Citizen Participation and Public Petitions Committee
Wednesday 5 March 2025
4th Meeting, 2025 (Session 6)

PE2067: Improve data on young people affected by conditions causing Sudden Cardiac Death

Introduction

Petitioner Sharon Duncan

Petition summary Calling on the Scottish Parliament to urge the Scottish Government to commission research to establish how many people aged 14-35 are affected by conditions that cause Young Sudden Cardiac Death; clarify the number of people who die annually in Scotland from these conditions; and set up a pilot study to establish if voluntary screening can reduce deaths.

Webpage https://petitions.parliament.scot/petitions/PE2067

1. The Committee last considered this petition at its meeting on 20 March 2024. At that meeting, the Committee agreed to write to CRY - Cardiac Risk in the Young, Save a Life for Scotland, the British Heart Foundation, St John Scotland, Chest, Heart and Stroke Scotland, the UK National Screening Committee, the Network for Inherited Cardiac Conditions (NICCS), and the Scottish Government.

2. The petition summary is included in **Annexe A** and the Official Report of the Committee's last consideration of this petition is at **Annexe B.**

3. The Committee has received new written submissions from the Network for Inherited Cardiac Conditions (NICCS), the Minister for Public Health and Women's Health, Cardiac Risk in the Young (CRY), the British Heart Foundation, Chest, Heart and Stroke Scotland (CHSS), the UK National Screening Committee, and the Petitioner, which are set out in **Annexe C.** St John Scotland declined to provide a formal response on this occasion.

4. Written submissions received prior to the Committee's last consideration can be found on the petition's webpage.

5. Further background information about this petition can be found in the SPICe briefing for this petition.

6. The Scottish Government gave its initial position on this petition on 13 December 2023.

7. Every petition collects signatures while it remains under consideration. At the time of writing, 2,081 signatures have been received on this petition.

Action

8. The Committee is invited to consider what action it wishes to take.

Clerks to the Committee
February 2025

Annexe A: Summary of petition

PE2067: Improve data on young people affected by conditions causing Sudden Cardiac Death

Petitioner

Sharon Duncan

Date Lodged

14 November 2023

Petition summary

Calling on the Scottish Parliament to urge the Scottish Government to commission research to establish how many people aged 14-35 are affected by conditions that cause Young Sudden Cardiac Death; clarify the number of people who die annually in Scotland from these conditions; and set up a pilot study to establish if voluntary screening can reduce deaths.

Previous action

I have written to, and met with, my MSP, Oliver Mundell, regarding the lack of clarity in data currently available.

I have also introduced an MSP Pledge urging MSP's to support a national strategy to prevent Young Sudden Cardiac Death and help save the lives of at least 12 healthy young people who die every week. Along with other bereaved parents, I have raised money to provide screening and publicised this issue in national press and television.

Background information

On 19 March 2022, my son, David Hill, died while playing for the Parliament's rugby team in Dublin. Almost a year after his death we found that he had died from an undiagnosed genetic condition which stopped his heart.

There is no screening programme for young people with these conditions and current estimates are that there are at least 12 preventable deaths each week in the UK.

Cardiac Risk in the Young (CRY) support and fund research as well as providing screening, which is mostly funded by bereaved families. Through this, CRY believes the incidence of young people identified with a potentially fatal cardiac condition (if untreated) to be 1:300, with another 1:100 to be found with a condition that could cause serious issues later in life if not monitored. The National Screening Committee (NSC) believe the incidence to be approx. 1 or 2:100,000. This discrepancy makes it difficult to establish the benefit of funding a national strategy. With accurate data from Scotland, the NSC could revisit their decision.

Screening costs £65 per person, and initially consists of an ECG, with follow-up by cardiologists.

Annexe B: Extract from Official Report of last consideration of PE2067 on 20 March 2024

The Convener: Our next new petition is PE2067, on improving data on young people who are affected by conditions causing sudden cardiac death. The petition, which was lodged by Sharon Duncan, calls on the Scottish Parliament to urge the Scottish Government to commission research to establish how many people aged 14 to 35 are affected by conditions that cause young sudden cardiac death; clarify the number of people who die annually in Scotland from those conditions; and set up a pilot study to establish whether voluntary screening can reduce deaths.

Members will be aware that the petition has been lodged by the mother of parliamentary staffer David Hill, who tragically passed away while playing in an inter-parliamentary rugby match two years ago almost to the day, on 19 March 2022. I understand that members of the family have joined us in the public gallery, and we extend our condolences and a warm welcome to them.

As the petition notes, there is currently no screening programme for young people for conditions that put them at risk of sudden cardiac death. The SPICe briefing that we have received notes that there are difficulties in reaching agreement on the number of young people who are affected by sudden cardiac death. Those include the way in which deaths are classified and the fact that research focuses on athletes rather than the general population.

In responding to the asks of the petition, the Minister for Public Health and Women's Health notes the Government funding that has been provided to the west of Scotland inherited cardiac conditions service and the network for inherited cardiac conditions to deliver a sudden cardiac death project, with a focus on improving clinical pathways for families and enhancing data quality. The minister has also made inquiries to the UK National Screening Committee about plans to review its position on population screening for conditions that are associated with sudden cardiac death in the young.

We have received a submission from the petitioner, Ms Duncan, emphasising the importance of understanding the incidence of those conditions to developing treatment pathways. Ms Duncan also seeks clarity on the coding that is used to inform data on incidence, and highlights that no account is taken of deaths such as David's, where the death is registered as being from natural causes, despite the post-mortem and follow-ups confirming a previously undiagnosed genetic cardiac condition.

Do members have any comments or suggestions?

Oliver Mundell: I should say that I know David's family well and it is lovely to see them in the public gallery. I have the utmost admiration for Sharon, his mother, who in very difficult circumstances has sought to see what she can do to help other families.

I have seen the SPICe briefing but, for me, it comes back to a point that Mr Ewing made on a previous petition: what if Sharon Duncan, the wider Hill family and some of the organisations that they are working with are right, and the National Screening Committee is wrong? Certainly, if it were my child, I would want to know that that question had been exhausted.

I would be keen for the committee to write to organisations with a relevant interest—Cardiac Risk in the Young, Save a Life for Scotland, the British Heart Foundation, St John Scotland and Chest Heart and Stroke Scotland—to seek their views and expertise on what is called for in the petition, and to find out about any work that they may be undertaking on conditions affecting sudden cardiac death.

I would also be keen for the committee to write to the UK National Screening Committee to ask when it expects to review the evidence for screening for sudden cardiac death, and to write to the network for inherited cardiac conditions seeking further details and an update on its sudden cardiac death project.

In addition, I would be keen to go back to the Scottish Government. It has provided quite a helpful response on the petition, but I would be keen to interrogate further its role in informing the National Screening Committee's work. It is one thing to ask questions and make representations, but I do not know how much more it can do.

Certainly, David Hill's family and Sharon Duncan, his mother, are not in a unique position. There are families like them in every part of Scotland, as we have seen through activities that have been undertaken in Parliament since David's death. The least that those people deserve is for us to try to understand how the process works and be absolutely sure that all the evidence has been taken into consideration.

Fergus Ewing: I entirely agree with everything that Oliver Mundell has just said. As he said, the minister, Jenni Minto, gave a fuller and more useful reply than some of the replies that we get, which should be acknowledged, but there are many complex issues raised here.

I want to make one point on the record. The SPICe document refers to a UK Government blog that gives reasons as to why there should not be a screening programme. Those include that people might be unnecessarily anxious, that false reassurance might be provided, or that they might be encouraged to get treatments that may be inappropriate.

I felt uneasy about that reply. There must be many screening programmes where not that many people will be detected as having the particular problem for which the screening is designed, but that does not mean that we do not have screening. I just want to put on the record that those arguments seem very weak and actually pretty offensive to people who have lost a loved one because of the condition. I hope that the minister will take that into account.

In addition to the points that Mr Mundell raised, could we ask for high-level information on what screening programmes are undertaken, to find out whether some are undertaken where there is a serious risk of death but, statistically speaking, not many people in the population are at risk?

Foysol Choudhury: I agree with both of my colleagues. I understand that the UK National Screening Committee is conducting a review and that the next review is expected to be completed in 2024. Do we know exactly when in 2024 that will be, and will we be informed of the recommendations?

The Deputy Convener: Thank you for that. Is the committee agreed with all those recommendations?

Members *indicated agreement*.

The Deputy Convener: Thank you. I thank the members of David Hill's family for attending.

Annexe C: Written submissions

Network for Inherited Cardiac Conditions Scotland written submission, 5 April 2024

PE2067/D: Improve data on young people affected by conditions causing Sudden Cardiac Death

The Network for Inherited Cardiac Conditions Scotland (NICCS) was established in 2008 (formerly, the Familial Arrhythmia Network Scotland) to address some of the challenges in caring for families affected by sudden cardiac death (SCD) or sudden unexpected death (SUD). The network has agreed guidance in Scotland, but despite this, we know from lived experience that more needs to be done and we continue to hear from families that have had a challenging journey following SUD/SCD to get to the right services at the right time.

When a death is sudden, or unexpected, there are legal requirements to ascertain the cause of death. In 15-20% of SCD/SUD cases unascertained at post-mortem a concealed electrical or heart muscle disorder may be identified through molecular genetic testing (references 1 and 2). At such a devastating time, the process can feel confusing and complicated, and investigations can take a long time (reference 3).

The West of Scotland Inherited Cardiac Conditions Service, based in NHS Greater Glasgow and Clyde received 18 months funding in 2023 for a cardiac nurse specialist to evaluate areas of need and develop better support for families in Scotland. This project has 3 work streams:

1. **Data quality and regional / national audit**. Working with NICCS, the Scottish Cardiac Audit programme (SCAP) and regional genetics and pathology services, we aim to standardise data collection and develop a robust national audit process.

2. **Pilot an integrated care system for SUD/SCD**. By appointing a cardiac nurse specialist we will facilitate early contact with families, which we know is critical to perception of risk and engagement with screening. Focus groups will gather feedback on service needs.

3. **Education, information and bereavement support.** Grief associated with SUD/SCD is particularly complex when people die at a young age and/or when other family members may be affected. We are working with educational and public information to improve access to resources.

The nurse specialist post commenced in February 2024 and has already made important progress by gathering information best practice and registry data form across the world. We look forward to working closely with families and 3rd sector organisations to drive improvement in this area.

References

1. Lahrouchi et al. Utility of Post-Mortem Genetic Testing in Cases of Sudden Arrhythmic Death Syndrome, JACC 2017, Vol 69 (17) p2134-44

2. Stiles et al. 2020 APHRS/HRS expert consensus statement on the investigation of decedents with sudden unexplained death and patients with sudden cardiac arrest, and of their families' Heart Rhythm Journal, 18 (1)

3. Allan et al. A Qualitative Study of Families' Experiences When a Young Relative Dies of Sudden Cardiac Death. Circ Cardiovasc Qual Outcomes. April 2023, p279-287

Minister for Public Health and Women's Health written submission, 19 April 2024

PE2067/E: Improve data on young people affected by conditions causing Sudden Cardiac Death

Thank you for your consideration of our previous response and your further correspondence asking for clarification on the Scottish Government's role in informing the work of the UK National Screening Committee (UK NSC), specifically whether the Scottish Government can make recommendations to the UK NSC. I hope the information provided below is helpful.

As you know, the UK NSC does not currently recommend systematic population screening of people under the age of 39 for cardiac conditions associated with sudden cardiac death (SCD). This recommendation was last reviewed in December 2019, and details regarding the reasons they did not recommend screening can be found on their website: Sudden cardiac death - UK National Screening Committee (UK NSC) - GOV.UK (view-health-screening-recommendations.service.gov.uk)

However, the UK NSC will review this recommendation in due course, and will take account of any new evidence available to them at that time. Should they recommend a screening programme following this review, then the organisations with responsibility for screening in Scotland will advise Scottish Ministers on how to take that recommendation forward.

The UK NSC is an independent, expert advisory group which advises all four UK nations on aspects of screening. It is sponsored by the Department of Health and Social Care on behalf of the other UK countries, and is accountable to the four Chief Medical Officers of the UK, who also agree the workplan for the UK NSC and have visibility on any decisions made.

Designated representatives, including those from the Scottish Government, develop, support and deliver the UK NSC strategic plan to enable and ensure national policy alignment across the UK. Representatives from Scotland attend all UK NSC meetings and clinicians and policy officials remain in close contact to discuss any emerging issues. However, officials cannot vote on committee decisions and at all times must respect and uphold the independence of the committee in formulating its advice. For the same reason, the Scottish Government and the other governments of the UK cannot tell the UK NSC which issues it should consider or review.

The UK NSC formulates its advice and recommendations based on the best and most up to date evidence available. The process involves a robust, transparent, and comprehensive appraisal of the evidence available. Reviews provide an overview of

the best quality and informative studies relevant to the internationally agreed criteria for assessing the viability, effectiveness and appropriateness of a screening programme.

Guidance on how the UK NSC assesses the evidence base against its criteria can be found here - UK NSC evidence review process - GOV.UK (www.gov.uk)

Kind Regards,

Jenni Minto MSP
Minister for Public Health and Women's Health

Cardiac Risk in the Young (CRY) written submission, 21 April 2024

PE2067/F: Improve data on young people affected by conditions causing Sudden Cardiac Death

Thank you for raising this issue in Scottish Parliament and the comments which were raised on 20th March 2024 in response to the petition.

Our response to the document will be around two concerns; how the incidence of young sudden cardiac death (YSCD) is **calculated** and **understood.**

How the incidence of YSCD is calculated

The focus of this petition is to correctly establish the incidence of Young Sudden Cardiac Death (YSCD) and the most appropriate measures to identify those at risk prior to a cardiac arrest or death. This is of utmost importance because government advice has consistently failed to address the first, and most important, question of how many young people this affects and therefore grossly underestimated the impact these conditions are having on families, communities and wider society. This point is clearly demonstrated in the SPICe briefing, where an FOI in June 2023 reporting 19 myocardial infarctions in 0-29 year olds in 2022 has been referenced. This is an example of how the data informing policy, and being given in response to politician's questions, underrepresents the incidence and the impact of these deaths. It is understood by specialists that there are inaccuracies in the way YSCDs are recorded within the Office of National Statistics (ONS), with the mortality statistics largely derived from documentation on death certificates which may under-report the true incidence of cardiac arrhythmias. This was discussed by Papadakis et al., (2009) (acknowledged in the SPICe response), which evaluated the ONS data and reported cardiac death rates in England and Wales.

The recent paper of the largest SCD cohort with autopsy findings ever reported (N=7,214), between 1994 and 2021, (Sheppard et al, 2023) presents SCD figures based on examinations (macroscopically and microscopically) by two expert cardiac pathologists. When compared to the FOI in June 2023 it highlights the significant disparity between government advice and actual death rates. The paper presents data on referrals to the centre by year, age groups, gender and conditions. 49% of the cases referred to the centre were under the age of 35 (N=3,547) and in 2021 there were 499 hearts were referred to the centre, 634 hearts in 2020. It is important to note this centre does not see all cases of SCD in the UK.

How the incidence of YSCD is understood

Whilst the SPICe briefing has provided an explanation of the statistical expression of "number of deaths per 100,000 people/year" it is unclear why they have used a hypothetical scenario of diabetes to illustrate this. Furthermore, notwithstanding the error in the 3rd bullet point, the way they have presented this does not enable the following question to be answered, ***based on the evidence which is supporting the NSC's policy and understanding of the incidence of Young Sudden Cardiac Death, how many of the young people who are aged 14 today are anticipated to die before their 36th birthday of a heart condition.***

When trying to evaluate the impact of YSCD it is necessary to understand the incidence of YSCD. Many people appear to believe there are only 2 per 100,000 people dying of these conditions. However, they are confusing incidence, incidence rate and cumulative incidence. These are common measures used in Epidemiology. The confusion is when 2 different statistics are conflated, "XX per 100,000 people" with "XX per 100,000 people **per year**". 2 per 100,000 people **per year** is the **incidence rate** (also known as **incidence density rate** or **person-time incidence rate**). However, in the case of young sudden cardiac deaths, the person is at risk of dying over many years, not just one year. In the case of CRY's screening programme it is testing people between the age of 14 and 35, covering a 22 year period of risk. It is therefore important to calculate the **cumulative incidence** (or **incidence proportion**).

A crude approach to extrapolating an estimated incidence rate of 2 deaths per 100,000 people per year, over 22 years of being at risk, would estimate approximately 44 deaths per 100,000 people, 1 in 2,272 people, dying of the conditions between the age of 14 and 36. This is oversimplified on a number of accounts, including incidence rates are likely to differ significantly by age, gender, lifestyle and ethnicity. However, it highlights how a person may perceive the risk of SCD to be very differently if they understood it to be 1 or 2 per 100,000 people, as opposed to 1 in 2,272 people, or more for higher risk groups (e.g. male athletes).

In Italy, the incidence rates of young people involved in organised sport (prior to the implementation of their screening programme) was 3.6 per 100,000 per year (Corrado et al, 2006). In the case of screening elite footballers Malhotra et al, (2018) reported an incident rate of 6.8 per 100,000 per year, post screening. This illustrates that male elite athletes are at significantly higher risk (compared to the general population), and this figure was calculated after cardiac screening had identified 42 high risk cases.

Whilst this analysis has evaluated the risk of dying before the age of 36, it is important to recognise there is a **lifetime risk** from these conditions, with sudden deaths from conditions causing SADS and cardiomyopathies occurring after the age of 35 (Sheppard et al, 2023).

The error, or misunderstanding of the incidence of YSCD, is why we have made repeated requests to meet with the National Screening Committee to clarify this issue, so politicians are correctly briefed. We have also requested for the NSC to transparently publish the pre-screening and post-screening incidence death rates for other conditions which meet the NSC screening criteria.

There are a number of issues and inaccuracies raised in the SPICe briefing which have been addressed previously in our response(s) to the National Screening Committee (for instance much of the research is not limited to “professional” athletes, it is young people involved in organized sport). These can be read on the CRY website.

British Heart Foundation (BHF) Scotland written submission, 22 April 2024

PE2067/G: Improve data on young people affected by conditions causing Sudden Cardiac Death

Thank you for your invitation for British Heart Foundation Scotland to give views in response to PE2067: Improve data on young people affected by conditions causing Sudden Cardiac Death.

I would like to first share BHF Scotland’s condolences to the family of David Hill. We applaud their work to raise awareness of sudden cardiac death and have welcomed the opportunity to hear from them at the Cross-Party Group on Heart and Circulatory Diseases, to which BHF provides the secretariat.

We note the briefing from the Scottish Parliament’s Information Centre (SPICe) which highlights the uncertainty around the incidence of Sudden Cardiac Death in those aged under 35.

BHF Scotland supports the position of the Scottish Government to follow the recommendation of the UK National Screening Committee on population screening for cardiac conditions. We note that the current recommendation is against a population level screening programme, with the reasons laid out in both briefing from SPICe and the response from the Minister for Public Health and Women’s Health.

Research is vital to improving our understanding of both the prevalence of the conditions that cause sudden death and how to best identify those at risk.

BHF Scotland would be supportive of any high-quality research studies into both the prevalence of conditions that cause sudden cardiac death in those under 35, as well as pilot projects that could provide further evidence around the effectiveness of screening at a whole population level.

BHF Scotland is involved in funding research into the causes of sudden cardiac death and screening for related conditions. Research funded by the BHF, alongside Cardiac Risk in the Young (CRY) has improved the interpretation of ECG tests in the screening of young athletes and demonstrated significant reductions in false positives[1], and influenced protocols around testing for cardiac conditions following a sudden cardiac death[2].

[1] Sharma et al. 2017. International Recommendations for Electrocardiographic Interpretation in Athletes.
[2] Anastasakis et al. 2016. Sudden unexplained death in the young: epidemiology, aetiology and value of the clinically guided genetic screening

However, we recognise that despite this progress the methods of screening currently developed are not evidenced to be precise enough to be accurate at a population level.

In addition to efforts to identify and prevent sudden cardiac events, BHF Scotland also believes that action is critical to improve survival from sudden cardiac arrest. Currently, only 1 in 10 people in Scotland survive a cardiac arrest. Whilst this figure is double the level in 2015, there are more actions to be taken and evidence shows that CPR and timely defibrillation can more than double the chances of survival.

RevivR is a new online CPR training course developed by the BHF that allows people to learn CPR in just 15 minutes without any specialist equipment – all you need is a smartphone and a cushion. To specifically support CPR skills in young people we have developed Classroom RevivR, a tool designed to teach CPR skills to 11 to 16-year-olds and is free to all educational settings.

Additionally, The Circuit is a first-of-its-kind national defibrillator network developed by the BHF and its partners that connects all defibrillators in the UK to a single network. Registering defibrillators on The Circuit allows ambulance services to quickly direct people to their nearest defibrillator. As of February 2024, there were more than 7,600 defibrillators registered on the circuit in Scotland.

BHF Scotland is committed to reducing the impact of sudden cardiac death in Scotland through research, encouraging CPR training and registering defibrillators.

BHF Scotland supports the UK National Screening Committee in their recommendation not to implement population level screening based on the most up to date available evidence. We would, however, support the funding of further research to better understand the prevalence of these conditions and how best to effectively screen for them on a whole population basis.

We would be happy to provide further details or evidence if the Committee would find this useful and look forward to any opportunities to further explore this important and urgent topic.

Chest, Heart and Stroke Scotland (CHSS), written submission, 26 April 2024

PE2067/H: Improve data on young people affected by conditions causing Sudden Cardiac Death

Chest Heart & Stroke Scotland (CHSS) would like to thank the Citizen Participation and Public Petitions Committee for the opportunity to respond to PE2067: Improve data on young people affected by conditions causing Sudden Cardiac Death.

Firstly, we send our sincere condolences to Sharon Duncan and all of David Hill's family for their tragic loss. We welcome Sharon's work to draw parliamentary attention to this important issue.

Chest Heart & Stroke Scotland is Scotland's largest organisation supporting people with chest, heart, and stroke conditions, including Long Covid. We ensure that

people across Scotland get the support they need to live full lives, including through community-based prevention and early detection services.

As part of our prevention work, we have recently introduced the use of portable ECG monitors which can identify Atrial Fibrillation. Our Health Defence team based in Maryhill, Glasgow have also worked on community detection of other conditions, such as hypertension, supporting hundreds of people to understand health risks and to be referred on for further support where required. We also deliver an Out of Hospital Cardiac Arrest (OCHA) Aftercare project to support those who witness or provide CPR to someone who experiences cardiac arrest at home or in the community.

Early detection of heart conditions saves lives and allows for the greatest chance to access successful treatment that reduces disability. As such, CHSS wants to ensure that Scotland's approach to heart condition detection and prevention reaches as many people as possible. This approach must be both comprehensive and informed by the best available evidence.

While the most recent UK National Screening Committee review does not currently recommend systematic population screening of people under the age of 39 for cardiac conditions associated with sudden cardiac death (SCD), it is important to note that this review did also conclude that 'further research is necessary to understand whether screening is effective'.

As addressed in the Petition, the SPICe briefing provided to the committee, and in the NSC review of the evidence, the true incidence of Sudden Cardiac Death in the young is uncertain and there is a clear need to establish stronger data on this.

Since much of the available research on focuses on young athletes, there is also a clear need to understand the appropriateness of screening at a general population level, consider ways to mitigate potential unintended negative consequences of screening such as false positives, and ascertain the most effective form of screening programme. For example, while resting electrocardiograms (ECGs) may improve diagnostic yield, research in young athletes found 25% of people affected by a disease that may lead to sudden cardiac death would still remain undetected.

CHSS believes Scottish Government commissioned research, including a pilot study on voluntary screening, could provide crucial insight and offer a valuable contribution to the current evidence base. In particular, any research undertaken should address:

- Reliability and accuracy of testing.
- Effectiveness of screening to the general population.
- Condition treatment and management when identified at screening.
- Potential unintended consequences of screening (including false positives).

Ultimately, Chest Heart & Stroke Scotland supports the petition's calls for further research, including a pilot study on voluntary screening. It is crucial that we build a robust evidence base on the impact of screening. We are very happy to engage further with the Committee on this important issue and discuss in greater detail.

UK National Screening Committee written submission, 9 May 2024

PE2067/I: Improve data on young people affected by conditions causing Sudden Cardiac Death

Thank you for your letter addressed to Professor Sir Mike Richards in respect of the petition PE2067. I am responding on his behalf as the head of the UK National Screening Committee (UK NSC) secretariat.

The UK NSC reviewed population screening for Sudden Cardiac Death (SCD) in 2019 and concluded that there was not enough evidence to recommend screening because there is:

- not a sufficiently predictive test for risk of SCD
- insufficient understanding of the genetic risk for SCD
- no agreed treatment for those detected through screening

The UK NSC's article alert and horizon scanning function and its open discussions with stakeholders have not indicated there is significant new work on whole population screening that would suggest a different outcome to that of the 2019 review. However, the UK NSC plans to review the evidence relating to population screening for SCD within the next three years, in line with their current work plan.

The committee will wish to note that since the review in 2019, the committee's terms of reference have been expanded to include consideration of targeted (groups of people identified as being at elevated/above average risk of a specific condition), and stratified screening programmes.

The UK NSC has not been asked to consider targeted or stratified screening for SCD, but it can be alerted to any new published peer reviewed evidence which may suggest the case for a new screening programme.

Proposals to change or review a topic early can be submitted via the UK NSC's annual call which will open in July 2024. More information can be found at UK NSC annual call: submitting a screening proposal.

There are currently 11 national screening programmes (in England), covering 36 health conditions.

The NHS newborn blood spot (NBS) screening programme enables early identification, referral and treatment of babies with 9 rare but serious conditions. These are sickle cell disease (SCD), cystic fibrosis (CF), congenital hypothyroidism (CHT), phenylketonuria (PKU), medium-chain acyl-CoA dehydrogenase deficiency (MCADD), maple syrup urine disease (MSUD), isovaleric acidaemia (IVA), glutaric aciduria type 1 (GA1) and homocystinuria (HCU). Ministers in England have also agreed this year to add tyrosinemia (HT1) to the conditions tested for at this stage.

More directly related to the issue of SCD, is the physical exam offered to babies at birth and again at 6-8 weeks. This tests for (among other things) congenital heart disease. This test was introduced in 2014, meaning children under 9 years of age should have been tested for this condition.

There are also adult screening programmes for bowel, breast and cervical cancers, abdominal aortic aneurysms (AAAs) and for diabetic retinopathy.

The UK NSC does not make decisions regarding which conditions to screen for based on how rare a condition is or is not. Rather, it assesses evidence against a set of internationally recognised criteria covering the condition, the test, the treatment options, and the effectiveness and acceptability of the screening programme.

More information on the UK NSC and NHS England's screening programmes can be found https://www.gov.uk/government/organisations/uk-national-screening-committee and https://www.gov.uk/government/publications/population-screening-applying-all-our-health/population-screening-applying-all-our-health.

We hope this information provides you with what you were looking for.

Yours sincerely,

Professor Anne Mackie
Head of the UK NSC Secretariat

Petitioner written submission, 18 February 2025

PE2067/J: Improve data on young people affected by conditions causing Sudden Cardiac Death

I thank the Committee for this opportunity to add to the information heard last March and for this review relating to my petition.

I am unable to find evidence of the NSC review due in 2023/24. There appears to be universal agreement about a lack of clarity regarding the numbers of young people affected, and even after Jenni Minto, Minister for Public Health and Women's Health, contacted the NSC, they simply said "in due course". Considering the last review is now 6 years old, I believe their data and information is significantly outdated, and hope the Committee might be able to shed light on this situation. Their review appears overdue, and young people are still dying.

I welcome the response from NICCS, noting they agree that "more needs to be done". The importance of genetic testing is recognized, and my son David falls into the 15-20% diagnosed, as a group who would never have been identified, being asymptomatic and with no family history. However, this is only instigated after a death for relatives of the deceased. Perhaps if screening had been available, David might still be here. However, in the 3 years since he died, assuming 12 deaths weekly, 624 have died each year – over 1,800 young people lost and families devastated.

The response from CHSS is appreciated, and I am grateful for their work to support those suffering a cardiac arrest out of hospital, and provision of defibrillators. However, a defibrillator is "a treatment of last resort" (Dr Steve Cox, CRY CEO) and can still be fatal, even if the defibrillator gets them to hospital. Survival rates are increasing, but this is population wide, not specific to young people with no apparent underlying disease. CHSS noted from the previous NSC review that "further

research is necessary" and support my petition. Analysis from Dr Steve Cox (CRY CEO) shows how difficult it is to accurately show the numbers affected by conditions causing SCD, and why a common language is needed in describing the number of deaths. I commend his analysis to this Committee.

I had a meeting last March with then First Minister Humza Yousaf, along with Health Secretary Neil Gray, Oliver Mundell MSP, and 2 cardiologists, I believe from Queen Elizabeth Hospital in Glasgow – all of whom were supportive of the aims of this petition to clarify the impact of SCD. The First Minister reassured me how seriously the Scottish Government takes the issue of SCD in the young, and agreed to discuss with the Chief Scientific Officer commissioning or supporting research into the impact of diseases leading to SCD in Scotland. I await news from Neil Gray MSP regarding this.

After meeting with the First Minister, I attended a round-table meeting, "One Voice, Many Hearts", at Westminster, chaired by a parent who sadly lost her daughter last year, where published research was shared, discussions held about what effective preventative strategy might look like, and how to build a policy for a screening programme. This involved MPs, cardiologists, associated professionals, bereaved families and survivors of diseases that cause SCD.

The impact of SCD on young people was again discussed only a few weeks ago at a meeting reported in the press with the then Parliamentary Under-Secretary of State at the Department of Health and Social Care, Andrew Gwynne MP, also attended by Dr Steve Cox, Professor Mary Sheppard (Director of Cardiovascular Pathology, CRY), Professor Ann Mackie (Director of Screening for Public Health England) and a fellow CRY parent who lost her son to SCD some years ago. At this meeting, Professor Sheppard pointed out that 12 deaths a week is an outdated figure and shared that she receives at least 16 hearts a week for detailed post-mortem, knowing there are others she doesn't receive. If we inform our estimation on Professor Sheppard's baseline number, then we are losing 832 young people every year, and over 2,500 since my son has died. Professor Mackie simply expressed her sorrow at my friend's loss of her daughter, and remained steadfast in her opinion that losses of 12 a week are an overestimation, in spite of hearing clear evidence to the contrary from Professor Sheppard. It is disappointing that Public Health England and the UK National Screening Committee appear unwilling to adjust their positions in light of updated evidence being shared with them.

The same friend also attended a meeting at the Italian Embassy in London, coordinated by the same bereaved parent; to review the impact of their screening decisions and how it could inform changes in practice here in the UK. The meeting included the Italian Ambassador, the President of the International Federation of Sports Medicine, the President of both Italian and European Federation of Sports Medicine, the Cardiology President of the Royal Society of Medicine, as well as several prominent cardiology experts from institutions including St George's University Hospitals, Guy's and St Thomas' hospitals, and Cambridge University – who spoke about the Cambridge screening programme. The discussion covered aspects of how other countries manage this issue, what the UK needs in order to meaningfully reduce the incidence of young SCD, and how to get there. There is in Italy mandatory screening for all young people involved in organised sport (backed by research) including school groups who are not professional athletes, where the

deaths from SCD have been reduced by almost 90%. While “mandatory” isn’t what we advocate, this shows a need to consider some kind of voluntary screening for our population.

SCD has been highlighted so often, especially over the last year and there is data showing that screening saves hundreds of lives every year. I urge the Committee to push the Scottish Government to take the lead in securing new research regarding voluntary population screening to clarify the extent of SCD in Scotland’s population, and the incidence of conditions that cause it, which impact on cardiac outcomes beyond the age of 35, without relying on the death of a young person to instigate screening of the surviving family.

Petitioner written submission, 25 February 2025

PE2067/K: Improve data on young people affected by conditions causing Sudden Cardiac Death

With my second submission I would like to address the statements from the British Heart Foundation (BHF) and the UK National Screening Committee (NSC).

I am pleased to note the agreement from BHF regarding the uncertainty surrounding the incidence of Sudden Cardiac Death (SCD) and the conditions which can cause SCD.

I am heartened by BHF’s commitment to raise the knowledge of CPR in the population at large, and the increase in the availability of defibrillators. We all know these together can, and do, save lives. However, the BHF report a survival from cardiac arrest as only 1 in 10, and these figures are for the population in general of all ages, many of whom have underlying conditions, and doesn’t take into account the number of asymptomatic young people affected. If anything, this highlights the perspective that CPR and use of a defibrillator is still a treatment of last resort, and to prevent such a catastrophic situation we must intervene before this point.

The BHF submission clearly states “[r]esearch is vital to improving our understanding” and I welcome their offer of support in high-quality research to “better understand the prevalence of these conditions and how best to effectively screen for them on a whole population basis.” This is precisely what my petition asks for – research with pre-agreed parameters and reporting of incidence of both prevalence of conditions and number of deaths, which is essential to avoid any ambiguity regarding the scope of these conditions.

There is already collaboration between BHF and CRY, which could form the basis of the pilot study research I am seeking.

I acknowledge the response from Professor Anne Mackie, on behalf of the UK NSC Secretariat, and recognise her conclusions from the 2019 review.

I am disappointed that the NSC now plan to review evidence relating to population screening for SCD in the next three years – after the 2019 review, it was clear that the next review would be expected in 2023/24, which has not yet happened, and now it will be up to a further three years before this will happen. How many young

people will die because of such a delay? Perhaps if a review had been conducted when expected there would be recognition of the body of research that has been published since the last review, the increased effectiveness of the testing available and the progress in treatment pathways.

I am grateful the NSC will now consider targeted screening. This might provide an agreeable starting point for the conversation regarding development of a screening programme, I struggle with the idea of stratified screening for SCD. By its very nature, SCD is impacting young, generally fit and healthy people with no underlying cardiac concerns, and I fail to see how identifying their risk factors will ensure screening is effective, as they are the very ones who don't have identified risk factors!

I am sure the physical exam of babies will improve detection of congenital heart disease, but as it was only introduced in 2014, there are many in the population who have not benefitted from this and remain asymptomatic while potentially at risk. Additionally, congenital heart disease is only one aspect of the conditions leading to SCD, and takes no account of the other causes including genetic conditions, or the impact of the number of people in the population who will die from the accumulated causes of SCD.

I believe that the pilot study I seek will provide critical evidence to present to the UK NSC whenever the next review happens, and with the backing promised from the Scottish Government and collaboration with all agencies offering support, will not only save the lives of young people in Scotland, but lead to a four nation approach towards the development of effective screening for Sudden Cardiac Death.