Reforming Rare Diseases

ALBIE
LIVING WITH LAL-D

This report was developed and funded by Alexion UK

Job number: M/UK/UNB-U/0066
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About this report

This report was developed to outline the current perspectives and experiences of care from the patient and rare disease community to reflect the impact of the 2013 UK Strategy for Rare Diseases[1] and provide insight and recommendations for the implementation of the anticipated new Rare Disease Framework.

The data presented in this report details findings from a patient experience survey of 1,020 people living with a rare disease or caring for someone with a rare disease, run in summer 2020. This survey was funded by Alexion UK and Genetic Alliance UK.

Alexion UK also convened an Expert Policy Group – including leaders of patient associations and clinicians working within the rare disease space in the UK. The group provided guidance into the development of a survey of clinicians within the rare disease community. This group was comprised of the following members:

- **Dr Jayne Spink**, Chief Executive, Genetic Alliance UK
- **Dr Robin Lachmann**, Consultant, Charles Dent Metabolic Unit, University College London Hospital
- **Professor Jemima Mellerio**, Lead Adult Epidermolysis Bullosa Team, St John's Institute of Dermatology
- **Kerry Leeson-Beevers**, National Development Manager, Alström Syndrome UK
- **Sinclair Dunlop**, Managing Partner, Epidarex Capital

Alexion UK funded the writing of the report and were supported in its drafting by Hanover Communications. The report has been reviewed in line with the ABPI Code of Practice.


JOE
WITH HIS FAMILY
LIVING WITH PNH


CHELSEY
LIVING WITH NMOSD


RACHAEL
WITH HER SISTER
LIVING WITH gMG


MARTINA
LIVING WITH LAL-D

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Foreword

A rare disease is defined by the European Union (EU) as a disease affecting five or fewer people in 10,000 of the general population.[2] Collectively, however, they are not rare. In the UK, one in every 17 people will be affected by a rare disease at some point in their life.[3] They can affect people of all backgrounds at all stages in their life, with half of new cases in children.[1]

Yet we know that the experiences of care for people with rare diseases can differ dramatically from person to person. In spite of the UK Strategy for Rare Diseases, published in 2013, those living with a rare condition still face significant challenges in getting a diagnosis, accessing treatment and receiving coordinated care. They also face challenges in relation to employment, education, social life and mental health. Unfortunately, the patients we represent tell us that, in general, the more complex the condition they have, the more challenges they encounter.

Progress across the sector and with policymakers to determine what should follow on from the 2013 Strategy have been encouraging. I have no doubt the new Rare Disease Framework will be welcomed by the rare disease patient community. Our key focus for the years ahead will be ensuring implementation is prioritised across the UK. The findings in our reports show why it is so important to take opportunities to listen and learn from patients and their experiences.

Jayne Spink

Jayne Spink, PhD, Chief Executive,
Genetic Alliance UK

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 are 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 available for families of children and young adults with undiagnosed genetic conditions in the UK.

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Foreword

At Alexion, people living with rare and devasting diseases are our Guiding Star. When you work with treatments for very small patient populations you are affected by the individual patient experience and the impact the diseases have on them and their families. The proudest moments are when you hear about the difference to people's lives made by the innovation you helped deliver.

Today, more patients are being given the possibility of receiving transformative therapies than ever before. In recent decades across the world there has been a major increase in the levels of research and investment going into understanding and treating rare diseases.

However, seven years on from the 2013 Strategy, we know that the patient experience of care in the UK is still a great cause for concern. From diagnosis to accessing treatment and coordination of care, more needs to be done to improve how people living with rare diseases receive their care. The imminent publication of the new Rare Disease Framework is a welcome first step.

This is a shared ambition for industry and the wider rare disease community and Alexion is pleased to have prepared this report following extensive collaboration with Genetic Alliance UK and contributions of others, including over a thousand people living with a rare disease, the patient organisations to which many belong, our clinical partners and those working to fund the discoveries of tomorrow. Our aim now is to make a contribution towards ensuring that the new Rare Disease Framework is focused on delivering for people affected by a rare disease and improving their experience of care.

Resolving the problems that rare disease patients experience in accessing diagnosis and good, coordinated care have often been parked in the "too difficult" box because there is no one size fits all solution for people affected by rare disease. However, if any lessons have been learned from the COVID-19 pandemic, it is that, when we work together we can rapidly overhaul entire patient pathways and implement technological solutions that we previously thought would take years for the NHS to grapple with. In other words, we learnt with the right mission and commitment, anything is possible. It is time we apply these lessons to help the 3.5 million people in the UK affected by a rare disease,[4] looking across the entire patient journey so that people with a rare disease have access to patient centred care and a robust plan of action is implemented, with every stakeholder assigned its accountability for delivering.

Sean Richardson, Vice President & General Manager, Alexion UK & Ireland

ALEXION®

About Alexion

Alexion is a global biopharmaceutical company focused on serving patients and families affected by rare diseases and devastating conditions through the discovery, development and delivery of innovative medicines, as well as through supportive technologies and healthcare services. We believe it is our responsibility to listen to, understand, and change the lives of patients and those who work tirelessly to help them. People living with rare and devastating diseases are our inspiration and our Guiding Star.

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Executive summary

Rare diseases collectively affect around 3.5 million people in the UK and the impact they have on patients, families and society is profound. Of the estimated 6,000 rare diseases, many are severe, chronic and progressive.[4]

The 2013 UK Strategy for Rare Diseases – which comes to an end in 2020 – has ensured tangible progress for the rare disease community. Yet there are still major changes that need to be made to improve the patient journey and overall standard of care. The principle aim of the 2013 UK Strategy for Rare Diseases "to ensure no one gets left behind because they have a rare disease"[1] was the right intention, but seven years on, the lack of consistent health system prioritisation given to addressing the challenges facing people with a rare disease means this aim can only be considered a work in progress rather than complete.

Alexion UK partnered with Genetic Alliance UK to assess the current experiences of care, now and in recent years, from the patient and rare disease community perspective. The survey of the patient community highlights that in spite of the 2013 Strategy, there are still major unmet needs in the care of patients living with a rare disease, across the entire patient pathway, from diagnosis through to treatment access and ongoing disease management and care.

Overall quality of care: One in two patients believe that the quality of their care has not improved in the past five years[5] and over one third rated their overall experience of care as poor or very poor.[6]

Diagnosis: The 'diagnostic odyssey' remains for people with rare diseases. Over half of patients diagnosed with a rare disease in the past five years waited over two years for their diagnosis.[7] A half of all patients have also received at least one misdiagnosis.[8]

Coordination of care: One in two patients believe their care is not coordinated effectively.[9] The more complex a condition a person has, the less they feel their care is coordinated.[10]

Access: Almost two thirds of patients believe the system for making medicines available to patients is unfair to people living with rare conditions.[11]

A new UK-wide Rare Disease Framework is set to be published by the end of the year and is a welcome step for the rare disease community. The new Framework must be backed up by robust implementation plans and accountability across the NHS and Government. Ensuring robust implementation plans deliver on the ambition of the new Framework will be crucial to ensuring that people with a rare disease have faster diagnosis, improved care, including better coordinated care, and timely access to new innovative therapies.

The findings in this report highlight why it is so important to listen to and learn from patients and their experiences. These perspectives have been translated into a series of key recommendations aimed at helping to improve the quality of life for people in the UK affected by rare diseases.

> ***More involvement from a GP or specialist would be immensely helpful.***[12] *Patient workshop*

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Recommendations

The below recommendations from Alexion UK and findings from the report must be considered in the implementation of the new Rare Disease Framework. These recommendations, if implemented, would ensure that the new Framework aligns with the strategic direction, achievements and possible shortfalls of the 2013 Strategy.

Rare disease focused recommendations

- The Government and NHS should ensure that the implementation plans for the new Rare Disease Framework should be based on an evaluation as to why previous initiatives have failed to improve care in the last five years for all patients living with a rare disease.
- The NHS must ensure it has the infrastructure in place to ensure that all patients suspected of having a rare disease are identified and able to access all the necessary diagnostic capabilities that the Genomic Medicine Service can offer.
- The Government and the NHS should focus on improving whole person care for patients with more complex conditions.
- The NHS should provide every patient with a rare disease with (i) a dedicated care coordinator, (ii) access to a specialist centre if available and (iii) a care plan if desired by the patient.
- The NHS should put metrics and standards in place to ensure that decision making is shared with the patient, including improving access to relevant information about their condition.
- The Government and the NHS must ensure the new Rare Disease Framework recognises the changing landscape and considers how future care for rare disease patients may be impacted by an ongoing pandemic.

Policy recommendations

- The Department of Health and Social Care should lead on the development of the implementation plans for the new Rare Disease Framework.
- The NHS and Government should establish clear accountability for the implementation of the new Rare Disease Framework from the outset.
- The NHS and Government should review the delivery of the commitments in the Rare Disease Framework on an annual basis.
- The NHS should appoint a dedicated National Clinical Director for rare diseases whose responsibility should be to provide leadership, strategic guidance and support to NHS England and NHS Improvement on the transformation of services for patients with rare diseases.
- The NHS and Government should ensure that rare diseases are always considered when developing future national and regional health plans and aligned with other strategic health priorities, such as the establishment of the Innovative Drugs Fund, review of the NICE methods and processes and establishment of Integrated Care Systems.
- The Government should ensure there is a clear allocation of funding made available at a national and local level for the full implementation of the new Rare Disease Framework.

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Key findings

The Reforming Rare Disease report details findings from an extensive survey of 1,020 people living with or caring for people with a rare disease on their experience of care along the patient pathway, now and in recent years, as well as clinicians involved in the delivery of their care.

68% 68% of patients believe that they have sufficient knowledge of their rare or undiagnosed condition.

42% Only 42% of patients felt that the hospital based professionals involved in their care had sufficient information about their condition.

Access to information for patients with rare diseases and their clinicians can help strengthen patient empowerment.

Diagnosis → Coordination → Empowerment → Treatment

Patients continue to report dissatisfaction with time to diagnosis as well as high levels of misdiagnosis.

52% **41%** 52% diagnosed within the last five years had to wait over two years for their diagnosis, with 41% of patients waiting over five years.

61% **56%** Among those without a definitive diagnosis 61% had been searching for a diagnosis for over 5 years and 56% have been misdiagnosed.

The 2013 Strategy has not led to the effective coordination of care for significant numbers of rare disease patients.

74% Despite being a commitment of the 2013 Strategy 74% of patients with a rare disease do not have a care plan.

- 50% of patients believe their care is not effectively coordinated.
- 30% of patients believe their care is effectively coordinated.
- 20% of patients are unsure.

Availability and access to care and treatments remain a problem for many people living with a rare disease.

48% 48% of patients state that there is a specialist centre to treat their disease, however, only 52% of these respondents access care at the specialist centre.

52% **64%** 64% of patients believe that the system is unfair on people living with rare diseases.

Overall care

A third of patients with a rare disease believe their care is poor.

Half of patients have seen no improvement in their care in the last five years while one in five patients has seen a deterioration.

Reference for all statistics: Genetic Alliance UK patient survey 2020

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About the UK Strategy for Rare Diseases

In November 2013, the UK Department of Health and Social Care (DHSC) published the UK Strategy for Rare Diseases. The Strategy's publication followed a 2009 Council of the European Union recommendation for all member states to have a strategy in place by 2013.[13] The strategy included 51 objectives set across five priority areas for change:

- **Empowering those affected by rare diseases**
- **Identifying and preventing rare diseases**
- **Improving diagnosis and early intervention**
- **Coordinating care more effectively for all patients**
- **Stimulating research and development (R&D) into rare diseases**

> ***The UK Strategy aims to ensure no one gets left behind just because they have a rare disease. We want to put the patients' needs first. To do this, we will bring together the talent, skills and professionalism of all relevant sectors. This will bring real, positive change in how we deal with rare diseases and how we help people with complex conditions.***
>
> **2013 UK Strategy for Rare Diseases.**

The Strategy's implementation period was 2013-2020 and devolved administrations were given responsibility for delivery of commitments. Scotland's plan was published in June 2014,[14] Northern Ireland's in October 2015,[15] Wales' in July 2017,[16] and England's two separate implementation plans (NHS England[17] and the DHSC[18]) in January 2018, four years after the 2013 Strategy had been published.

The NHS has evolved considerably since 2013. Structural reform has led to a realignment in healthcare policymaking and commissioning at national and regional levels, while the impact of COVID-19 has transformed how many patients receive their care and interact with the health system. **As demonstrated in the NHS Long Term Plan, the coordination of services, the integration of care and a holistic personalised approach to treatment is seen as a priority.[19]**

Since 2013, technological innovation has also created new opportunities for researchers, clinicians and patients. Many of these advances have had a profound impact on patients across the UK. The 100,000 Genomes Project has set the framework for the establishment of a world-leading Genomic Medicine Service. And European Reference Networks for rare disease have facilitated the sharing of expertise, knowledge and resources across the EU.

These ambitions for technology and innovation align with the recommendations in the Life Sciences Industrial Strategy (LSIS) which identifies the importance of strategic investment to further the development of the next generation of medicines and deliver growth for the economy.[20] Implementing the LSIS is essential to ensure the UK remains at the forefront of genomic research and clinical trials to provide UK patients earlier chances to benefit from these advances.

The UK Government and the NHS have recognised the need for a new Rare Disease Framework and work began on priorities in 2019.[18] Set to be published by the end of 2020,[21] there is an expectation that implementation plans will follow. Ensuring these implementation plans deliver on both on the ambition of this new Framework and priorities of the patient community will be crucial. This report sets out the priorities for the new Rare Disease Framework and implementation plans from across the rare disease community.

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Methodology

To develop data and insight for this report, Alexion has worked with two groups.

Genetic Alliance UK

In partnership with Alexion UK, Genetic Alliance UK fielded a survey to understand how patient and carer experiences of navigating the rare diseases patient pathway had been impacted by the 2013 Strategy.

1,020 people responded to the survey

82% were patients and **18%** carers of people with a rare disease.[22]

83% of respondents were female; **16%** were male (in line with anticipated responses).[23]

87% of responses were from/on the behalf of people over the age of 18.[24] The most common age of respondents was between the ages of 45-54.[25]

83% of respondents were from England; **9%** from Scotland, **7%** from Wales and **1%** from Northern Ireland.[26]

88% of respondents had received a definitive diagnosis, **9%** remain undiagnosed and **3%** were unsure. [27]

15% of patients had one aspect of health affected by their condition. **21%** had 2-3 aspects of health affected, **22%** had 4-5 aspects of health affected, **20%** had 6-7 aspects of health affected. **22%** had 8 or more aspects of health affected.[28]

Following the survey, Genetic Alliance UK conducted two half-day patient workshops with 11 attendees. These workshops allowed patients to discuss the key themes of the patient experience survey in more depth. Quotes from these sessions are used throughout this report. More information can be found in the report that Genetic Alliance UK has published on its survey findings,[29] as well as a proposed forthcoming journal article.

An Expert Policy Group convened by Alexion UK

Alexion UK convened an Expert Policy Group, bringing together five experts across the rare disease community including clinicians and patient organisation leaders.

An initial meeting was held on the 9th October, where members of the Group provided their own insights into the success of the 2013 Strategy and identified areas for improvement in the new Rare Disease Framework.

Survey questions were agreed with members of the Expert Policy Group following this meeting. These were structured around the priority themes of the 2013 Strategy.

This survey was distributed among networks of Expert Policy Group members. The survey was live for two weeks and received the majority of responses from rare disease clinicians.

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Overall experience of care

For many patients, the quality of their care is not improving, while the COVID-19 pandemic has also brought additional challenges.

PATIENT PERSPECTIVE

Our survey of patients with Genetic Alliance UK indicated that:

- **37%** of patients view their care as poor or very poor.[6]
- **35%** of patients view their care as good or very good.[6]
- **28%** of patients have a neutral view of their care. [6]

- **50%** no change in the quality of care over the past five years.[5]
- **20%** reported a negative change in the experience of their care.[5]
- **16%** reported recent improvements in their care.[5]

Data varies depending on the time of diagnosis and the complexity of a patient's condition:

- **43%** of those diagnosed within the last five years rate their overall experience of care as very poor or poor, compared to **28%** of those diagnosed more than six years ago.[30]
- Patients whose conditions affect multiple aspects of their health are consistently more likely to have a negative experience of care.[31]

COVID-19

The survey also assessed how COVID-19 has impacted experience of care:

- Almost two thirds of patients have reported that their care has been disrupted due to COVID 19,[32] while **73%** reported changes in their care.[33]
- Those with multiple aspects of health affected by their condition were more likely to experience changes to their care during COVID-19.[34]
- Only **24%** of patients have used video consultations or appointments,[35] which **64%** found useful.[36]

Unsurprisingly, the majority of clinician respondents felt that COVID-19 had negatively impacted the coordination of care for patients with rare diseases and that it had negatively impacted access to treatment.[37]

Whilst rare disease patients and clinicians are not alone in feeling this impact, it is important context for the new Rare Disease Framework implementation plans to note that many hospitals will be working to restore services to full capacity.

CLINICAL PERSPECTIVE

Over half of the clinicians consulted by Alexion UK via the survey felt that, overall, there has been an improvement in care for rare disease patients since 2013.[38] However, only a small minority of clinicians reported that they felt that the prioritisation placed on the investment in and the commissioning of rare disease services was currently adequate.[39]

The Government should seek to evaluate the current standard of care to ensure lessons are learned and the patient experience of care can be improved by the new Rare Disease Framework. It is important that the Government takes the impact of COVID-19 on rare disease patients into consideration in the new Rare Disease Framework and that future services are designed to be more resilient in the face of disruption to the health service.

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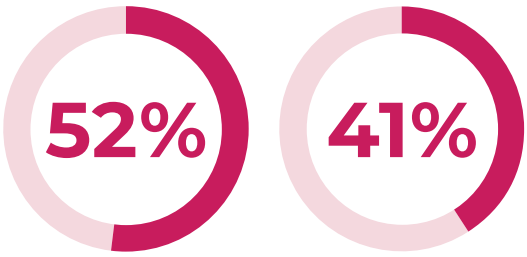
Diagnosis

Patients continue to report dissatisfaction with time to diagnosis as well as high levels of misdiagnosis.

A core focus of the 2013 Strategy was on improving time to diagnosis and certainly, progress has been made. The establishment of the Genomic Medicine Service and the completion of the 100,000 Genomes Project led to one in four participants with a rare disease receiving a diagnosis for the first time.[40] For the patients with a definitive diagnosis who responded to the Genetic Alliance survey:

- **46%** had had their diagnosis confirmed by genetic testing. Of those, **35%** were offered whole genome or exome sequencing.[41]
- Over **50%** were satisfied with the speed that testing was offered.[42]

Despite the greater availability in genetic testing, overall time to diagnosis remains an issue for many patients.



52% diagnosed within the last five years had to wait over two years for their diagnosis, with 41% of patients waiting over five years. [7]

The number of patients receiving a diagnosis within one year appears to be declining.

- Between three and five years ago, **41%** of patients received a diagnosis within a year of the onset of symptoms. In the last two years, this figure has fallen to **31%**.[43]

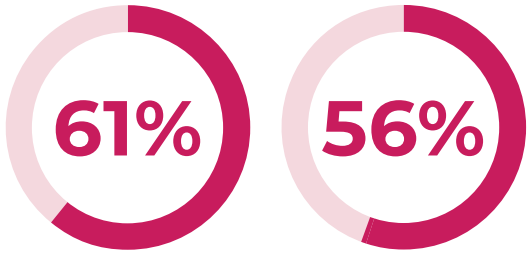
Patients with both diagnosed and undiagnosed conditions continue to experience high levels of misdiagnosis.

- For those with a definitive diagnosis, **49%** had previously been misdiagnosed at least once. **32%** had previously been misdiagnosed two or more times.[8]
- For those diagnosed within the last five years, **56%** had received misdiagnoses for their condition.[44]

> ***As I am undiagnosed, I get sent off to various hospital departments with no coordinated approach... So frustrating, feel like a hamster on a wheel, going nowhere.***[45]
>
> *Patient workshop*

The experience of finding a diagnosis was also more challenging for patients whose condition affects multiple aspects of their health or who are still undiagnosed:

- **67%** of patients whose condition affects six or more aspects of their health have been misdiagnosed in the past, compared with **37%** of those with up to five aspects of health affected.[46]



Among those without a definitive diagnosis 61% had been searching for a diagnosis for over five years[47] and 56% have been misdiagnosed.[48]

With the growing capabilities of genomic testing, the NHS should utilise the Genomic Medicine Service to ensure all patients suspected of having a rare disease are identified and able to access the full range of diagnostic capabilities. This aligns with the ambition in the NHS Long Term Plan to be the first national healthcare system to offer whole genome sequencing as part of routine care.[19] Alongside a clear desire from patients to improve the time to an accurate diagnosis, respondents to the survey of clinicians identified accelerating diagnosis and referral for specialist treatment as the most important factor to consider in the new Rare Disease Framework.[49]

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Empowering those affected by rare diseases

Access to information for patients with rare diseases and their clinicians can help strengthen patient empowerment.

The first theme in the 2013 Strategy was the commitment to patient empowerment by improving patient information and ensuring that patients can be active partners in their care.[1] The DHSC has shown a commitment to engaging patients over the course of the 2013 Strategy, including its work with the Rare Diseases Forum and the Patient Empowerment Group.

Engagement with patients and patient groups continues to be vital, particularly as online groups and patient organisations were identified, amongst those surveyed by Genetic Alliance UK, as the main sources of information and support for a rare or undiagnosed condition.[50]

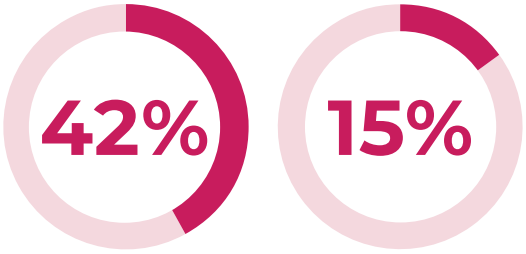
- **68%** of patients believe that they have sufficient knowledge of their rare or undiagnosed condition.[51]
- For those who are still undiagnosed, only **30%** hold this view.[52]

However, patients' views of the knowledge and information provided by their healthcare professionals (HCPs) vary at different stages of the patient pathway:

- **49%** of patients were unsatisfied or very unsatisfied with the information provided by healthcare professionals pre-diagnosis.[53]
- At the point of diagnosis, **58%** of patients were satisfied or very satisfied.[53]
- Levels of satisfaction about the information provided by their HCPs decrease as condition complexity increases, at all stages of diagnosis (before, during and after).[54]

> ***Part of the reason I have not sought answers is because in the past, healthcare professionals have looked completely blank when I brought up my long-term condition.***[55] *Patient workshop*

Many patients also question the levels of information about their condition that hospital staff have access to:



Only 42% of patients felt that the hospital based professionals involved in their care had sufficient information about their condition, this falls to below 15% across HCPs outside of hospital settings .[56]

- As the complexity of the condition increases, confidence and patients' belief that the professionals involved in their care at hospitals had sufficient information about their condition falls.[57]
- **64%** of patients with a rare disease do not have access to an 'alert card' with information about their condition, treatment regime and contact details for the expert involved in their care.[58] Additionally, the more recent the diagnosis, the less likely it is a patient will have an alert card.[59]
- **75%** of adults are involved in decisions about their treatment,[60] but for those people who are not, **84%** would like to be.[61]

The NHS should put metrics and standards in place to ensure that decision making is shared with patients where that is wanted by the patient. The varying satisfaction that patients reported with the information provided by HCPs should be addressed by ensuring every patient has a dedicated care coordinator, access to a specialist centre, a tailored alert card (a 2018 recommendation from NHS England that providers of rare disease care services should make these available to all patients)[17] and care plan available to them if desired. Access to the appropriate specialists should guarantee that patients feel they have adequate information and are appropriately supported with their care.

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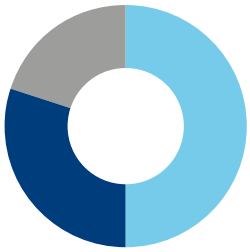
Coordination of care

The 2013 Strategy has not led to the effective coordination of care for significant numbers of rare disease patients.

PATIENT PERSPECTIVE

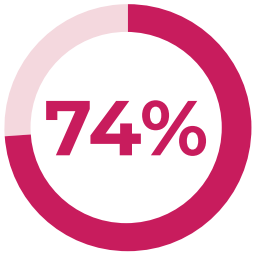
The coordination of care was highlighted as a priority in the 2013 Strategy, stating that specialist clinical centres should coordinate care for patients and all patients should be provided with an appropriate care plan.[1]



- **50%** of patients believe their care is not effectively coordinated.[9]
- **30%** of patients believe their care is effectively coordinated.[9]
- **20%** of patients are unsure.[9]

However, the coordination of care remains a core challenge for patients with a rare disease. The survey results suggest that key reasons for half of all patients[9] feeling that care is not effectively coordinated are that:

- **71%** of patients coordinate their own care[62] and only **26%** of this group of patients believe that their care is effectively coordinated. This stands in contrast to patients who have a dedicated care coordinator, **71%** of whom rate their care as effective.[63]
- **66%** of patients who believe their care is not coordinated think the professionals providing their care do not 'work as a team'.[64]



Despite being a commitment of the 2013 Strategy 74% of patients with a rare disease do not have a care plan.[65]

> ***My partner is the care coordinator, has gone down to part time work to fit this in and spends hours phoning and chasing for appointments etc.[66]*** *Patient workshop*

Patients with complex conditions[10] and those who are yet to receive a definitive diagnosis[67] also report lower levels of care coordination.

- The more areas of health which a condition affects correlates with a lower probability of having a care coordinator,[68] thus patients are left to coordinate their care on their own, even if these patients are more likely to have a care plan.[69]
- Those yet to receive a definitive diagnosis were also more likely to state that the professionals providing their care did not work as a team.[70]

> ***I organise all of my care. I had to push for every service I got.[71]*** *Patient workshop*

A core reason why patients with rare diseases require specific care coordination relates to their frequent interactions with the health service, often within different settings, e.g. clinics, admissions and emergency admissions at hospitals, GP appointments and home care visits. The Genetic Alliance UK survey found that undiagnosed patients and those with more complex conditions had significantly different interactions with the health service than those with less complex conditions.

- Those patients without a definitive diagnosis tend to have used the health service more frequently compared to those who have received a diagnosis. They are over twice as likely than those with a diagnosis to have used the health service over 20 times between March 2019 and March 2020.[72]
- **31%** of the of the patients that had used the health service over 20 times between March 2019 and March 2020 have a condition that affected eight or more aspects of their health, which represents **22%** of all respondents.[73]
- For patients with one aspect of their health affected, just **5%** had used health services more than 20 times in the same period.[73]

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CLINICAL PERSPECTIVE

The survey of rare disease clinicians had a specific focus on their views regarding the coordination of care for patients. There were a number of key areas of improvements seen in recent years:

- Improved ability to share patient information between primary and secondary care.[75]
- Improved coordination between specialists.[75]
- Improved access to rare disease registry and ability to register patients on them.[75]
- Higher levels of appropriate patient referrals.[75]

However, there were still a number of challenging areas and key recommendations for the development of the new Rare Disease Framework:

- Just over one quarter of clinicians are satisfied with the level of care coordination they see offered to patients.[76]
- A majority still felt that access to shared patient information remains inadequate.[77]
- Less than one quarter of clinicians found that care coordination between primary and secondary care had improved since 2013, despite improved access to data.[75]
- Just under half of clinicians report that patients within their service are usually assigned an appointed care coordinator.[78]
- The majority felt that the new Rare Disease Framework should make provision for patients to have access to a dedicated care coordinator by the time they have reached a diagnosis.[79]
- Just over one quarter of clinicians see patients receive an alert card within their service, reflecting the responses from the patient experience survey.[78]

One clinician raised an important challenge faced within their service, which although funded directly by NHS England, they have experience at a local level that administrative and nursing staff have their time diverted by Trust management to other parts of hospital services that are under pressure. Further research is needed to establish if this represents a national issue, but it will be essential for rare disease services to have the appropriate workforce in place to provide care coordination for patients and engage in clinical research activities.[80]

There was a unanimous view that the implementation of the new Rare Disease Framework must be accompanied by appropriate funding,[81] and a majority opinion that the Department of Health and Social Care should retain central accountability for the implementation of the strategy.[82]

> ***It isn't coordinated at all, none of the health professionals liaise with each other and each aspect of my care is handled by a different team.***[74] *Patient workshop*

The very nature of rare diseases makes personalised care essential, as identified by the commitments in the 2013 Strategy.[1] The fact that a large majority of patients coordinate their own care and do not have a care plan should be looked in to further, as it is important to understand if this is out of preference or habit due to inefficiencies in the system. The clear correlation between frequency of health service use and whether a patient has a diagnosis or not, and whether their condition affects multiple aspects of their health, suggests an apparent need to ensure care is more effectively integrated both for patients suspected of having a rare disease and with complex conditions. The reduction in interactions with the health service which could result from a more integrated service would be beneficial for patients, in their experience of care, and the NHS, in reducing demand for services and lowering the costs of care.

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Good practice case study

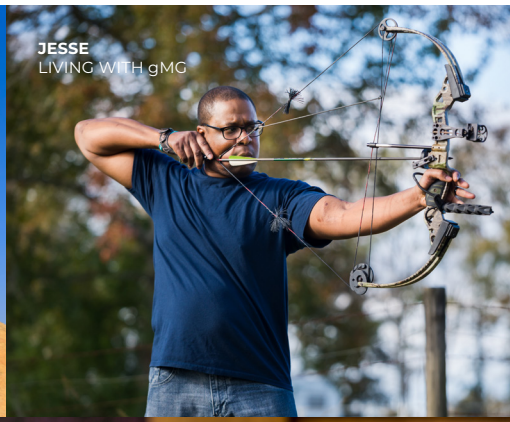
Xeroderma pigmentosum is a rare inherited condition that affects approximately 100 people in the UK. Patients are not able to repair the normal damage that occurs from exposure to ultraviolet light and they develop skin cancers from early childhood, severe eye disease, and neurological degeneration. Most patients will die of skin cancer with a life expectancy of 32 years.[83]

Patients tended to be seen by consultants who had never seen the condition before. Meaning they were given inadequate care with unnecessary, inappropriate investigations and surgery, until a new service was established in 2010 to coordinate care.

Patients come annually to a multidisciplinary clinic where they can see a dermatologist, dermatological surgeon, ophthalmologist, neurologist, neuropsychologist, geneticist, and specialist nurse. Nurses also supervise an outreach programme and liaise with the patients' GP, as well as visiting the patient at home to provide advice on preventative measures. A patient support group is an integral part of the project.

The service has reduced the number of appointments and the amount of unnecessary surgery, saving the NHS valuable capacity and £80,000 a year.[83]

Case study suggested by a clinician responding to the rare disease clinical survey.



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Access to specialist care and treatment

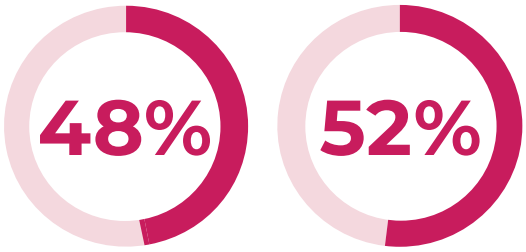
Availability and access to care and treatments remain a problem for many people living with a rare disease.

Access to clinical experts and specialist centres

The Genetic Alliance UK survey found that patients with a rare disease have contrasting access to clinicians for their condition.

- **46%** of patients with a diagnosis have access, while another **46%** do not.[84] Of those patients without a definitive diagnosis, only **11%** report having access to an expert.[85]
- There is a clear correlation between condition complexity and access to an expert: those with more complex conditions (more than six areas of health affected) are twice as likely not to have a doctor who is an expert in their condition than those with less complex conditions (one aspect of health affected).[86]

Access to specialist centres, defined as one able to provide expert advice on diagnosis, assessment and treatment of a particular condition, is also mixed with patients whose conditions affects more aspects of their health again at a disadvantage:



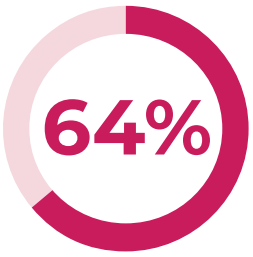
48% of respondents state that there is a specialist centre to treat their disease,[87] however only 52% of these respondents access care at the specialist centre. 43% are able to and 5% are unsure.[88]

- Those with eight aspects of health affected are twice as unlikely to access centres than those with just one area of health affected.[89]
- There is a clear correlation between satisfaction with services overall and access to a specialist centre. **85%** of patients with access to a specialist centre reported that their overall experience of care as very good, compared to just **11%** of respondents who do not have access to a specialist centre.[90]

> ***Many other countries around the world don't have this limitation on use of [product] and it's very frustrating that I can't receive the same treatment because of funding.***[91] *Patient workshop*

Access to treatment

For patients living with a rare disease, access to medicines and treatment for their conditions is a clear concern.



64% of patients believe that the system for making treatments available to patients is unfair on people living with rare diseases,[11] as demonstrated by the following statistics:

- Only **10%** of patients surveyed are satisfied with the process used to decide on funding rare disease medication in the NHS.[11]
- Only **3%** agree that enough money is allocated to rare disease medicines.[11]
- **65%** of patients believe the system is too slow to make treatments for rare diseases available to patients.[11]
- **58%** believe decision-making and pricing is not transparent.[11]

Access to specialist services and clinicians as well as the provision of treatment are highly important for patients and are significant factors in shaping their overall experience of care. As a consequence, enabling greater access to specialists and specialist centres should be a specific focus of the new Rare Disease Framework and the development of other relevant healthcare policies. Alignment with opportunities such as the development of the Innovative Drugs Fund and review of NICE methods and processes should be made in the Rare Disease Framework, while these policies should be harnessed to their full extent.

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Discussion

The patient survey findings and insight from clinicians set out in this report have highlighted four important issues which should form the heart of Government and NHS-led plans to develop and implement the new Rare Disease Framework.

Quality of care

Too many people living with a rare disease are still not satisfied with the overall level of care they are receiving for their condition. Progress against the commitments in the 2013 Strategy should not be ignored, but neither the experience of patients nor the views of clinicians indicate widespread satisfaction with the real-world outcomes of the Strategy. Indeed, a majority of patients take the view that, since 2013, there has been no discernible change in the quality of care they have received.[5] Crucially, the causes of this dissatisfaction appear to lie across the patient pathway.

Diagnosis

Improvements in diagnostic capabilities have led to higher numbers of patients receiving earlier and more accurate diagnoses. These technological advances need to be harnessed to their fullest extent in the coming years, because overall, patients still report multiple years of waits for diagnosis and high levels of misdiagnosis.

Coordinated care

For patients living with a rare disease, the coordination of their care is a particular concern. This is clear from analysing patients' specific views on how coordinated they believe their care is, and how those patients with particularly complex conditions interact with the health service. On both counts, improvements are required. At the same time, on almost every metric surveyed, from diagnosis through to the level of information their consultants have access to, those people whose conditions affected multiple aspects of their health, and therefore interact with multiple services, have a markedly poorer overall experience of the care pathway. The struggle that people with complex conditions face with care coordination suggests that there is still a long way to go for the NHS to achieve its ambitions for personalised care for all patients as set out in the Long Term Plan.[19]

Access to treatment and care

The most decisive views held by people living with a rare disease collectively relate to access to treatment for their condition. The survey results demonstrate clear dissatisfaction with decision-making processes, allocation of resources, transparency and speed of access to treatment. Overall, this leads to almost two thirds of patients believing the system in general is unfair on them.[11] On access to care, there is still a significant number of patients reporting they are unable to access expert clinicians or specialist centres for their conditions.

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The development and implementation of the new Rare Disease Framework will be a welcome first step for many of the 3.5 million people in the UK who live with or will be affected by a rare disease in their lifetime. But, to ensure that this enables the improvements in care sought by patients and clinicians across the whole patient pathway, it is vital policymakers identify and learn from where the 2013 Strategy succeeded and where it fell short.

- Accountability for the implementation of the recommendations should be clear from the outset, a point which could be enhanced through the establishment of a National Clinical Director for Rare Diseases.
- Where necessary, recommendations should also be backed up by requisite funding pledges.
- Progress against delivery of the commitments in the new Rare Disease Framework should be regularly reviewed and publicly documented.
- Rare diseases should be considered specifically in the development and delivery of other healthcare policies, including how the NHS responds to the COVID-19 pandemic. The test for future strategies, such as Integrated Care System plans and the Innovative Drugs Fund, should be assessing the impact they will have on rare disease patients.
- Alignment with the recommendations in the Life Sciences Industrial Strategy[20] will also be important. Strategic investment will further the development of the next generation of medicines for patients with a rare disease, ensure the UK remains at the forefront of genomic research, and through clinical trials, will provide UK patients with an earlier chance to benefit from these advances, at no extra cost to the NHS.

If backed up by the right policy prioritisation, development and implementation, the skill and dedication of healthcare professionals across the UK, combined with technological and scientific advances, give the NHS the tools to transform the experience of care of people living with a rare disease. Within this challenge, addressing the coordination of care for patients is possible and should be an early priority.

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Conclusion

Improving the lives and experience of care for patients is a unifying ambition of all stakeholders involved in the patient journey, from researchers and biopharmaceutical companies pushing the boundaries of science; healthcare professionals and NHS staff who provide care for millions; and organisations who provide vital support for people living with rare diseases.

The research conducted for this report has identified some of the key challenges which face people living with rare diseases, as well as the clinicians who provide their care. It has also set out clear recommendations which could help bring about these changes. If addressed through the implementation of the new Rare Diseases Framework, these changes will help to improve the overall experience of the care pathway for over 3.5 million people in the UK.

Alexion looks forward to continuing to work alongside the wider rare disease community, government and health system to ensure the potential of the new Rare Disease Framework is fully realised. Collectively we hope to see this potential reflected in improvements reported by patients in the future.

MONIKA
WITH HER HUSBAND
LIVING WITH gMG

SUMARIA
LIVING WITH NMOSD

ANNA
WITH HER SISTER
LIVING WITH HPP

AIRA
LIVING WITH HPP

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ALBIE
WITH HIS MUM
LIVING WITH LAL-D

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