

# Caring for children and young people with inherited cardiac conditions: the evolving role of specialist nurses

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None declared

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## Abstract

Inherited cardiac conditions (ICCs) encompass a range of rare genetic heart diseases and require the expert care of a skilled multidisciplinary team. Increased awareness of these conditions and advances in genetic testing have led to a rise in demand for specialist ICC services. The Centre for Inherited Cardiovascular Diseases at Great Ormond Street Hospital in London is a tertiary centre for the diagnosis and management of ICCs in children and young people. Specialist nursing roles have developed at the centre in recent years in parallel with the growth of ICC nursing. There are increasing opportunities for advanced and autonomous nursing practice supporting the provision of high-quality care for children and young people affected by ICCs and their families.

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## Introduction

Inherited cardiac conditions (ICCs) encompass a range of genetic heart diseases including inherited cardiomyopathies, inherited arrhythmias and aortopathy syndromes. Many patients with an ICC are at increased risk of sudden cardiac death (SCD). Increased awareness of ICCs has led to an increased demand for specialist ICC services nationally (Alway et al 2024).

The incidence of ICCs is variable but all are considered rare diseases in children – the accepted definition of ‘rare’ being  $\leq 1$  in 2,000 (Department of Health and Social Care 2021). In a systematic review of the incidence of SCD among people aged 12–39 years, Couper et al (2020) found that, in most studies, the incidence was between one and two cases per 100,000 person-years. The incidence of ICCs in childhood is not well known and requires further research (DeWitt et al 2019).

Table 1 gives an overview of ICCs.

| <b>Group</b>               | <b>Examples of conditions</b>                                                                                                                                                                                                                                                                                                                                                                                                                            | <b>Symptoms and presentation</b>                                                                                                                                                                                                                                    | <b>Treatment</b>                                                                                                                                                                                                                                                                                                                                                                                                                                                                             |
|----------------------------|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| Inherited cardiomyopathies | <ul style="list-style-type: none"> <li>» Hypertrophic cardiomyopathy</li> <li>» Arrhythmogenic cardiomyopathies including: <ul style="list-style-type: none"> <li>– Arrhythmogenic right ventricular cardiomyopathy</li> <li>– Arrhythmogenic dilated cardiomyopathy</li> <li>– Non-dilated left ventricular cardiomyopathy</li> </ul> </li> <li>» Rare presentations such as Carvajal syndrome, RASopathies, metabolic and storage disorders</li> </ul> | <ul style="list-style-type: none"> <li>» Syncope</li> <li>» Presyncope</li> <li>» Palpitations</li> <li>» Chest pain</li> <li>» Shortness of breath</li> <li>» Reduced exercise tolerance</li> <li>» Atrial arrhythmia</li> <li>» Ventricular arrhythmia</li> </ul> | <ul style="list-style-type: none"> <li>» Pharmacological treatment, for example betablockers</li> <li>» Implantable cardioverter defibrillator (ICD) for life-threatening arrhythmia</li> <li>» Electrophysiology procedures, for example ablation, to target electrical pathways causing ventricular ectopy or arrhythmia</li> <li>» Septal myectomy for the relief of left ventricular outflow tract obstruction</li> </ul>                                                                |
| Inherited arrhythmias      | <ul style="list-style-type: none"> <li>» Long QT syndrome</li> <li>» Brugada syndrome</li> <li>» Catecholaminergic polymorphic ventricular tachycardia</li> <li>» Short QT syndrome</li> <li>» Rare presentations such as Andersen-Tawil syndrome, Jervell and Lange-Nielsen syndrome</li> <li>» Sudden Arrhythmic Death Syndrome (SADS)</li> </ul>                                                                                                      | <ul style="list-style-type: none"> <li>» Syncope</li> <li>» Seizures (including febrile seizures)</li> <li>» Presyncope</li> <li>» Palpitations</li> <li>» Ventricular arrhythmia</li> </ul>                                                                        | <ul style="list-style-type: none"> <li>» Pharmacological treatment, for example betablockers or anti-arrhythmic medicines</li> <li>» Electrophysiology procedures, for example ablation, to target electrical pathways causing ventricular ectopy or arrhythmia</li> <li>» ICD for life-threatening arrhythmia or pacemaker for cardiac conduction abnormalities</li> <li>» Left cardiac sympathetic denervation (LCSD) to reduce risk of life-threatening ventricular arrhythmia</li> </ul> |
| Aortopathy syndromes       | <ul style="list-style-type: none"> <li>» Marfan syndrome</li> <li>» Loeys-Dietz syndrome</li> <li>» Vascular Ehlers-Danlos syndrome</li> <li>» Familial thoracic aortic aneurysm</li> <li>» Turner syndrome</li> <li>» Bicuspid aortic valves</li> </ul>                                                                                                                                                                                                 | <ul style="list-style-type: none"> <li>» Syncope</li> <li>» Presyncope</li> <li>» Chest pain</li> <li>» Shortness of breath</li> <li>» Reduced exercise tolerance</li> </ul>                                                                                        | <ul style="list-style-type: none"> <li>» Pharmacological treatment, for example betablockers or angiotensin receptor blockers (ARBs)</li> <li>» Prophylactic aortic root surgery to repair or replace the dilated aortic root and reduce risk of aortic dissection</li> </ul>                                                                                                                                                                                                                |

(Adapted from Erbel et al 2014, Stiles et al 2021, Zeppenfeld et al 2022, Arbello et al 2023)

The Centre for Inherited Cardiovascular Diseases at Great Ormond Street Hospital (GOSH) in London, England, was established in 2005 and has developed into a highly specialist service providing care for children and young people diagnosed with an ICC or who are at risk of having inherited an ICC. Concurrent with the evolution of the ICC nursing subspeciality, nursing roles have developed which offer opportunities for autonomy, growth and innovation in practice.

This article describes the development of the Centre for Inherited Cardiovascular Diseases – referred to throughout as ‘the ICC service’ – and of specialist nursing roles at the service. It discusses the challenges specific to the care of children and young people affected by ICCs and ways in which nurses can help address these challenges. Nurse-led approaches to patient-centred care are described and areas for professional development are identified.

## Development of the service

GOSH is a specialist tertiary paediatric hospital. It has a cardiac day ward, a cardiology inpatient ward including a high-dependency unit, and a cardiac intensive care unit. The ICC service is one of several cardiology subspecialties at GOSH. It provides care for children and young people diagnosed with an ICC and those at risk of having inherited an ICC. It also sees children and young people presenting with out-of-hospital cardiac arrest, many of whom are subsequently diagnosed with an ICC.

**The service has experienced exponential growth over the last 15 years. Today, it cares for over 6,000 patients and receives over 1,000 referrals per year.** This rapid growth is due to several factors, including greater awareness of ICCs and an increase in genetic testing due to the growing knowledge of causative genes.

## How the service works

The ICC service is primarily outpatient-based. Disease-specific clinics run daily, with a total of ten outpatient clinics per week. The service also runs specialist outreach clinics in Scotland and Ireland. Inpatient admissions under the care of the ICC team account for around 500 inpatient days per year.

The service receives referrals from across the UK and Europe, primarily from GPs and paediatricians, including paediatricians with expertise in cardiology. Around 20-30% of referrals are received from other tertiary and specialist centres in the UK requesting a second opinion. Children and young people diagnosed with an ICC undergo a range of cardiac investigations to evaluate clinical risk and symptoms so that adequate management strategies can be implemented. Children and young people with a family history of ICC or with confirmed genetic predisposition are regularly screened to detect signs of disease onset.

In line with international guidelines and contemporary evidence (Ommen et al 2020, Stiles et al 2021, Arbelo et al 2023), the service sees patients from birth to the age of 18 years, when they transition to adult services. Screening children from an early age is vital due to the risk of adverse events such as SCD; research has demonstrated that in ICCs, acute presentations, although rare, do occur at early ages (DeWitt et al 2019, Norrish et al 2019a).

### **Clinical expertise, staffing and training**

Caring for children and young people affected by ICCs requires knowledge and experience across cardiology and genetics. Much of the current literature relating to ICCs is based on studies in adults (Stiles et al 2021, Arbelo et al 2023) and paediatric guidelines are often lacking, so clinical expertise is essential.

The ICC service strives to incorporate the three aspects of evidence-based practice in the care it provides to patients: clinical expertise, research evidence and patient preferences (Haynes et al 1996). Centralising the care of children and young people diagnosed with an ICC creates large cohorts of patients with the same diagnoses, enabling staff to cultivate essential clinical expertise and undertake research that will guide care – for example, the ICC team has developed an SCD risk calculator for children with hypertrophic cardiomyopathy (Norrish et al 2019b). Finding out about patients' preferences and tailoring care accordingly is considered vital, and feedback from patients and families is regularly sought to check that their preferences are taken into account.

The NHS service specifications for ICC services (NHS England 2013) due to be updated in 2024, set out staffing requirements which are reflected in the service's multidisciplinary team. In addition to specialist nurses, the team includes specialist doctors, geneticists and genetic counsellors, cardiac physiologists, clinical psychologists, social workers, family support workers, patient pathway coordinators and administrative support workers. This wide-ranging skillset allows the service to provide high-quality, holistic care.

The NHS service specifications for ICC services (NHS England 2013) recommend that all professionals working with children and young people affected by ICCs are appropriately trained and carry out sufficient levels of activity to maintain specialist knowledge. Training should include taking assent for, or consent from, children and young people as well as informing them about, and supporting them with, their care and treatment (NHS England 2013).

### **Role of nurses within the service**

#### **Clinical nurse specialist team**

The CNS team at the ICC service comprises band 6 and band 7 nurses filling 7.2 whole-time equivalent (WTE) posts. Each CNS is embedded within one area of the service but can provide cover in other areas as needed. The role of CNSs at the service is complex and varied, encompassing many areas of patient care, and the input of CNSs is crucial in patients' journey through the service from initial referral to transition to adult services.

**Figure 1 illustrates the role of CNSs at the service.**

#### **Figure 1. Role of clinical nurse specialists at the service**

The CNS team coordinates a fortnightly ICC psychosocial meeting attended by cardiac psychologists, social workers and family support workers to address psychosocial and safeguarding concerns. In paediatric settings, such concerns can be identified, for example, if a patient is not being brought to their appointments or if there is a lack of adherence to treatment or disengagement from services (Chambers et al 2021).

Patients under the care of the ICC service may be followed up throughout childhood into their late teens. It is therefore vitally important to make any interaction with the service a positive experience from the outset. As children grow older, the CNS team may be involved in supporting them with symptoms, treatment regimens, restrictions on physical exercise and education choices. The duration of the nurse-patient relationship in lifelong conditions such as ICCs fosters a partnership that can be key in supporting patients to overcome challenges as they arise.

Patients with an ICC often have similar experiences (Potterton et al 2024) and psychosocial concerns (Ingles et al 2013). Specialist nurses have an important role in providing holistic patient-centred care, which includes managing anxiety, explaining risks and providing support with symptoms. One example of the patient-centred care provided to patients at the ICC service is the organisation of support days for children who live with an implantable cardioverter defibrillator (ICD). These events bring children together for educational sessions, information from cardiac charities and crafts and games activities. The CNS team is well placed to organise these events, since their relationships with patients and families give them a unique understanding of their needs. Patients are invited to provide feedback and suggest topics for future events, helping to ensure that these respond to their needs and wishes. Families value the opportunity to meet other people with similar experiences to theirs.

### **Advanced clinical practitioner role**

The ICC service employs one advanced clinical practitioner (ACP) (band 8a). The role developed from a previous CNS position, which demonstrates the potential for nurses working in specialist areas to progress to advanced practice. A master's degree in advanced clinical practice enables the ACP to autonomously offer consultations, assess patients and prescribe treatments. The ACP has expertise in cardiac implantable electronic devices and oversees a nurse-led pathway to ensure consistent and high-quality care for patients who use such a device. This includes counselling and education before implantation, inpatient management during implantation and follow-up.

As the most senior nurse in the ICC team with oversight of all three areas of the service (cardiomyopathy, inherited arrhythmia and aortopathy), the ACP supervises the professional development and education of all specialist nurses who join the ICC team and existing team members. This helps standardise the quality of nursing care and identify areas where further development may be needed.

### **Research nurse role**

In 2017, the ICC service received charity funding for a new research nurse post (band 7). The large and varied cohorts of patients seen at the service provide unique opportunities for research into ICCs in children and young people. The research nurse works within a multidisciplinary research team – including research assistants and research fellows – to develop research projects, explain research to patients and support patients through the consent process. The research nurse also assists with the collection and analysis of data and the drafting of articles submitted for publication.

The research nurse facilitates a wide range of projects including retrospective cohort studies, case studies and the investigation of new diagnostic and monitoring tools – see for example Norrish et al (2019b), Bueno-Beti et al (2023), Boleti et al (2024) and Lawley et al (2024). Research is integrated into routine clinical care and disruption to patients is kept to a minimum. Furthermore, the research nurse supports the CNS team with clinical audits and service evaluations, helping to improve practice by gaining systematic patient feedback (Field et al 2018).

### **Patient and public involvement and engagement**

Nurses are uniquely placed to advocate for patients and can help bridge the gap between patients and service providers. The ICC nursing team has organised and led, with clinical psychology support, focus groups with patients and families affected by different ICCs with the aim of listening to their experiences. The information collected is used to guide research and service development (NIHR 2019). Although ethical approval is not required for patient and public involvement and engagement (PPIE) activities, the team always ensures that they are underpinned by ethical principles (Tollit et al 2023).

The value of the information collected is immeasurable, since these focus groups allow the voice of patients and families to be heard in the development of services and research that directly concern them. Improvements that have been made following PPIE activities include the introduction of a model heart for teaching during clinic appointments and the creation of educational videos showing the

clinic process and various cardiac investigations being undertaken. Focus groups have also informed the development of new research projects (Bueno-Beti et al 2023) and the awarding of research grants, notably a PhD Fellowship for one of the nurses on the team.

## **Challenges of care**

Caring for children and young people affected by ICCs presents multiple challenges, two of them being genetic testing and counselling and the transition to adult services.

### **Genetic testing and counselling**

Due to the hereditary nature of ICCs, genetic testing is a crucial part of the services offered to families. It can be used to confirm a suspected clinical diagnosis, to confirm the pathogenicity of a genetic anomaly, to conduct predictive testing in asymptomatic patients and to support patients with future reproductive choices.

Genetic testing in children has historically been contentious due to concerns about autonomy and consent, the potential negative psychological effects and potential genetic discrimination (Borry et al 2006, Fryer et al 2000, Twomey et al 2006). However, evidence increasingly suggests that, in a specialist setting and with appropriate support, genetic testing for ICCs in children and young people can be beneficial for the child and family (Landstrom et al 2021). The identification of a pathogenic variant can aid in risk stratification and management, while a pre-symptomatic genetic diagnosis can lead to earlier diagnosis and more timely management (Ormondroyd et al 2014).

Living with an ICC can be challenging for families, requiring them to understand complex genetic concepts and make decisions that are right for their child, for themselves and for the wider family. Genetic counselling aims to help patients and families understand and adapt to the medical and psychological consequences of inherited conditions (Patch and Middleton 2018). The ICC service has benefited from the inclusion of a genetic counsellor role since 2018.

CNSs at the ICC service coordinate genetic testing for families, sharing information (with the family's consent) and facilitating testing for relatives where appropriate. CNSs also offer informed, objective and professional advice and support in addition to the information provided by the genetic counsellor, distilling complex topics in a way that makes them easier to understand for patients and families. Understanding the genetic basis of one's condition is likely to enhance self-management. When no genetic cause is identified, CNSs support patients and families to manage the uncertainty around the child or young person's condition. Finally, CNSs may themselves offer genetic counselling to patients (Buaki-Sogo and Percival 2022), if resources allow and if they have received appropriate training. University modules in genomics and counselling skills are available for nurses with a special interest in genetics; however, having undertaken these modules is not requirement for CNSs joining the service.

### **Transition to adult services**

The transition to adult services can be daunting for patients and families, particularly since some ICCs, for example cardiomyopathies, tend to present or progress during adolescence. Additional factors such as puberty, education, relationships, independent living and peer pressure can combine to create a period of substantial change for the young person (Potterton et al 2024).

Entering adult services may have advantages, such as autonomy and a more appropriate setting. However, when patients have been regularly followed up since childhood, leaving the service they are familiar with brings a sense of loss (Kovacs and McCrindle 2014). The ICC service aims to offer optimal education and support throughout the transition process to make it as smooth as possible.

At the time of transitioning, it is important to ensure continuity of care and the safe transfer of patient records. However, preparing patients for the transition is a long-term process that must start much earlier (Strijbosch et al 2016). This process should equip young people with the knowledge and understanding to manage their health as independently as possible. Patients are affected by their condition differently, have differing attitudes and levels of understanding, and will reach independence at different ages (Moons et al 2021). A tailored preparation process is therefore needed, which requires considerable resources.

The CNS team has an essential role in preparing young people for transition through individualised education and transition consultations. Transition work is undertaken by all CNSs in the ICC team. Regular meetings between CNSs and their counterparts in adult services support communication and enable effective handovers. Special transition clinics where patients have the opportunity to meet the CNSs from the adult service they are transitioning to are ideal for establishing links, with the aim to minimise the number of patients lost to follow-up as they transfer between services. The ICC service at GOSH has close links with adult services at St

Bartholomew's Hospital. A majority of patients who have been under the care of the GOSH ICC service transition to Barts when they reach the age of 18 years. Regular team meetings are held between the two services to coordinate their care.

## **Areas for development**

There are significant areas for development in ICC nursing, both for the ICC nursing team and ICC service at GOSH and for the subspecialty as a whole.

## **Competency framework**

ICC nursing is a relatively young subspecialty of cardiac nursing and is therefore behind other areas in terms of developing national standards of care and professional competencies. There is no national framework outlining care standards and staff competencies in ICC nursing, resulting in considerable variation nationally. Skills and responsibilities such as genetic counselling and prescribing are a part of role of clinical nurse specialists (CNSs) in some ICC centres but not in others.

In the absence of a national framework, managing an ICC specialist nursing team requires both a deep understanding of the demands of the service and effective staff appraisals to identify the skills and learning needs of each team member and support their professional development. A recently created national network of ICC nurses provides a valuable resource for peer support and education.

The ICC nursing network is in the process of developing national care standards and professional competencies, which will outline the skills and experience required for nurses to operate as specialist ICC CNSs. It is hoped that this future competency framework will structure ICC nursing roles in a similar way to the competency framework on congenital cardiac nursing for children and young people (Royal College of Nursing 2021), and that it will provide a clear pathway for professional development from beginner to expert across the domains of clinical competency, education, research and leadership.

## **Service models**

Nationally, it is proposed that ICC services move towards a 'hub and spoke' model of care to better manage the geographical coverage of expertise and the increasing number of patients seen in tertiary centres (Alway et al 2024). Specialist nurses are well-placed to facilitate this change, providing education and support to regional centres and helping to standardise care.

One long-term plan at the ICC service at GOSH is the development of a young people's service that would bridge the gap between children and adult services. The aim is to share care between the services at GOSH and Barts for patients aged between 14 and 25 years, allowing a gradual transition process. This young people's service will require significant funding, but could ensure that young people affected by ICCs receive comprehensive support at an important time in their life. In keeping with a patient-centred approach, a series of patient focus groups and interviews will be used to identify areas for improvement in the transition process and inform the development of resources that are more engaging for adolescent patients.

It is widely regarded as a challenge for nurses to work with true autonomy in the care of children and young people. Often, only the most senior specialist nurses who have undergone additional assessment training are able to lead clinics. The ICC team at GOSH is working to develop nurse-led clinics, which will enable experienced nurses on the team to manage their own cohort of patients. Nurses will be required to undertake specific training and demonstrate their professional development, based on the national framework that is being developed at the moment.

## **Conclusion**

ICCs require the expert care of a skilled multidisciplinary team. Within that team, specialist nursing roles are vital to holistic patient and family care. Nurses working in specialist ICC services are uniquely placed to identify and address the needs of children and young people affected by ICCs and their families. Due to increased awareness of ICCs and advances in genetic testing, the ICC service at GOSH and its nursing team have rapidly grown over the past two decades. Specialist nursing roles are evolving as the knowledge of ICCs increases and new treatments emerge. Specialist ICC nursing for children and young people presents increasing scope for advanced, autonomous and innovative practice.

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