National Specification for Inherited Cardiac Conditions Services in Scotland
1 Population Needs

1.1 National context and evidence base

Inherited Cardiac Conditions (ICCs) are a group of largely monogenic disorders affecting the heart, its conducting system and vasculature. The first indication is sometimes sudden cardiac death (SCD) often in adolescents or early adulthood. When an ICC is diagnosed there are implications for the relatives.

The last decade has seen dramatic advances in our understanding of the molecular pathology of ICCs. More than 50 ICCs have been recognised and genetics tests are increasingly available for the more common disorders such as hypertrophic cardiomyopathy (HCM) and for some rarer disorders, for example, Long QT syndrome (LQTS). The conditions are highly heterogeneous, both genetically and clinically.

Epidemiological evidence is incomplete, but suggests a combined total prevalence for ICCs (cardiomyopathies and arrhythmias) of about 10,000 in Scotland (excluding individuals affected by familial hypercholesterolaemia or FH). Risks associated with these conditions are highly variable, depending on the gene variant and the spectrum of clinical risk factors. The average annual risk of SCD is about 0.1% for LQTS, and the annual mortality for HCM about 0.3-1%. Risks are substantially higher for patients with the most severe symptoms or in those who have experienced a resuscitated cardiac arrest. Gene variant carriers with no identifiable clinical risk factors have lower absolute risks. The research also suggests that ICCs are responsible for the deaths of around 50-60 young people in Scotland each year.

Due to the complex and diverse ways in which ICC services are currently delivered, it is difficult to provide accurate data on current average caseloads and the annual throughput of services in Scotland. This, coupled with uncertainty regarding the prevalence and incidence of ICCs, make it difficult to estimate the capacity required for the future. However, it may be the case that in some specific areas, provision is inadequate.

In 2008 the Network for Inherited Cardiac Conditions Scotland (NICCS) (previously known as Familial Arrhythmia Network Scotland – FANS) was created to oversee improvements to services in Scotland and ensure a better approach to the management of ICCs. The network brings together stakeholders involved in the management of ICCs from specialist regional centres, district general hospitals (DGH), primary care, the third sector, pathology services and the Crown Office.

A fuller description of the networks activity can be found in Annex 2 & 3 on pages 10 & 11.

The scope of ICC services includes the following main categories of ICCs:

- **Arrhythmia syndromes** caused by gene variants in the proteins involved in generating the action potential; these are mainly the proteins making up sodium, potassium or calcium conducting channels in the membranes of the cardiac myocytes, but also some proteins that affect ion conduction indirectly. The
arrhythmia syndromes include conditions such as LQT syndrome, Brugada syndrome and catecholaminergic polymorphic ventricular tachycardia (CPVT).

- **Cardiomyopathies**, caused mainly by gene variants in the proteins making up the contractile system of the myofibrils, such as actin, myosin and troponin. The cardiomyopathies include hypertrophic cardiomyopathy (HCM) characterised by asymmetrical thickening of the heart muscle, dilated cardiomyopathy (DCM), which weakens the heart muscle resulting in heart failure, and arrhythmogenic cardiomyopathy, which is a primarily arrhythmogenic heart muscle disorder. These conditions can lead to heart failure, stroke and arrhythmia.

- Families afflicted by Sudden Arrhythmic Death Syndrome (SADS), in some cases will be found to have an inherited cardiac condition.

2 Scope – What is an Inherited Cardiac Condition (ICC) Service?

2.1 Aims and objectives of service

**Aims**
The aim of ICC services is to improve the diagnosis, treatment and outcome of patients with inherited cardiac conditions. Improving outcomes in patients with ICC requires specialist clinical management to improve both life expectancy and quality of life.

**Objectives**
The service will deliver these aims by establishing defined models of care with robust shared/network care arrangements where appropriate to ensure:

- Timely diagnosis with appropriate counselling and psychological support to the patient and their family.
- Provision of high quality proactive treatment and care.
- A smooth and managed transition from children’s care to adult care.

2.2 Service description/care pathway

The ICC services provide care/treatment for infants, children, young people and adults. Cases of sudden death in infants less than one-year-old should be referred to the paediatric service.

The ICC service will need to investigate suspected index cases. Where an index case is identified, screening should be offered to first degree relatives, and cascaded to others as deemed necessary on the basis of risk.

The ICC service will function as a multi-disciplinary team (MDT). The MDT will have oversight of the whole service, which includes the following:

- Receiving referrals
- Provision of advice about possible referrals
- Diagnosis and assessment
- Treatment or advising on treatment
- Identification of family members at risk and recommending appropriate evaluation
- Referral to genetic counselling
- Discharge
- Prioritisation of services for patient, such as access to genetic testing, according to clinical need
Input to specialist care provided by other specialities (for example ophthalmology, rheumatology, cardiac surgery)

Input to long term monitoring, surveillance and care, including shared care arrangements with other hospitals and support provided by voluntary organisations

Sonography

All ICC services must be represented on the NICCS Steering Group, with consultants in cardiology and clinical genetics from each specialist service as part of the network. Specialist staff in nursing and genetic counselling should also be represented. MDT meetings must be minuted and case records kept securely.

Staff, training, qualifications and experience

The core team of staff will include:

- Consultant Cardiologists and Consultant Paediatric Cardiologists with specific expertise and experience in the management of ICCs.
- Consultant Clinical Geneticists and Genetic Counsellors, with specific expertise in the management of ICCs, to provide pre- and post-test counselling and to co-ordinate DNA testing, aid in genetic data interpretation and cascade testing of at-risk family members.
- ICC Nurse specialists in the evaluation and management of adults and children with inherited cardiovascular conditions.
- Cardiac physiologists with specific training and expertise in the evaluation of inherited cardiovascular conditions.
- Cardiac physiologists with specific training and expertise in the evaluation of inherited cardiovascular conditions in children.

All of these specialists may not necessarily be present on the same site or available at the same visit, but will be accessible to the ICC service.

ICC services will ensure that:

- The MDT meets on a defined and regular basis. This will be no less than once a month.
- The MDT includes input from cardiology, genetics and, when necessary, from paediatric cardiology.
- The outcomes of MDT discussions are clearly recorded.
- The MDT oversees transition from the paediatric to the adult components of the service.
- There is robust evidence of clinical audit.
- Nationally agreed clinical protocols (through NICCS) are followed.

Diagnostic Facilities

Regional centres providing the service will have access to the following:

- Dedicated echo service.
- Access to and experience with cardiac magnetic resonance imaging in cardiomyopathy.
- Exercise testing (risk stratification and diagnosis).
- Ambulatory electrocardiographic monitoring.
- Signal averaged electrocardiograms.
- Facilities for non-invasive or minimally invasive electrophysiology investigation, e.g. Ajmaline testing.
• Genetic testing, including specific blood tests which relate directly to ICCs.

**Discharge criteria**
The following groups of patients may be considered for discharge from the ICC service:
• Patients at no risk.
• Patients at low risk for whom the risk unlikely to increase and who are unlikely to benefit from intervention. This group may be discharged to a GP or secondary care cardiologist for continued monitoring. Nurse-led surveillance clinics should also be considered as a more cost effective way of follow up.
• Adult patients for whom no further intervention is required at this time, e.g. HCM with no associated risk factors and symptomatically well. This group may be discharged to a secondary care cardiologist for continued monitoring.

In addition:
• At risk children will be followed up until they reach adulthood, when assessment of the most appropriate future care will be made.
• Patients with a progressive condition who could potentially benefit from treatment at a later date will not normally be discharged.
• Patients will usually be discharged to the care of their General Practitioner or local Hospital Cardiologist. Clear discharge plans are required in these circumstances and the discharge process must be central to clinical pathways.

**2.3 Population covered**
The service outlined in this specification is for patients ordinarily resident in Scotland. The ICC services provide care/treatment for infants, children, young people and adults. Cases of sudden death in infants less than one-year-old will be referred to the paediatric service.

Anyone with any of the conditions listed in section 1.1, or with a first degree relative with one of these conditions, will be able to access the ICC service. This will include the next of kin in cases of sudden cardiac death.

The service will be delivered at times that meet individual’s needs. The service will be accessible and in an appropriate setting. The service will be delivered in a format and style, which recognises the needs and preferences of all patients, which include:
• different cultural or faith groups
• physical/learning disabilities
• Ethnic minorities

The provider will make information available to patients and the public on their services, provide patients with suitable and accessible information on the care and treatment they receive and, where appropriate, inform patients on what to expect during treatment, care and after-care. NICCS produces information to support this, as does recognised third sector organisations.

The onus upon services to monitor the ethnic origins of their patients might also have important clinical consequences, given emerging epidemiological evidence suggesting that for some conditions, such as cardiomyopathy, there is an increased predisposition for the development of disease in certain ethnic groups.
2.4 Referral Criteria

Referral Criteria
Anyone with any of the conditions listed in section 1.1, or with a first degree relative with one of these conditions, shall be able to access the ICC service. This will include the first degree relatives in cases of sudden cardiac death.

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. They should not be managed by non-ICC cardiologists, geneticists or other specialties alone, without comprehensive ICC service involvement.

Patients with an ICC will have the initial diagnosis made or confirmed within a specialist ICC service. The ICC service will determine the initial management plan, assess the pedigree, set up genetic testing and arrange family evaluation and cascade screening where appropriate.

The follow up plan may involve the ICC service or may be devolved to local hospital cardiologist or primary care depending on patient risk and complexity.

The ICC service will need to investigate suspected index cases. Where an index case is identified, screening shall be offered to first degree relatives, and cascaded to others as deemed necessary on the basis of risk.

Non-specialist secondary and primary care services need to be able to recognise individuals at risk, refer appropriately, support patients and families in interpreting and acting on specialist advice, and provide a partnership for shared care and follow-up. This requires that all professionals in cardiology, primary care, paediatrics and any of the secondary and tertiary specialties that interface with inherited cardiac disease have a necessary basic level of understanding and competence.

2.5 Interdependencies with other services

The Regional ICC service will be available and accessible to all its catchment population. Whilst it is usually necessary that it should be sited geographically in a major tertiary centre (often where there is a regional genetics service) the service will find means to liaise with its district general hospital health board cardiology services to ensure that individuals with ICCs are diagnosed and managed effectively.

Where outreach services are agreed, assurance will be given that the same quality of service can be delivered and that the designated service retains responsibility for the overall delivery of the service including clinical governance. There will be clear arrangements to define responsibility for service quality, clinical governance (including, but not restricted to, clear arrangements for dealing with complaints, serious untoward incidents and clinical indemnity).

Co-located services
Certain elements of the ICC service need to be co-located including the Consultant ICC Cardiologist, ICC Nurse Specialists and basic non-invasive testing facilities such as Electrocardiogram (ECG), Holter monitoring, exercise stress testing and echocardiography. Ideally the cardiology elements of the ICC service should also be co-located with those interdependent cardiology specialties given below. The genetic elements of the ICC service, Consultant Clinical Geneticist and Genetic Counsellors, should also be co-located.
It is accepted that ICC Cardiology specialist services and Genetic Services may not necessarily be physically housed in the same building or on the same site, but they should function as a co-located service. Due to the arrangement of paediatric services across Scotland, paediatric ICC cardiology or metabolic disease specialties may also not be physically present on the same site, but the ICC service should have established pathways of access to these services.

**Forensic Pathology**
Specialist cardiac pathology should ideally be co-located with the main ICC centre. Where this is not possible, the link should be well-established such that the two elements of the service are functionally co-located. Molecular genetic testing laboratories may be remote from the ICC service. Regular MDT meetings with pathology services are highly recommended. A national approach via NICCS should be in place to facilitate robust processes and reduce variation in practice.

**Other Health and Statutory Agencies**
Education is crucial for general practice, the police service and Crown Office Procurator Fiscal Service (COPFS), to make sure the system works and sudden cardiac death being a potential cause of death is investigated appropriately. Without these processes in place relatives of the deceased may not be picked up by screening services. There should be an ICC/NICCS representative on the national Death in the Community committee which looks at the processes facilitating the management of sudden out of hospital deaths.

**Interdependent services**
In addition to the core team, ICC service should have close links with electrophysiology, diagnostic angiography, interventional cardiology, cardiothoracic surgery and cardiovascular imaging departments.

**Related services & the Third Sector**
Individuals and families accessing the ICC service shall also have access to dedicated bereavement counselling services, through either their regional ICC service or national support groups. Third sector organisations such as Cardiomyopathy UK (www.cardiomyopathy.org), British Heart Foundation (www.bhf.org.uk/heart-health/conditions/inherited-heart-conditions) and the Arrhythmia Alliance (www.heartrhythmalliance.org) are invaluable sources of information and support to people with inherited cardiac conditions. The BHF, for example, provide the Genetic Information Service, a telephone hotline for people with, or suspected of having, an ICC.

**3 Applicable Service Standards**

**3.1 Applicable national standards**
The core team of staff will include:

- Consultant Cardiologists and Consultant Paediatric Cardiologists with specific expertise and experience in the management of ICCs.
- Consultant Clinical Geneticists and Genetic Counsellors to provide pre- and post-test counselling and to co-ordinate DNA testing, aid in genetic data interpretation and cascade testing of at-risk family members.
- Nurse specialists with training in counselling, and in the evaluation and management of adults and children with inherited cardiovascular conditions.
- Cardiac physiologists with specific training and expertise in the evaluation of inherited cardiovascular conditions in both adults and children.

NICCS and ICC services will ensure that:

- The MDT meets on a defined and regular basis. This will be no less than once a month in the specialist centre.
• There is robust evidence of clinical audit.

Diagnostic Facilities
Regional specialist centres providing the service will have access to the following:
• Dedicated echo service.
• Access to and experience with cardiac magnetic resonance imaging in cardiomyopathy.
• Exercise testing (risk stratification and diagnosis).
• Ambulatory electrocardiographic monitoring.
• Signal averaged electrocardiograms.
• Facilities for non-invasive or minimally invasive electrophysiology investigation, e.g. Ajmaline testing.

Audit
NICCS oversees the development of audit of inherited cardiac conditions in Scotland. It also monitors, where possible, activity. This includes levels of genetic testing for arrhythmias and cardiomyopathies in Scotland, activity at multi-disciplinary team meetings and works with individual clinicians or services on ad hoc audits.

Guidelines and guidance
NICCS has developed a suite of protocols and guidelines for inherited cardiac conditions.
http://www.niccs.scot.nhs.uk/protocols-standards/

Clinical Quality Indicators
Clinical quality indicators (CQIs) can encompass process or outcome driven aspects of the ICC management to monitor clinical care and support improvement. Clinical audit can assess these CQIs and should be done in partnership between NICCS and clinical services. Examples of CQIs that may be utilised include on the length of time to appointment, named person in place, evidence of transition (where appropriate), referral to specialist staff and patient feedback. These should be set up and monitored by the NICCS network.

4 Key Service Outcomes
Service provision is focused on achieving optimum clinical management. For many ICCs, careful clinical management will reduce risk of worsening clinical conditions or death. For arrhythmia syndromes (such as LQT) and HCM, treatments can include a variety of drugs, lifestyle advice to avoid triggering events, fitting of implantable defibrillator devices or, in some cases, surgical options.

Genetic testing can aid clinical management by enabling more accurate diagnosis and risk assessment, and in some cases guiding the choice of treatment. Importantly, identification of a gene variants in an affected person enables their relatives to be offered targeted testing for the same gene variants. Those who carry the gene variant can then be assessed and offered appropriate surveillance and / or treatment, while those who do not carry the gene variant can be spared further investigation. This approach is known as cascade testing.
ANNEX 1: Provision of services to children

Service description/care pathway
All paediatric specialised services have a component of primary, secondary and tertiary elements. The efficient and effective delivery of services requires children to receive their care as close to home as possible dependent on the phase of their disease.

In 2016 the NICCS network produced a clinical pathway and guidance for inherited arrhythmia paediatric services in Scotland.

Paediatric services should aim to provide multi-disciplinary specialist expertise in regional centres, similar in scope to adult services. Electrophysiology specialist care should be available across all regions through outreach clinics.

Other Paediatric Issues to Consider
- Transition
- Imaging
- Mental Health
- Family Support
ANNEX 2: Network for Inherited Cardiac Conditions Scotland

NICCS is a Scotland-wide network established to improve the diagnosis and care for individuals and their families affected by inherited cardiac conditions. The management of a person with an inherited cardiac condition has traditionally been complex due to the number of consultations with different healthcare professionals. Together with the challenges in establishing a diagnosis, this can lead to delays in care. A person's care for these conditions involves a multidisciplinary approach by many healthcare care professionals. For example:

- Cardiologists
- Geneticists
- Genetic Counsellors
- General Practitioners
- Specialist Nurses & Allied Health Professionals
- Paediatricians
- Pathologists

Since its inception in 2008 NICCS (then FANS) has worked across clinical and regional boundaries to facilitate lasting improvements in the management of inherited cardiac conditions. This has been done through service developments including the initiation of joint clinics between cardiac, genetics and specialist nursing services, consistently well received education events, an active and involved public involvement subgroup group, an extensive toolkit of protocols and guidelines and by working with stakeholders across organisational and professional boundaries. NICCS has gone beyond its original remit of familial arrhythmia to incorporate inherited cardiomyopathies, conditions which affect greater numbers of people across Scotland.

NICCS has authored this specification and will oversee further developments to ICC services in Scotland. It links to the rest of the UK via membership of the Association of Inherited Cardiac Condition (AICC).

Acknowledgement

The National Specification has been made possible via the permission of Professor Nigel Wheeldon Honorary Professor of Inherited Cardiac Conditions at the Northern General Hospital in Sheffield, to use his NHS England Commissioning Specification as a basis for this document.
ANNEX 3: Current Overview (Autumn 2019) of ICC Services in Scotland

NICCS encompasses patients and their families affected by two strands of conditions. The first is collectively known as familial arrhythmias. Specifically:

- Long QT Syndrome (LQTS)
- Short QT Syndrome (SQTS)
- Brugada Syndrome
- Arrhythmogenic Cardiomyopathy
- Idiopathic Ventricular Fibrillation (IVF)
- Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT)

The second disease area are the related, inherited cardiac condition called cardiomyopathy (of which arrhythmogenic cardiomyopathy is an arrhythmogenic variant) creating a family of conditions known as inherited cardiac conditions (ICC’s). The main form of inherited cardiomyopathy is Hypertrophic Cardiomyopathy (HCM).

The table below relates to the NICCS related regional centres, where specialist care takes place. In addition, non-specialist patient management occurs in all district general hospital sites in Scotland within cardiology departments.

<table>
<thead>
<tr>
<th>Specialist Centre</th>
<th>Health Board</th>
<th>DGH Health Board Linkages</th>
</tr>
</thead>
<tbody>
<tr>
<td>Glasgow</td>
<td>Greater Glasgow &amp; Clyde</td>
<td>Dumfries &amp; Galloway, Ayrshire and Arran, Lanarkshire, Forth Valley</td>
</tr>
<tr>
<td>Edinburgh</td>
<td>Lothian</td>
<td>Fife, Borders, Forth Valley (partially)</td>
</tr>
<tr>
<td>Aberdeen</td>
<td>Grampian</td>
<td>Highland, Orkney, Shetland, Western Isles</td>
</tr>
<tr>
<td>Dundee</td>
<td>Tayside</td>
<td>Fife (north east part only)</td>
</tr>
</tbody>
</table>

The table below relates to the NICCS multi-disciplinary team infrastructure in the specialist centres across Scotland. These clinics and meetings (which act as virtual clinics) discuss all ICC cases including diagnosis, testing, family screening, management and follow up. MDT clinics often include close family members in one clinic visit. None of these services existed prior to 2008.

<table>
<thead>
<tr>
<th>REGION</th>
<th>MDT NICCS (FANS) Clinic (per month)</th>
<th>MDT NICCS (FANS) Meeting (per month)</th>
</tr>
</thead>
<tbody>
<tr>
<td>West</td>
<td>4</td>
<td>1</td>
</tr>
<tr>
<td>East</td>
<td>1 (Cardiac genetic clinic, not MDT)</td>
<td>1</td>
</tr>
<tr>
<td>North</td>
<td>1</td>
<td>0.5</td>
</tr>
<tr>
<td>Tayside</td>
<td>1</td>
<td>2</td>
</tr>
</tbody>
</table>
ANNEX 4: Clinical Quality Indicators for Inherited Cardiac Conditions

The following CQIs would be formalised and implemented through the NICCS network. They will be used to monitor the effectiveness of services. NICCS will oversee the measurement and analysis of these CQIs each year and report to commissioners via its annual reporting processes. Some of these CQIs are not quantifiable and will require qualitative evidence.

1. That each regional specialist service has a single point of contact for all referrals
2. There is evidence of a mechanism for patient feedback and involvement in service development
3. Evidence of a formal transition process in place (from paediatric to adult care) according to agreed guidelines
4. A multidisciplinary team meeting is held at least once per month and minutes are recorded
5. Evidence of ongoing service evaluation (audit, quality improvement, research)
6. Evidence that individuals at risk of developing an ICC e.g. “gene positive relatives” have an appropriate clinical surveillance in place. This should not be specific to HCM but relevant to all ICCs
7. Timely review of families requiring investigations relating to Sudden Unexplained Death (SUD)/Sudden Cardiac Death (SCD)
8. Provide evidence that ensures there is a systematic approach to managing patients who DNA from ICC clinics, referral and follow up