Bristol Myers Squibb™

## More than Inherited Cardiac Conditions:

# Driving Policy Reform in ICCs

This report has been developed and funded by Bristol Myers Squibb.

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# STATEMENTS OF SUPPORT

Cardiomyopathy UK
the heart muscle charity

## Statement from Anna Tomlinson on behalf of Cardiomyopathy UK

*As Chief Executive of Cardiomyopathy UK, I am pleased to welcome this important report and the insights it provides. It is vital that the voices and experiences of those affected by cardiomyopathy and ICCs continue to inform service development across the UK.*

*We know that many people face significant barriers in accessing timely diagnosis, consistent care, and specialist support. Variability in services and limited awareness contribute to gaps that impact the quality of care patients receive. Yet alongside these challenges are real opportunities for progress. By raising awareness, improving diagnostic pathways, expanding access to genetic testing, and strengthening specialist services, we can transform the experience of individuals and families living with cardiomyopathy.*

*The recommendations in this report set out a clear roadmap for change. If acted upon, they have the potential to deliver meaningful improvements in care, outcomes, and support. Cardiomyopathy UK is committed to working closely with clinicians, policymakers, and the patient community to ensure these opportunities are realised, so that everyone affected by cardiomyopathy receives the care and support they deserve.*

> ***This report makes clear both the challenges in accessing consistent, specialist care and the real opportunities to improve services. By acting on its recommendations, we can ensure better diagnosis, support, and outcomes for everyone affected by cardiomyopathy across the UK.***

**Anna Tomlinson,**

**Chief Executive, Cardiomyopathy UK**

Anna Tomlinson has been Chief Executive of Cardiomyopathy UK since March 2025. Cardiomyopathy UK is the national charity for anyone affected by the heart muscle disease, cardiomyopathy. The charity's vision is for everyone affected by cardiomyopathy to lead long and fulfilling lives. Anna has more than 20 years of experience in the charitable, healthcare, and professional services sectors, including holding senior leadership positions at Genomics England and LifeArc.

# Statement from Dr Stephen Page on behalf of the Association of Inherited Cardiac Conditions (AICC)

*The delivery of specialist services has evolved rapidly over the last twenty years but has been limited by the lack of a national strategy for service delivery, the lack of structured data collection, and the lack of an infrastructure to coordinate care across geographical regions. This has resulted in marked geographical variation in service provision and access to specialist care.*

*This policy reform report for Inherited Cardiac Conditions (ICCs) is a vital initiative in our attempt to address these issues. The Association for Inherited Cardiac Conditions (AICC) is committed to leading this transition, towards a more equitable, higher quality range of services across the UK. The AICC fully endorses this report with specific reference to improving awareness of ICCs amongst healthcare professionals, the empowerment of primary care, and a desire to improve access to specialist services such as self-referral pathways. These goals, along with the prioritisation of diagnostic genetic testing, are perfectly aligned with defined NHS priorities.*

*This policy reform report for Inherited Cardiac Conditions (ICCs) is a vital initiative in our attempt to address these issues.*

**Dr Stephen Page – President, Association of Inherited Cardiac Conditions**

Dr Stephen Page is a Cardiologist and Electrophysiologist at Leeds General Infirmary. He is the clinical lead for the regional ICC Service for West and North Yorkshire and the President of the AICC. As President, he has led the development of the AICC National Survey and is on the Writing Group for the new ICC Service Specification. Dr Page's main interest is in service development, and he hopes to strengthen the ability of the AICC to promote and support equitable service delivery across the UK.

# Statement from Professor Daniel Augustine on behalf of the President British Society of Echocardiography

*The BSE welcomes this report and strongly supports the recommendations within. ICCs can have devastating consequences and early detection is paramount. Early diagnosis will ensure that people living with ICCs can access timely treatment, long-term monitoring and maintain a good quality of life.*

*Echocardiography is pivotal to the diagnosis of these conditions and this report supports our own work in highlighting the current challenges faced by the UK echocardiography workforce, as well as the recommendations we have made to address them. If we are to make early diagnosis a reality for this community, we need policy makers to prioritise retention within the workforce. Developing advanced roles and physiologist led services will support retention of our most experienced echocardiographers and increase capacity in ICC clinics.*

*We are delighted that these issues are being raised on behalf of the ICC community and we urge policy makers to act. The report offers new insights and reiterates previous recommendations which could, if implemented, result in significant improvements in care for these individuals and their families. As a key stakeholder, the BSE is committed to doing what it can.*

***The report offers new insights and reiterates previous recommendations which could, if implemented, result in significant improvements in care for these individuals and their families. As a key stakeholder, the BSE is committed to doing what it can.***

**Professor Daniel Augustine - President, British Society of Echocardiography**

Prof Daniel Augustine is a consultant cardiologist and became President of the British Society of Echocardiography (BSE) in October 2023. He has published widely on the assessment of heart pump function and the use of echocardiography, which has included national guidelines. The BSE represents clinical echocardiography professionals working at all levels and in all areas of the field. With over 5000 members, its aim is to provide them with the necessary support to deliver the highest standard of care in echocardiography. An internationally recognised provider of accreditation for both individuals and departments/service providers and education, the Society also produces guidelines to promote best practice, support the continuing advancement of cardiac ultrasound and reduce variation.

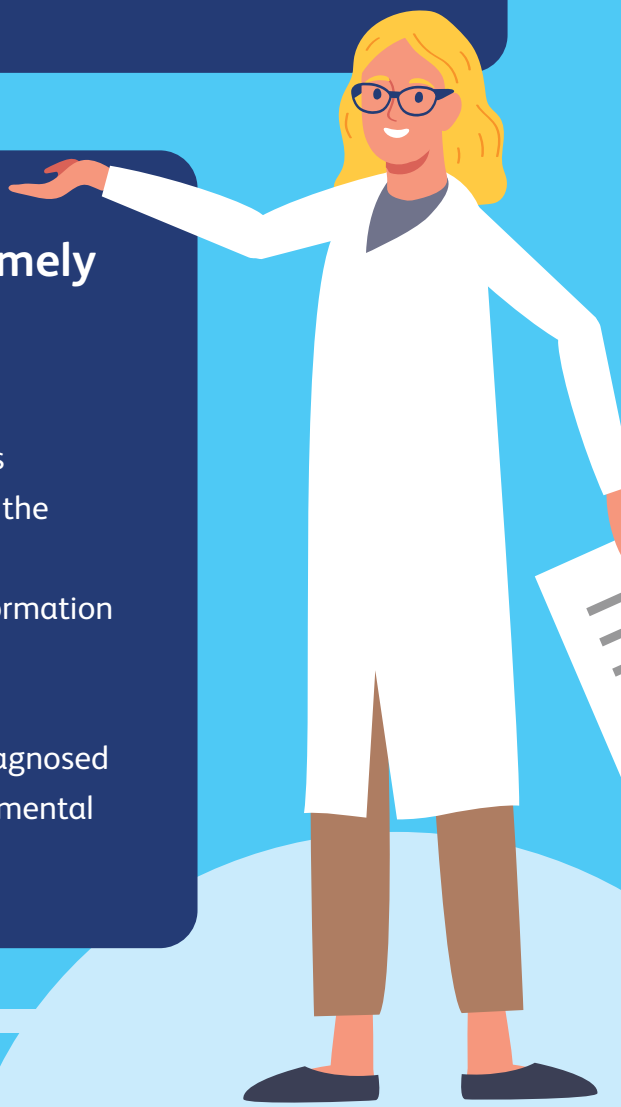
# LIST OF RECOMMENDATIONS

## 1. Creating a system that works for patients, clinicians, and innovation

- To develop the existing service model for ICC, either based on a traditional hub and spoke concept or, based on the recently introduced heart failure system. The implementation of this model would lead to a greater empowerment of the ICC service network.
- To review the current use of payment models/funding streams and consider the best type of contract to use reflecting the specific nature of ICCs.
- To identify areas of best practice and adoption of innovation and encourage the rollout of these across the NHS and the devolved nations.
- To gain consensus about the frequency of clinical examinations for suspected patients in England, based on the areas of best practice.

## 2. Ensuring patients have access to timely and effective diagnosis and care

- To ensure that patients can access echocardiograms and electrocardiograms in a timely manner through the expansion of the sonography workforce.
- To ensure that patients are sign-posted towards information about ICC after a diagnosis, and in-between clinic appointments.
- To ensure that there is bespoke support for those diagnosed with ICC, with individualised care plans, including a mental health plan.



## 3. Education of Healthcare Professionals

- Greater empowerment of healthcare professionals to refer those with suspected ICCs to specialist cardiologists through a newly created red-flag system, which will identify patients who have symptoms commonly associated with ICCs.
- Improve awareness amongst healthcare professionals about the prevalence and specific symptoms of ICCs.

## 4. Improving access to genetic testing for high-risk patients

- Greater use of self-referral for family members of those with genetic cardiac conditions to the ICC service.
- Ensuring that primary and secondary care clinicians are requesting a family's history of ICCs or sudden unexplained death and following up to secure this information.
- Improved access to genetic testing to the family members of those with suspected ICCs to provide patients and their extended family with a potential diagnosis.
- Increased support and information for patients to have conversations with their extended family who will need to be educated on their risk and need to be screened.
- To ensure that patients can access appropriate pharmacogenomic tests, which have not been freely available, as well as any other innovations within the therapy area.
- To ensure that clear referral processes to mental health professionals are present within the ICC centres.

*"I was in the gym, I was fit and healthy, and I had a cardiac arrest, I was resuscitated successfully and was subsequently diagnosed with HCM. It was a real shock. My heart wasn't going to be the thing that was going to let me down. But it has. And it's changed my life totally."* [xvii]

# CONTEXT

## (Inherited Cardiac Conditions today)

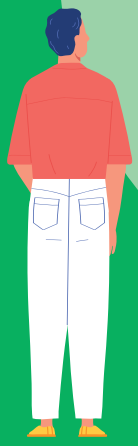
Inherited cardiac conditions (ICC) is an umbrella term covering a wide variety of relatively rare diseases of the heart. They are also referred to as genetic cardiac conditions.[i] These conditions do not always have symptoms, which can sometimes mean that the first time a family is aware of being affected is after a sudden cardiac death (SCD).[i] However, great improvements are being made in the detection of ICCs as well as some great improvements in treatment options; if people are diagnosed early and treated optimally, they can have a fulfilling life.[i]

People who are living with an ICC that is not well managed may find it difficult to work, struggle to be active, and therefore the impact on their day-to-day life can be huge.[ii] Symptoms can include shortness of breath, dizziness, palpitations, and blackouts; however, these symptoms may not be symptoms of ICC and **therefore a healthcare professional should be consulted to confirm.**[iii]

**An estimated prevalence of**

# 340,000 people with ICCs in the UK[iv]

# HCM affects 1 in 500[vi]



***"We know that many people face significant barriers in accessing timely diagnosis, consistent care, and specialist support."*** **– Anna Tomlinson, Chief Executive, Cardiomyopathy UK**

***"This policy reform report for Inherited Cardiac Conditions (ICCs) is a vital initiative in our attempt to address these issues."*** **– Dr Stephen Page, President, AICC**

**If you have any questions about your symptoms or ICCs, please consult your healthcare provider for advice**

Evidence is incomplete, but data from 2013, suggests a combined total prevalence for ICCs of about 340,000 in the UK[iv] with Cardiomyopathies and Channelopathies being the most common. [v] Hypertrophic Cardiomyopathy (HCM) is the most common genetic cardiomyopathy,[v] affecting around 1 in 500 people in the UK,[vi] and there is a significant unmet patient need in HCM.[vii] Patients with HCM may be at risk of a range of adverse outcomes and the development of debilitating symptoms that impair quality of life.[viii] Obstructive hypertrophic cardiomyopathy is associated with increased risk of arrythmias, stroke, heart failure and sudden death.[ix]

ICCs can lead to heart failure, and fewer than half of all people diagnosed with heart failure are alive five years later – a worse survival rate than most cancers.[x] It is therefore essential to achieve earlier diagnosis. In addition, the unpredictability of the condition causes much anxiety to patients.[xi]

***"The report offers new insights and reiterates previous recommendations which could, if implemented, result in significant improvements in care."***

**– Prof Dan Augustine, President, BSE**

While there have been significant advancements in treatment options for ICCs, inequalities persist throughout the patient pathway.[xii] As this report will explore, opportunities for improvement exist at every stage - from diagnosis to treatment. It will be crucial that these recent advancements within both diagnosis and treatment are realised and maximised by policymakers. Given the profound impact that ICCs have on patients, their families, and society, it is essential that key stakeholders and policymakers understand and address the disparities that exist across race, gender and socio-economic status. [xiii] [xiv] [xv] [xvi]

# ABOUT THIS REPORT

The following report has been co-created with stakeholders from the British Society of Echocardiography (BSE), Association of Inherited Cardiac Conditions (AICC), European Society of Cardiology (ESC), Cardiomyopathy UK (CMUK) to highlight, drive, and accelerate policy and service reforms in ICCs.

While we welcome the recent cardiology report published by GIRFT, there were some aspects that need greater focus on the differences associated with inherited cardiac conditions, leading to the initiation and development of this report. The recent report contained few mentions of ICCs, as well as limited reference to cardiomyopathy.[xviii]

During the last year, we have supported the establishment of an Expert Working Group from leading clinicians, charities, and patients. The first meeting was chaired by Virendra Sharma MP, Chair of the Heart Valve Disease APPG, which took place in the Houses of Parliament.

This event led to the creation of a dedicated Expert Working Group for the co-creation of this policy and service reform report.

This Expert Working Group included voices from across the cardiovascular community, with representation from cardiologists, patients, and cardiac scientists from leading organisations such as the BSE, CMUK, ESC, and the AICC.

The group's work included 1 to 1 meetings to gain insights into case studies which could act as best practice examples to be rolled out nationally.

This report has been reviewed and considered by relevant stakeholders before publication and secured strong support for both the overall direction of travel and specific recommendations.

## An Expert Working Group with representatives of:

British Society of Echocardiography

ESC European Society of Cardiology

Cardiomyopathy UK the heart muscle charity

Association for Inherited Cardiac Conditions AICC

# FINDINGS AND RECOMMENDATIONS

- Diagnosis
- Screening
- Genetics
- Familial Mapping
- Creating a workforce for the future
- Treatment

# DIAGNOSIS

***"My journey started three years ago, but if the respiratory team did more checking earlier then I could have been diagnosed earlier. I have been told they can't do anything surgery wise now because it's too dangerous. But if I had been diagnosed earlier, perhaps they could have."*** [xvii]

The patient experience of securing a diagnosis for ICCs is mixed, and examples can be seen in the varied HCM patient experiences.[xix] The key crucial element in improving outcomes for people living with ICCs is securing an early diagnosis, allowing them to start treatment sooner.[xx] In 2013, NHS England suggested that all patients with an ICC, or potentially at risk of having an ICC, should be offered assessment in a specialist ICC service, or advice taken from that service, and should not be managed by non-ICC cardiologists, geneticists or other specialties alone, without comprehensive ICC service involvement.[iv] Currently, patients with ICCs or suspected ICCs present in four different arenas: primary care, A&E, through screening programmes, or are self-referred. In primary care, those with suspected cases or family history of ICCs or that present with symptoms will be referred to the local ICC clinic through secondary care for testing, with subsequent treatment when required. In A&E, patients more commonly present with symptoms, such as syncope, and will undergo testing to secure a diagnosis. If the ECG is abnormal, clinicians will then refer patients to specialist care and an ICC hub. Those patients that are diagnosed with an ICC through family or routine screening

programmes commonly do so in ICC clinics because of a recommendation by their cardiologist, or a referral as the result of a sudden unexpected death within their family. These two final groups will then undergo clinical assessments, genetic evaluation to understand their condition fully. [xxi]

Within the current pathway for those living with ICCs, there are several issues which may be resulting in a lower standard of care. For example, when patients attend their primary care appointment, their GP is supposed to request a full medical history (including their heart history), to provide a fully informed referral to the patient. This will also ensure that primary care clinicians are referring patients with ICC symptoms to the correct specialist within secondary care to secure a prompt and accurate referral.

Results from a recent survey from Cardiomyopathy UK, which represents patients living with the most common ICCs, found that 29% were asked about a family history of heart disease.[xxii] The current system is not working as it was designed to; a clearer implementation or a re-design of the pathway for ICCs is required to achieve the best possible care for those living with these diseases.

Patients often spend time in primary care being treated for the incorrect disease, potentially owing to symptoms presented to the HCP at the time but also because of this inconsistency in information requested. GPs struggle to diagnose ICCs such as cardiomyopathy - according to a survey by CMUK, 53% of people who first went to their GP with symptoms were diagnosed initially with a non-heart related condition.[xxiii]

While a key part of this is improving awareness in primary care and non-ICC cardiologists about ICCs. Developing a medical education programme that could be used across Primary Care, non-ICC Cardiologists, and other interested specialties, would provide significant gains in the identification of patients.

Support in the development and delivery of this

**53%** [xxiii]

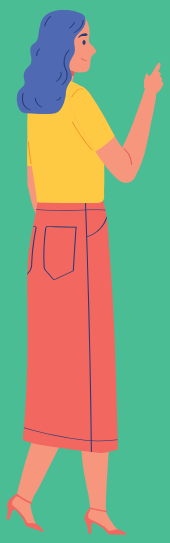
**with symptoms were diagnosed with a non-heart related conditions**

***"I feel like explaining your diagnosis doesn't always refer to what is wrong with you. My GP thought I had angina and things like that."*** [xvii]

would be beneficial in the period that the Hubs are established and could support the more in-depth education suggested for cardiac physiologists within local hospitals. There is a growing opportunity for working with ICC spokes and local hospitals to provide additional skills and understanding for cardiac physiologists. This would ensure that patients did not always have to travel repeatedly to the Hub for access to tests. However, in the specific case of Cardiopulmonary Exercise Testing (CPET) and stress Echoes in general, the lack of common access to this may mean that it remains in Hubs.

Once diagnoses have been made, there are several ongoing challenges which may be affecting the system and preventing improvements in care for patients. There is currently insufficient data being collected and shared on the diagnosis and treatment of ICC, due the lack of requirements to compel trusts to do so. For example within HCM, one of the more common ICCs, there is currently no measurement of how many people have HCM, how many cases are undiagnosed (for example, due to a lack of recorded family history), and how services and outcomes can be improved for patients.[xxiv] The Department of Health and Social Care recently confirmed that they do not "hold specific information on how many people were diagnosed" with HCM.[vi] To achieve an improved standard of care for patients living with ICCs, we must acknowledge and understand the challenge that the NHS currently faces, and that begins with tracking the data to understand the issues in more detail.

Diagnosis is further complicated by significant variability in how an ICC presents, with symptoms often vague or attributable to numerous other conditions.[xxv] Research by Cardiomyopathy UK found



**73%**

**did not associate their symptoms with heart problems** [xxiii]

***"Echocardiography is pivotal to the diagnosis of these conditions and this report supports our own work in highlighting the current challenges faced by the UK echocardiography workforce"***

**– Prof Dan Augustine, President, BSE**

that 73% of patients surveyed did not associate their symptoms with a heart problem at the time of diagnosis.[xxiv]

In the UK, there remains significant variation in both the time it takes to receive a diagnosis, and the area of the healthcare ecosystem in which they are diagnosed with ICCs.[xxvi] Often these variations and disparities are directly linked to patients' geographic location, with the time it takes to get a confirmed diagnosis once you have sought help differs depending on which UK nation you live in.[xxvii]

**Fewer new cardiovascular drug prescriptions during the pandemic**

# 470,000[xxiii]

As a result, there is an urgent need to standardise diagnostic pathways to ensure equitable access to timely and effective diagnostic services.[xxviii] Without such measures, delays in diagnosis can result in higher risks of sudden cardiac death, and lead to a worsening of the condition's severity, further endangering patients' lives.[xxix]

We propose that the European Society of Cardiology's guidelines are introduced within the NHS, to improve the speed at which patients are diagnosed, and to ensure that they are referred on to be seen and cared for by the appropriate specialist once their condition is identified, as many patients can be misdiagnosed with anxiety or respiratory conditions when they seek medical support.[xxv xxx]

Diagnosis of ICCs was significantly impacted by the COVID-19 pandemic, which has caused serious disruption across all CVD pathways.[xxxi] Analysis suggests that there have been an estimated 23,000 missed heart failure diagnoses during the pandemic. At the same time, there were 470,000 fewer new prescriptions of preventative cardiovascular drugs during the pandemic.[xxxii]

Currently around 80% of heart failure diagnoses in England are made in hospital, despite 40% of patients having symptoms that should have triggered an early assessment.[xxxiii] Less than half of all people diagnosed with heart failure are alive five years

> ***"A desire to improve access to specialist services such as self-referral pathways … along with the prioritisation of diagnostic genetic testing, are perfectly aligned with defined NHS priorities"***
>
> **– Dr Stephen Page, President, AICC**

# 80%

**of heart failure diagnoses are made in hospital**[xxxiii]

later – a worse survival rate than the majority of cancers[x] – and cardiomyopathy can lead to increased risk of disease progression, arrythmias, stroke, heart failure and mortality[ix], so achieving earlier diagnosis of the condition is vital.[xxxii] This is particularly true in the most deprived populations where people are four times more likely to die prematurely from cardiovascular diseases, compared to those in affluent populations.[xxxiv]

At present, to secure a diagnosis for an ICC, patients rely on a GP referral to access specialist treatment, unless, in the unlikely circumstances that the patient requests a private referral to a cardiologist. Thus, patients rely on their GP identifying the signs and symptoms of ICC to make the referral, which results in education of GPs being a key part of improving a diagnosis for the patient community.

Self-referral forms are an option in certain hospitals,[xxxv] whereby those with identifiable risk factors such as a close relative with ICC should be able to bypass GPs and book directly with a cardiology service. However, this relies on patient support and empowerment to access services, alongside robust education, and awareness.

To ensure that there is a joined up diagnostic service between the various elements of the NHS that address ICCs, it is key that ICC clinics can provide links between all the Nurse Specialists, Cardiac Physiologists, Diagnostic services, Cardiac Screening Clinics, Pharmacy, and Genetic services and Counselling. The current system is uncoordinated with multiple computer systems used within the Trusts in the NHS, and this issue needs resolving.[xxxvi] This limited co-ordination and potential risk of underdiagnosis can have a knock-on effect to the patient's family, particularly if the genetic marker means that the patient and their family are at a higher risk of sudden death if not managed.[xxxvii]

Despite this, there are clear examples where NHS Trusts are tackling this with great effect.

## Case Study: Leeds Teaching Hospital

Leeds Teaching Hospital have developed a nurse and clinical scientist-led screening programme which independently assesses patients for ICC and other inherited cardiac conditions. This programme involves cardiac scientists and nurses working collaboratively to deliver more efficient and cost-effective care for patients living with inherited cardiac conditions. These clinics allow symptoms to be identified, a diagnosis to be made, and subsequently, treatment to begin. These services could be rolled out nationally, offering more efficient and cost-effective care to patients that are living with ICCs.[xxxviii]

## Case Study: National Triage Guidance by The British Society of Echocardiology (BSE)

The National Triage Guidance, released by the BSE in July 2024, facilitates the effective and timely triage of patients, which is a vital step in securing appropriate diagnosis.[xxxix] An improved method of triage would contribute to patients receiving a diagnosis more quickly.[xl] Further, a 2024 study highlights the benefits of implementing a clinical scientist-led screening clinic for hypertrophic and dilated cardiomyopathies, as this approach provides an efficient review of patients, and, crucially, helps save appointments with specialist ICC consultants.[xli]

# SCREENING

Patients may be frequently misdiagnosed or undiagnosed since some patients with ICCs can be asymptomatic and may not present until later in life. As a result of frequent misdiagnosis and lack of diagnosis mentioned in the previous section, patients may often unknowingly pass an ICCs onto their children.[xlii] There should be a greater emphasis on genetic testing ensuring that ICCs are diagnosed in a timely manner.

Thus, screening for the condition is vital to identify at risk patients and monitor them accordingly. Recent developments in the national screening landscape have elevated the prioritisation of screening within the NHS agenda.[xliii] Crucially, there is not currently a national approach to screening for ICCs. [xliv]

***"Echocardiography is pivotal to the diagnosis of these conditions"***

**– Prof Dan Augustine, President, BSE**

The high number of ICC diagnoses made in A&E following cardiac arrests underscores the inconsistent screening of at-risk individuals.[xlv] Some vulnerable patients do not receive the screening they need to identify their conditions before emergencies arise and they only receive a diagnosis post-cardiac incident.[xlvi] While there are areas of good practice across the UK, they are not universally available.[xlvii] Ensuring timely screening and management for everyone at risk of ICCs is essential to prevent avoidable crises and improve health outcomes.

To address these issues that are currently creating inequality in the care of people with an ICC, we would recommend that the NHS ensure that patients can access echocardiograms and electrocardiograms in a timely manner through the expansion of the sonography workforce.

***"Variability in services and limited awareness contribute to gaps that impact the quality of care patients receive."*** **– Anna Tomlinson, Chief Executive, Cardiomyopathy UK**

**In addition, we would recommend improved access to genetic testing to extended family of those with ICCs by increasing the numbers of genetic councillors that can provide patients and their extended family with an earlier diagnosis.**

***"It took quite a few visits before I got an ECG. And the clinician who did it said 'oh, when did you have your heart attack?' I told him I hadn't. And he said, 'according to this, you've had 2 heart attacks' , which really stuck with me."*** [xvii]

While a long-term approach for screening is clearly required, there are several solutions that can help to deliver diagnoses and, crucially, interventions for patients. The Government needs to ensure the NHS has the resources and staff it needs to reduce waiting times for heart care.[xlviii] There are further ways that policymakers can ensure that patients with ICCs receive more timely diagnosis, such as utilising private sector capacity to clear existing backlogs in screening programmes.[xlix] In the longer term, the BSE has recommended the need for implementation of a formal national career pathway for sonographers to make services sustainable as well as new roles which help distribute work such as administrators, data managers and support workers.[l]

Genetic testing may be beneficial for some patients, which have not previously been available, as well as any other upcoming screening innovations within the therapy area.

A clear example of Government in the UK prioritising CVD, and specifically the role of genetic testing, can be seen through the Heart Disease Action Plan published by the Scottish Government.

## Scotland's Heart Disease Action Plan[li]

**The Scottish Government published The Heart Disease Action Plan in March 2021. This sets out the Government's priorities and actions to minimise preventable heart disease and ensure equitable and timely access to diagnosis, treatment, and care. It states that heart disease is a major cause of death and disability in Scotland with over 9,000 deaths per year, and that approximately 28,000 people in Scotland are living with an inherited heart condition. The Plan highlights HCM as an example of a nurse-led model of specialist care, citing the genetic testing service which could be replicated in England.**

# GENETICS

There is wide variation in genetic testing for ICCs, with HCM being an example of this, which affects patient care.[lii] During a recent survey of those living with cardiomyopathy, 37% of patients had not been offered a genetic test for the condition.[xxiv] Furthermore, there is a lack of awareness of the genetic nature, with nearly one in five not informed by their clinician that the condition may be genetic.[xxiv]

The seven NHS Genomic Medicine Service Alliances currently play an important role in supporting the strategic and systematic embedding of genomic medicine in clinical pathways and clinical specialities,[liii] supporting the achievement of equitable access to standardised care, inclusive of genomic testing. We consider there is a requirement for greater resource and prioritisation of this so that patients can receive the timeliest diagnosis possible. We acknowledge that these alliances play a crucial role in raising awareness among clinicians and the public of the genomic testing available through the National Health Service. However, this should go further across the entire country for all those living with ICCs. Collecting and joining up patient data throughout the pathway, specifically within genetics, would help drive improvements in ICC care.

Despite the genomic medicine service set-up in 2018 that has been rolled out in England, there are still significant waiting times for clinical genetic services, and these were highlighted as a key barrier.[liv]

***"The tests seem to happen and take an awfully long time to get the results... I was basically denying there was anything wrong with me."*** [xvii]

Whilst the Department for Health and Social Care recently published a pledge to achieve more genetic testing for patients with a rare disease, no such similar commitment exists for ICCs.[lv]

There is still further education required for policymakers and stakeholders about the impact of these conditions both on current and future generations, as a set of genetic diseases. Education is essential - not only for individuals at risk of ICCs but also for general practitioners (GPs), who play a pivotal role in early identification and intervention.,[lvi] [lvii] Clinicians, and particularly GPs as the first-point of contact for many, are instrumental in facilitating access to genetic testing and subsequent treatment. By enhancing awareness and empowering GPs with[lviii] the necessary knowledge and resources, we can aim to ensure that more individuals benefit from timely diagnosis and appropriate management of ICCs.

In addition to genetic testing, familial mapping and screening family members of suspected ICC patients could also result in a quicker journey to accurate diagnosis.[lix] While genetic testing can identify potential disease-causing genes, this process involves a blood test or mouth swab, and so improved familial mapping and screening would cut back on the complexity of testing and associated costs.[lx]

According to a survey by CMUK, the proportion of people who go to their GP with symptoms first, compared to those who go straight to A&E has changed since the pandemic. 11% of the people in their survey (632 people) who were diagnosed before the pandemic told CMUK that they went to A&E first compared to 42% of people who were diagnosed in the last two years in the post pandemic period.[xxiii] Patients being diagnosed in A&E results in significant impact on both the system and patients that are living with ICCs. The system needs to ensure that primary care is the area which is making the diagnoses for this, to ensure efficiency within the pathway and that patients are not presenting with more severe symptoms.

***"My son is in London, he had MRI, genetics, the works at St Georges, but it's taken me two and a half years to get all those tests done."*** [xvii]

# FAMILIAL MAPPING

Familial mapping simply consists of a shortlist of questions that can be easily asked by a healthcare professional. It is crucial that Primary and Secondary Care clinicians request familial information of history of cardiac conditions and following up with a referral or follow up of the wider family if there are any risks to the patient.

Familial mapping is a crucial tool for identifying whether people within the population carry the gene for an ICC.[lxi] Despite its importance, access to this service remains inconsistent across the UK, leaving many without the opportunity to benefit from early diagnosis and intervention.[lxii] This disparity amounts to a postcode lottery, where access depends on geographic location rather than clinical need. To ensure effective management of ICCs across the UK,[lxiii] familial mapping must be universally available, with appropriate facilities and timely testing for all who require it, regardless of where they live.

There are already examples of where this takes place effectively, such as the pilot study on Familial

Hypercholesterolaemia (FH), in which increased FH detection rates in participating practices increased by up to 25%.[lxiv] These case studies of best practice should be rolled out across the NHS, to other conditions which would benefit from this efficient and effective approach. There will of course be instances where familial mapping is not sufficient and genetic testing is necessary, hence the need for genetic testing to be widely available as well.

Finally, having access to detailed patient data is important for genetic conditions like HCM because, as noted earlier, new genetic variants are still being identified and it is not always immediately clear whether variants are associated with the disease or not. Sharing patient data allows experts to gather the evidence needed to make those links and could potentially help to identify more ICC patients in future. Patient data should be collected at a national level, with ICCs and NHS Genomic Medicine Service Alliances working together to ensure that this data is captured. This, alongside the expansion of self-referral forms, would improve the current situation facing patients with ICCs.

Another key recommendation made by this report, is for NHSE to gain consensus about the frequency of clinical examinations for suspected patients in England, based on the areas of best practice. This may result in the increased frequency of testing for potential patients that have family members with ICCs, to confirm diagnosis, as called for by European Society of Cardiology and their guidelines. It will be crucial to ensure this change to every 1–2 years[xxx] is managed without any profoundly negative effect on patients, clinicians, and the systems.

## Welsh Heart Conditions Delivery Plan 2017

The 2017 Delivery Plan highlighted inherited or idiopathic cardiac conditions, including cardiomyopathies as 'major heart conditions' which will be addressed by measures contained in the plan.[lxv] One such measure was that people with a family member who died suddenly of a suspected cardiac cause and those with family members with inherited heart conditions should have access to appropriate genetic advice and testing.[lxvi] For patients with HCM, a genetic testing service located at Swansea Bay was established in 2018 through the British Heart Foundation and the Miles Frost Fund.[lxvii] It was announced this service was to receive NHS funding and become a permanent service. Following the successful pilot, there is an opportunity to expand similar services throughout Wales.

# CREATING A WORKFORCE FOR THE FUTURE

There is currently a limited awareness about ICCs, what these conditions are, and how they present. Similarly, there is a broader lack of understanding about CVD more generally – with almost a third of patients given the wrong initial diagnosis after a heart attack.[lxviii]

The creation of a national leadership role for ICCs offers the ability to raise awareness of inherited cardiac conditions amongst HCPs and key stakeholders.

There is a need to improve clinicians' education and raise public awareness of the hereditary nature of ICCs, but also the severity of the disease, including encouraging people to have conversations with their families and GPs about their heart history and symptoms. There should be a greater education of primary care, so that they are able to refer those with suspected ICCs effectively to specialist cardiologists through a newly created red-flag system.
This will enable patients to be referred for an electrocardiogram (ECG) which will support a fast and precise diagnosis and allow for an experienced healthcare professional to interpret the data to see if a patient needs an echocardiogram or an MRI more quickly.

> ***"If we are to make early diagnosis a reality for this community, we need policy makers to prioritise retention within the workforce."***
> **– Prof Dan Augustine, President, BSE**

Some GPs are well-informed about cardiovascular disease, and in certain cases, individuals with ICCs are diagnosed during GP visits for unrelated issues. However, this is not the norm. As outlined in the 'Diagnosis' section of this report, ICCs are frequently misdiagnosed or remain undiagnosed until they present as emergencies in A&E. To address this, it is crucial to ensure that the entire NHS workforce is equipped with the knowledge to identify those most at risk of an ICC and to monitor them effectively. Early detection and intervention can significantly improve outcomes and reduce avoidable emergencies.

In addition, cardiologists could also empower GPs. While GPs have an enormous decision-making burden to triage patients effectively, the production of written instructions with clear triggers and next steps through the red-flag system will help patients in accessing the care and treatment they need.

This red-flag system would aim to alert GPs when a certain combination of symptoms is identified at the same time. This would encourage primary care healthcare professionals, in unison with requesting the correct medical history, to ensure that the correct referral is made in secondary care. This system could take shape in many forms and would be led by existing best practice within the system but would ensure that the current system is streamlined and more efficient in giving accurate and prompt diagnosis to patients.

In order for patients to receive a timelier diagnosis it will be important to ensure GPs understand the patients' symptoms, refer patients quickly and share the relevant information with the cardiologist within the local ICC network.

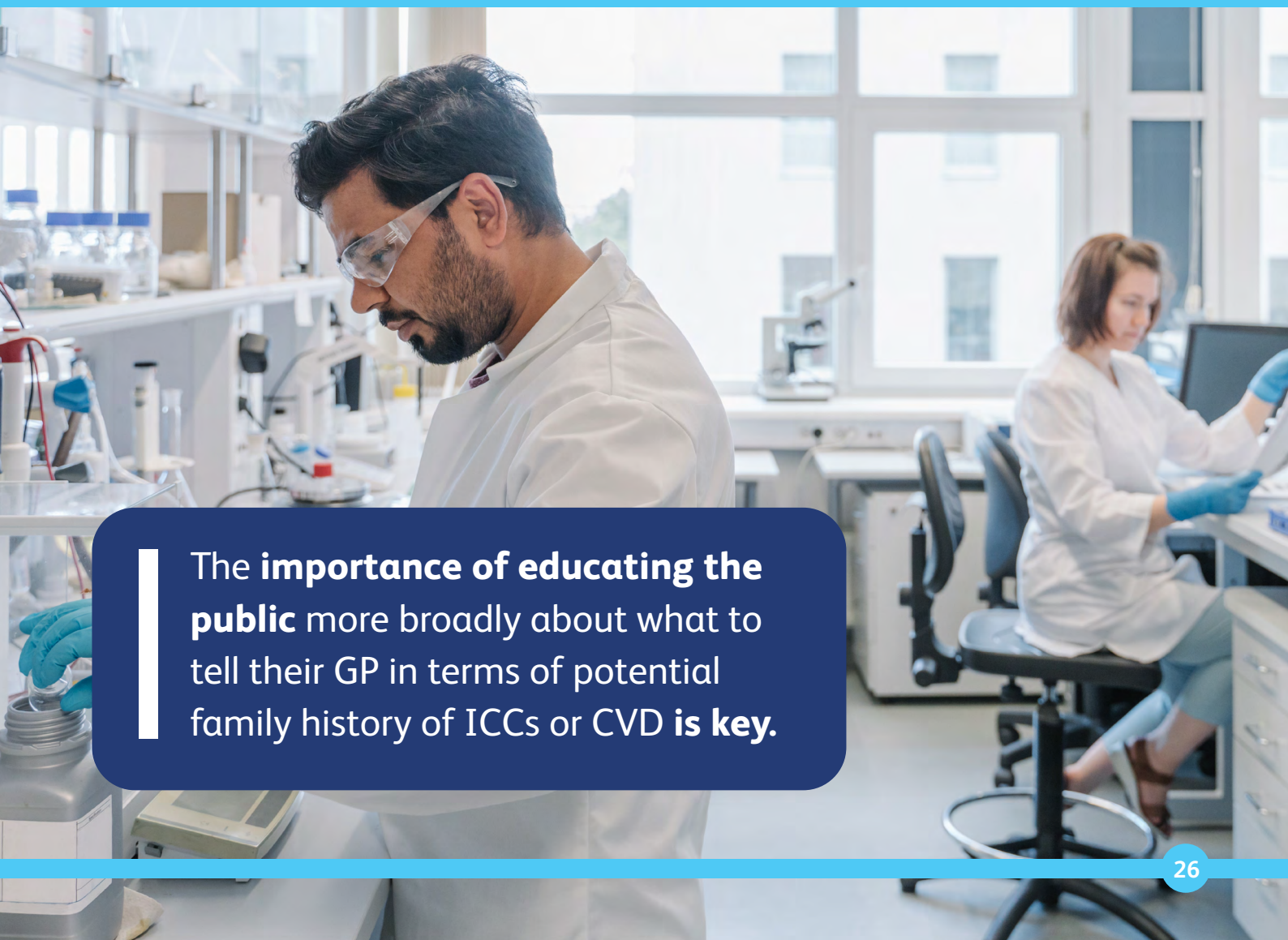
***"Developing advanced roles and physiologist led services will support retention of our most experienced echocardiographers and increase capacity in ICC clinics."*** **– Prof Dan Augustine, President, BSE**

To improve care for those living with ICCs, we would recommend having an internal list of cardiologists, which GPs can access, that are ICC specialists in each NHS Trust within their area; to ensure the referrals from General Practitioners are easily facilitated. Any changes to the system would need to focus not only on GPs, but also on community, mental health, social care, pharmacy, hospital, and voluntary services within primary care networks (PCNs). This awareness raising would focus on engagement with PCNs which have recently been rolled out. This would have the aim of leading to the standardisation of care as well as improved implementation.

Alongside this, the importance of educating the public more broadly about what to tell their GP in terms of potential family history of ICCs or CVD is key.

Improving patient education is crucial in overcoming the barrier of anxiety and uncertainty which prevents patients accessing appropriate services.

The National Institute of Cardiovascular Outcome Research (NICOR) publishes an annual audit containing data about heart failure patients admitted in England and Wales, but it does not include any ICC-specific data.[lxix] The inconsistency across the UK regarding the data leads to limited ability to understand, and subsequently tackle, the problem within ICC. A key example of this can be seen through the lack of data that is published in relation to the causes and origin of heart failure. Healthcare systems need to understand the causes of heart failure to understand the impact that ICCs are having at a national level.



The **importance of educating the public** more broadly about what to tell their GP in terms of potential family history of ICCs or CVD **is key.**

# TREATMENT

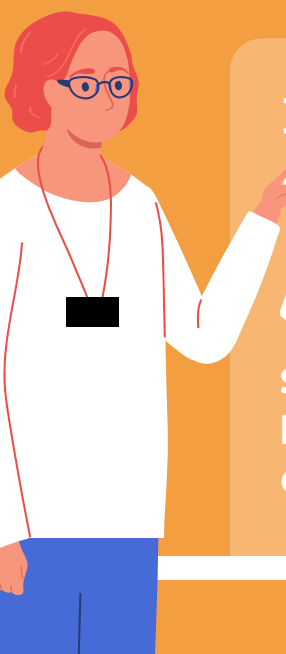
For many ICCs, such as cardiomyopathy, early diagnosis and intervention will reduce risk of worsening symptoms or death.[lxx] Despite the significant impact of ICCs on people, some of these conditions have limited treatment options for patients.[lxxi, lxxii] While open-heart surgery remains a non-therapeutic option, this may still lead to disease progression and need for re-intervention later. For some ICCs, there have been recent developments that are available which is highlighting problems in other aspects of the pathway. These interventions can include a variety of drugs, lifestyle advice to avoid triggering events, or fitting of implantable defibrillator devices.

***"ICCs can have devastating consequences and early detection is paramount. Early diagnosis will ensure that people living with ICCs can access timely treatment."***

**– Prof Dan Augustine, President, BSE**

Current ICCs services remain based on previous models where no new developments had been introduced over the past three decades. This is no longer the case as knowledge and awareness about ICCs is improving, services need to be updated proactively so they can include appropriate and timely management to help improve outcomes and experiences. To address these issues effectively, policymakers must update the problems within the system which are limiting patient access to these innovations.

With new innovations emerging to manage ICCs, it is vital that the healthcare system is equipped to deliver these advancements to patients who need them. Currently, some areas of the UK are leading the way in providing timely access to these interventions, setting a standard of excellence. However, in other regions, access is delayed, creating unacceptable geographic disparities. Every NHS Integrated Care System (ICS) must prioritise equitable and prompt management of ICCs, ensuring that all patients, regardless of location,



**In HCM,only** [xxiii]

**36%**

**surveyed by CMUK have a plan in place for cardiomyopathy care.**

can benefit from the latest advancements in ICC care. While therapeutic approaches may control symptoms, and improve the quality of life, this alone will not result in substantial improvement for people living with ICCs. It is important that the system has been redesigned and has ample capacity, to benefit the patient community.

Finally, there needs to be a more holistic approach to treatment of ICCs, with acknowledgement of the mental health impact of the conditions. It will be crucial to ensure that there is bespoke support for those diagnosed with ICCs, with individualised care plans, including a mental health programme.

# CONCLUSION

In conclusion, this report on Inherited Cardiac Conditions (ICCs) underscores the imperative for systemic enhancements to ensure optimal care provision for all those affected by ICCs. Key recommendations focus on education, access to genetic testing, timely diagnosis, and patient-centred care.

Education initiatives, including both the establishment of a national position within the NHS dedicated to raising awareness of ICCs and the empowerment of primary care for early referral through a red-flag system, are essential steps to improving early detection.

Enhancing access to genetic testing, particularly for high-risk patients, demands systematic recording of familial ICC history and the expansion of genetic counselling services to facilitate informed decision-making.

Timely diagnosis and effective care hinge on improving access to diagnostic services such as echocardiograms and electrocardiograms, along with tailored support for patients and their families' post-diagnosis.

Creating a patient-centric healthcare system entails refining service models, exploring alternative payment schemes, and fostering innovation adoption to streamline care pathways and optimise resource allocation.

In essence, implementing these recommendations may help enact systemic improvements, ensuring equitable, patient-centred care for individuals and families affected by ICCs. Collaboration among stakeholders is crucial to realise this vision, ultimately enhancing outcomes and quality of life for those impacted by these conditions.

**This report offers an opportunity for policymakers to improve outcomes for patients and clinicians across ICCs.**

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