

THE RARE REALITY OF COVID-19

July 2020



**GENETIC
ALLIANCE** ^{UK}

ABOUT GENETIC ALLIANCE UK



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



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



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

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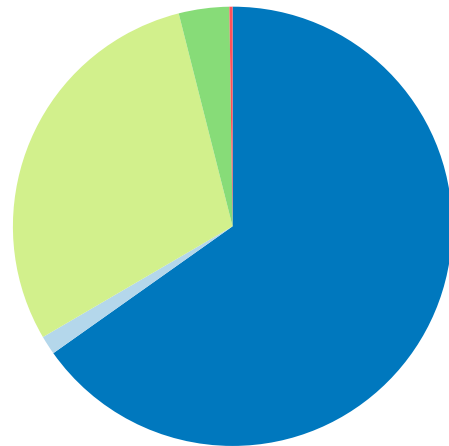
EURORDIS Rare Barometer Covid-19 survey

The data presented in this document was collected through the EURORDIS Rare Barometer Covid-19 survey which launched on 18 April. Only UK data is presented.

There were 431 respondents, 65% were patients, 1% patient representatives, 29% parents, 4% spouses, 1 respondent was a sibling.

Note

In this report we have chosen to refer to rare conditions, and people living with rare conditions. This is because all undiagnosed conditions, and almost all genetic conditions are rare, and because the negative impact upon our community has its roots in the rarity of the conditions affecting them. UK policy documents refer to 'rare diseases', our community prefer the term 'rare condition'. The two terms are interchangeable and have no difference in meaning.



- Patient
- Patient Representative
- Parent of adult or child
- Spouse
- Sibling

EXECUTIVE SUMMARY

People living with rare conditions have been placed under immense pressure by the Covid-19 pandemic. Access to appropriate support, information, care and treatments has become more difficult and levels of social isolation have been increased.

In this report, we present findings from the EURORDIS Rare Barometer Covid-19 Experience Survey, and from our weekly community meetings during the months of April, May and June 2020.

Routine rare condition healthcare has been interrupted – 66% of respondents say that the interruption has been probably or definitely detrimental to their wellbeing.

Hospital care is severely affected – 80% of our community receive care in hospitals, but 40% reported closure of units and for 16% necessary equipment for their hospital care was absent because it was needed for Covid-19 care.

Access to medicines is being disrupted – 1 in 5 people affected by a rare condition have experienced disruption to access to their usual medication.

Support for people living with rare conditions has been slashed – support from neighbours, family, psychological services, home care, respite care and day care have all been reduced or taken away.

People living with rare conditions are scared and worried – more than 40% of respondents thought Covid-19 posed a very high level of threat to themselves or the person with a rare condition they cared for.

There has been a sudden switch to remote consultations – delivering mixed results for people living with rare conditions – this has been a good stopgap for many, but much of the change has been without consultation with the community.

Access to PPE has been difficult – only a small minority of people living with a rare condition had adequate access to PPE for the person they care for, for themselves or for their social care professionals.

Local authority support for people in the home is no longer an obligation – many children have lost all or a portion of their education support.

Tests have not been available for those who need them – only 9% of people affected by a rare condition who thought they should have been tested for Covid-19 actually accessed a test.

The sudden and, for many, devastating changes to our community's lives came quickly and out of necessity. The relaxation of lockdown and changes to healthcare can be performed with greater consideration and consultation. It is crucial that further shocks to our community be avoided to prevent additional serious impact to people living with rare conditions.

The following recommendations will mitigate the negative impact of Covid-19 on people living with rare conditions and prepare the UK better to support our community in the case of another pandemic.

RECOMMENDATIONS

Continuing to deal with Covid-19

- In monitoring the spread of Covid-19, data should be collected that will enable assessment of the impact, in terms of morbidity and mortality, on people living with rare conditions.
- People living with a rare condition who have a legitimate reason to ask for a test for Covid-19 (either for diagnosis or in order to assess future risk) should be given priority access.
- Access to PPE (personal protective equipment) should be guaranteed for people with rare conditions and those providing care and support to rare condition patients.
- When a vaccine or other prophylactic treatment becomes available, patients with rare conditions should be among those with priority access, provided their health condition allows.

Successful transition from crisis state

- For those with rare conditions that confer high risk from Covid-19 who wish to continue protecting themselves through isolation, the support mechanism established should continue to be provided – no one should be forced to stop their protective isolation.
- A clear and short timetable for the reintroduction of services should be published to give people living with rare conditions clarity as to when they can expect a full service from the NHS.
- The provision of remote consultations should be continued. Care should be taken to integrate telemedicine into routine care practice with the necessary clinical assurance and data protection safeguards.
- Schools and educational facilities should be supported to ensure that they have the necessary flexibility to respond to individual needs and to adapt practices for children with rare conditions.

Learning for the future

- The challenges and failures in delivering letters giving shielding advice should be examined, and actions should be taken following this analysis to ensure the UK is able to rapidly provide tailored public health advice to specific populations with specific conditions.
- The development of the new UK framework for rare diseases has been delayed by this crisis – it is now more necessary than ever. Learnings from this crisis should be incorporated into the new framework before the end of 2020.

INTRODUCTION

A rare condition is defined as one that affects fewer than 5 in 10,000 of the general population. There are between 6,000 and 8,000 known rare conditions, and collectively they are not rare. One in 17 people will be affected by a rare condition at some point in their lives. This equates to approximately 3.5 million people in the UK. Often, rare conditions are chronic and life-threatening.

Genetic Alliance UK is the national charity working to improve the lives of patients and families affected by genetic, rare and undiagnosed conditions. We are an alliance of over 200 patient organisations. We are home to Rare Disease UK – the national campaign for people with rare conditions 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. In this report we have chosen to refer to rare conditions, and people living with rare conditions. This is because all undiagnosed conditions, and almost all genetic conditions are rare, and because the negative impact upon our community has its roots in the rarity of the conditions affecting them. UK policy documents refer to ‘rare diseases’, our community prefer the term ‘rare condition’. The two terms are interchangeable and have no difference in meaning.

Those living with a rare condition can face significant challenges in getting a diagnosis, accessing treatment and receiving coordinated care, as well as challenges with employment, education, social life and mental health. Social isolation is a significant challenge, as is getting access to the necessary information and support. Caring for a child or adult with a rare condition can impact parents’ and carers’ emotional wellbeing and impact on the wellbeing of the wider family, including siblings. An earlier survey on the impact of living with a rare condition on mental health conducted by Genetic Alliance UK found that 88% of individuals feel emotionally exhausted by living with a rare condition.

Having a rare condition can place a person at increased risk of contracting Covid-19 and/or at increased risk of mortality from the virus. The pandemic has left many families isolated and struggling to access support, as well as experiencing problems in accessing ongoing care and treatment for their condition.

In March 2020, Genetic Alliance UK launched its Covid-19 hub and has been hosting ‘safe space’ online meetings on a weekly basis for organisations that support patients and families with rare conditions. Through these meetings and through our online communities we have learned more about the immense and escalating practical, emotional, social and financial impacts of Covid-19 for people affected by rare conditions and their families.

This report captures these experiences at a time when our four nations are emerging from lockdown and while the pandemic still poses a significant risk to our community. It brings together the learning from our Covid-19 programme and UK data from a major multi-national study undertaken by EURORDIS (Rare Diseases Europe).

IMPACT ON HEALTHCARE FOR RARE CONDITIONS

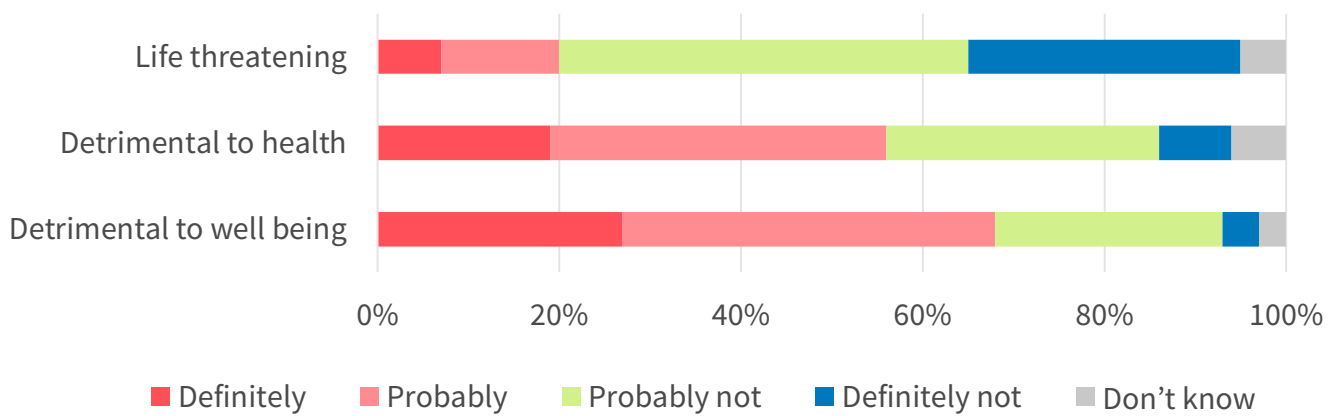
Every challenge inherent in living with a rare condition has been exacerbated by Covid-19 and by the necessary measures taken to contain the pandemic.

More than half of UK respondents to the Rare Barometer Covid-19 experience survey judged the impacts of interruptions caused by the pandemic to be detrimental to health and two thirds reported negative impacts on their wellbeing.

Some elements of the pandemic’s disruption to care for conditions will not have been felt yet. Cancelled appointments, missed tests, delayed access to treatments and paused physiotherapy will all have knock on effects for months or years into the future.

Would you say that interruptions related to the Covid-19 pandemic you are experiencing are:

Data from EURORDIS Rare Barometer Covid-19 survey



‘I’m tired now. I’ve been at home three full months on my own with three children. Two have a learning disability, visual impairment, mental health needs and challenging behaviour. And I have a toddler. My partner can’t visit as he is a key worker and would put the boys at risk. Our carers can’t come in. There is no school, no nursery, no clubs, no groups or activities, respite is closed. Normally we have three carers, a tutor, a cleaner, respite and support from family and friends. Normally we get by. I’ve had to give up working because I simply couldn’t do it and there is no end in sight for us.’

Sarah (name changed), a mum of two children affected by a rare genetic condition, shares her experiences of Covid-19.

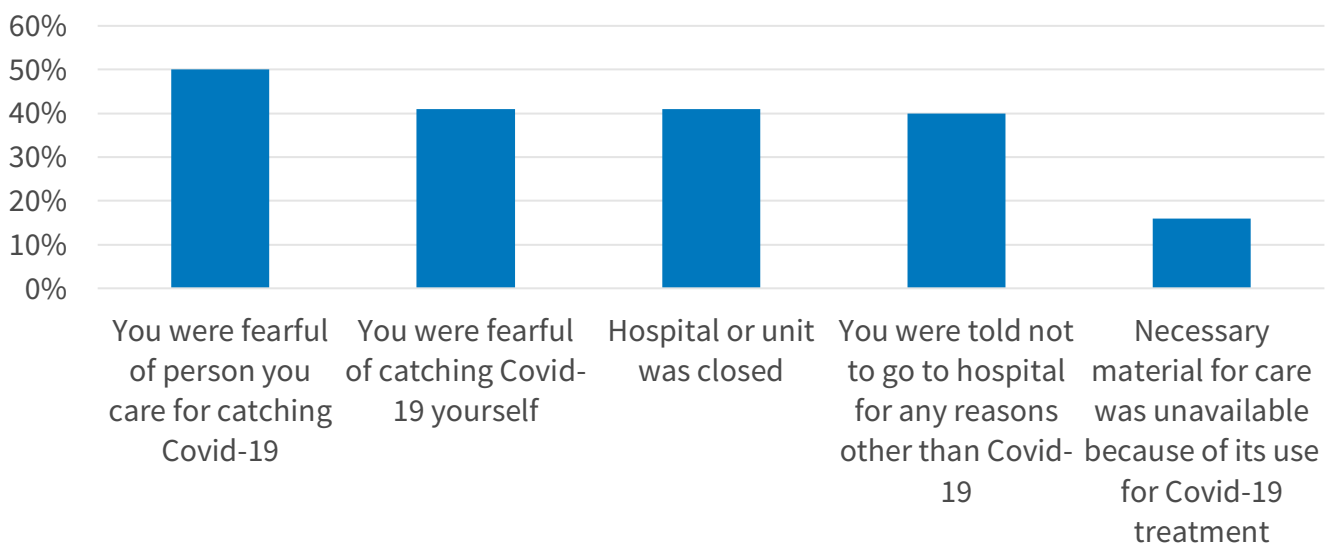
Hospital care

Around 80% of patients with rare conditions receive all or part of their care through a hospital. Disruption in access to routine care has been substantial, with 40% reporting that their hospital or unit had closed and 40% reporting that they have been told not to go to hospital for any reason other than Covid-19. Also of concern are reports that for those who did attend routine appointments, equipment necessary for care was absent because it was in use for patients affected by Covid-19.

A further barrier to accessing care has been the fear of catching Covid-19, with more than half avoiding hospital for fear of contracting the virus. This avoidance of risk is not surprising given that the majority of people affected by rare conditions believe the level of threat to themselves or the person they care for to be high or very high.

Percent answering 'yes' to the question, were you unable to go to the hospital because:

Data from EURORDIS Rare Barometer Covid-19 survey



There are a number of highly damaging potential impacts from reductions in access to healthcare for people with rare conditions. For many, ongoing monitoring and adjustments to care and treatment prevent or delay progression of the condition, or prevent or lessen the severity of symptoms. Contact with health teams and specialists also serves to support the overall mental health and wellbeing of patients and their families and can serve as a bridge to sources of social, community and wider support.

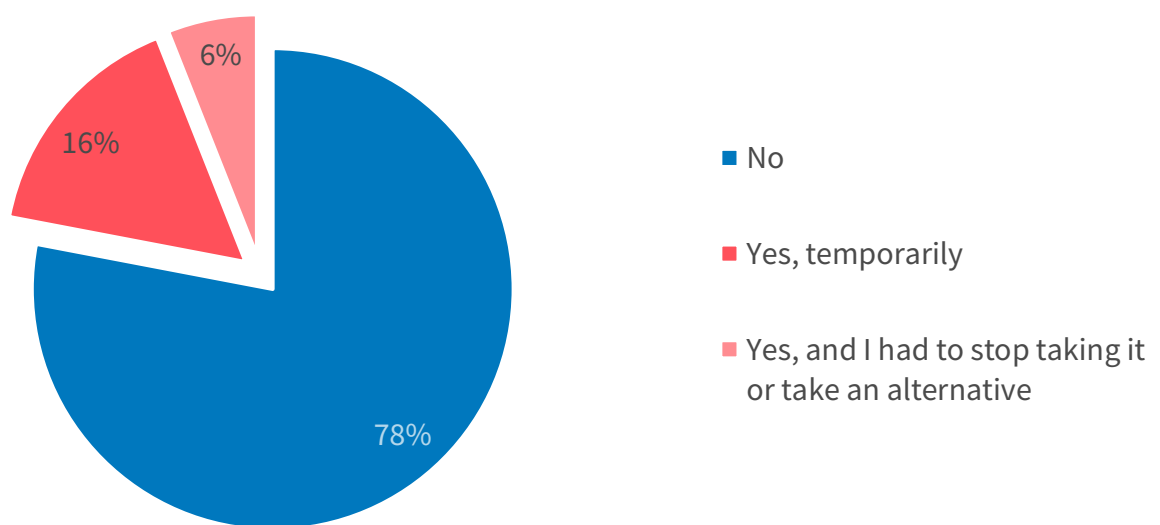
Access to medicines and treatments

During the pandemic, the National Institute for Health and Care Excellence (NICE) has had to prioritise work that is either therapeutically critical or relates to addressing Covid-19 diagnostic or therapeutic interventions. We received notification from NICE that assessments for nine rare condition medicines have been, or are likely to be, paused.

Regarding the supply chain for prescribed medications, 1 in 5 rare condition patients or carers report disruption to access that has either been temporary or has required a switch to an alternative medication.

Since the pandemic started, has one of the medicines or treatments needed for the rare condition been unavailable?

Data from EURORDIS Rare Barometer Covid-19 survey



There is evidence that some NHS Trusts have effectively deprioritised patients with rare conditions as resources have been diverted to patients who have contracted Covid-19. Such resources include anti-bacterial ventilator filters which would normally be provided to individuals with progressive muscle-wasting conditions who are reliant on ventilators part or full time.

The widespread discontinuation of clinical trials due to Covid-19 has further impacted people affected by rare conditions. These disruptions directly impact those patients enrolled, meaning that for some with progressive conditions the opportunity to participate may be lost entirely. There will be negative impacts on the availability of research funding that will include third sector investment in biomedical research and early trials.

Remote care

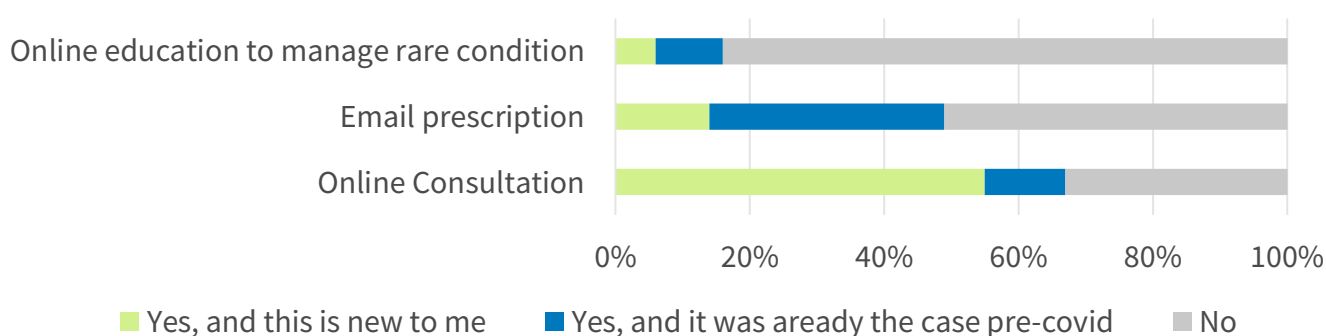
‘Unless there are clinical or practical reasons, all consultations should be done by telemedicine.’

Matt Hancock, Secretary of State for Health (England)

During the Covid-19 pandemic all four nations of the UK have moved to adopt phone or video consultations in order that patients can consult with healthcare professionals without risking exposure to the virus at clinics and hospitals.

Since the beginning of the pandemic and in relation to your rare condition care have you experienced the following?

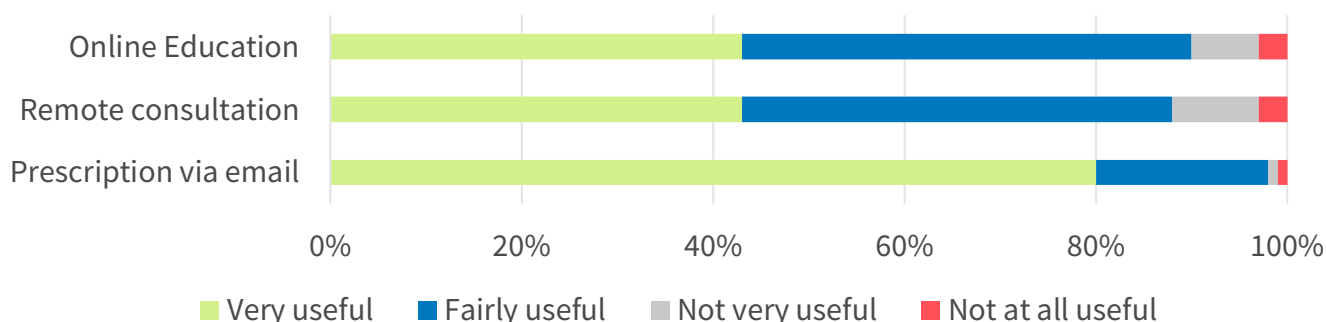
Data from EURORDIS Rare Barometer Covid-19 survey



The findings of the Rare Barometer Covid-19 survey indicate that the proportion of UK rare condition patients attending online consultations has increased markedly since the beginning of the pandemic, rising almost seven-fold, with 9 out of 10 patients who have received care in this way rating the experience positively (very or fairly useful).

How would you qualify your experience with:

Data from EURORDIS Rare Barometer Covid-19 survey



This rapid and necessary change has brought both benefits and significant disbenefits to our community, reflecting its heterogeneity. Many service users have been calling for more access to telemedicine for a portion of their clinical appointments, usually because of condition related challenges with travelling or the expense and inconvenience of travelling. This portion of our community has found that their needs have been met due to Covid-19 emergency measures, and would be sorry to see these gains rolled back when services revert to their pre-Covid-19 state.

We know of no part of our community who would want no more face-to-face clinics though. While it is possible to deliver some types of ongoing care remotely, and some home testing is possible where families have the necessary equipment, full examinations can't be done remotely. Initiating new medicines / treatments or continuing treatments that require careful monitoring are also not supported by remote consultations. Some clinicians are reluctant to share test results over the phone. Building new relationships through remote consultations is more difficult than during face-to-face consultations as they do not lend themselves so easily to nuanced conversations.

Telemedicine does not remove all barriers to accessing healthcare, it can also create new barriers which lead to inequity. Patients need phones, computers and Wi-Fi to access remote consultations, and not everyone will be able to afford or access these goods and services. For some, sensory or cognitive disabilities may mean remote consultations are intrinsically more difficult or even impossible.

Case Study: Alström Syndrome UK

The Highly Specialised Service for Alström Syndrome at Birmingham Children's Hospital switched to a telemedicine service. Normally running 11-12 clinics a year, the service postponed appointments from March 2020 in response to the Covid-19 pandemic. In this context telemedicine clinics give families contact with experts that will bridge the gap until clinics resume.

Alström Syndrome UK is developing a telemedicine information sheet with advice for families on preparing for appointments and is helping families get used to the technology involved. They are also working on accessibility and translation services and attending telemedicine clinics to support families. As an interim measure, the remote consultations are clearly beneficial. Families are able to receive diet and nutrition advice and can avoid challenging journeys and risk of exposure to Covid-19. For now, the consensus is in favour of remote consultations, but there are mixed feelings about continuation of remote consultations in the longer term.

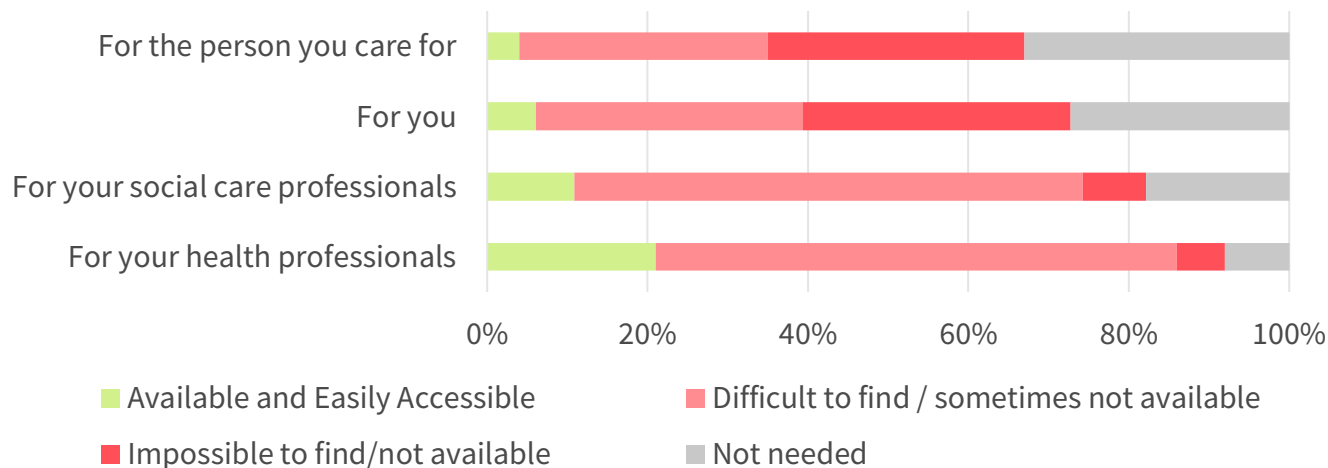
Challenges identified include the impossibility accessing some tests, such as blood tests that would take place during a face-to-face consultation. Many families do not have the equipment necessary to take blood pressure or weight measurements at home. In this way, the value of the consultation is constrained or limited.

There are undoubtedly advantages to remote consultations, particularly where these supplement face-to-face consultations and it is important to capture and retain these benefits. At the same time, we are learning from the experiences of our community that telemedicine cannot yet, and most probably never will, entirely replace the benefits of being physically in the same room as your healthcare professional.

Access to personal protective equipment

How available is personal protective equipment?

Data from EURORDIS Rare Barometer Covid-19 survey



A minority of respondents to the Rare Barometer Covid-19 survey who require access to personal protective equipment (PPE) for themselves, the person they care for or for care professionals coming to their home, have found ready access to the equipment they need. Around 60% report that PPE is difficult or impossible to source for themselves and for the professionals providing care in the home setting. Around half of those who have lost some or all access to care at home report a lack of PPE as the primary reason.

NHS England's guidance on Direct Payments for Healthcare places the responsibility for managing the risks for carers on Clinical Commissioning Groups (CCGs), rather than the family budget holder. During the pandemic families have been left to deliver this CCG responsibility, creating potential risks for themselves and carers coming into their home.

'I fear for Cameron [my son], my family and my carers. If the authorities appear to be unconcerned about something as fundamental as PPE, what will be their attitude about everything else?'

Karen, a mum of a young adult affected by a rare genetic condition, shares her experiences of Covid-19.

Covid-19 Treatment

Those patients who have rare conditions must not be discriminated against in access to Covid-19 treatment and care. In its first iteration, the NICE Covid-19 guideline on critical care said that all adults on admission to hospital should be assessed for frailty using the Clinical Frailty Scale (CFS) and that the score should be used to guide decisions about access to ICU and ventilation. Application of this score to people with disabilities is highly problematic and Genetic Alliance UK welcomed the subsequent update of this guideline to stipulate that the CFS should not be used to assess those under 60 years old.

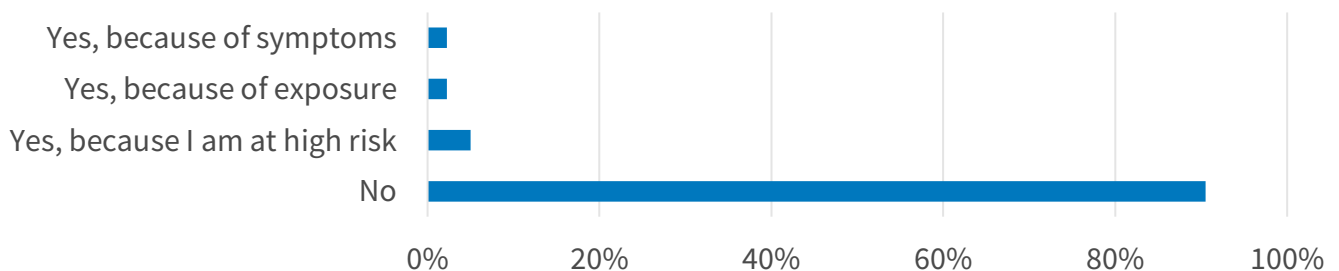
This example demonstrates how unintentional discrimination against people living with rare conditions can easily occur without careful consideration and consultation with the community.

The term ‘pre-existing condition’ has been used frequently when describing deaths caused by Covid-19. Members of our community are concerned that this term potentially hides the impact of Covid-19 on our community. We would be keen to understand how many people living with rare conditions lost their lives in this epidemic, and to understand whether their experience of treatment for Covid-19 was affected at all by their rare condition.

We note that this kind of surveillance will be more easily performed, and to a better level of detail, with greater information infrastructure within the NHS for recording specific rare conditions.

If you felt you needed a Covid-19 test, were you able to access one?

Data from EURORDIS Rare Barometer Covid-19 survey

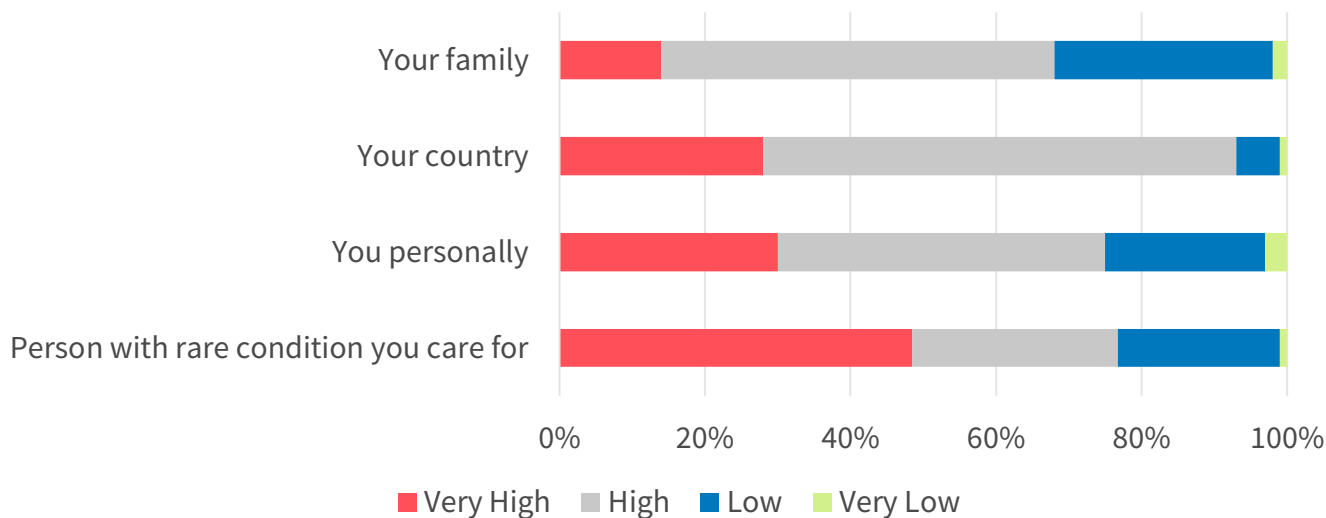


Of those members of our community that wanted themselves or their family member to be tested for Covid-19, 91% could not access a test.

WIDER IMPACT ON PEOPLE LIVING WITH RARE CONDITIONS

What level of threat do you think Covid-19 poses to:

Data from EURORDIS Rare Barometer Covid-19 survey



Alongside healthcare challenges, the community of people living with rare conditions has had to face isolation and withdrawal of care and education support. All of this has been made much worse, by the genuine fear the community has faced from the virus itself. More than half of respondents thought that the person in their family with a rare condition was at very high or high risk from Covid-19.

Access to Educational Support

The closure of schools has placed significant pressure on the parents of children with special educational needs and disabilities (SEND). Those children who receive SEND support at school but do not have an education, health and care (EHC) plan, will have stayed home. Among those who do have an EHC plan will be those who, due to the nature of their rare condition, are vulnerable to Covid-19 and these families will have been shielding. This means that only a minority of children with rare conditions and SEND will have been attending school.

In England, the obligations on Local Authorities (LAs) were reduced on 1 May 2020 such that those unable to deliver the usual level of support are required to make 'reasonable endeavours' to meet their obligations, though the level of 'reasonableness' is not prescribed. This has meant that some LAs and CCGs have delivered better than others, with some children losing either all or a substantial proportion of their education entitlement.

The reopening of schools: The timing of reintroduction of children to school will require careful and tailored planning and support. For those returning there remains considerable concern regarding safe staffing levels, particularly for children who require one-to-one support. For some children with SEND, maintaining routine and consistency is crucially important to their health and wellbeing. An abrupt re-introduction, for example where a child has sensory needs or challenges, could have considerable negative impact.

Living with shielding

The main form of protection for people with rare conditions during this pandemic has been either to shield or to stay at home as much as possible and take particular care to minimise contact with others. These two approaches are largely the same, but the distinction arises from whether a person living with a rare condition was placed in the 'clinically extremely vulnerable category' (those advised to shield) or the 'clinically vulnerable category' (those advised to stay at home as much as possible). Shielding came with government and NHS support to isolate, the other group received support only from volunteers.

This arrangement has caused a great deal of worry and confusion in our community, for a great number of reasons:

- Guidance in the four nations of the UK has been different, and has been changed at different times, sometimes suddenly, on the basis of opaque reasoning. The announcement of 22 June was an improvement in this regard, coming in advance of the change and with relatively clear justification.
- Letters conferring the 'extremely clinically vulnerable' status came late, or not at all, and covered different sectors of our community unevenly. People with the same condition received different advice in different parts of the country.
- The distinction between the two tiers of people told to protect themselves has led to those in the less protected tier feeling abandoned and isolated without support.
- Many elements of real life have not been covered by the guidance – families with a child who must be shielded have not had clear guidance on what to do about a parent needing to go to work, or a sibling being invited back to school.
- The basis for the categorisation of conditions' and patients' risk levels has been slow to be explained.

To an extent some of these challenges are beginning to be addressed, but too late, and after families have faced the extremely difficult challenges of shielding themselves without an adequate explanation as to why and how. If shielding must be reactivated, it is crucial that messages are clearer and communicated more effectively.

For those with rare conditions that confer high risk from Covid-19 who wish to continue protecting themselves through isolation, the support mechanism established should continue to be provided – no one should be forced to stop their protective isolation.

Stepping down from the pandemic

More than 78% of 132 of our members (charities and voluntary groups supporting people affected by rare, genetic and undiagnosed conditions) believe that this announcement came at a time when it was too soon to lift strict shielding advice in England; it would have been safer for those shielding to go out during lockdown.

The step down of shielding advice left people living with rare conditions in a very difficult position. With support measures for those shielding at risk of being stepped down, we run the risk of forcing patients and families out of shielding prematurely.

Lessons need to be learned from the sudden announcement of the lifting of shielding arrangements in England and Wales (June 2020) that left many in the rare condition community feeling increasingly exposed. The inconsistency of approach between nations is concerning and the seeming contradiction of lifting lockdown when at level 4 (Covid-19 in general circulation with transmission high or rising exponentially) has damaged confidence among our community.

The pressures of shielding and continuation of more rigid social distancing are likely to continue for patients with rare conditions and their families for some time after the bulk of our society returns to (near) normality. It is crucial that those with rare conditions are not forgotten or left behind. When a vaccine or other prophylactic treatment becomes available, rare condition patients should be among those with priority access, provided their health condition allows.

Recommendation: It is crucial that those shielding are not forced to lift their shield if they believe it is unsafe to do so. It is crucial that all the measures that local governments and essential services have put in place, remain for as long as our community needs them.

CONCLUSIONS

The pandemic has significantly disrupted the lives of people affected by rare conditions. The overall impact for these clinically vulnerable and clinically highly vulnerable members of our society has been immense and will continue to be felt long after the immediate crisis has passed.

It is hard to look at how this crisis has played out for our community without dwelling on missed opportunities from the past seven years of the UK Strategy for Rare Diseases. If the UK had done more about rare condition data than ‘monitor the development of ICD-11’, could we have communicated to families and patients more quickly and more effectively? Would we know more about ‘underlying health conditions’ and risks for people living with rare conditions from Covid-19?

If care coordination had been delivered for rare conditions in the NHS, would the implementation of telemedicine (mentioned four times in the Strategy document, but not in any of the progress reports) been smoother, and with fewer long-term negative impacts?

At the beginning of 2020 we were already making the case that the UK framework for rare diseases is more necessary now than the UK Strategy for Rare Diseases was in 2013. The damaging experience of Covid-19 to our community only underscores that point.

Our recommendations are broken into three categories, how to better deal with Covid-19 now, how to move out of this crisis period without losing gains or creating more disadvantages, and the longer term.

Continuing to deal with Covid-19

- In monitoring the spread of Covid-19, data should be collected that will enable assessment of the impact, in terms of morbidity and mortality, on people living with rare conditions.
- People living with a rare condition who have a legitimate reason to ask for a test for Covid-19 (either for diagnosis or in order to assess future risk) should be given priority access.
- Access to PPE (personal protective equipment) should be guaranteed for people with rare conditions and those providing care and support to rare condition patients.
- When a vaccine or other prophylactic treatment becomes available, patients with rare conditions should be among those with priority access, provided their health condition allows.

Successful transition from crisis state

- For those with rare conditions that confer high risk from Covid-19 who wish to continue protecting themselves through isolation, the support mechanism established should continue to be provided – no one should be forced to stop their protective isolation.
- A clear and short timetable for the reintroduction of services should be published to give people living with rare conditions clarity as to when they can expect a full service from the NHS.
- The provision of remote consultations should be continued. Care should be taken to integrate telemedicine into routine care practice with the necessary clinical assurance and data protection safeguards.
- Schools and educational facilities should be supported to ensure that they have the necessary flexibility to respond to individual needs and to adapt practices for children with rare conditions.

Learning for the future

- The challenges and failures in delivering letters giving shielding advice should be examined, and actions should be taken following this analysis to ensure the UK is able to rapidly provide tailored public health advice to specific populations with specific conditions.
- The development of the new UK framework for rare diseases has been delayed by this crisis – it is now more necessary than ever. Learnings from this crisis should be incorporated into the new framework before the end of 2020.

