion:

- It is important to look at the wider picture and not just the facts and figures of published papers. Waiting for new data will cost more lives.

- Do not underestimate the hidden costs to the NHS that result from bereaved families.

- Early screening would not only save young lives now but also the lives of people over 39 years of age.

- Early screening could result in future savings to the NHS.

- Currently there is no readily available choice for 12 – 39 year olds to be screened for cardiac abnormalities by the NHS which means over the next seven years thousands of unsuspecting young adults will lose their lives to SCD.

- Screening should be available to all young people regardless of whether or not they play sport and wherever they live in the UK.

- The NHS has to manage its current resources in a responsible manner but it also has to look to its future costs by investing in preventative measures now.

- No one at UKNSC appears to be prepared to take the long view if the data and statistics are not available.

- The NHS is in the main a reactive organisation; it should become more of a proactive organisation to reduce costs in the future.

- In an ideal world cardiac screening should be available to all.

We know only too well that it is neither a fair nor an ideal world. However, none of us should be complacent and we all have an obligation to look after our young people to the best of our ability.

They are the future and the UKNSC has it within its power to effect change.

Thank you again for the opportunity to respond to this report.

XXXX XXXX January 2015

In memory of our daughter and the hundreds of young people who lose their lives every year from undiagnosed sudden cardiac death.

UK National Screening Committee

14/365
XXXX XXXX
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10th February, 2015

Dr Anne Mackie
Director of Programmes
UK National Screening Committee
Floor 2, Zone B
Skipton House
80 London Road
London, SE1 6LH

Dear Dr Mackie

I have been sent your letter to **XXXX XXXX**, which replied to her letter to you regarding possible screening for Sudden Cardiac Death after I contacted her regarding this after our youngest son died **XXXX XXXX** of a sudden a completely unexpected heart attack aged 41. You say that the UK NSC is currently consulting on this and would welcome feedback. I am not exactly sure of what you require, but give below a review of our family history and what happened to our son **XXXX XXXX**.

As a family my husband suffers with atrial fibrillation. This first manifested itself at the age of 39 after he went diving for the first time. At that time his heart went back into normal sinus rhythm of its own accord. However, 10 years later, after an extremely stressful time working on an aid project in Kenya the AF returned. This time it needed treatment and he had several electric shock sessions, none of which worked for long. However, aged 74 he has now been on a regime of Flecainide and warfarin for 26 years and has had no further problems.

XXXX XXXX was the youngest of our sons, and he and also our eldest son had never had any heart problems of which we or they were aware. However, our second son had AF aged 17 while still at school (unbeknown to us) when playing strenuous games – he was exceptionally good at all sports. Now aged 47, he has been on Flecainide ever since. However he tells me that he sometimes goes into AF when playing squash or running a short distance for a bus, although oddly enough when having a stress ECG (which he says left him shattered!) the result was completely normal. So it only seems to occur when a sudden burst of energy/stress is required, not when building up to it slowly.
Our third son also has AF problems, starting when he was about 29 after a wedding party at which he consumed too much wine followed by a lot of coffee! He was prescribed Pruprafenone, but only very rarely takes this if he has a problem – now usually caused more

by stress than the combination of alcohol and caffeine. The doctor at the hospital in **XXXX XXXX** who first treated him in 1999, was keen to do some tests on all the family to see if a gene could be discovered that might have been causing the problem. But with the relevant family members living in **XXXX XXXX** at that time it was sadly too complicated to organize and our second son now lives in **XXXX XXXX**.

So far as **XXXX XXXX** was concerned, he had been complaining of a stuffed up nose and "wheezyness" since October 2013 and had seen a doctor in **XXXX XXXX** who had diagnosed some sort of allergy. **XXXX XXXX** Christmas that year he went to see one of the doctors at the surgery in **XXXX XXXX** who also considered that he was suffering from an allergy. He returned to **XXXX XXXX** in January 2014 and moved **XXXX XXXX**, where we visited him last February and he was still having the same problems. I found out the names of the two doctors in the town and gave them to him, but suspect that he never got around to seeing either of them (being a man!). In July he had a recurrence of the malaria that he had first had in 2000 and self-medicated – we did not know about this at the time and do not know if might have had any relevance, but suspect that it may have, although we do not think that he would have known that this might affect his heart.

In the middle of August, having apparently spent a perfectly normal day - walking his dogs before breakfast, going shopping and working at his computer and then walking his dogs again in the late afternoon – he was in the middle of his supper when he must have found he could hardly breathe, rushed out of the house, opened his gates, reversed his car out into the road, shut the gates again and then driven about ½ mile to the nearby clinic, where he collapsed on the floor saying "Please help me, I can't breathe, I think I'm dying". He was helped (half walking/half being carried) to the bed with oxygen and all the resuscitation equipment right beside it. The doctor lived next door and was there within 2 minutes she said, but in spite of all their best efforts he could not be revived. The doctor told us she had never seen anyone die so quickly.

When we were first told the news **XXXX XXXX** we assumed that he must have had a severe asthma attack, although he had never been diagnosed with asthma, but his "wheezyness" suggested that. However, after a post mortem had been carried out three days later it transpired that he had died from cardiac infarcation. The doctor who did the PM told us that he had been having a heart attack for at least 12 hours before he actually died, but presumably he was totally unaware of this, or presumably he would have sought help earlier in the day. This doctor also told us that he had been suffering from untreated heart disease for a number of years, probably caused by stress.

This made a certain amount of sense, as **XXXX XXXX** had nursed his wife of 2 years through ovarian cancer 5 years earlier and had suffered a lot of stress and depression during that time and ever since. He was really only coming out of this in the few months before he died, having realised after moving **XXXX XXXX** that a lady he had met there soon after his wife died, and who had also lost a fiancé in a car accident and so understood what he was going through, was actually more than just a good friend. He was in the process of buying another

house in **xxxx xxxx** to be with her when he died so unexpectedly - leaving all of us devastated and in total shock. He should have been moving into his new home with her on the day of his funeral.

The doctor who did the PM also told us that a simple ECG or even a chest X-ray, which would have revealed an enlarged heart, would have shown that he had a heart problem which could have been treated. Due to this I feel that a national screening programme could save so many (often quite young) lives and save so many families from the anguish we have all suffered. We are relatively lucky – at least he did not leave any orphaned children to be cared for; nor did he kill or injure anyone on his drive to the clinic. Only last month, here in **xxxx xxxx** a good friend (aged 91) suffered a heart attack while driving his car. Fortunately he was in a small, quiet, residential road and, having just turned the corner into it, he was not going at all fast. His car simply veered off the road into a wall. But if this had happened to him just 2 minutes later he would have been on one or other of the two steep main roads into the town and he could have unwittingly caused endless damage and possibly injury or even loss of life to other people.

I hope that the UK NSC will now review carefully this problem of SCD, especially in men over the age of 40 – this I understand is the main criteria for having heart disease: to be male and over 40. Hopefully more accurate tests for SCD are now available than were possible in 2008 when a screening programme was last considered. However, it does seem to me that if a young man's life could be saved by, at the age of 40, having an ECG (available in most surgeries) or a simple chest X-ray, which cannot cost much and is available in even small hospitals, then overall a huge amount of money could be saved by the State not having to look after that man's family, possibly now with no wage earner in the family, quite apart from the distress that his death would have caused to so many people.

If I can help with any further information, then please do not hesitate to contact me. I am sorry that this is rather long-winded, but I do not know exactly what sort of details would be helpful to the Committee.

Yours sincerely,

xxxx xxxx