IMPROVING CARE FOR RARE CONDITIONS IN SCOTLAND

A report by the Cross Party Group of Rare, Genetic and Undiagnosed Conditions

GENETIC ALLIANCE UK

ABOUT GENETIC ALLIANCE UK

Genetic Alliance UK is the national charity working to improve the lives of patients and families affected by all types of genetic conditions. We are an alliance of over 200 patient organisations. We undertake various initiatives to improve health service provision, research and support for families. These initiatives include:

Rare Disease UK, a multi-stakeholder coalition brought together to work with government to effectively implement the UK Strategy for Rare Diseases.

SWAN UK (syndromes without a name), the only UK-wide network providing information and support to families of children and young adults affected by undiagnosed genetic conditions.

Published by: Genetic Alliance UK
Email: contactus@geneticalliance.org.uk
Website: geneticalliance.org.uk

Registered charity numbers: 1114195 and SC039299
Registered company numbers: 05772999
Published: 22 March 2021
Author: Natalie Frankish
Cover photograph by: Claire Tennant Photography

Genetic Alliance UK provide the Secretariat to the Cross Party Group on Rare, Genetic and Undiagnosed Conditions.

The CPG would like to thank the many people living with a rare condition, their families and carers, patient organisations and health professionals who have attended CPG meetings to share their experience and views on how care for rare conditions in Scotland can be improved. The CPG is grateful to those who have given their time to contribute to meetings and to this report and its recommendations.

CONTENTS

INTRODUCTION

The Cross Party Group on Rare Genetic and Undiagnosed Conditions

Cross-Party Groups (CPGs) provide an opportunity for Members of the Scottish Parliament (MSPs) to engage with external stakeholders on a particular subject. Although CPGs do not have power to introduce issues formally into the parliamentary or government systems, they are an important forum for the consideration of particular issues and can seek to inform policy and service development.

The Cross Party Group on Rare, Genetic and Undiagnosed Conditions (The CPG) was recognised by the Standards, Procedures and Public Appointments Committee of the Scottish Parliament in 2017.

The CPG was formed to:

- Act as a channel of communication between the Scottish Parliament and families affected by rare, genetic and undiagnosed conditions.
- Act as a channel of communication between the Scottish Parliament and those working in the fields of research, treatment, care and prevention of rare, genetic and undiagnosed conditions.
- Monitor and contribute to the implementation of the Scottish Plan for Rare Diseases in Scotland.
- Identify areas where inequalities exist in provision of care for rare, genetic and undiagnosed conditions and campaigning for improvement.
- Examine areas of health and social care policy or service provision relating to rare, genetic and undiagnosed conditions.

The CPG is Co-Convened by Bob Doris MSP and Mark McDonald MSP.

The Secretariat is provided by Genetic Alliance UK, the national charity working to improve the lives of patients and families affected by rare, genetic and undiagnosed conditions. Genetic Alliance UK is an alliance of over 200 patient organisations. Genetic Alliance UK is home to Rare Disease UK – the national campaign for people with rare diseases and all who support them – and SWAN UK (syndromes without a name), the only dedicated support network for families of children and young adults with undiagnosed genetic conditions in the UK.

A full list of the current membership of the CPG on Rare, Genetic and Undiagnosed Conditions, in addition to details of meeting and associated papers, can be found on the Scottish Parliament website – parliament.scot/msps/rare-genetic-and-undiagnosed-conditions.aspx

Policy context

The UK Strategy for Rare Diseases (2013-2020)

The UK Strategy for Rare Diseases was published in 2013. The Strategy, agreed by all four health departments of the UK, aimed to 'ensure that no one gets left behind just because they have a rare disease'.

Key features of the Strategy include; a personal care plan for every patient that brings together health and care services, ensuring patients, their families and carers have the information they need and developing better methods of identifying and preventing rare diseases. The Strategy also committed to improving diagnosis, providing better education and training for health and social care professionals and building on research to improve personalised approaches to healthcare for those with a rare disease.

In 2014, the Scottish Government laid out its approach to implementing the Strategy in 'It's Not Rare to Have a Rare Disease: The Implementation Plan for Rare Diseases in Scotland', committing to implementing the 51 commitments of the UK Strategy. The Scottish Implementation Plan was set within the strategic context of the NHS Scotland 'Quality Strategy' and the associated '2020 Vision and its accompanying Route Map', with a strong focus on safe, person-centred, effective care.

Both the UK Strategy and the Scottish Plan were introduced without funding. The commitments within the Scottish Implementation Plan were intended to be met through existing funding arrangements.

The UK Strategy and Scottish Plan came to an end on 31 December 2020.

The 'new' UK Rare Diseases Framework

On 9 January 2021, a new UK Rare Diseases Framework was published.[1]

The Framework addresses key challenges faced by people living with a rare condition and identifies four priorities: faster diagnosis, increased awareness of rare diseases, better coordination of care, and improving access to specialist care, treatments and drugs.

The four nations of the UK are expected to develop and deliver Action Plans, highlighting steps they will take to meet the aims of the framework in accordance with their own arrangements. In order to ensure cross-border collaboration and maximise the benefits of the framework for the rare disease community, each nation will follow the below core principles when delivering action plans and implementing the framework. Each nation will:

- Deliver the aims of the UK Rare Diseases Framework under each of the priorities and underpinning themes.
- Consider where Action Plans can contain specific and measurable commitments under each focus area and regularly review commitments (every 1 to 2 years).
- Develop policy commitments with expertise, in close collaboration with patients and others living and working with rare diseases.
- Ensure any impacts on health inequalities are considered when developing action plans.
- Ensure that the experiences of rare disease patients during the COVID-19 pandemic are reflected in the development of action plans and implementation of framework priorities and themes.
- Ensure that the voice of the rare diseases community is recognised across the system and that work, as part of the UK Rare Diseases Framework, is aligned with other relevant policy development, such as mental health and social care.
- Work collaboratively across nations to share knowledge and best practice.
- Review progress made towards the aims of the framework every 5 years and update priorities when necessary.

The UK Framework states that 'where possible, each nation will aim to publish the action plans in 2021.'

This report

This report provides a narrative of the evidence and experiences presented to the CPG from people living with a rare condition, their families and the patient organisations that support them.

Whilst it is clear that there has been some progress in the implementation of the Scottish Plan for Rare Diseases, particularly in the areas of diagnosis and early intervention, testimony provided at CPG meetings demonstrates that people living with a rare condition continue to face fundamental challenges in relation to their care.

Evidence presented at meetings suggests that access to information, access to specialist care and research and support for mental health and emotional wellbeing is still difficult for people living with rare conditions in Scotland. Whilst there have been positive examples of care that is well coordinated, fragmented care across rare conditions remains commonplace. Patient organisations have regularly raised examples of when poorly coordinated care has contributed to people feeling lost within the healthcare system, unable to effectively manage their condition and in some particularly concerning situations, failing to receive correct care and treatment. The opportunity to ensure efficient use of NHS resources, at a time when this is required most, has been missed as a consequence of a lack of improvement in care coordination for rare conditions.

The recommendations contained in this report are intended to inform the development of the Scottish Government's Rare Disease Action Plan.

RARE DISEASE CARE IN SCOTLAND

Overview

Rare conditions are defined by the European Union as a condition that affects less than 1 in 2,000 of the general population.[2] One in 17 people will be affected by a rare condition at some point in their lives. This equates to approximately 420,000 people in Scotland.[3]

The Orphanet database contained details of 6,172 unique rare conditions at the beginning of 2020. The most common rare conditions in the UK, such as Sickle Cell and Cystic Fibrosis, can affect a little over 10,000 people each. The rarest can affect just one family, with maybe only a few recorded cases in the world.

Whilst there are a wide range of rare conditions, there are common challenges faced by many living with a rare condition. These include; significant challenges in obtaining a diagnosis, accessing appropriate care and treatment and receiving coordinated care, as well as challenges with employment, education, managing a social life and emotional and mental wellbeing.

Most rare conditions affect multiple body systems, meaning that many professionals from different specialities and disciplines need to be involved in the person's care and treatment. Coordinated care across these specialities and disciplines is essential for ensuring people receive the best possible care.

In Scotland, health services for people living with a rare condition are delivered by NHS Scotland through 14 NHS Health boards. For some rare conditions, specialist services are commissioned in the rest of the UK on behalf of all Scottish NHS Boards by NHS National Services Scotland. Rare conditions touch every aspect of NHS service delivery.

There is no rare disease service or centre within NHS Scotland, nor is there an individual within NHS Scotland who is responsible for overseeing the delivery of services for rare diseases. Information is collected on an individual rare condition basis, but there is no central collection or analysis of rare conditions across Scotland. Rare Disease Policy in Scotland is the responsibility of the Scottish Government.

It is important to acknowledge that many people living with a rare condition report that they receive excellent care from NHS Scotland. However, the CPG is a forum to explore areas of care were NHS Scotland services are not meeting the expectations of people living with rare conditions.

The CPG has held meetings on a variety of topics including; genomic sequencing, access to medicines for rare conditions and the impact of COVID-19. There are, however, two issues regularly raised with the Cross Party Group:

- Lack of awareness of rare conditions amongst health professionals.
- Poor care coordination.

Cross Party Group members attending Rare Disease Day reception in the Scottish Parliament

AWARENESS OF RARE CONDITIONS AMONGST HEALTH PROFESSIONALS

People living with, or caring for someone with, a rare condition have frequently told the CPG that health professionals lack sufficient awareness of rare conditions and that this has had a detrimental impact on their experience of care.

Patient group representatives point to examples of delayed diagnosis, lack of information and signposting, failure to involve patients in decisions about their care and delays in receiving appropriate specialist care or treatment, to demonstrate the impact of health professionals having insufficient expertise to appropriately manage a person with a rare condition. This can lead to a loss of confidence in healthcare services, the necessity to seek second opinions or file formal complaints, and can have an extremely detrimental impact on the emotional and mental wellbeing of the person living with the condition and those that support them. A recent report by Rare Disease UK identified that 88% of individuals feel emotionally exhausted because of their rare disease and that their struggle to have their needs met can lead to anxiety, low mood and depression.[4]

The impact of the lack of awareness of rare conditions amongst healthcare professionals on outcomes for patients can include:

Delay in reaching a diagnosis

People who have shared their story with the CPG frequently use language such as 'fight', 'battle', 'struggle' or 'ordeal', to describe their experience of reaching a diagnosis for themselves or a person that they care for. People have described not being believed when presenting symptoms to healthcare professionals for the first time, or healthcare professionals not considering a series of different symptoms presenting over a period time as being connected. This can have a significant impact on the time it takes to reach a diagnosis.

> **'I feel like it was one continuous fight to get his diagnosis – no one would listen to my concerns. I knew there was something, I just needed to get in front of the right person – but I had to keep pushing and complaining until I got there. Eventually I got the referral to a specialist. Even though it still took a little bit more time, things began to fall in to place... Now we have a proper care package, but so much time was wasted and we had so much heartache getting here.'**

Delay in access to the correct care and/or treatment

The 'battle' does not necessarily end with a diagnosis. It can often be the case that a diagnosis is of a condition so rare, that the healthcare professional has not heard of it, does not know how to manage the condition and is unaware of when to, or how to, access appropriate specialist or expert support. This can lead to people living with a rare condition being left to research the details of their condition and how to manage it themselves. It can result in delays to accessing appropriate care and treatment, often resulting in a person's health deteriorating unnecessarily and in some cases, people receive inappropriate interventions or treatment.

> **'I had been taken by ambulance and ended up in hospital. Although the doctor treating me had heard of my condition, he was completely overwhelmed by my medication needs – I was given the wrong medication which rendered me completely immobile and unable to function. I was having spasms and in a lot of pain – all of which could have been avoided.'**

Frustration at repeating medical history

Patients with rare conditions often come into contact with health and social care professionals who know little or nothing about their conditions and find themselves needing to explain complex details of their medical histories over and over again. People have complained that this takes away time from medical appointments and can lower a patient's confidence in their healthcare professional. Failure to update and share patient records or care plans across different health specialities can exacerbate this problem. Healthcare professionals have shared their frustrations of the limitations of the NHS IT

system which restricts the ability to share patient information and records accurately and in a timely manner between different NHS Health Boards, or between specialist and local services.

Inappropriate treatment being administered during emergencies

Rare conditions unknown to emergency medical staff can pose a particularly serious problem. If a patient is unable to communicate in an emergency situation (e.g. if they are unconscious) mistakes can be made in the accurate diagnosis, treatment and care of an individual; in some cases, this can have fatal consequences. For example, a person with a rare condition may have a different response to a medicine than patients with a similar, more common condition.[5]

Patient organisations have shared best practice examples of anticipatory care plans (such as Healthcare Improvement Scotland's "My Anticipatory Care Plan" and the PAMIS Patient Passport) and condition-specific medical alert materials which allow for the communication of hidden information, such as if someone has a rare disease, whether they have any allergies to medicines or any specific needs. However, there is inconsistency in approach across NHS Health Boards in which plans and alert cards are actively looked for and followed, and patient representatives call for consistent guidance on the templates used for care plans across Scotland and the development of a rare disease specific, and NHS Scotland branded, alert card to improve recognition.

Inadequate provision of information and signposting

Patient organisations have expressed frustration at the lack of information materials provided to people living with a rare condition, their parents and/or carers at the time of diagnosis. Too often people are given a diagnosis and little else, leaving patients, parents and carers with no option but to source the information themselves, often finding out-of-date, frightening or inaccurate information online.

> **'I was given no information at all, just a very long name on a piece of paper. I had to Google it all myself.'**

A recent report by Rare Disease UK[6] found that 49% of respondents to a survey did not feel that they were given enough information after their diagnosis. In addition to a lack of information about the condition they have been diagnosed with, people also report a lack of appropriate signposting to support organisations or peer networks. This can result in people feeling overwhelmed by many unanswered questions and uncertainty about what the future may hold, it can delay people accessing appropriate social care and financial support and can lead to feelings of isolation.

Lack of information on opportunities to participate in research

Patient organisations have spoken of the value of research in rare conditions, citing the potential to improve understanding of conditions, improve diagnosis and chances of developing a treatment. People living with a rare condition have spoken of the value of learning more about research opportunities, and health professionals have spoken about the challenges of finding people to recruit for studies for rare conditions. A study by Rare Disease UK demonstrates that although 80% of patients would be interested in participating in research, only 25% felt that they had been provided enough information on the opportunities available.[7] Many people living with rare conditions rely on their health professionals to share information of research opportunities, when their healthcare professional is not an expert in their condition, or familiar with how to access information on available studies and trials, patients can miss important opportunities to participate in research.

Patients and/or carers are not considered partners in decision making

Patient organisations have repeatedly called for people living with a rare condition (or their parent/carers) to be treated as the experts in their care. Citing their tendency of detailed research into their own condition and efforts to seek information from experts or others living with the same, or similar condition, patient groups attest that in many cases, a person with a rare condition will know more about their condition than a health professional with limited experience of it. However, people living with rare conditions frequently report that they are not listened to, or involved in decisions about their treatment or care.

> **'I am the expert – I've read everything ever written about my son's condition, I've spoken**

to anyone who I can find who is in the same boat and I live everyday with him, managing his care and his behaviour – but I am still just seen as Mum and I still have to fight to be taken seriously.'

The CPG recognises that healthcare professionals want to be able to provide the highest standard of care for their patients. However, rare conditions can pose some significant challenges to a healthcare professional's ability to do that. These include:

Lack of information available to health professionals

Information on rare conditions can be scarce. The nature of rare conditions means that there may not be a recognised patient organisation to produce and distribute high quality information. A condition may be so rare that there are few references within medical literature or journals. Where information does exist, it can be hard to find. In the Scottish context, this can be demonstrated by the comparative lack of information on rare conditions available on NHS Inform, Scotland's national health information service and online resource, with information typically only available for the most 'common' rare conditions.

Health professionals may have limited knowledge of trusted third sector organisations and support services for rare conditions, and in some cases there can be a reluctance to refer to patient organisations that are less well known or do not have direct affiliation with NHS Scotland.

'There is no central repository of information on rare conditions for patients and people working in the NHS in Scotland – no one-stop place they can go to access links and downloadable information, or find out about services available to their patients, there is no one in charge of rare diseases that they can ask for help. It means clinicians have to do the leg work – and some simply do not have the time, and others may not even have the inclination, to do that.'

Some health professionals may be unaware of how to source information on expert centres and specialist services for rare conditions, or how to make referrals. This is particularly true when such services exist outside of Scotland. There is not a resource within NHS Scotland, or a person or department responsible for, mapping rare conditions services and experts in Scotland. Indeed, information on the number of people living with a rare condition and the conditions that they have is not collected by NHS Scotland in a coherent way that allows for better understanding of the rare disease community. Work is underway to develop CARDRISS (the Congenital Anomalies and Rare Disease Registration and Information Service for Scotland), however this work is in its infancy and will not begin collecting comprehensive details of rare conditions for a number of years.

Insufficient opportunities for training

Patient organisations have called for greater opportunities for training health professionals in rare conditions, in particular paediatricians and general practitioners. Some patient organisations have shared examples of training modules that they have developed and delivered. However, much frustration is also expressed as a result of the lack of take up of training by health professionals, and specifically, general practitioners.

The introduction of mandatory rare conditions training in general practice and paediatric training has been recommended, as has the development of a well-publicised programme of Continuing Professional Development (CPD) training for all health professionals. Such a programme should include a generic training module on rare conditions and also include a range of condition-specific sessions which health professionals could utilise when necessary. To combat the problem of apathy, CPD points should be attached to the training modules, providing an incentive for health professionals to take the time to complete the training.

It is the view of the Cross Party Group that NHS Scotland must be better equipped to support healthcare professionals to support those living with a rare condition and their families. Consideration should be given to developing and introducing a central point of information in Scotland, including information on rare conditions and their management, details of available specialist services and appropriate patient organisations to refer or signpost to, information on clinical trials or research and training resources for healthcare professionals should exist within NHS Scotland.

CARE COORDINATION

Overview

Many rare diseases affect multiple systems of the body, which means several different professionals need to be involved to deliver effective care and treatment. The quality of overall care received is often affected by the quality of coordination between these healthcare professionals.

Without properly coordinated care, people affected by rare diseases often have to devote significant periods of time juggling and attending numerous appointments at different hospitals, sometimes travelling long distances to access the care they need. In addition to caring and medical tasks, time is spent making telephone calls to various providers and services, organising meetings, working with support groups, and researching information. With poor care coordination, management of a rare condition becomes a full-time role, taking up all a patient or carer's personal time, causing many people to decide to leave employment.[8]

> **'I think it's easy to forget about the impact on the whole family – we are travelling up and down to see an expert in London so often and that means unpaid time off work for my husband. We don't have much of a social life anymore and home life can be really stressful too. Our daughter ends up having to stay with her grandparents a lot. She understands and is a wonderful big sister, but it is really hard on her and she misses out on family time together and a lot of the 'normal' life experiences her friends have. I'm really worried about how it will affect her education when she gets to high school.'**

Results of a recent study[9] show that the experiences shared with the Cross Party Group are common in rare conditions. The study found that uncoordinated care typically resulted in an additional burden for patients and barriers or delays to accessing care. The impacts described by study participants either attributed to or exacerbated by uncoordinated care, included: impact on physical health (including fatigue), financial impact (including loss of earnings and travel costs), and psychosocial impact (including disruption to school, work and emotional burden).

Coordination of care can be defined as care that is delivered by 'working together across multiple components and processes of care to enable everyone involved in a patient's care (including a team of healthcare professionals, the patient and/or carer and their family) to avoid duplication and achieve shared outcomes, throughout a person's whole life, across all parts of the health and care system, including:

- Care from different healthcare services (e.g. different medical disciplines – medical, mental health, behavioural, health promotion).
- Care from different healthcare settings (including primary and secondary; community settings e.g. social care) and locations (e.g. rural/urban).
- Care across multiple conditions, or single conditions that affect multiple parts of the body.
- The movement from one service, or setting, to another.

Coordination of care should be family-centred, holistic (including a patient's medical, psychosocial, educational and vocational needs), evidence-based, with equal access to coordinated care irrespective of diagnosis, patient circumstances and geographical location.'[10]

Coordinated care can bring the benefits of:

- Ensuring all clinicians caring for someone with a rare condition consider each other's proposed interventions.
- Ensuring there is synergy in the patients' overall care and treatments do not interact negatively or adversely affect other body systems.
- Reducing the burden of a high frequency of clinics by scheduling them efficiently and with account of how results of tests and scans can feed into all aspects of care.
- Ensuring primary and secondary care providers

understand all aspects of care and can play their role appropriately, involving specialist teams appropriately.[11]

The COordiNated Care Of Rare Diseases (CONCORD)[12] study which began in 2018 will deliver a comprehensive description of the elements of care coordination and how they can be incorporated into models of service delivery. The taxonomy will form the basis of future cost analyses of different models of care. The findings of the CONCORD project should inform the Scottish Rare Disease Action Plan.

Care coordinators

The CPG has heard that coordinated care is best achieved when a care coordinator is in place to oversee care and facilitate collaboration between medical specialities, and where appropriate, between health and social care.

Care coordinators are trained professionals responsible for ensuring a care plan is in place and carried out. They may be known by other titles such as specialist nurses, key workers, patient navigators or care advisors. Their role is to ensure that people have a positive experience of the care that they receive by providing support and information to facilitate effective self-management and care is delivered smoothly.

Parents and carers of children with a rare condition report that when they have no one to support the coordination of their child's care, the responsibility falls on them to become an expert for the condition and a full-time advocate for their child.

'We have been talking about the need for care coordinators and patient navigators for years, it is just unreasonable to expect a parent to look after their sick child, do their jobs, manage all their appointments and on top of that, be the person who is making sure the different departments are talking to one another and that referrals and test results are being shared and followed up.'

Despite patients and carers demonstrating a preference for having a dedicated care coordinator, a recent survey[13] has shown that 71% coordinate their own care and only 4% have a dedicated care coordinator.

'It's chaotic & relies on me organising and chasing, which is exhausting...There is nothing I would love more than a dedicated person to coordinate my care for me.' [14]

Benefits of care coordination

There is evidence to show that when a care coordinator is in post, outcomes for patients are generally improved. The CPG has heard a number of examples of this.

A 2012 study of Sickle Cell and Thalassaemia Specialist Nurses in NHS England demonstrated the important role the specialist nurses had played in coordinating and managing care and providing psychosocial support. The impact of the nurses included patients reporting being better able to manage anxiety, a reduction in emergency hospital admission and patients and families indicating that having a 'key accessible professional' had improved their quality of care.[15] The CPG has heard that a similar service does not yet exist in Scotland, but would be welcomed by Scottish patients who report the detrimental impact of emergency hospital admissions on the quality of family life. Specialist nurse services for cystic fibrosis, haemophilia and primary immunodeficiency have also been cited by CPG members as examples of improving a patient's care experience.

Care coordinators can also play an important role in linking and coordinating health and social care services. The CPG heard how the former Tuberous Sclerosis Advisor post supported families in Scotland by providing and attending multi-disciplinary meetings with families, education, health and social work professionals with the aim of encouraging effective collaboration and communication to ensure a care plan is agreed and carried out.

It is not always the case that a care coordinator needs to be an expert in the person's particular condition. The Clinical Nurse Specialist based in East of Scotland Genetic Service provides valuable support to people living with a wide range of rare, genetic conditions. The Clinical Nurse Specialist in post has considerable experience working with a range of rare conditions and has the skill set to track down scarce information, research and identify specialist services and sources of support regardless of the rare condition. The role is also

pivotal in ensuring care plans reflect the needs of the person and their families and the role supports multi-disciplinary collaboration to ensure the smooth delivery of care.

Care coordinators play an important role in ensuring people with a health condition have a positive experience of the care that they receive. They ensure effective coordination of services, providing a vital source of information and support and assist in supporting effective self-management. Care Coordinators, by becoming a trusted point of contact, can also reduce the sense of isolation often reported by those living with a rare condition.

A recent study suggests that the improvements in care can be achieved with the support of a professional coordinator, using MDTs, care plans, technology and/or a point of contact to improve communication, and organising appointments to meet the needs of patients and families (by providing them locally or virtually where possible, offering a range of services in one visit, and scheduling them at a convenient time for the family.[16]

Care coordinators provide person-centred care that is responsive to individual personal preferences, needs and values and assuring that patient values guide all clinical decisions and as such, have the potential to deliver the key objectives of NHS Scotland's Realistic Medicine Policy.

Despite the benefits that they bring, very few people with a rare condition have a named care coordinator. Genetic Alliance UK's Rare Experience 2020 Report (2020) found that only 4% of people with a rare condition have access to a dedicated care coordinator[17] the majority of conditions represented at the CPG do not have a coordinator in post in Scotland. Consequently, poorly coordinated care continues to be a major issue for many people living with rare conditions.

Cost effectiveness of care coordination

The CPG acknowledges that introducing new care coordinator posts in Scotland would require investment, however there is evidence that care coordinators benefit the wider health and social care system due to their cost effective nature.

Costs associated with providing care coordinators have the potential to be offset in the short or medium term by increasing efficiency in the way services are utilised, and in the long term by the benefits gained from improving patient outcomes.

The advice, support and information provided by care coordinators for various conditions, both rare and common, have been shown to deliver cost benefits through saving GP and consultant time, preventing unplanned hospital admissions, reducing the length of hospital stays and enabling better management of conditions.

There are numerous studies of existing care coordination services which demonstrate potential cost savings:

- A study by Parkinson's UK on the impact of Parkinson's nurses in England shows that one nurse can save an average of £80,000 in unplanned hospital admissions by supporting patients in their home and in the community.[18] In other parts of the UK, nurses have reduced hospital admissions by 10%. In Scotland, for example, a Parkinson's nurse team has saved around £39,000 over 18 months in avoided hospital admissions, by providing early stage intervention.[19]
- A study by the Royal College of Nursing found that Rheumatology Nurse Specialists represented good value for money by reducing costs in primary care and saving consultants time. Per annum, outpatient work by the Rheumatology Nurse Specialists is worth £72,128 per nurse, and saves £175,168 per nurse by freeing up consultant appointments. In addition, telephone consultation reduces the number of GP appointments, saving £72,588 per nurse.[20]

It is clear that care coordinators have the potential to deliver excellent value for money, whilst playing an important role in improving patient outcomes – resulting in both short and long term savings to NHS Scotland.

THE FUTURE OF RARE DISEASE CARE IN SCOTLAND

Policy and funding

With the development of a new Rare Disease Action Plan for Scotland, there is opportunity to consider how best NHS Scotland can serve the needs of patients with rare conditions and the healthcare professionals who support them. For the new Action Plan to be a success, we must learn from the limitations and missed opportunities evident from the implantation of the 2014 Plan.

It is true to say that there were successes delivered under the Rare Disease Implementation Plan for Scotland (2014) including developments in genomics which resulted in improvements in diagnosis, the development of the Congenital Anomalies and Rare Disease Registry and Information Service for Scotland (CARDRISS) and the implementation of new care pathways, developed in collaboration with patients and services users. However, evidence provided to the CPG indicates that there has been no significant improvement in the experiences of healthcare reported by people living with rare conditions.

There are two factors which undoubtedly limited the success of the Rare Disease Implementation Plan (2014)

- The implementation of the Plan was the sole responsibility of the Scottish Government. No department or body within NHS Scotland was responsible and accountable for the delivery of the implementation plan, and
- The Plan was enacted with no dedicated funding to deliver its objectives.

To ensure greater success and improved patient outcome results, we cannot afford to repeat this mistake.

For rare disease policy to be truly effective in delivering better patient experience and outcomes, there must be a vehicle within NHS Scotland to drive forward its implementation. By funding a service for rare conditions within NHS Scotland an infrastructure can be built to deliver the priorities of the UK Framework for rare conditions and the Rare Disease Action Plan for Scotland, to monitor and measure progress and be accountable for policy delivery.

The new Rare Disease Action Plan for Scotland will require funding to deliver meaningful change, specifically investment in developing an infrastructure around rare conditions within NHS Scotland to make rare conditions more visible to health professionals, to provide a source of information and support for both health professionals and people living with a rare condition and deliver quality care to patients.

The future of care for rare diseases in Scotland

The CPG has heard testimony demonstrating where care for rare conditions has fell short of expectation but has also focused on considering what is considered 'good care' across the spectrum of rare conditions. During a meeting in March 2020, attendees were asked to consider what elements comprise good care. The following features were identified:

Health professionals that are knowledgeable about rare conditions: People living with a rare condition recognise that it is not possible for health professionals to know about every individual rare condition. However, it would be helpful if health professionals held an understanding of rare conditions in general, knew where to access reliable information and support and understood the challenges faced by people living with a rare condition. Health professionals should be supported by NHS Scotland to undertake training to better understand rare conditions. NHS Scotland should consider how healthcare professionals are supported to access reliable information on rare conditions, sources of expertise and information and support.

Access to high-quality condition-specific information: People living with a rare condition want to know as much as possible about their rare conditions, however information can be limited and/or hard to find. Healthcare professionals can also struggle to source reliable and accessible information for people in their care. It would be helpful if NHS Inform had a dedicated resource on rare conditions, providing general information about rare conditions and links to sources of condition specific information.

Access to information on research and clinical trials: People with rare conditions value information on research and clinical trials that they can participate in, either for their own benefit or for the benefit of others. People can find it difficult to source information on research relevant to them. In rare conditions, a relevant research project or clinical trial might exist in a different country. It would be helpful for NHS Scotland to have a research coordinator for rare conditions – someone who can collate and share information on clinical trials available and inform and support patients with decision making around taking part in research.

Improved protocols in emergency care: Anticipatory Care Plans (ACPs) are considered an essential tool for ensuring the appropriate care is given when a patient presents in an emergency care setting. Consideration should also be given to producing an NHS Scotland issued rare conditions 'Alert Card' containing key information about the patient's condition and healthcare needs, which could be carried by a patient and produced to health professionals when required. To be effective, ACPs and alert cards should be uniform across NHS Scotland Health Boards to improve recognition. ACPs should be developed through collaboration between patients and their health care professionals and their implementation should be supported by care coordinators.

Effective collaboration between professionals involved in care: People living with a rare condition, who may have to see multiple healthcare professionals across a range of specialties, expect the professionals involved in their care to work together. Effective collaboration ensures that a patient's needs are managed and that the people living with a rare condition do not spend their healthcare appointments repeating their symptoms and providing detailed information about how their care has been managed by other health professionals – collaboration between professionals, supported by a dedicated care coordinator, can ensure that accurate and up to date information is available to a healthcare professional ahead of their appointment with the patient.

A named person, responsible for overseeing care and support and providing a continued point of contact: People living with a rare condition regularly share that the impact of managing a health condition includes having to travel to attend multiple appointments, liaising with the many healthcare professionals involved in their care, uncertainty over who is their 'lead' clinician, chasing up referrals and test results and researching their own condition. The impact of this can affect day to day life sometimes resulting in loss of social life, difficulties remaining in work or education, financial hardship and an impact on emotional and mental wellbeing. It would be helpful for people living with a rare condition to have access to a care coordinator to assist in the management of their condition. A dedicated care coordinator, responsible for overseeing care and providing holistic support, should be available to every person with a rare condition.

A Rare Conditions Coordination Service for Scotland

The CPG recommends that an NHS Scotland, 'Rare Conditions Coordination Service' for Scotland (RCCSS) be funded.

Such a service would be an appropriate vehicle for delivering meaningful patient outcomes, in line with the definition of 'good care' as provided above.

Service model

The CPG proposes that a 'Rare Conditions Coordination Service for Scotland' (RCCSS) would suit a model similar to that of the former Single Gene Complex Needs Network, a short term project delivered under the Calman Review into Genetics in Scotland. The objectives of the RCCSS should be:

- To develop strategic approach to delivering services to people living with rare conditions

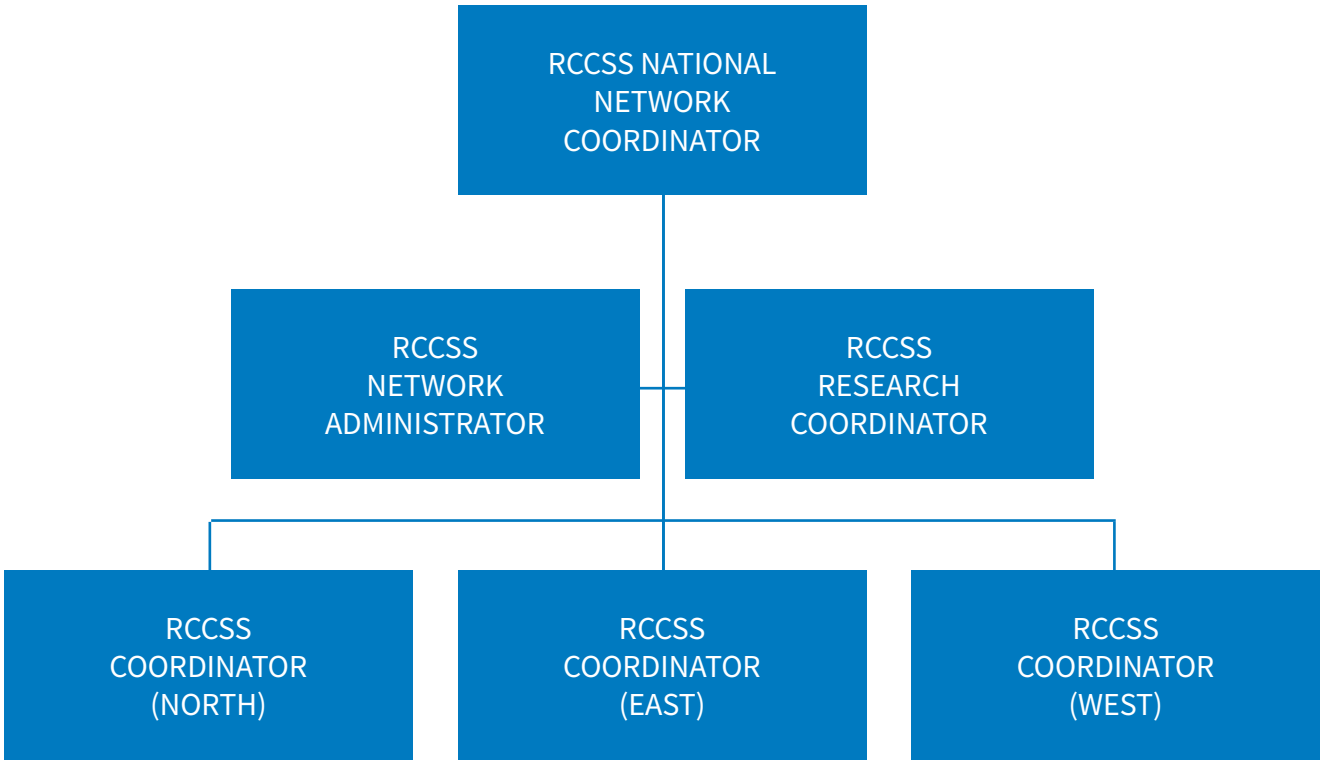
in Scotland through the identification of need, improving service delivery and working in collaboration to optimise patient outcomes.
- To provide direct support and information to families living with a rare condition.
- To support health professionals in NHS Scotland through the provision information on rare conditions, the support and services available to patients and the delivery of training and education to raise awareness of rare conditions.

The RCCSS requires there to a balance of nationally coordinated, but regionally delivered, care. This could be achieved by a network of team members, working regionally but coordinating nationally.

The CPG envisage that an initial service would consist of six team members:

1. **A National Network Coordinator:** Responsible for facilitating collaboration across the service, collating and interpreting data to identify patient populations and patient need, participating in discussion around service development, and promoting the service widely across NHS Scotland.
2. **Three Regional Care Coordinators:** Each care coordinator would be based within a health board region, but would be part of a wider RCCSS network to facilitate case discussion and best practice sharing. Their role would require partnership working with families to develop anticipatory care plans to manage their care and needs. In addition, they would work collaboratively with partners from the third sector, health and social care to ensure patients receive coordinated care.
3. **Research Coordinator:** Responsible for monitoring research opportunities and working closely with patient communities to inform and promote the benefits of research, strengthening the position of NHS Scotland to attract high quality research and clinical trials.
4. **Network Administrator:** Required to provide high quality secretarial and administrative service to the service team, including responding to enquiries, directing and prioritising queries, collating data, managing patient files and coordinating and managing team communication.

Further information on the proposed model for the Rare Conditions Coordination Service for Scotland can be found in Annex One.



Value for money

A Rare Conditions Coordination Service for Scotland would require moderate investment, and the CPG does recognise that seeking new investment at a time when there is already significant pressure on health budgets is ambitious. However, the CPG is of the view that investment in such a service will be justified in the medium to long term by improvement in patient outcomes and the reduction of wasteful spend.

For example, the advice, support and information provided by care coordinators for various conditions, both rare and common, have been shown to deliver cost benefits through saving consultant time, preventing hospital admissions and enabling better management of conditions. Evidence, outlined earlier in this report, has shown that where care is coordinated, and managed by a care coordinator, significant cost savings can be achieved.

A Rare Conditions Coordination Service would not, certainly in the first instance, require physical infrastructure to be built. Recent developments in the field of telehealth, including the roll out and success of NHS Near Me, will provide opportunities for the service to work 'virtually' were necessary, reducing overhead costs.

To ensure a Rare Conditions Coordination Service does provide value for money, it would be necessary to ensure that its development, implementation and impact are closely monitored and evaluated. It would be appropriate to undertake a three to five-year pilot study in the first instance, to measure success and provide justification for future long-term funding.

Delivering Scottish Government policy objectives

A Rare Conditions Coordination Service will be responsible for the delivery and experience of care for people living with a range of rare conditions in line with the principles of the Realistic Medicine Policy, namely that care will feature:

- Shared decision making.
- A personalised approach to care.
- The potential for reducing harm and waste.
- The reduction of unwarranted variation.

Importantly, the service will create a vehicle to drive forward the implementation of the UK Rare Disease Framework and the Scottish Rare Disease Action Plan and provide the necessary infrastructure to monitor the progress of implementation and evaluate its success.

And finally, by having a role in promoting and supporting research into rare conditions – and through its role in improving the understanding of Scotland's rare conditions population – the service, over time, will make Scotland a desirable location for research. Through its potential to collaborate with academic institutions, the service could become a driver for innovation and research – meeting the priorities of the Life Sciences Strategy for Scotland.

Cross Party Group members attending Rare Disease Day reception in the Scottish Parliament

Next steps

It is out of the scope of this Cross Party Group to present and recommend a fully-costed and prescriptive model and proposal for a Rare Conditions Coordination Service for Scotland. An expert-led Short Life Working Group would be required to be established to determine the scope of such a service and how it would operate within NHS Scotland.

The Short Life Working Group would also be required to consider models for sustainable funding of this service, as funding for services for rare conditions, and in particular locally delivered but nationally coordinated services, do not sit comfortably within existing local or regional planning structures of the NHS. Options for service funding could include (but are not limited to):

- Investment from Scottish Government, aligned with the new Rare Disease Action Plan.
- An extension of the Specialist Nursing Fund, established by the Scottish Government in 2015, to incorporate this service.
- Research project funding to support a pilot of the RCCSS Project. This would have a benefit of a required, robust evaluation of the service.
- A funding model involving pump-primed funding from a charity or third sector organisation is not considered an appropriate source of funding. The broad range of rare conditions and the typically small nature and limited resources of third sector organisations would result in a disproportionate burden on these organisations and pose a risk to sustainability of a service.

The COordiNated Care Of Rare Diseases (CONCORD) study which began in 2018 will deliver a comprehensive description of the elements of care coordination and how they can be incorporated into models of service delivery. The taxonomy will form the basis of future cost analyses of different models of care. The findings of the CONCORD project should inform the Scottish Rare Disease Action Plan.

Cross Party Group members attending Rare Disease Day reception in the Scottish Parliament

SUMMARY AND RECOMMENDATIONS

The CPG is encouraged by the Scottish Government's demonstration of its continued commitment to improving the lives of people living with a rare condition by supporting the development of the new UK Rare Diseases Framework and is keen to see continuity for people living with a rare condition in Scotland, with the swift implementation of the revised UK Framework in the form of a Scottish Rare Disease Action Plan.

There is an opportunity for the Rare Disease Action Plan to significantly improve patient experience and health outcomes, however we must recognise that to do so, requires a commitment to listening to the needs of people living with rare conditions in Scotland and where necessary, providing sustainable funding for developments that can deliver meaningful change and improvement.

The Cross Party Group has identified two key priorities for improving patient care in rare diseases:

- **Improving awareness of rare conditions amongst health professionals** to reduce delays in obtaining a diagnosis, accessing appropriate care and treatment, to ensure people living with rare conditions are provided with high quality information, support and access to research.
- **Improving care coordination, and where appropriate providing access to a care coordinator** to improve patient experience of healthcare, improve patient outcomes and deliver cost benefits through saving GP and consultant time, preventing unplanned hospital admissions, reducing the length of hospital stays and enabling better management of conditions.

Cross Party Group members attending Rare Disease Day reception in the Scottish Parliament

Although it would require new investment, the CPG believes an 'easy win' for the Scottish Government would be to fund NHS Scotland to develop a Pilot project for a Rare Conditions Coordination Service Scotland. Such a service would:

- Contribute to the delivery of two of the four priorities listed in the UK Rare Diseases Framework.
- Provide an infrastructure to implement the Rare Disease Action Plan, monitor progress towards implementation and evaluate success.
- Provide infrastructure to collect information on rare conditions across Scotland to be used to inform service planning.
- Provide a central point of information and support for health professionals to access so that they can better support their patients.
- Provide education and training materials for NHS Scotland healthcare professionals.
- Provide every patient with a rare condition access to a care coordinator, when this role is not being provided elsewhere.
- Provide people living with rare conditions, and their families, access to comprehensive holistic information and support.

Acknowledgements

The CPG would like to thank the many people living with a rare condition, their families and carers, patient organisations and health professionals who have attended CPG meetings to share their experience and views on how care for rare conditions in Scotland can be improved. The CPG is grateful to those who have given their time to contribute to meetings and to this report and its recommendations.

RECOMMENDATIONS

The Cross Party Group makes the following recommendations to the Minister for Public Health and Sport and the Scottish Government:

1. That Scottish Government must ensure the swift implementation of the UK Rare Disease Framework by committing to publishing a Scottish Rare Disease Action Plan by 31 December 2021.
2. The Scottish Government should establish a Short Life Working Group to explore delivery models for a pilot project for a Rare Conditions Coordination Service in Scotland.
3. The Scottish Government's Rare Disease Action Plan should include a commitment to fund NHS Scotland to deliver a pilot project for Rare Conditions Coordination Service. This would provide people living with a rare condition, whose needs are not being met by an existing service for their condition, access to comprehensive, holistic support and care coordinator; and provide an information and training resource to health professionals within NHS Scotland.

ANNEX ONE

A Rare Conditions Coordination Service for Scotland (outline proposal for a pilot project)

Project overview

The Pilot Rare Conditions Coordination Service for Scotland (RCCSS) will support the key objectives and aims of the UK Rare Disease Framework and Scottish Rare Disease Action Plan by:

- Developing a strategic approach to delivering services to people living with rare conditions in Scotland through the identification of need, improving service delivery and working in collaboration to optimise patient outcomes.
- Providing direct support to families living with a rare condition and information and support the health and social care professionals that support them.

Project outcomes	
Person Centred Care support	People living with a rare condition, whose needs are not being met by an existing service, will have access to comprehensive and holistic assessment and support.
Person Centred Care	Improved coordinated care for individuals living with a rare condition.
Person Centred Care	Those living with a rare condition, their families and carers, will have a better understanding of their rare condition and be confident about coping with the challenges of living with the condition.
Identifying Need	Existing service gaps for many patients living with a rare condition will be identified and addressed.
Access to care, treatment and research	Individuals living with a rare condition will receive information concerning relevant research and clinical trials, treatment and healthcare opportunities.
Raising awareness of Rare Conditions	NHS Scotland healthcare professionals will have an improved understanding of rare conditions and will report improved access to information on rare conditions.
Service Planning and Delivery	The service will inform service planning and strategic policy development, resulting in improved services for individuals living with a rare condition and their family.
Multi-stakeholder Collaboration	A collaborative network of stakeholder, including health and social care professionals and third sector organisations will be formed and will share information, best practice and would collaboratively support people living with rare conditions and their families.

Service delivery

The RCCSS Pilot project, based on the model for the former Single Gene Complex Needs Network, would achieve a balance of nationally coordinated, but regionally delivered, care.

The service would consist of six team members – A National Network Coordinator, three Regional Care Coordinators, a Research Coordinator and a Network Administrator.

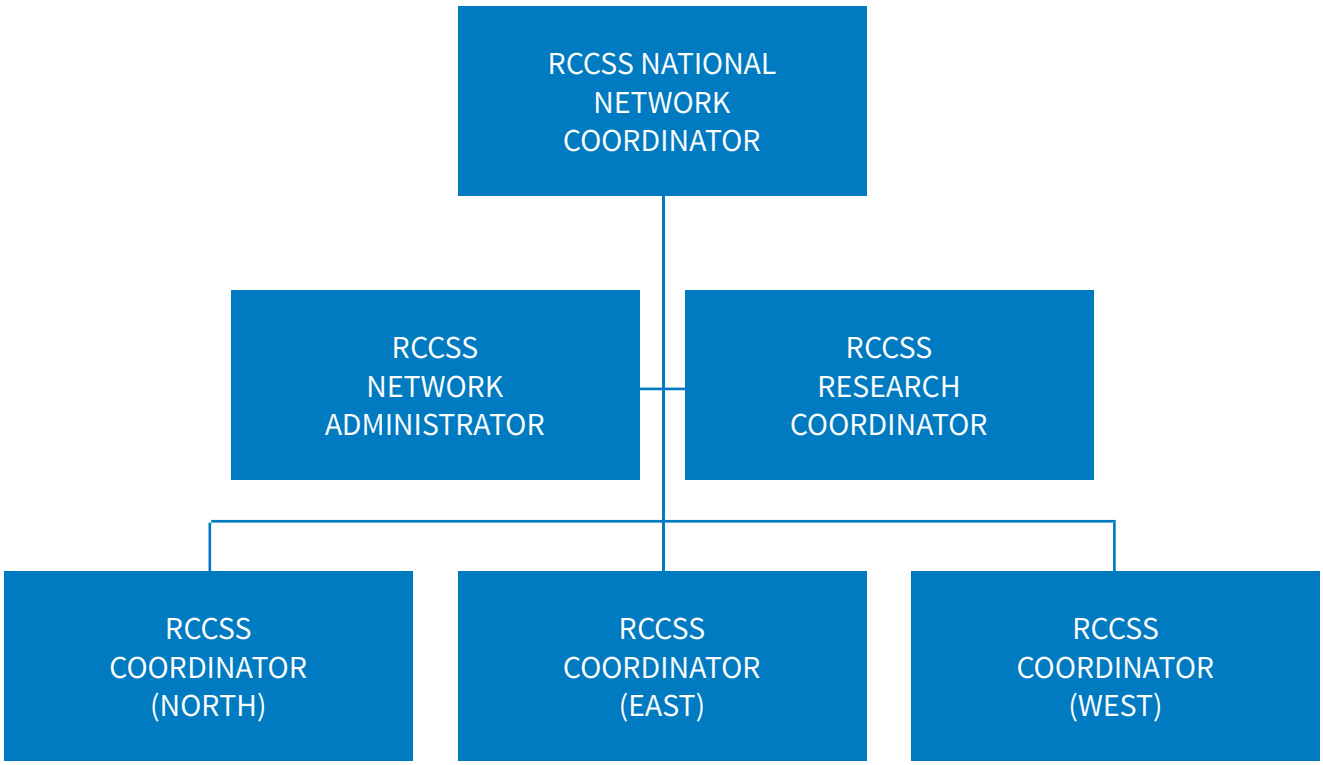
Each RCCSS care coordinator would be based within a health board region, but would be part of a wider RCCSS network to facilitate case discussion and best practice sharing. Their role would require partnership working with families to develop anticipatory care plans to manage their care and needs. In addition, they would work collaboratively with partners from the third sector, health and social care to ensure patients receive coordinated care.

The RCCSS Network Coordinator would play a role in promoting the service within NHS Scotland. The service would become a central point for rare conditions – a recognisable service for health professionals to engage with to obtain training and information on rare conditions.

The RCCSS Network Coordinator would also be responsible for facilitating collaboration across the service, collating and interpreting data to identify patient populations and patient need. They would have a role in shaping future service planning and delivery through participation on relevant planning and strategic committees.

The Administrator role would be required to provide high quality secretarial and administrative service to the service team, including responding to enquiries, directing and prioritising queries, collating data, managing patient files and coordinating and managing team communication.

The service would benefit from a Research Coordinator, responsible for monitoring research opportunities and working closely with patient communities to inform and promote the benefits of research. This position would strengthen the position of NHS Scotland to attract high quality research and clinical trial development and the associated investment that accompanies this.



Job summaries

Network coordinator	Care coordinator	Administrator	Research coordinator
Responsible for the strategic approach to service delivery Promoting the RCCSS Service through NHS Scotland Developing and promoting online training for health professionals Ensure collaboration across network and promote information and best practice sharing Responsible for monitoring and evaluation of the project Collect data and monitor trends across all regions Inform service planning discussions at a regional and national level Budgetary responsibility for project delivery	Manage a caseload of individuals living with a rare condition in their respective region Provide expert clinical care and professional advice to patients, carers and their families Be responsible for advising on clinical and support services available to patients and their families Make referrals to clinical and/or social care services as appropriate Become the principal point of contact for the family Ensure all patients have care plans Build relationships with the families and the clinical teams that support them – ensuring communication between clinical teams if necessary. Contribute to data collection, performing audits to identify trends and support needs Become a point of information for rare conditions for health professionals in their region and contribute to the education of health professionals and students Have a role in supporting research Work collaboratively with other RCCCs across Scotland to produce information, coordinate support and share expertise	Respond to telephone and face-to-face enquiries from all staff groups, patients, carers and families Providing information, directing and prioritising queries as appropriate Collating data and ensuring efficiency and effectiveness of service delivery Responsible for appointing RCCCS Coordinators referrals to appropriate clinics Manage specialist/team patient's files, ensuring information is accurate and up to date Coordinate and manage team communication	Liaise closely with patient communities to establish research priorities Provide people living with rare conditions information on clinical research and support informed decision making Collating information on existing and upcoming research opportunities Develop working partnerships with clinical leaders in the major academic institutes and clinical speciality leads in NHS Scotland Liaise and explore partnership opportunities with industry Contribute to the Scottish Research Nurse and Co-ordinators Network

Skill level

The CPG consider that, in the short term, the roles of national network coordinator and care coordinators and research coordinator, be filled by clinical nurse specialist.

The CPG accepts that the care coordinator roles could be equally well carried out by individuals with relevant experience from other roles within the NHS or the third sector. However, to establish and introduce this service would require the skills of individuals who hold an in-depth knowledge of NHS Scotland, experience of delivering high standards of care, knowledge and understanding of rare diseases and the authority to command collaboration from health and social care professionals.

REFERENCES

1 UK Framework for Rare Diseases – gov.uk/government/publications/uk-rare-diseases-framework/the-uk-rare-diseases-framework

2 European Commission – European Commission. (2020). Rare diseases. [online] Available at: ec.europa.eu/info/research-and-innovation/research-area/health/rare-diseases_en [Accessed 27 Jan. 2020].

3 Scottish Government – gov.scot/policies/illnesses-and-long-term-conditions/rare-diseases/

4 Rare Disease UK, Illuminating the Rare Reality Report (2019) raredisease.org.uk/wp-content/uploads/sites/7/2019/02/Illuminating-the-rare-reality-2019.pdf

5 Rare Disease UK, Rare Alert: Alert Cards for Rare Disease Patients (2020) raredisease.org.uk/wp-content/uploads/sites/7/2020/02/Alert-cards-report-online.pdf

6 Genetic Alliance UK, Rare Experience 2020 (2020) rareexperience2020.geneticalliance.org.uk/wp-content/uploads/2020/12/Rare-Experience-2020-Report-.pdf

7 Rare Disease UK, Illuminating the Rare Reality (2019) raredisease.org.uk/our-work/illuminating-the-rare-reality-2019/

8 Rare Disease UK: Illuminating the Rare Reality (2019) raredisease.org.uk/our-work/illuminating-the-rare-reality-2019/

9 Simpson, A., Bloom, L., Fulop, N.J. et al. How are patients with rare diseases and their carers in the UK impacted by the way care is coordinated? An exploratory qualitative interview study. Orphanet J Rare Dis 16, 76 (2021). doi.org/10.1186/s13023-020-01664-6

10 Walton H, et al. Defining Coordinated Care for People with Rare Conditions: A Scoping Review. International Journal of Integrated Care, 2020; 20(2): 14, 1–16. DOI: doi.org/10.5334/ijic.5464

11 Genetic Alliance UK (2020) Rare Experience 2020 rareexperience2020.geneticalliance.org.uk/wp-content/uploads/2020/12/Rare-Experience-2020-Report-.pdf

12 Simpson, A., Bloom, L., Fulop, N.J. et al. How are patients with rare diseases and their carers in the UK impacted by the way care is coordinated? An exploratory qualitative interview study. Orphanet J Rare Dis 16, 76 (2021). doi.org/10.1186/s13023-020-01664-6

13 Genetic Alliance UK – Rare Experience 2020 (2020) rareexperience2020.geneticalliance.org.uk/wp-content/uploads/2020/12/Rare-Experience-2020-Report-.pdf

14 Genetic Alliance UK – Rare Experience 2020 (2020) rareexperience2020.geneticalliance.org.uk/wp-content/uploads/2020/12/Rare-Experience-2020-Report-.pdf

15 'Understanding the contribution of sickle cell and thalassaemia specialist nurses', Prof Elizabeth Anionwu & Dr Alison Leary, supported by Roald Dahl's Marvellous Children's Charity, July 2012

16 Simpson, A., Bloom, L., Fulop, N.J. et al. How are patients with rare diseases and their carers in the UK impacted by the way care is coordinated? An exploratory qualitative interview study. Orphanet J Rare Dis 16, 76 (2021). doi.org/10.1186/s13023-020-01664-6

17 Genetic Alliance UK, Rare Experience 2020 (2020) rareexperience2020.geneticalliance.org.uk/wp-content/uploads/2020/12/Rare-Experience-2020-Report-.pdf

18 'Protect Parkinson's nurses in England', Parkinson's UK, June 2011

19 'Parkinson's nurses in Scotland: providing effective, safe, person-centred care', Parkinson's UK, April 2012

20 'Clinical nurse specialists: adding value to care – An executive summary', Royal College of Nursing, April 2010


GENETIC
ALLIANCE UK