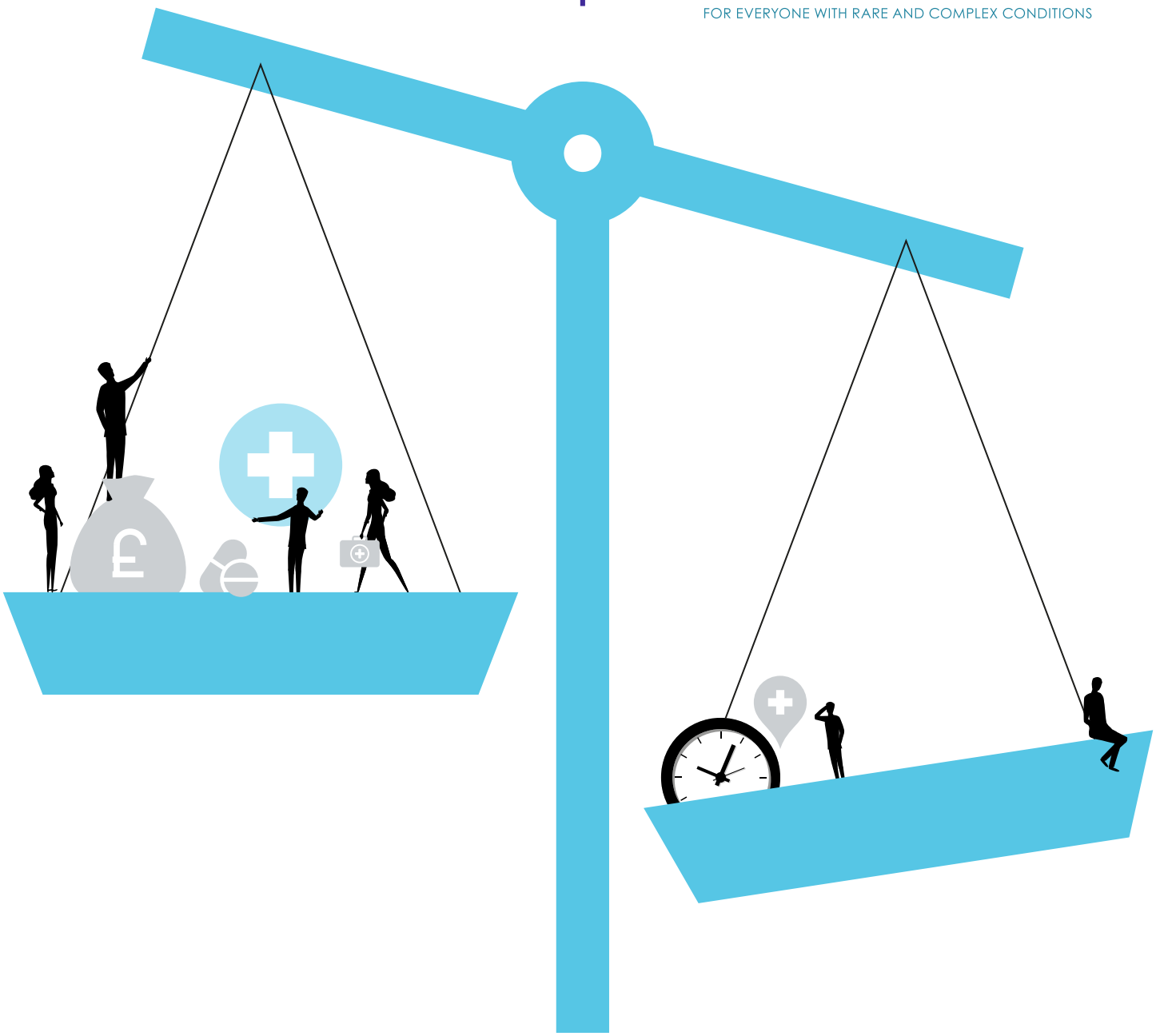
Specialised Healthcare Alliance
FOR EVERYONE WITH RARE AND COMPLEX CONDITIONS



Rare diseases, common inequalities: Bringing rare diseases into the health inequalities agenda

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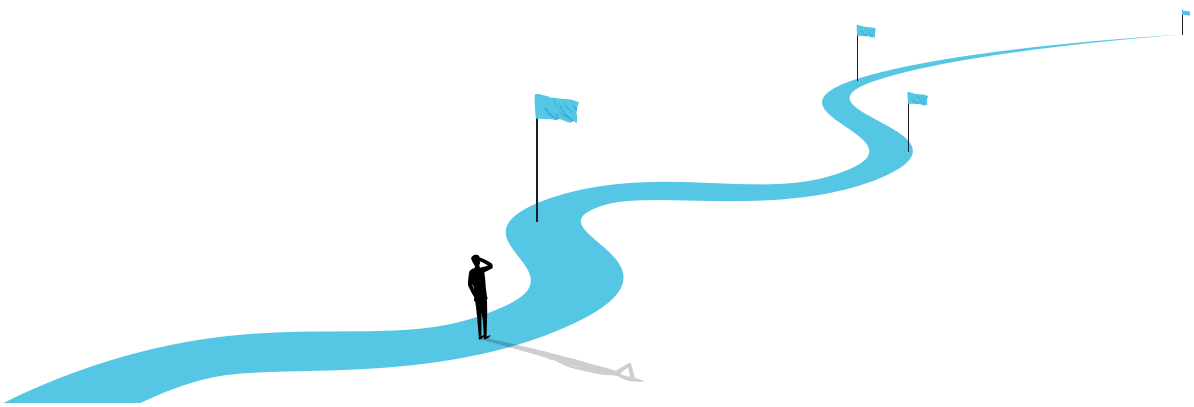


1. Recommendations

These recommendations aim to improve access to services for people with rare diseases, reducing inequalities related to physical, mental, and social wellbeing.

Taking these recommendations forward and bringing rare diseases into the health inequalities agenda would help to improve the lives of those living with rare diseases today, as well as to ensure further progress for the rare disease patients of the future.

Access to care

Lack of knowledge, understanding and awareness among HCPs

1 NHS England should provide and promote resources similar to Medics4RareDiseases' (M4RDs) 'Rare Disease 101' to ensure that healthcare professionals have a general awareness of rare diseases, including the common challenges patients face in accessing care. This information should be easily accessible and readily available for all healthcare professionals, not just those involved in the care of people with rare diseases.

Poorly coordinated care

2 The UK Government should consider the importance of a named worker/care coordinator for people with rare diseases and commit to funding to enable the implementation of potential models of care coordination from the CONCORD study.

Scarcity of specialised centres/services across the UK

3 The NHS should better support patients to access specialist centres/services regardless of where they live. Work should be done to include virtual consultations in new service specifications, to be used where appropriate for those facing challenges in reaching specialist centres, and the implementation of virtual consultations should be assessed and reported regularly.

Access to medicines

Lack of funding for rare disease research

4 The UK Government, and the devolved nations, should take steps to accelerate funding and prioritisation of rare diseases in research to ensure that there are more opportunities for rare disease treatments to be developed.

Accessing clinical trials

5 The UK Government and the devolved nations should make an assessment of the barriers to clinical trial participation for people with rare diseases, and report on progress of current related actions within their plans to assess if they are improving access.

Delays in medicines approval

6 The National Institute for Health and Care Excellence (NICE) should consider inequalities in access to medicines, reviewing whether their processes are fit for purpose for rare disease medicines. This should include working with the rare disease patient community to understand and develop further opportunities to increase participation in medicine approvals processes, as well as undertaking deliberative engagement to review societal attitudes to rare disease treatments, updating past Citizen's Council conclusions.

Access to support for wider issues

Mental health

7 The Department of Health and Social Care (DHSC) and the devolved nations should ensure that tailored mental health support is built into service planning for people with rare diseases to meet their unique needs and reduce inequalities related to accessing quality mental health support.

Financial impact of a rare disease

8 The Department of Health and Social Care and the devolved nations should acknowledge the financial impact of rare diseases, worsened by inequalities in access to social care support, and work with other departments to address these issues, including setting out commitments in this area as part of future Action Plans.

2. Introduction

Everyone deserves equal opportunity to live as healthily as possible, regardless of their condition. This is not currently the case for people with rare diseases, as they encounter a range of obstacles to accessing care and support that could help to improve their health outcomes. This report, produced by the Specialised Healthcare Alliance (SHCA), explores the set of challenges unique to people with rare diseases, in comparison with more common conditions, and illustrates how they unequally impact on the health of the rare disease community.

Through a review of the literature on health inequalities and rare diseases, and a series of one-on-one interviews with SHCA charity members, grounding this work in the experiences of people with rare diseases, we have identified three key areas in which people with rare diseases face challenges that drive health inequalities:

This report seeks to shine a spotlight on the health inequalities experienced by people with rare diseases, across the community. Our research shows that, compared to people with more common diseases, it is much harder for people with rare diseases to access the treatment, high-quality care, and extra support they need. In turn, this drives poorer and ultimately avoidable differences in health outcomes and experiences of healthcare. We consider the implications of these findings for relevant rare disease and health inequalities policies, highlighting opportunities and making recommendations for a range of policymakers, with the aim of supporting equitable access to services for people with rare diseases.

It is important to recognise the heterogeneity of rare diseases and the implications that this has for the experiences of individuals – the exact nature of the inequalities they face will be shaped by their circumstances. However, despite the vast differences between individual rare diseases, there are similarities across the rare disease community in terms of the set of challenges faced when navigating health and care systems. This report therefore focuses on the shared experiences across the rare disease community, rather than exploring individual diseases.

The scope of this report is not wide enough to fully interrogate the inequalities within the rare disease community – such as racial, gender or socioeconomic disparities – however, the impact of such social inequalities on health, in terms of accessing care, cannot be separated from lived experience. It should be recognised that existing social inequalities are exacerbated further by, inequalities facing people with rare diseases.

3. Background

Rare diseases and health inequalities

Rare diseases are conditions that affect less than **1 in 2000** people.[1]

It is estimated that there are over **7000** rare diseases.[1]

Rare diseases are collectively common, with **1 in 17** people worldwide having a rare disease at one point in their lifetime.[1]

People with rare diseases are therefore a significant group in society, with **3.5 million** people in the UK living with a rare condition, making the common challenges they face all the more important to address.[1]

Health inequalities are the avoidable and systemic differences in health between different groups of people.[2] Discourse on health inequalities often focuses on differences in health between groups based on social inequalities related to gender, race or socioeconomic status – this is helpful to identify and support groups marginalised within society. Similarly, when health inequalities are discussed in relation to rare diseases, they are often referred to in terms of the inequalities for marginalised people that have a rare disease. While these are very important inequalities to address, it can also be argued that regardless of these social inequalities, there are common inequalities that face all people with rare diseases, and they are a marginalised group themselves. As such, this report frames rare diseases as a community that are all impacted by health inequalities, and makes the case that rare diseases should be considered collectively as part of the health inequalities agenda.

There are a wide range of inequalities experienced by people with rare diseases, but the impact on life expectancy is the clearest and most stark illustration of the challenge faced by the community:

More than **75%** of people with rare diseases are children and 30% of these children will die before they reach 5 years of age[1]

Only around **37%** of people with rare diseases have a 'normal' life expectancy[3]

Of course, various common conditions will also lead to poor life expectancies, but as this report details, there are barriers associated with having a rare disease that limit interactions with health and care systems, thus unequally limiting opportunities to improve health outcomes. Therefore, it is important that policymakers acknowledge rare diseases as a priority area within health inequalities policy to improve the health of a significant, yet marginalised group.

Policy environment

In January 2021, the Department of Health and Social Care (DHSC), in collaboration with the devolved nations, published the UK Rare Diseases Framework which sets out governmental priorities to address challenges facing people with rare diseases. The four overarching priorities include:

1. Helping patients get a diagnosis faster
2. Increasing awareness of rare diseases among healthcare professionals
3. Better coordination of care
4. Improving access to specialist care, treatment and drugs

The UK Government and the devolved nations make a commitment to reduce health inequalities as part of phase two of the framework; we believe that addressing the challenges set out in this report will help to reduce health inequalities across all four priorities by improving access to medicines, care and wider support.

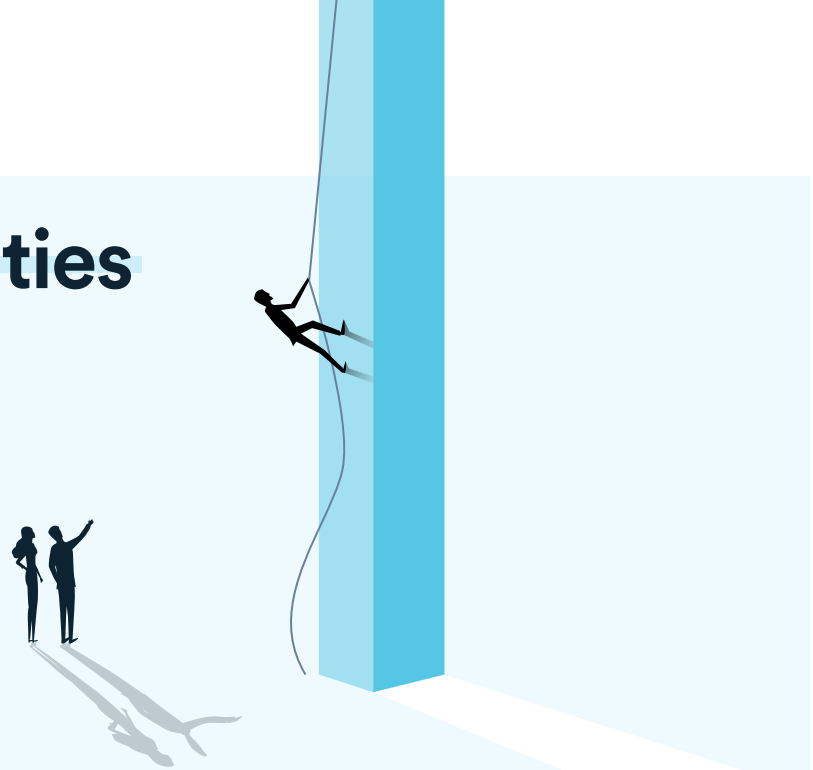
In the two years following the publication of the Framework, all four UK nations have published their own Action Plans, setting out individual approaches to delivering on its priorities. The England Rare Disease Action Plan 2023[4] includes tackling health inequalities for people with rare diseases as an underpinning theme, and we hope that England and all of the devolved nations will take the inequalities discussed as part of this report into consideration for future Action Plans.

One specific aim within the England Rare Disease Action Plan that cuts across health inequalities policy is the action to include rare diseases within **NHS England's (NHSE)** Core20PLUS5 approach to reducing health inequalities, in some capacity.[1] Core20PLUS5 informs action on reducing health inequalities for target populations – 'Core20' refers to the most deprived 20%, 'PLUS' refers to various marginalised groups, and '5' refers to five clinical areas of focus.[5] Action 28 of England's 2023 Action Plan commits to gathering evidence to include rare diseases as an example of a 'PLUS' population. We support this action and the framing of rare diseases as a marginalised group that experience health inequalities in comparison to other groups. We hope that the findings of this report can be used as evidence towards this action, ultimately supporting greater inclusion of rare diseases within health inequalities policy.

The National Institute of Health and Care Excellence (NICE) – the regulatory body responsible for health technology assessments (HTA's) in England and Wales – has the aim of reducing health inequalities as one of their core principles.[6] As part of NICE's recent review of methods and processes,[7] they acknowledged the importance of reducing health inequalities, agreeing that there was a strong case for a modifier that would allow committees to give greater importance to medicines that would help to reduce health inequalities – however, NICE has yet to make formal progress towards the introduction of a modifier.

We welcome the increased focus among policymakers on health inequalities in recent years, but inequalities for rare diseases are not well recognised within policy and continue to persist. It is important that the gap between rare diseases and health inequalities within policy is closed and action is taken to bring more equal opportunity for people with rare diseases to access health and care services.

4. Inequalities



Access to care

We have heard from our members that a number of challenges exist for people with rare diseases in accessing care. Given the complexity of their conditions, people with rare diseases require multidisciplinary, specialist care to meet their complex health needs and manage their care effectively. A lack of access to joined-up, coordinated, high quality specialist services drives avoidable differences in healthcare experiences and outcomes.

One major barrier to accessing care is the delays to diagnosis experienced by people with rare diseases. This is a clear inequity for people with rare diseases that must be addressed, and the SHCA have recently published a report with comprehensive recommendations to improve rare disease diagnosis,[8] this is a further inequality that must be acknowledged and addressed.

Lack of knowledge, understanding and awareness among HCPs

While it is not realistic for every healthcare professional to have an in-depth knowledge of each rare disease, it is important that people with a rare disease are seen by a clinician that understands their condition, or has a general awareness of rare diseases, so that they can support access to high quality care and advice. However, the lack of knowledge, understanding and awareness of rare diseases among healthcare professionals is consistently reported as a significant challenge for rare disease patients.[9,10,11] In the National Conversation on Rare Diseases Survey – the survey that informed the UK Rare Diseases Framework – awareness of rare diseases among healthcare professionals was highlighted as the second-biggest challenge for people living with a rare disease.[12]

This is a problem particularly applicable to healthcare professionals outside of specialist care, and particularly relevant when accessing primary care. We heard from one member that for one specific rare neuromuscular condition, it isn't uncommon for people to visit the GP ten times before they are referred for specialist diagnosis. One survey found that 56% of people with rare neurological conditions did not believe their GP or practice understands their rare disease.[13] We heard from our charity members that this can result in delays in treatment, misinformation leading to incorrect management of the disease, misdiagnosis and missed referrals for specialist services – all of which impact the health of the individual. It is important to acknowledge the fact that GPs are under extreme pressure, and cannot be expected to have an in-depth knowledge of all rare diseases, but the impact of a lack of awareness is apparent and must be addressed.

CASE STUDY: Impact of a lack of knowledge of rare diseases

We heard from one charity member about the experiences of people accessing non-specialist care with a rare inherited blood disorder.

This rare disease has a higher population size and is generally more well-known than other rare diseases, and even has NICE guidelines. However, the experience of people when accessing non-specialist care varies greatly and has been described as 'problematic'. In comparison, the specialised services they have access to are described as being 'very good'.

When admitted to general wards, or A&E, patients feel that a lack of knowledge or understanding of their condition is a barrier to good quality care, and a lack of awareness of the care needed for rare diseases prevents correct referrals being made – leading to acute health crises.[14] In some cases, a lack of awareness and understanding of this rare condition has even led to avoidable and untimely deaths.

Undoubtedly, the lack of knowledge and understanding is linked to a shortfall in rare disease research, resulting in a limited pool of knowledge from which to pull from and a lack of general awareness of rare diseases. However, even for the more well-known and better researched rare diseases, people have experienced a lack of understanding among non-specialist healthcare professionals that has impacted their health acutely.

One example of an initiative to increase awareness and understanding of rare diseases among healthcare professionals is Medics4RareDiseases (M4RD), a charity that provides 'Rare Disease 101' training, among other resources, for medical professionals with little prior knowledge in rare diseases.[15] 'Rare Disease 101' provides basic information on rare diseases that can help professionals understand the common challenges faced across rare diseases, meaning they can provide more appropriate care.

RECOMMENDATION

NHS England should provide and promote resources similar to M4RDs' 'Rare Disease 101' to ensure that healthcare professionals have a general awareness of rare diseases, including the common challenges patients face in accessing care. This information should be easily accessible and readily available for all healthcare professionals, not just those involved in the care of people with rare diseases.

Poorly coordinated care

As people with rare diseases often see a wide range of healthcare professionals, both specialist and non-specialist from across different settings, it is important to have well-coordinated care. Coordination of care involves all functions of a person's care working together and communicating effectively towards achieving shared outcomes for the patient.[16]

The CONCORD study – a major review of the coordination of care of people with rare diseases funded by the National Institute for Health Research (NIHR) – found that, for the majority of people with rare diseases, their care is not well coordinated and this has a negative impact on all aspects of their lives, including their physical, mental and financial wellbeing.[17] A survey conducted as part of the CONCORD study found that only 12% of respondents have a care coordinator, and only 10% have a care plan in place.

The findings of the CONCORD study were reflected in our conversations with charity members, reporting little to no care coordination for a range of rare diseases. This can lead to delays in receiving treatment or support, with clinicians not being kept up to date with the other aspects of a patient's care. Without coordination of care, the burden falls on the patient, further compounding the stress of having a rare disease.

> ***"I really do think having a rare condition is like having a part-time job, if not a full-time job. I know of one of our enquirers who actually has a separate mobile phone that is listed for all of her NHS appointments, and she keeps it separate from her personal life. That just shows how much of a job managing something that is rare or complex can be for some people."***
>
> – Quote from an SHCA charity member

As well as detailing the negative impact of poor coordination, the CONCORD study developed taxonomies of care coordination that can be used to inform models of care: these were in various domains including ways of organising care, ways of organising teams, responsibilities, and how often care appointments and coordination take place.[17] Well coordinated care provides patients with efficient, high-quality support across multiple settings and services, something that is essential for the complex health needs of rare diseases. This can be seen in practice below:

CASE STUDY: The positive impact of care coordination for rare diseases

The East of Scotland Clinical Genetic Service provides care coordination for patients with rare conditions on a long-term basis through the work of two specialist nurses.

The clinical nurse specialists act as named workers across a wide range of rare diseases, remaining in regular contact and filling gaps in care coordination for patients, including through: providing one-to-one patient support throughout care; developing care plans; managing appointments and referrals; and sharing information between clinicians involved in care.

Their work delivers positive outcomes for people with rare diseases, making day-to-day life easier and care more efficient for patients, and ensuring that all appointments, tests and referrals are carried out.

The specialist nurses acknowledge that, although they are not experts in every rare disease they are involved in, they understand the challenge of managing care across multiple specialties, especially for rare diseases, and support patients to control their own care. This model highlights how care coordination can be provided across multiple rare diseases without requiring extensive resources.[16]

RECOMMENDATION

The UK Government should consider the importance of a named worker/care coordinator for people with rare diseases and commit to funding to enable the implementation of potential models of care coordination from the CONCORD study.

Scarcity of specialised centres/services across the UK

Given the small population size of each rare disease, and the fact that there is limited research, specialist knowledge for individual rare diseases is often limited and not spread equally across the country. We heard from our charity members that specialist knowledge for rare conditions is often centralised within a limited number of locations across the UK. In some cases, specialist knowledge can often be held by only a handful of clinicians nationwide. This concentration of expertise creates inequality in access to specialist services and, when care can be accessed, quality can differ.

One member told us that the only specialist centre in the UK for their rare disease is found in London, and that there are only around five clinicians with an in-depth knowledge of the condition. This means that for anyone living outside of London, reaching the specialised centre with the highest standard of care for this particular condition is extremely challenging, especially with the physical implications of their condition. This is common across rare diseases, with patients not being able to access a specialist centre because of distance. The CONCORD survey revealed that only 32% of respondents were accessing a specialist centre, and 60% responded that the main reason they would choose to not use a specialist centre was the travelling involved.[17] If patients are not using specialist centres then they are not receiving the highest quality of care available, meaning their health is likely impacted by the scarcity of specialised centres, worsening health outcomes.

> ***"I'm lucky that my centre of excellence is close by to me. I wouldn't want to visit my local district hospital for monitoring [of my condition] because I don't think that the understanding is the same. When you go to the A&E department of the same hospital it is hours of looking at a screen reading up on your records and your condition before you can be seen properly."***
>
> – Quote from an SHCA charity member who also lives with a rare disease

The impact of scarcity of specialised centres (with patients living far from high-quality specialist care) is acknowledged within the UK Rare Diseases Framework, with the potential solution of virtual consultations included in some of the associated Action Plans.[18] While virtual consultations do provide access to specialist centres/services for patients who would otherwise have to travel, they are not always appropriate and are not a replacement for local specialised services.

RECOMMENDATION

The NHS should better support patients to access specialist centres/services regardless of where they live. Work should be done to include virtual consultations in new service specifications to be used where appropriate for those facing challenges in reaching specialist centres, and the implementation of virtual consultations should be assessed and reported regularly.

Access to medicines

Access to medicines for people with rare diseases is often challenging for a number of reasons, not least the fact that only around 5% of rare diseases have a treatment approved for use.[19] The limited range of medicines available for rare diseases is the result of a range of factors, in particular a lack of rare disease research, difficulties accessing clinical trials, and delays in approval.

Lack of funding for rare disease research

A strong research base is essential for generating a good understanding of a health condition and is an integral part of developing treatment options for any disease area. As such, research efforts are particularly important for rare diseases given the pressing need for more treatment options. However, research is limited across rare diseases and this is often cited as a significant barrier in terms of how it limits knowledge and understanding; this then drives inequalities in access to medicines for people with rare diseases.[20]

One charity member told us that there was less appetite for academic researchers to go into rare diseases in comparison to common conditions, in part because the lack of previous research makes new research more difficult, as well as a lack of funding for research in this space. Historically, an inequality in research between rare diseases and more common conditions has been reflected in the amount of funding allocated; one analysis from 2013 found that of the top four funders of clinical research in the UK, three allocated less than 1% of their total funding to rare diseases.[21] Recent research by the NIHR, in collaboration with the Medical Research Council (MRC), has mapped the rare disease research landscape, finding that there are a small number of rare conditions for which there is a large amount of research, and a large number of rare diseases with no research.[22]

> ***"Researchers go where the funding is and where there is already existing research – getting new research generated is a big challenge for rare diseases."***
>
> – Quote from an SHCA charity member

With a lack of accessible funding, there is less opportunity for clinical academics to research rare diseases and treatments, disproportionately impacting progress made for new treatment options. Similarly for the life sciences industry, the costs of research and development, paired with issues in pricing and access, make rare diseases research and pharmaceutical development a challenging and often unviable option,[12] further limiting access to medicines for people with rare diseases.

RECOMMENDATION

The UK Government and the devolved nations should take steps to accelerate funding and prioritisation of rare diseases in research to ensure that there are more opportunities for rare disease treatments to be developed.

Accessing clinical trials

Conducting a successful clinical trial is an essential step towards pharmaceutical development, and participating in clinical trials is an important way for people with rare diseases to access innovative treatments, as there are often no other options available. However, rare disease clinical trials are challenging to run; small patient population sizes mean that recruitment is difficult and they are often logistically complex, often spanning multiple countries.[9]

For rare disease patients, accessing traditional clinical trials is equally as challenging because they often require that participants travel to a study site, meaning significant time and cost for individuals who might also find it physically taxing to travel.[23] These challenges can potentially discourage participation, limiting the number of people who access rare disease clinical trials, on top of an already limited patient population.

One specific issue also identified in our discussions with charity members in terms of accessing available clinical trials was that patients were generally unaware of clinical trials that were taking place despite their interest in participation. According to Rare Disease UK, 80% of rare disease patients said they would be interested in research participation, but only 25% felt that they were given enough information about ongoing research.[10]

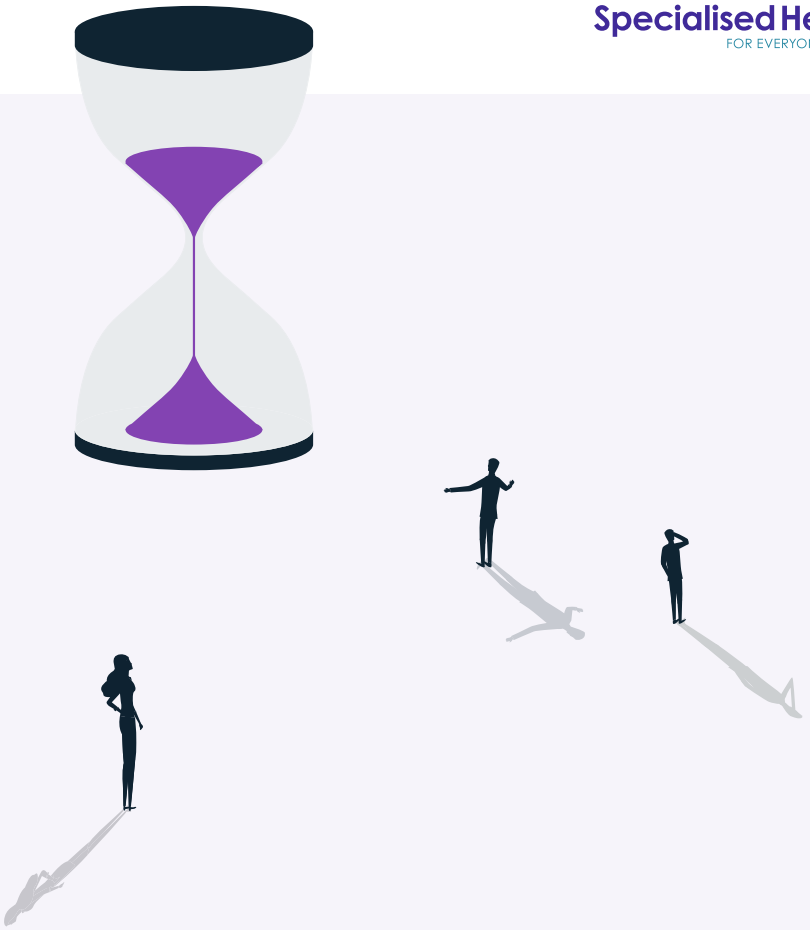
> ***"Less than a fifth of people with [rare disease] have ever been offered a clinical trial, many of these might've not even been pharmaceutical trials but may have been trials to gather data."***
>
> – Quote from an SHCA charity member

One charity member noted that if patients do not have a proactive clinician who is knowledgeable about rare diseases and the clinical trial environment, then they are immediately disadvantaged. They also said that they were increasingly finding that if patients are not connected to a specialist service, then they do not have a good connection to clinical trials and are much less likely to understand their benefits or access them.

Within the UK Rare Diseases Framework, and to a varying degree within the different devolved nations' Action Plans, there is an acknowledgement that work is required to improve access to clinical trials, but challenges remain to real-world access.

RECOMMENDATION

The UK Government and the devolved nations should make an assessment of the barriers to clinical trial participation for people with rare diseases, and report on progress of current related actions within their plans to assess if they are improving access.



Delays in medicines approval

Even when treatments are developed, access can be delayed. For rare conditions that do have a potential treatment in development, or treatments that have received regulatory approval but are not yet approved for use in a clinical setting, it can be months or years before this treatment becomes available to patients, due to challenges in approval processes.

According to Rare Disease UK, in 2019 only half of licensed rare disease treatments were available on the NHS which has been attributed to the UK's approval processes being slower and stricter than other international HTA systems.[10] Furthermore, a study analysing the impact of rarity on NICE HTAs found that appraisals of orphan medicines in comparison to non-orphan medicines were significantly longer and less likely to be recommended for use: the average time for rare medicines in the approvals process was 370 days compared with 277 days for non-rare medicines.[24] The authors of this analysis suggest that the absence of an appropriate mechanism to factor inequalities between rare and non-rare medicines into evaluations of medicines can impact decision making.[24]

This point was echoed by one of our charity members who felt that there is a question around whether QALYs – a measurement of the health benefits of treatments being assessed – are fit for rare diseases, and that medicines for rare diseases should be reviewed differently to ensure equity in decision making. Recent research from the UK BioIndustry Association (BIA) also suggests that NICE's processes for evaluating rare disease medicines are not reflective of the social value of rare disease treatments, limiting equitable access to medicines for patients.[25]

Inequalities in medicines approvals for rare diseases directly impacts access to medicines for people with rare diseases. In a survey of more than 1,000 people with a rare disease and their carers, 66% of respondents agreed that 'the system was too slow to make decisions' on rare disease medicines, and 64% agreed that it was 'unfair on people living with rare conditions'.[11]

CASE STUDY: International example of early access to rare disease medicines

In 2021, the French Government introduced new mechanisms that allow early access to medicinal products and off-label use of authorised medicinal products, with the goal of improving access to treatment for conditions with high unmet needs, including rare diseases.

One of the new mechanisms, 'autorisation d'accès précoce' or 'AAP',[26] grants early access authorisation for innovative medicinal products if: the drug is for patients who are affected by a serious, rare, or disabling disease; no other appropriate treatment exists; and, there is a strong presumption that the product is effective and safe based on trial data. This mechanism means that rare disease patients can access vital treatments quickly, with assessment processes then determining the terms on which access is provided in the long-term.

Delays in access to rare disease medicines while waiting for approval can impact on the health of patients and, given the fact that many rare diseases do not have alternative treatment options, this delay is even more damaging. In 2021, it was announced that an Innovative Medicines Fund (IMF) would be set up as a new mechanism to speed up access to innovative non-cancer medicines,[27] which was welcomed across industry, patient, and professional organisations.

The prospect of the IMF is particularly important for rare disease medicines because it would allow patients to access medicines before they are approved for hospital-use whilst more evidence is gathered for full approval. However, since it was announced, no medicines have entered the IMF. It remains unclear why the IMF is not being fully utilised, however the absence of entries means that it is not delivering as anticipated. In comparison, the Cancer Drugs Fund (CDF) – a similar mechanism allowing early access to innovative cancer medicines – has enabled access to medicines for tens of thousands of patients.[28]

Improving access to available medicines is becoming increasingly important, as the pipeline for upcoming innovative personalised therapies with the potential to transform care for some rare diseases looks promising in the near future.[29]

RECOMMENDATION

The National Institute for Health and Care Excellence (NICE) should consider inequalities in access to medicines, reviewing whether their processes are fit for purpose for rare disease medicines. This should include working with the rare disease patient community to understand and develop further opportunities to increase participation in medicine approvals processes, as well as undertaking deliberative engagement to review societal attitudes to rare disease treatments, updating past Citizen's Council conclusions.

Access to support for wider issues

Accessing treatment and care is a significant part of living with a rare disease but by no means the whole picture. The impact of having a rare disease stretches far beyond the implications for people's physical health, whether it be social, emotional, financial or educational. This has not necessarily been reflected in the support they receive, further exacerbating inequalities.

In the National Conversation on Rare Diseases Survey, respondents identified the availability and provision of wider support for rare diseases as a concern for people with rare diseases, as well as for their family and carers.[12] It was apparent in our discussions with SHCA members that they shared the same concerns, particularly around challenges in mental health support and the financial burden of rare diseases, compounded by the cost of living crisis.

Mental health

Mental health is influenced by a range of factors, not least the experience of illness – this is further compounded by the unique experience of managing a rare disease.[30] One survey of around 1,800 patients and carers, from across the rare disease community, reported that over 90% of respondents had felt low, depressed, worried, anxious or stressed as a result of a rare disease and the challenges associated with it.[31] The lengthy and exhausting process of seeking a diagnosis, coming to terms with a rare condition, not being able to find information about your condition, and feeling like you are not completely understood or believed, are some of the common experiences of people with rare diseases described by our charity members.

> ***"They've all got problems with their mental health, of course they do, they've all got a rare disease. They all struggle with it, particularly when they feel there is very little hope."***
>
> – Quote from an SHCA charity member

Despite the pronounced impact of rare diseases on mental health – highlighted by every charity member we spoke to – there are inequalities in the provision and quality of support for mental health, compounding poor mental health. In another survey, conducted by one of the SHCA's charity members, over 70% of people living with their particular rare disease reported issues with mental health, with 63% reporting the mental health support provided to be poor or very poor.[13] With a national shortage in formal mental health support,[32] lack of access is not specific to people with rare diseases, but the increased impact on mental health means that access to support is crucial and often unfairly falls on charities to address.

Furthermore, with the unique challenges that rare diseases present, it is important that affected individuals can access tailored mental health support,[33] although these services are extremely scarce, further limiting access. We heard from our charity members that specialist mental health support is valued by people with rare diseases, with some describing experiences with general mental health services as 'unhelpful' to patients' mental health, in part because they had to relive their experiences when describing their rare disease to the professional.

CASE STUDY: Tailored mental health support

The positive impact of tailored mental health support for people with rare diseases is exemplified through the work of Rareminds, a charity that advocates for, and provides access to, specialist mental health support. Rareminds describe a tailored service as one that is aware of the generic issues involved in being diagnosed and living with a rare condition, and also of the specific rare condition. They provide support in the form of counselling, workshops and group programmes.

Following accessing Rareminds services:

- 83% of people felt more resilient after counselling
- 90% felt better equipped to manage their mental health after counselling
- 92% felt less isolated after attending workshops

We also heard from charity members that they feel clinicians are reluctant to ask rare disease patients about their mental wellbeing or offer support, because a) the support doesn't exist and b) clinicians don't feel they can personally address patients' concerns. This can be seen in the results of a patient experience survey comparing rare and non-rare neurological conditions, finding that only 33% of people with rare conditions felt that healthcare professionals had asked them about their mental wellbeing, compared with 44% of people with non-rare conditions.[34]

There is a need for increased and tailored mental health support for people with rare diseases given the unique challenges they are faced with. It is also important that all healthcare professionals involved in the pathway of care for people with rare diseases are aware of the impact of rare diseases on mental health so that they can identify problems and offer support where appropriate.

Mental health is an area of focus highlighted in England's 2022 and 2023 Rare Disease Action Plans. Specifically, in action 21 of the most recent Action Plan, the need for mental health support for people with rare diseases – due to the profound impact that rare diseases have on mental health – was recognised and supported by DHSC.

The Action Plan commits to embedding mental health support within future care coordination actions, which we support as a solution to ensure that care pathways for people with rare diseases are inclusive of physical and mental health needs. However, the need for *tailored* mental health support that meets the specific needs of people with rare diseases must also be acknowledged and actioned.

RECOMMENDATION

The Department of Health and Social Care and the devolved nations should ensure that tailored mental health support is built into service planning for people with rare diseases to meet their unique needs and reduce inequalities related to accessing quality mental health support.

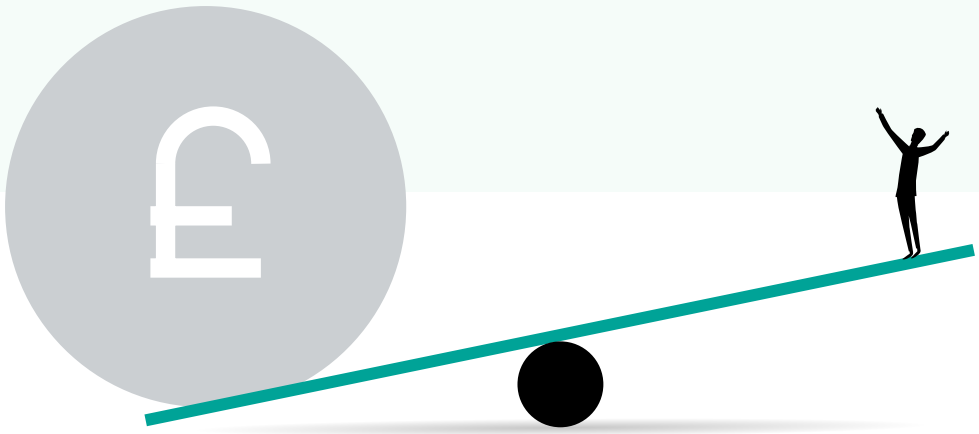
Financial impact of a rare disease

In the UK, access to free healthcare at the point of use is a fundamental principle of the NHS. However, individuals still face many different forms of direct and indirect costs associated with managing both short and long-term health conditions, for example; direct costs of travel and personal medical supplies; and indirectly with loss of employment and education impacting income. While these sometimes apply to both rare and non-rare conditions, we heard from charity members how additional challenges in accessing support for rare diseases can worsen the financial impact.

Balancing work and life with a rare disease can be challenging, leading to reduced working hours or lower quality of employment; for example 76% of respondents in a rare disease survey said that their condition has limited their professional choices.[35] Because of this, among other reasons, people are more likely to experience loss of income and financial hardship with a rare disease.[20]

We heard from members how the increased likelihood of financial hardship facing people with rare diseases and their families means the extra costs associated with accessing care have an amplified impact. Given the low numbers of specialist centres for rare diseases, the likelihood of living further away from specialised services is high for people with rare diseases, and so too are the costs associated with this. A feasibility study carried out by the Genetic Alliance UK in 2016 revealed hidden costs of rare diseases including travel expenses (including petrol, parking, or train fares), temporary accommodation costs and the cost of taking time away from work as some of the financial implications of travelling for specialist care.[36]

There are some mechanisms in place to reimburse patients for travel expenses, but Genetic Alliance UK reported that in many cases these could not be accessed for people with rare diseases,[36] while our charity members said they are sometimes not sufficient enough to cover the additional costs.



CASE STUDY: Cost of living

The recent cost of living crisis has revealed the impact of increased financial burden associated with rare diseases on the health of the individual.

We heard from one charity member how it affects people within their community, who must maintain a specialised diet to manage their symptoms – but due to increasing financial constraints, this has become difficult to sustain. In a cost of living survey, 93.3% of respondents stated that they had been directly impacted by the costs of food and groceries needed to maintain their diet.

This is just one example of how the financial burden of a rare disease can indirectly impact health and drive inequalities.

Further worsening the financial impact of rare diseases is the lack of access in support for people with rare diseases. We heard from a number of our charity members that although people with rare diseases are eligible for benefits to support them financially, they are hard to access. The process for claiming benefits for people with rare diseases is said to be "lengthy", "stressful" and "startling",[37] and some members said that the process of submitting medical evidence is especially difficult with a rare disease – with or without a diagnosis. Due to challenges in accessing benefits for people with rare diseases, one survey found that 35% of respondents did not claim benefits and those that did, didn't receive their full entitlement.[37]

The UK Rare Diseases Framework acknowledges as part of 'wider policy alignment' that rare diseases have an impact beyond the physical and that people living with rare diseases must be supported with the wider impact. However, there are no specific actions or priorities in place within the Framework, or any of the Action Plans, that aim to address challenges related to financial support.

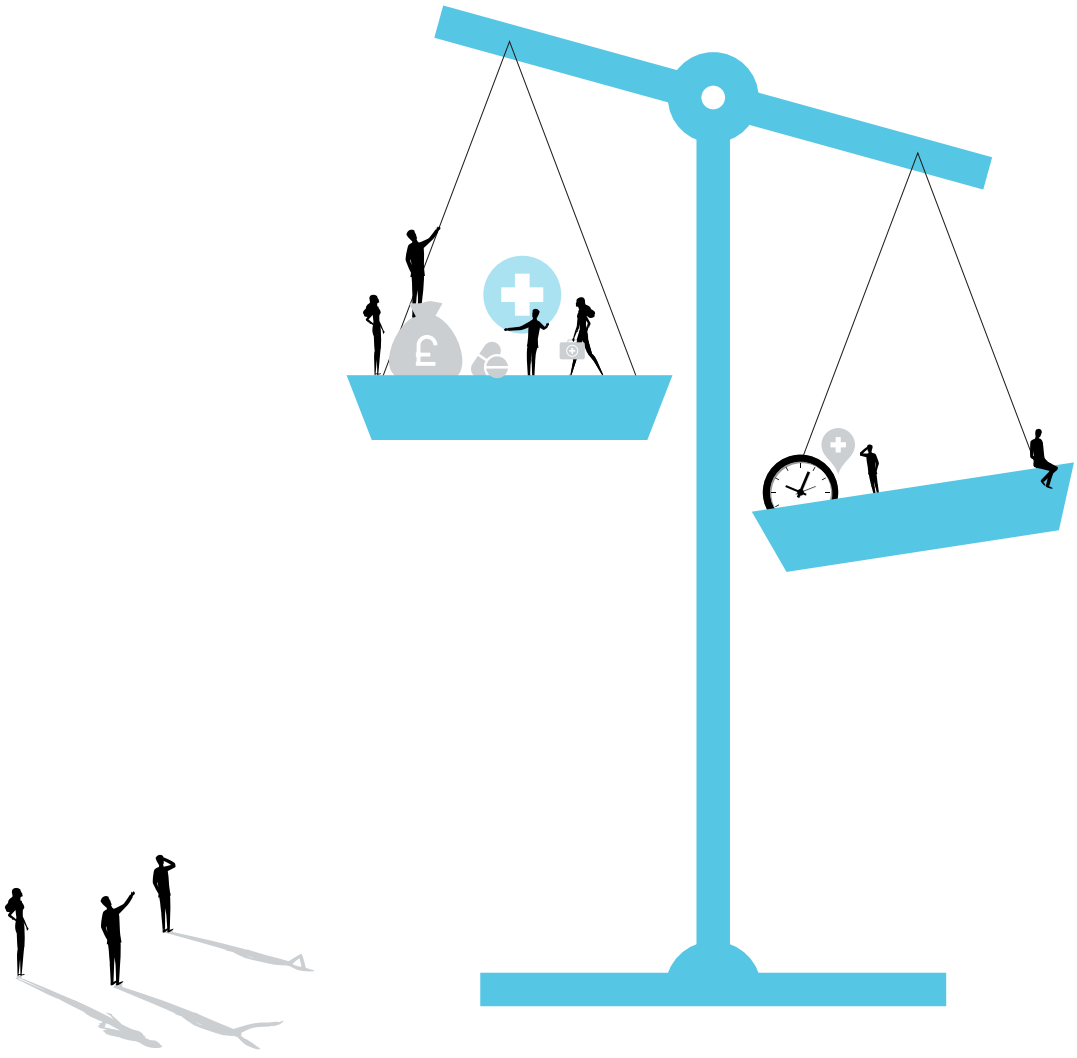
RECOMMENDATION

The Department of Health and Social Care and the devolved nations should acknowledge the financial impact of rare diseases, worsened by inequalities in access to social care support, and work with other departments to address these issues, including setting out commitments in this area as part of future Action Plans.

5. Conclusion

It goes without saying that every person with a rare disease deserves equal opportunity to live as healthy a life as possible. Receiving high-quality care, treatments and wider support while having a rare disease is extremely important to meet the complex needs of the individual. This report has highlighted the inequalities that exist in each of these three areas, resulting in limited access for people with rare diseases, which drives avoidable differences in health outcomes and experiences of healthcare.

We hope the findings and recommendations in this report have highlighted opportunities within health inequalities and rare disease policy to ensure equitable access to care, treatments, and wider support for people with rare diseases. To address challenges in access would be to address avoidable differences in health outcomes and fulfil commitments to reducing health inequalities.



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Specialised Healthcare Alliance
FOR EVERYONE WITH RARE AND COMPLEX CONDITIONS

About the SHCA: The Specialised Healthcare Alliance (SHCA) is a coalition of over 130 patient groups organisations and charities, who support people across the UK living with rare and complex conditions. Our work is funded by 9 corporate supporters. We advocate for better services for patients, keeping rare diseases at the forefront of the national conversation. The SHCA focuses exclusively on overarching policy and does not address individual therapeutic issues. The SHCA has no political affiliation and seeks to work across party lines.

For more information about the work of the SHCA, please visit www.shca.info or email SHCA@incisivehealth.com