A Rare Resources Guide for Healthcare Professionals in Scotland

Rare Resources

This Rare Resources guide is for health professionals involved in the care of people with a rare condition in Scotland. It may also be useful for social care professionals, education providers and family support workers.

This guide includes:

- Information to help professionals better understand the needs of people with rare conditions.
- Information on how to assist families to access appropriate care, information and support.
- Links to reliable sources of information and support.
- Links to training resources for health professionals.

GENETIC ALLIANCE UK

This guide has been produced by Genetic Alliance UK as part of the Rare Resources Scotland Project.

Rare Conditions

A disease or condition is defined as rare if it affects fewer than 1 in 2,000 people within the general population.

There are over 7,000 known rare conditions. Although individually rare, rare conditions are collectively common.

1 in 17 people will be affected by a rare condition at some point in their lives. This amounts to approximately 320,000 people in Scotland.

Rare conditions include rare cancers such as childhood cancers and some other well known conditions, such as cystic fibrosis and Huntington's disease.

Rare conditions can be chronic and affect multiple body systems. Consequently, these conditions often require complex care from a range of health professionals. Evidence shows that often basic care standards are missed due to professionals being unprepared to deal with such conditions.

Despite the differences in their particular conditions, people living with rare conditions experience similar challenges.

These often include:

- Delayed diagnosis, misdiagnosis and inappropriate intervention
- Uncertain prognosis
- The majority of rare conditions currently have no effective treatment
- Lack of understanding from professionals involved in their care
- Limited medical literature relating to their rare condition
- Poorly coordinated care with lots of appointments and different healthcare professionals involved
- Difficulties in accessing information and support from an expert, or patient group, for their condition
- Experiencing feelings of isolation
- Significant impact on their emotional wellbeing and mental health

People living with a rare condition recognise and understand that health professionals may not be experts in their condition, however they do expect that those involved in their care will have an understanding of what it means to have a rare condition.

It is important for professionals to recognise the additional needs of people living with rare conditions and their families and support them to access appropriate care and support.

Improving Care for People with Rare Conditions

For people living with, or caring for an adult or child, with a rare condition, managing lots of appointments and interactions with different health and social care professionals can be an emotional, confusing and often stressful experience. Poor experiences of care can lead to distrust and disengagement from NHS Scotland services.

Genetic Alliance UK's Rare Experience 2020 survey asked people with rare conditions to rate confidence and trust in healthcare professionals.

41%	41% reported that they lacked confidence and trust in hospital staff involved in their care
32%	Only 32% had confidence and trust in GP staff
30%	30% had confidence and trust in paramedics and staff in emergency departments
23%	Only 23% had confidence and trust in professionals working in social care

You may be one of many professionals involved in your patient's care and they may have already spent a number of months or years fighting to get a diagnosis or their needs addressed.

People living with rare conditions value having health professionals who work in partnership with them to get the answers that they need.

'I wasn't treated as an individual, but as a nuisance. This has affected my mental health seriously.'
(Rare Experience 2020)

'My GP had never heard of my condition before, no one in the surgery had. He has been very helpful though and has done a lot of research to try and find some information that can help me.'
(Rare Experience 2020)

'My doctor has made no effort to understand my condition, she has never heard of it before and seems to think that because it is so rare, she doesn't have to. I repeat myself at every appointment and have to beg for tests and referrals. I have no confidence in the care I am getting – I wish she'd just say she doesn't know enough and try to find someone who does.'
(Rare Experience 2020)

'Rare and unknown conditions not being taken seriously by medical staff without prior knowledge of them is dangerous and I rarely feel safe with a care provider who is not aware of my condition, or who is making medical or medication decisions that are not appropriate for someone who has said condition.'
(Rare Experience 2020)

'I've had a paramedic google my current diagnosed condition.'
(Rare Experience 2020)

'I have to make all the decisions about my care as my GP knows nothing about my rare diseases and I'm not under the care of any consultants.'
(Rare Experience 2020)

Rare Resources Scotland

Living with a rare condition can be overwhelming. Juggling multiple appointments, processing sometimes difficult and complex information, all while unwell, can leave people with rare conditions feeling isolated and unsupported.

Support organisations play an integral role in providing high quality support and information to people throughout their journey. People with rare conditions need to be made aware of available reliable sources of information that they may want to use to research their condition or access support and information. These sources should include the relevant support organisations where they exist. These organisations often have forums and helplines where people with a rare condition are able to discuss their concerns with others who have experienced similar situations. This can be a simple, effective way to address some of the support needs that people with rare conditions may have.

Healthcare professionals may need help to do this as they may not be aware of support organisations, may not know how to find them, and in some cases there may not be an organisation at all.

Genetic Alliance UK have developed the Rare Resources Scotland project to support healthcare professionals to identify reliable sources of information and support.

Rare Resources Toolkit

Genetic Alliance UK have developed a collection of guides to help support people living with rare conditions in Scotland. The Rare Resources guides contain a wide range of general information on rare conditions as well as information on how to access reliable information, care and support.

Professionals unable to source condition-specific information for their patients, may find it helpful to provide patients with the relevant Rare Resources Guide.

The guides can be downloaded from bit.ly/rareresourcesscotland

Rare Resources for Scotland

Genetic Alliance UK's Rare Resources webpage provides links to a wide range of materials to help healthcare professionals support and signpost people with rare conditions.

Rare Resources Awareness Materials

The Rare Resources Scotland project has developed materials for raising awareness in healthcare settings. These include leaflets, posters, information cards and items to increase visibility of rare conditions, such as name badge clips and lanyards.

Genetic Alliance UK: Professional Engagement Network for Scotland

Genetic Alliance UK coordinates a virtual Professional Engagement Network for health and social care professionals in Scotland who support people living with rare conditions. Members of the Network will receive regular e-newsletters containing information about rare conditions, training opportunities and details of new services and support organisations.

Visit the Rare Resources Scotland webpage to access information, awareness raising materials and join the Professional Engagement Network for Scotland, bit.ly/rareresourcesscotland **or contact** rareresources@geneticalliance.org.uk

Reliable Sources of Information and Support

1 in 17 people in the UK will be affected by a rare condition in their lifetime. Therefore it is very likely that you will work with families with a child with a rare condition throughout your career.

Although it will not be possible to have knowledge of all rare conditions, it is essential that you recognise the additional challenges families face and be aware of the many organisations that provide reliable information and support.

Where to find reliable information:

Genetic Alliance UK is a national alliance of organisations with a membership of over 200 charities that support children, families and individuals affected by genetic conditions. You can search a list of member organisations on the Genetic Alliance UK website.
bit.ly/geneticallianceuk
0300 124 0441

Genetic Alliance UK has a permanent presence in Scotland and has developed the Rare Resources Scotland project designed to improve information and support for people living with rare conditions, and the professionals involved in their care.
rareresources@geneticalliance.org.uk

Rare Disease UK is a national campaign for people with rare diseases and all who support them. Rare Disease UK provides a united voice for the rare disease community by capturing the experiences of patients and families.
raredisease.org.uk
scotland@raredisease.org.uk

SWAN UK supports families affected by a syndrome without a name – a genetic condition so rare it often remains undiagnosed. SWAN UK is the only dedicated support network for these families in the UK and is run by the charity Genetic Alliance UK.
undiagnosed.org.uk

Unique provides support for families affected by rare chromosome disorders or autosomal dominant gene disorders. They provide free guides on specific chromosome and gene disorders.
0188 372 3356
rarechromo.org

Office for Rare Conditions Glasgow works to support people and families who attend hospitals in Glasgow and the West of Scotland, and the healthcare professionals who support them. Funded through Glasgow Children's Hospital Charity, the office provides information and signposting, educational and patient support events and encourages participation in research.
officeforrareconditions.org
info@officeforrareconditions.org

Contact is a charity for families with disabled children. Their website provides a searchable A to Z directory of medical conditions with links to information and support organisations.
bit.ly/rrcontactinformation

Orphanet is an online portal that provides information on individual conditions.
orpha.net

PubMed links to medical literature, including journal articles about a wide range of conditions.
ncbi.nlm.nih.gov

NORD (National Organization for Rare Diseases) is an American rare disease database providing brief introductions to over 1000 rare conditions.
rarediseases.org

National Managed Clinical Networks for Scotland are developed where the full range of specialist care for patients with rare conditions or complex needs are not available locally or regionally. Each National Managed Clinical Network is responsible for designing pathways of care. A full directory of national managed clinical networks can be found on their website.
nss.nhs.scot

Highly Specialist Services
National Services Division commissions highly specialist services for people with rare conditions and/or complex needs. A full directory of commissioned services can be found on their website.
bit.ly/rrNationalServicesDivision

Training Resources for Healthcare Professionals

It is not possible for healthcare professionals to know about every rare condition, however people with rare conditions expect healthcare professionals to:

- Be open to exploring rare conditions as a possible diagnosis
- Acknowledge the limitations in their knowledge and understanding of rare conditions
- Be willing to undertake research and training to better understand rare conditions

There are excellent training and information materials available from a variety of organisations and sources.

Office for Rare Conditions Glasgow

The Office for Rare Conditions hosts education events and webinars. It also offers a six week placement in the care of people with a rare condition to one University of Glasgow student each year. The aim of this placement is to develop understanding of the challenges faced by people with rare conditions and the strategies used to improve care.
officeforrareconditions.org/

Institute of Genetics and Cancer

The ICG Shining a Light Series is a collection of webinars showcasing rare, genetic and undiagnosed conditions.
bit.ly/IGCSpotlightSeries

ScotGen

The Scottish Genetics Education Network (ScotGEN) is a network for all individuals involved in teaching genetics for healthcare in Scotland. The ScotGEN website provides access to a range of learning and teaching resources to assist healthcare professionals to better understand genetics.
scotgen.org.uk

Medics4RareDiseases – Rare Disease 101

Medics4RareDiseases is a registered charity driving an attitude change towards rare diseases amongst medical students and doctors in training. Equipping medical professionals to confidently manage their undiagnosed and diagnosed patients living with rare conditions.
learn.m4rd.org

Rare Disease 101 is an online and interactive education module created for medical professionals by M4RD with the aim of reducing the diagnostic odyssey and improving the rare patient experience. It contains tools, case studies and pragmatic tips for all doctors. While it is aimed at doctors it is relevant to all healthcare professionals.
bit.ly/rrMedics4RD

After completing Rare Disease 101, whilst you won't understand the details of every rare disease, you will be able to:

- Communicate to your colleagues that rare disease is relevant to mainstream medicine
- Recognise when someone might have an undiagnosed rare disease
- Quantify the cost of being undiagnosed and the benefit of having a diagnosis
- Appreciate the impact of having a rare disease on the patient and their family
- List trusted places to go for accurate information about rare disease
- Understand how you can facilitate the patient's journey through healthcare
- Rise to the challenge of coordinating your patient's care
- Support and be supported by patient advocacy groups
- Prepare yourself for the possibilities ahead in terms of diagnostics and treatment

Top Tips

For supporting people with rare conditions

Don't be afraid to say 'I don't know' – It is not possible to know about every rare condition. Patients understand this and value honesty. Be honest about your level of knowledge and demonstrate that you are prepared to learn or ask others for help. This can help build trust and a positive relationship with your patient.

Listen – People with rare conditions often report that they are not listened to or that their concerns are dismissed. People with rare conditions are typically experts in their (or their child's) condition and they are best placed to communicate how the condition affects them/their child and what their needs are.

Signpost to support groups – Wherever possible, assist families to access information and support from condition specific support organisations. Genetic Alliance UK can help you identify support organisations for your patients.

Communicate – Dealing with health and social care professionals can be a daunting experience. Help to build a positive relationship by communicating considerately and clearly; introduce yourself and explain your role in your patients care, avoid using jargon and consider using aids (such as diagrams or online videos) to describe complicated things. People can find appointments overwhelming and stressful, it is important to show empathy and understanding.

Coordinate care – Many patients with a rare condition require complex care from a range of different health professionals. It is in their best interest, and the best interest of those involved in their care, to ensure care is properly coordinated. Wherever possible, coordinate with other care providers and assist patients in obtaining a dedicated care coordinator and do not assume this is a job for someone else to do.

Consider the family – Having a child with a rare condition can significantly affect the lives of other members of the family. Consider the needs of parent carers and siblings in your interactions with the family – ask how they are coping or if there is any support they need. Where appropriate, signpost family members to sources of practical, emotional and financial support.

Living With A Rare Condition

Healthcare professionals must acknowledge the impact living with a rare condition has on a person's life, including their emotional and mental wellbeing.

'82% agree or strongly agree that being asked more frequently about their wellbeing and mental health by their healthcare professionals would improve it.'
(Rare Disease UK, 2018)

It can be helpful to hear directly from people living with rare conditions about their experience.

Rare Experience Reports

Genetic Alliance UK regularly produce experience reports which demonstrate the impact of living with a rare condition.
Recent reports include:

- Rare Experience 2020
- Good Diagnosis: Improving the experiences of diagnosis for people with rare conditions (2022)
- Coordinating Care: Learning from the experiences of people living with rare conditions (2023)

Rare Experience Videos

People living with rare conditions have shared their experiences in a collection of short videos available on Genetic Alliance UK and Rare Disease UK YouTube channels.

You can access rare experience reports and videos from the Rare Resources Scotland webpage.
bit.ly/rareresourcesscotland
or by using the QR code below

Living with a Rare Condition

When I was 24, I was diagnosed with an extremely rare genetic bleeding disorder called Dysfibrinogenemia. The diagnosis came as a surprise as there was no known previous family history of the condition. I had been through high-risk procedures such as having my tonsils removed with no bleeding complications, but if I cut my finger, it could take some amount of time to stop bleeding. Dysfibrinogenemia is an unpredictable condition, and each situation never presents in the same way.

When working as a nurse I became unwell and was admitted to a respiratory ward. Blood work showed abnormalities with my clotting which prompted referral to haematology for investigation.

Lack of recognition of Dysfibrinogenemia by health professionals out with the specialty of haematology, resulted in misdiagnosis and my bleeding disorder remaining undiagnosed for years despite me presenting with symptoms in my adolescent years. Knowledge gaps regarding the condition have resulted in mismanagement of care, including serious near-miss medication errors, delayed surgery and feeling isolated.

My daughter Eleanor also has the condition and I had a high-risk pregnancy due to greater risk of bleeding. Midwives were unsure of how to manage my bleeding disorder during pregnancy and I was apprehensive about having to have a delivery under general anesthetic, as I would not be able to self-advocate for myself. A bleeding disorder pregnancy care plan was created by haematology, this shared document was accessible to all involved in my care, because of this safe and effective care was delivered.

Having no newborn screening offered at birth to test Eleanor meant she was one year old before receiving a diagnosis. Bleeding disorders can be life threatening and can require immediate attention. A late diagnosis risked Eleanor not getting the appropriate treatment she needed.

Dysfibrinogenemia and the challenges it present can be managed effectively, but only if there is recognition and awareness of the condition amongst healthcare professionals.

Rare conditions are underestimated because they are invisible. It is important that healthcare professionals 'Think Rare' and help people with rare conditions access the care that they need.

Rare Resources: Supporting People with Rare Conditions in Scotland

1 in 17 people in the UK will be affected by a rare condition in their lifetime.

It is very likely that you will work with families with a child with a genetic, rare or undiagnosed condition throughout your career.

Although it will not be possible to have knowledge of all genetic, rare and undiagnosed conditions, it is essential that you recognise the additional challenges families face and be aware of the many organisations that provide reliable information and support.

Rare Resources Scotland is a project run by Genetic Alliance UK, compiling useful resources for people living with rare conditions and the health and social care professionals that support them.

Visit Rare Resources Scotland to access resources for healthcare professionals.

bit.ly/rareresourcesscotland

Access the Rare Resources Toolkit and materials for healthcare professionals.

THINK RARE

For further information on the Rare Resources Scotland project contact rareresources@geneticalliance.org.uk

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