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# Supporting positive patient experiences for rare disease care during disruptive times: findings from a national study

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**ABSTRACT**

**Aim:** We describe the perceptions and experiences of healthcare services during the pandemic of those newly diagnosed with a rare, chronic eye disorder. **Methods:** A cross-sectional mixed-methods study nested within a multi-center inception cohort study. Participants were UK families and adolescents newly affected by childhood uveitis. Using a validated tool (Health Foundation COVID-19 Survey), we captured quantitative (analyzed using descriptive statistics) and qualitative (analyzed using content and thematic analysis) data. **Results:** Responses received from 60 families (September 2020–March 2022), of whom 92% felt comfortable accessing healthcare services, despite 40% reporting challenges in accessing medication. Thematic analysis identified five themes: the value of protected spaces; the positive role of digital health tools, negative experience of immature telemedicine, disintegration of care; and dealing with uncertainty. **Conclusion:** Our findings will support ongoing developments in care with an aim to making services more robust to future periods of disruption.

**Plain language summary:**

**What is this article about?:** In this study, we explored how families with children newly diagnosed with a rare eye disorder and affected adolescents experienced UK healthcare services during the COVID-19 pandemic. We collected information from 60 families between September 2020 and March 2022 using a survey tool.

**What were the results?:** Despite some challenges in getting medications, most families felt comfortable accessing healthcare services. Interestingly, our findings suggest that these families had more confidence in accessing healthcare during the pandemic compared with the general population. We identified five main themes from responses: the importance of safe spaces for care, the positive role of digital health tools, the negative experience of telemedicine when it wasn't the right tool for what was needed, the breakdown of care coordination and how families dealt with uncertainty.

**What do the results of the study mean?:** While families with children facing rare health conditions seemed to navigate healthcare well during the pandemic, improvements in telemedicine and overall

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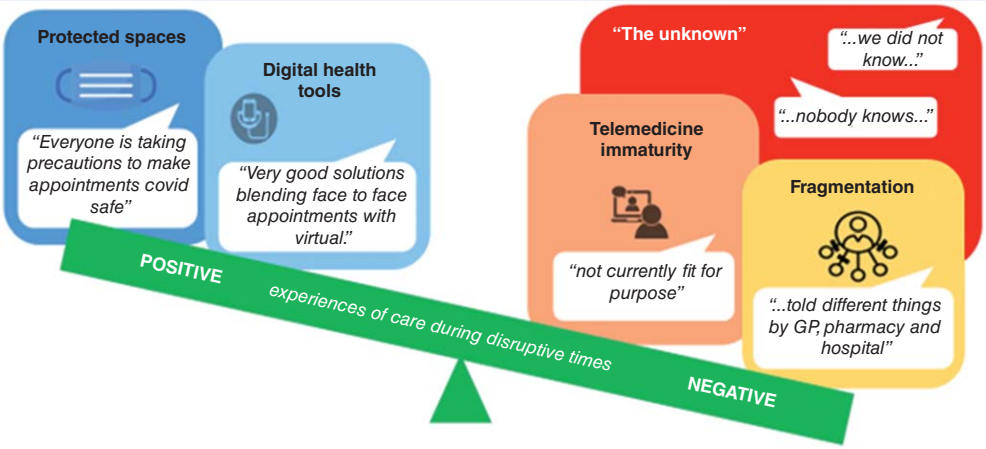
healthcare processes are essential. Learning from the experiences of those with rare diseases can help create better healthcare services for these populations in times of disruption. It is crucial to ensure that the entire healthcare team has the necessary information to support these families effectively.

**TWEETABLE ABSTRACT**

When the going gets tough, rare gets left out…but findings from a childhood eye disease study could improve how we care for rare disease in times of national and global disruption.

**GRAPHICAL ABSTRACT**

Thematic analysis of responses from n = 60 families of children with newly diagnosed, rare, chronic inflammatory eye disorder:




## 1. Background

The COVID-19 pandemic disrupted all elements of global healthcare services [1]. Lessons learnt from positive and negative patient experiences during this disruptive period, and the service-specific factors associated with those experiences, have been disseminated across care settings. This provides evidence to develop more robust services ahead of the further periods of healthcare disruption predicted by many [1,2]. Health service planning is typically informed by a patient population predominantly comprising older people, with common disorders, many of which are associated with aging. To be appropriate, redevelopment of robust services should be informed by the voices of two groups who are often overlooked: namely children, who, for example, comprise 1 in 5 of the global population [3] and those with rare disease, who collectively comprise 1 in 17 individuals [4].

Uveitis encompasses a family of rare disorders characterized by intraocular inflammation with or without accompanying systemic disease [5]. Uveitis of childhood onset is further characterized by chronicity [6], frequent contact with health service [6] significant (visual and quality of life) morbidity [7] and the need for immunosuppressive treatment and highly specialized multidisciplinary car [5,6,8] Children are usually diagnosed in secondary or tertiary care, may require input from quaternary care centers [5,6,8], use pharmacy or blood testing services in the community [6], depend on other primary care services for co-ordination of care and need hospital emergency services for disease flares or complications of immunosuppression [6,7]. These children and their families are well placed to describe experiences of interacting with and between different levels of healthcare. This study aimed to describe and explore the perceptions and experiences of healthcare service use by those children newly diagnosed in the UK with uveitis during the time of significant disruption caused by the global COVID-19 pandemic.

## 2. Materials & methods

We undertook a mixed-methods cross-sectional study embedded within the Uveitis in Childhood Prospective National Cohort Study (UNICORNS) [9]. This is a multicenter inception cohort study recruiting children newly diagnosed with non-infectious uveitis since March 2020, with recruitment active across 32 National Health Service (NHS) Hospitals. Within UNICORNS, participants are identified at diagnosis by the collaborating managing ophthalmologist. Following informed consent, data are collected from clinical records and directly from participants using validated patient reported outcome metrics.

### 2.1. Study participants

Families of children aged up to 18 years and newly diagnosed with non-infectious uveitis were invited to consent

to their or their child's participation. Young people aged 16 and over were invited to give their own consent to participation.

### 2.2. Data collection tool

The Health Foundation COVID-19 Survey was selected for use as a tool developed, piloted and validated by the Health Foundation, (a health policy non-governmental organization) [10] to evaluate the effect of the pandemic on the perceptions and experiences of health service use by members of the UK public. Its use therefore also allows comparison with the general UK population. The first section measures the impact on health and wellbeing and the second measures experiences of using NHS services and perceptions of how services are managing. The survey allowed a convergent mixed-methods design, with concomitant collection of quantitative (closed text responses) and qualitative (free text responses) data. The survey was adapted for use in this study (Supplement S1), with respondents being asked about experiences and perceptions of healthcare use for their child's, or their disorder(s) during the time around diagnosis. The young person version (Supplement S1) was further adapted so that the questions on 'Experience of using NHS services during the pandemic and perceptions as to how services are managing' was limited to their experience of eye care centers.

### 2.3. Survey administration & data collection

The questionnaire was sent via post to all eligible participants following recruitment into the UNICORN study. Children aged older than 14 years were asked to complete the survey themselves, while for younger children the survey was completed by the parent/carer. A member of the study team (SK) manually entered responses into study databases with entries reviewed by a second investigator (ALS).

### 2.4. Analysis

Quantitative data from closed ended questions were analyzed using descriptive summary statistics. Qualitative analysis was then applied to the responses to open ended questions in the survey. Firstly, content analysis [11] was undertaken on responses to open ended questions on the direct impact of the pandemic on health and wellbeing. In order to explore patterns and themes around the perceptions and experiences of healthcare use, thematic analysis [12] was undertaken on free text responses to those questions. Two investigators (SK, a non-clinical research assistant, ALS, a researcher and senior pediatric ophthalmologist) initially immersed themselves with all qualitative responses to gain understanding of the content and attitudes present. Qualitative analyzes were undertaken using Microsoft Excel with color coding labels [13].

For the content analysis of responses on the 'direct impact of the pandemic on health and wellbeing', two coders (ALS, SK) worked together in an iterative, negotiated process to develop a coding system to summarize the data. Following the drawing of initial conclusions around categories of data, data were re-interrogated to identify any characteristics not captured within those categories.

The thematic analysis of 'perceptions and experiences of healthcare service use' was undertaken using a hybrid of deductive and inductive approach, whereby the data were coded and categorized deductively as 'positive' or 'negative' experience and by generating new codes inductively to capture the issues emerging spontaneously from the data. This approach was used to condense data into meaningful categories and applied iteratively to develop and label the key overarching themes.

Themes were generated on a non-exclusionary basis, with some participants' comments able to provide data which belonged to more than one theme. All steps of the analysis were conducted independently by both researchers with peer discussion on completion of every step. Initial codes for labeling the data and emergent themes were identified independently by the two coders (ALS, SK) and then compared and agreed through wider group discussion (SK, ALS, VT). The resultant overarching themes were then reviewed to examine fit with the data, and to identify any characteristics not captured within the thematic map, with themes revised or relabeled as needed.

### 2.5. Patient & public involvement

Our patient research partner group, the Childhood Uveitis Study Steering Group (established in 2019 to support the UNICORN study) [14] was involved in the study's design, conduct and interpretation of the data.

### 2.6. Ethical disclosure

Ethics approval for this work was granted by the Health Research Authority London – London – Bloomsbury Research Ethics Committee, REC reference 20/LO/0661. Written consent was granted by participants or the parents/legal guardians of participants, including consent for use of anonymized responses. The researchers have followed the principles outlined in the Declaration of Helsinki for all human or animal research.

## 3. Results

Of 113 families first approached to take part in the survey, 53% responded. The analysis therefore included data

from 60 respondents (45 parents or carers, 15 young people) representing uveitis cases diagnosed between 1st March 2020 and 20th November 2021. Most participating families were from a white ethnic background, with over-representation (relative to national demographics) of those owning their homes or having two or more cars (Table 1). Similar demographic patterning was seen among those UNICORN cohort study participants who did not respond to the C-19 survey. However, there was a higher rate of missingness in returned demographic data among those cohort study participants who did not, versus those who did respond to the C-19 survey (3/60, 5%, versus 18/53, 34%; $\chi^2 = 15.5$; $p < 0.001$).

### 3.1. Impacts of the pandemic on health & wellbeing

Most respondents expressed concerns over the direct or indirect impact of the pandemic (Table 2), and more than half reported difficulty accessing basic food or household items. Compared with the general public [15], a higher proportion of responding families reported challenges in accessing essential medication, and in communicating with friends and family (Table 2).

Content analysis of open-ended responses revealed seven categories of concern around the health of the respondent and their family, specifically: the direct risk of contracting COVID-19, the lack of exercise, closure of leisure activities and weight gain. There were concerns that their child was put at additional risk due to their diagnoses:

> *Due to A's immune system being affected…we are concerned for A's health if A was to come into contact with the virus*.

Worries over the impact on mental health were described by many:

> *My main concern about the virus involves the impact of lockdowns and restrictions on mental health*". Families shared treatment specific concerns around the negative outcomes of immunosuppression, perceiving an increased risk of contracting SARS-Cov2, and increased risk of poorer outcomes following COVID infection:
> "*With B's immune system compromised due to the medication it's scary*"
> "*we are fearful C could get more ill or may not be able to fight virus if infected…*"
> "*…D is on methotrexate & I'm very concerned that it's…easier to catch the virus….*

The final category of concern was around the impact of the negative financial impact of the pandemic on wellbeing, with expectations of negative national economic impact, worsening public services, rises in the cost of living and job losses or poor job security, reported by the majority of respondents.

### 3.2. Perceptions around, & experience of, using NHS services

Most families described feeling comfortable when using NHS services (Table 3) during the pandemic for the management of newly diagnosed uveitis, with a lower proportion expressing discomfort with the use of local hospitals, or of accident and emergency departments when compared with the general population.

Thematic analysis of free text responses on perceptions and experiences of healthcare service use, provided in section two of the survey, identified five themes, of which two captured 'positive' (protected spaces and digital health tools), and three 'negative' experiences and perceptions (telemedicine immaturity, fragmentation of care and the unknown; Figure 1).

### 3.3. Positive experiences & perceptions

#### 3.3.1. Protected spaces

Many families and young people expressed gratitude for the precautions taken within hospital based clinical areas, with visible and consistent adoption of safety measures conferring a sense of being protected at a vulnerable time. The phrase 'safe' was used multiple times when respondents were describing positive perceptions on their health service environment:

> *Everyone is taking the right precautions to make the appointments covid safe…*"
> "*…precautions have been taken to prevent a spread of the coronavirus in the hospital*".
> "*Everyone we've encountered has been helpful, calm, informative and covid safe.…(YP, response from young person)*"
> "*a lot of safety measures were taken so I didn't worry about catching COVID while I was there (YP)*"
> "*They {doctors and nurses}…make me feel safe (YP)*

These precautions were, for those who had never before experienced specialist hospital-based services, a positive introduction to spaces which they knew would form a long term 'home' for the care of their chronic condition:

> *Didn't want to go but trust the people that work there (YP)*"
> "*…anyway, the risk of not being seen would be greater*.

**Table 1.** Characteristics of participants[†].

| | C-19 adult n = 45, n (%) | C-19 Child n = 15, n (%) | C-19 non respondents n = 53, n (%[‡]) | 2021 UK general population % |
|---|---|---|---|---|
| *Number of patients* | | | | |
| *Age group of respondent* | | | | |
| Less than 20 years | 0 | 15 (100) | ≤3 (≤6) | – |
| 21 to 30 years | ≤3 (≤7) | – | 5 (9) | – |
| 31 to 40 years | 17 (38) | – | 16 (30) | – |
| 41 to 50 years | 21 (47) | – | 28 (53) | – |
| More than 51 years | 6 (13) | – | 3 (6) | – |
| *Ethnicity of respondent* | | | | |
| White – English, Scottish, Welsh, Irish | 27 (60) | 9 (60) | 36 (68) | 76 |
| White other | 7 (16) | ≤3 (≤20) | ≤3 (≤6) | 6 |
| Black African | ≤3 (≤7) | 0 | 4 (8) | 3 |
| Indian | ≤3 (≤7) | 0 | | 3 |
| Pakistani | ≤3 (≤7) | 0 | ≤3 (≤6) | 3 |
| Bangladeshi | ≤3 (≤7) | 0 | ≤3 (≤6) | 1 |
| Chinese | 0 | 0 | | 1 |
| Asian other | ≤3 (≤7) | 0 | ≤3 (≤6) | 2 |
| Other | 4 (9) | ≤3 (≤20) | 4 (8) | 5 |
| *Gender of respondent* | | | | |
| Female | 39 (87) | 13 (87) | n/a | 51 |
| *Family structure* | | | | |
| Two parent family | 41 (91) | 13 (87) | 36 (69) | 83 |
| Single parent family | ≤3 (≤7) | ≤3 (≤20) | 14 (27) | 15 |
| Other | ≤3 (≤7) | ≤3 (≤20) | ≤3 (≤6) | 2 |
| *Car ownership of parents* | | | | |
| None | ≤3 (≤7) | ≤3 (≤20) | 6 (15) | 32 |
| One | 12 (27) | 5 (33) | 11 (27) | 48 |
| Two or more | 28 (62) | 7 (47) | 24 (59) | 20 |
| *Home ownership status of parents* | | | | |
| Owned | 34 (76) | 13 (87) | 10 (29) | 63 |
| Privately rented | 5 (11) | 0 | 9 (26) | 20 |
| Rented from a housing association or local authority | 5 (11) | ≤3 (≤20) | 4 (11) | 17 |
| Other | ≤3 (≤7) | 0 | 12 (34) | – |

[†]From UK Office of National Statistics database: www.ons.gov.uk/peoplepopulationandcommunity/culturalidentity/ethnicity/bulletins/ethnicgroupenglandandwales/census2021.
[‡]Percentage of all those with data.
Small numbers have been reported as <3 to avoid inadvertent disclosure.

#### 3.3.2. Adoption of digital health tools

Families described positive experience of the adoption of telemedicine and digital health tools (e.g., particularly those supporting synchronous, or live, real-time exchanges between patient and care provider and asynchronous care, where the provider reviews content which has earlier been reported or submitted by a patient) across all levels of care from primary care services to quaternary care services:

> *xxx {digital health application, DHA} service had been very good*"
> "*We contact the GP using xxx {DHA} which works well*"
> "*We've been using xxx {DHA} to contact the hospital team and get responses*"
> "*blending face to face appointments with virtual appointments…diagnosis, treatment and care were not compromised*

A much-mentioned benefit was the ability to share patient generated image data with clinical teams:

> *…send photos if necessary*"
> "*…easy to send photos*.

### 3.4. Negative experiences & perceptions

#### 3.4.1. Telemedicine immaturity

The absence of mature telemedicine systems underpinned many negative experience and perceptions. System immaturity was reflected in multiple ways, including reliance on voice telephone contact only due to the absence of video communication, and systems which had not yet been refined or informed by patient experience and were therefore overly complicated for families to navigate:

> *Telephone assessments…getting seen by GP only telephone was a worry*"

**Table 2.** Impact of pandemic on well-being and daily life of families.

| Area of concern for n = 45 parent responders | Fairly concerned n (%) | Very concerned n (%) | Total expressing concern %, (95% CI) | UK general public levels of expressed concern† %, (95% CI)[14] |
|---|---|---|---|---|
| Risk posed to personal/family health & wellbeing | 21 (47) | 10 (22) | 69 (53–81%) | 55 (53–57%) |
| Risk of knock-on impact to personal/family health & wellbeing | 25 (56) | 7 (16) | 71 (56–84%) | 73 (71–75%) |
| Risk posed to health & wellbeing of the nation | 26 (58) | 27 (12) | 84 (71–94%) | 86 (84–87%) |
| Risk of knock-on impact to the health & wellbeing of the nation | 29 (64) | 11 (24) | 89 (76–96%) | 94 (93–95%) |
| | A little harder % (n) | Much harder % (n) | Total %, (95% CI) | UK general public %, (95% CI) |
| Communicating with friends/family | 25 (56) | 4 (9) | 64 (49– 78%) | 47 (45–49%) |
| Being able to get essential medication | 14 (31) | 5 (11) | 42 (28–58%) | 24 (22–26%)‡ |
| Being able to get basic food items | 26 (58) | 4 (9) | 67 (51–80%) | 54 (52–56%) |
| Accessing green space locally | 12 (27) | 1 (2) | 29 (16–44%) | 33 (31–35%) |
| Being able to get household goods | 26 (58) | 2 (4) | 62 (47–76%) | 49 (47–51%) |

†From the Health Foundation Wave 1 survey of 2102 adults, November–December 2021.
‡Difference in proportions *z* score 2.8; $p < 0.01$.

**Table 3.** Reported levels of discomfort on using health services.

| Health Service | Quite uncomfortable % (n) | Very uncomfortable % (n) | Total expressing discomfort %, (95% CI) | UK general public levels of expressed discomfort† %, (95% CI) |
|---|---|---|---|---|
| GP practice (used by n = 55) | 7 (13) | 6 (11) | 24 (13–37%) | 15 (13–17) |
| Local hospital for a non-emergency or routine appointment (used by n = 56) | 5 (9) | 0 (0) | 9 (3–20%) | 22 (20–24%)‡ |
| NHS 111 telephone services (used by n = 44) | 1 (2) | 2 (5) | 7 (1–19%) | 10 (9–11%) |
| Pharmacies (used by all respondents) | 3 (5) | 0 (0) | 5 (1–14%) | 14 (13–16%) |
| Accident and emergency at hospital (used by all respondents) | 3 (5) | 2 (3) | 8 (3–18%) | 28 (26–30%)§ |

†From the Health Foundation Wave 1 survey of 2102 adults, November–December 2021.
‡Difference in proportions *z* score -2.4; $p < 0.05$.
§Difference in proportions *z* score -3.3; $p < 0.01$.

> "*…frustrated by the complicated appointments system*"
> "*App used for triage makes it more complicated to get a phone appointment.*"
> "*the appointments system [is]…not currently fit for purpose*"
> "[secondary care center]…*did not ask to see daughter's eyes…diagnosis + treatment were delayed*

### 3.4.2. Fragmentation of care

The negative impact of the perceived failures in coordinating care was reported by families and young people, with negative experience of synchronous consultations with primary and secondary care teams without access to clinical information from specialist centers.

> *…told different things about our medicines by GP, pharmacy and local hospital*"
> "*{local} hospital…did not take our concerns seriously and didn't have any knowledge about condition*

Families also reported that primary care teams were often unaware of the urgency or severity of the potential need of their child for care, despite the families attempt to communicate this:

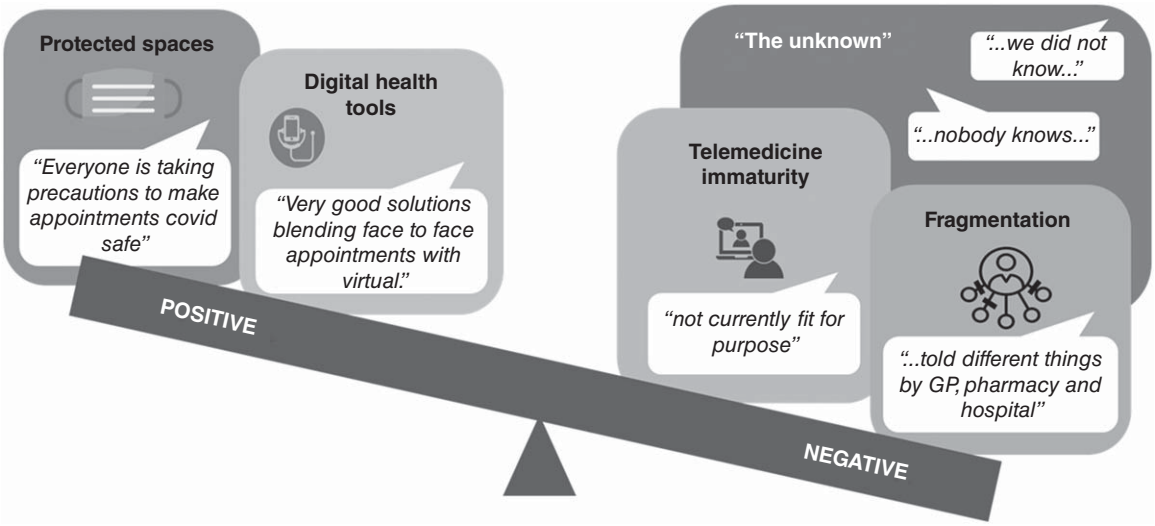



**Figure 1.** Themes around the positive and negative perceptions and experiences of healthcare service use.

*…told no hospital referrals {in} local area at present only urgent referrals"*
*"…seem to fob you off"*
*"…they did not listen to me…*

Young people in particular reported negative experiences of perceived gaps in the co-ordination of care, expressing concerns around the awareness of their primary and secondary care teams of the particular needs of their disorder:

*When I attended the xxx hospital I am not confident with them (YP)"*
*"…this makes me nervous to see xx again as I don't think xx knows what xx is doing (YP)"*
*"…didn't really listen as much…(YP)*

### *3.4.3. The 'unknown'*

The perception that they, their family, the country and the world were navigating uncharted space emerged frequently across responses from parents and young people and across different care settings. Families and children described not knowing how to judge their direct risk of contracting COVID-19, the impact of the use of immunosuppression, how their clinical course and access to care would be disrupted by the pandemic, and the risk to their wider communities. This was often anchored in their understanding of the potential uncertainties ahead with their child's journey through their first years following diagnosis of a complex rare disease.

*…new virus and {I'm} nervous…(YP)"*
*"Because nobody knows the {coronavirus} side effects*

## 4. Discussion

From this mixed methods study embedded in a national cohort, we report that families of children newly diagnosed with a potentially disabling chronic inflammatory eye disease during a time of considerable disruption maintained confidence in the UK National Health Service. They expressed a low level of discomfort in using those services despite concerns around the wider impact of the pandemic. Families' accounts of their experiences included positive experiences of protected spaces, informed clinicians and digital health tool implementation. However, there were negative perceptions around the impact of telemedicine immaturity on care services alongside perceived or actual failures in the co-ordination of care. The fear of the unknown was expressed by many, with the uncertainty of the rare disease journey ahead compounded by the uncertainties brought by the pandemic. These negative perceptions and experience were an additional burden for families already sharing the same fears around health and wellbeing as expressed by the national population.

Our study is strengthened by the use of a nested approach which took advantage of an already underway inception cohort study. The study provided a contemporaneous insight into perceptions and experiences for UK patients and families embarking on a journey of multi-system, multi-level care for a rare disease. Similar cross-sectional survey approaches have been used successfully in other rare diseases areas with an aim to capture the patient voice [16]. Study limitations include a small sample size and the likelihood of some elements of response bias. Families who are particularly vulnerable to negative experiences of healthcare service, specifically those from socioeconomically deprived or non-White

backgrounds [17,18], are under-represented within the respondent sample compared with the overall UK population. There was an overall decline in population mental health during the pandemic [19], attributed to the distinctive circumstances of societal lockdown related isolation, acute and acute-on-chronic financial strain, concerns around chronic health conditions [20] and changes in household dynamics [21]. This decline disproportionately affected people from a lower socioeconomic background [19]. The experience and perceptions of these families may be less positive, and grounded in different domains, with different lessons which need to be learned in order to develop equitable healthcare service provision during disruptive times. The sociodemographic healthcare access and provision inequities starkly highlighted by the pandemic [22,23] may have resulted in inequitable experiences of health services during the pandemic, which may not be captured by our survey findings because of the under-representation of families from more vulnerable socioeconomic strata, as these families may be less likely to take part in medical research [24] Another limitation was the absence of direct exploration of the determinants of family or care-giver mental health and well-being or their pandemic experiences through in-depth interviews. While the population for this study were affected by eye disease, families did not report specific themes around the additional impact of visual impairment on pandemic experiences. The study is therefore unable to explore the intersectional impact of accessibility concerns on the experiences of healthcare use. Additionally, the study used a survey tool which has not yet been validated for use in those aged under 18 years, with data from young people and parents/carers considered in combination [15]. Future work on the impact of periods of disruption may benefit from the collection of data only from children themselves, to gain a stronger sense of their perspective. Despite these limitations, the findings from our unique cohort show the value of our pragmatic approach. Using free text responses to add nuance to survey collected data, this study has generated useful evidence, informed by the experiences of families with rare disease, for service redevelopment.

The families and young people who participated in this study were all at the start of their rare disease odyssey, having been recruited due to a new diagnosis of childhood uveitis. Conceivably, many of them had little prior exposure to specialist healthcare services, and certainly, most of them would have little experience of Uveitis care. Our findings must therefore be considered in this context. Participants' low level of discomfort in using healthcare services, and positive experiences of protected spaces, informed clinicians and digital health tools may have arisen because of absent or low expectations around levels of service. Negative perceptions around telemedicine immaturity and co-ordination of care may have arisen due to overly high expectations from families new to these rare disease pathways. However, our qualitative analyzes consistently reveal that participants perceive a negative impact from those issues on care outcomes. Additionally, the negative impact of uncertainty is intuitively and evidentially greatest early in the rare disease journey [25,26]. Considering these findings in the context of the participants life course adds power to our conclusions, as the 'ripples' created during the first years of childhood onset disease can coalesce, swell and amplify over the years, with subsequent greater impact on the adult that child becomes.

Uncertainty as a driver of negative patient experience has arisen in other patient populations. Although there are no directly comparable studies for this examination of perceptions and experiences among those newly diagnosed, during the pandemic, with a rare chronic disorder, there are sufficiently similar studies involving pediatric care. For families of children with severe neurodisability, the clinical, financial and social uncertainties of the lockdown imposed in the UK in response to COVID-19 had a negative impact on mental health and anxiety [27]. These concerns were however reported to investigators by clinicians rather than by the families themselves. Families who did take part in another study, which used a focus group approach (with 20 participants) to gather qualitative data on experiences of healthcare, again reported on the negative impact of uncertainty, for example "*[clinicians], they didn't have any information themselves, so we were all just winging it and just doing what we felt was safe for our own, for our own children*" [27]. There was evidence of negative perceptions around telehealth, with participants stating that remote consultations were unable to replicate face-to-face contact with regards to 'proper' management of disease. Conversely, positive experiences of the convenience and accessibility afforded by telehealth were reported, alongside hopes that these services would remain accessible beyond the pandemic [27]. These positive experiences with telemedicine were also reported across the 20 families of children with complex medical conditions who took part in a US qualitative study [28]. Families within both settings also felt that pediatric healthcare services had been de-prioritized during the pandemic [28,29]. As these studies recruited families and young people at all stages of their disease course, they could not speak directly to the experience of those in the vulnerable phase of being newly diagnosed, although there were findings suggesting that parents of children who received their diagnosis during the pandemic were particularly affected. When considered alongside our findings, the accumulated evidence suggests a

real need for changes to the services provided for children with rare and or complex disorders to ensure that high quality, patient centered care pathways are robust to unpredictable but arguably inevitable disruptions to service delivery.

A key difference between this cohort and the general UK population, when it came to healthcare related experiences, was the greater proportion of Uveitis patients reporting difficulties in accessing medication. This may reflect the greater vulnerability of supply chains for medications for rare disease. Even during 'uneventful' times, access to essential medicine for rare disease patients can be challenged by the limited production of 'orphan' medicines, limited distribution and a limited number of pharmacies able to dispense, and the limited pool of informed clinicians able to prescribe [30,31]. These limitations are further exacerbated by evidence gaps which prevent the development of guidance and implementation recommendations for the commissioning of services for patients. Additionally, all these obstacles are particularly true for children affected by rare disease.

The adoption of face masks, protective personal equipment (PPE) and social distancing across hospital services was recognized and welcomed by families. Our findings on the feelings of safety expressed by families attending hospital appointments may offer vulnerable patients reassurance and encourage attendance during future pandemics. While the adoption of digital interventions during the pandemic was perceived as supporting some aspects of care, with health tools allowing families to share media or request actions or information directly from care providers, other aspects of care were not well served. Platforms used to organize and deliver telephone-based consultations, and the absence of video-based consultations to replace face to face appointments were perceived poorly by families seeking primary healthcare services for their child. The additional information conveyed through visual communication may be necessary to facilitate understanding for discussions around complex or rare disorders. This disadvantage has also been noted by other primary care providers in the context of telehealth consultations for a range of disorders, with reports of 'lack of confidence' in telemedicine platforms, limited access to adequate technology and the negative impact of the loss of non-verbal communication with remote consultations [32]. Although the provision of remote primary care consultations has found some favor with the UK public [33], our findings suggest that pathways enabling synchronous and asynchronous conveyance of visual information (e.g., physical appearance or behaviors) should be prioritized for families seeking care for rare or complex conditions. Additionally, the burden of rare disease often disproportionately falls on those families from socioeconomically and educationally disadvantaged backgrounds, those with limited or no access to the internet, or whose first language is not English [34–37]. Although quaternary care for patients with rare diseases is delivered in specialized centers, the need to devolve safely and effectively certain aspects of such multidisciplinary care to local providers such as general practitioners or community pharmacists remains.

Our findings also suggest that primary and secondary care teams require additional support in delivering and co-ordinating care for those with rare disease. This support is not simply needed during disruptive times. Managing the fear of the 'unknown' will necessitate sharing clinical experience and promoting further research. Answers emerging through these processes will need to be appropriately communicated to patients and their families to best support patients and avoid science misinformation and disbelief. Digital health innovations can be transformative for families with rare disease [38]. While further maturation of information and communication technologies is probably a matter of time, the responses of patients and their caregivers in our cohort emphasizes the core need to ensure optimal communication among healthcare providers. For this reason, it will be important to devise specific patient educational activities through the coordinated involvement of healthcare professionals and dedicated patient groups. An illustrative example from this cohort is the messaging around the protection of children on immunosuppressive agents. Early in the course of the COVID-19 pandemic, concerns around the risk SARS-COV-2 posed to immunocompromised individuals led to advice on 'shielding' those individuals from contact with others to avoid viral transmission. Later on, evidence emerged on the reduced risk of adverse outcomes following COVID-19 infection for children, even those on immunosuppressive therapies [39–42]. While dissemination of this information among specialists prescribing immunosuppression to children was timely, this may not have been the case for primary care providers, with general practitioners, pharmacists and patients appearing to give and receive conflicting advice. One approach to address this challenge will be empowering parents/carers to use their expertise to as advocates for the care of the child, and for health professionals to recognize that parents are 'experts' in many aspects of their child's care [43].

The need to restructure clinical pathways in response to the pandemic created opportunities to test the feasibility of novel approaches in healthcare provision [44]. These approaches could help increase throughput while dealing with clinical backlog in the aftermath of the pandemic, and serve as a template for dealing with future public health emergencies, with particular refer-

ence to the integration of primary and specialized care services. Appropriately mature and stakeholder attuned digital health tools should be adopted or implemented to ensure communication and collaboration between different branches and levels of the health service, including community services which are often omitted from such services, such as pharmacies. The exceptional circumstances of the pandemic have brought into stark relief the elements of healthcare which require uninterrupted service, and it is the apparently 'outlying' populations which may carry exemplars of best practice, or barriers to achieving good health outcomes. Our findings from this rare disease cohort suggest that these patients should have prioritized access to visual based communication with care providers, and that sources of information should be clearly signposted to patients and community care providers. Additionally, families need to be empowered to be advocates for their child's care e.g., through the use of parent information strategies [32]. These suggestions should inform and support the ongoing post-pandemic redevelopment of care.

## 5. Conclusion

Our findings suggest that while families of children with a rare chronic condition felt comfortable when accessing healthcare services during the pandemic, they experienced greater challenges in accessing essential medication. There were negative perceptions around the impact of telemedicine immaturity on care services alongside perceived or actual failures in the co-ordination of care. We call for more robust health services for such populations in future times of disruption, and for developments in telemedicine to be directly informed by the experiences of those with rare disease, as well as the other stakeholders across the primary, secondary and specialist care settings. The significant negative experience of these families is often driven by uncertainty, suggesting that the development of new healthcare processes which empower families, or which ensure the whole healthcare team has adequate information to support families, should be prioritized.

Article highlights

- Overall, despite challenges, families of children with rare disorders generally felt comfortable accessing healthcare services, with both positive and negative experiences highlighting areas for improvement in healthcare delivery.
- A higher proportion of them faced challenges in accessing medication for their childhood onset rare disease compared with the general public.
- Families were grateful for safety measures in hospital-based clinical areas, and the consistent adoption of precautions provided a sense of protection.
- Benefits of telemedicine and digital health tools included synchronous and asynchronous care and the ability to share patient-generated image data.
- Issues around telemedicine included over-reliance on voice telephone contact, difficulties with navigating complicated systems and fragmentation of care.
- The 'Unknown' was a key driver of negative experiences, specifically uncertainties about the direct risk of COVID-19, the impact of immunosuppression and the future of their child's health journey.
- Our findings emphasize the need for mature telemedicine systems informed by patient experience.
- We call for improvements in care coordination to avoid discrepancies in information.
- We stress the importance of addressing uncertainties to provide better support for families dealing with rare diseases.

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## Author contributions

S Kellett: data curation, formal analysis, project administration, resources, visualization, writing – original draft, writing – review & editing; V Tadic: formal analysis, writing – review & editing; H Petrushkin: investigation, resources, writing – review & editing; J Ashworth: investigation, resources, writing – review & editing; A Connor: : investigation, resources, writing – review & editing; E McLoone: investigation, resources, writing – review & editing; S Sharma: investigation, resources, writing – review & editing; E Agorogiannis: investigation, resources, writing – review & editing; P Watts: investigation, resources, writing – review & editing; E Hughes: investigation, resources, writing – review & editing; A Ritchie: investigation, resources, writing – review & editing; R Pilling: investigation, resources, writing – review & editing; J Benzimra: investigation, resources, writing – review & editing; C Marsh: investigation, resources, writing – review & editing; D Pharoah: investigation, resources, writing – review & editing; J Choi: investigation, resources, writing – review & editing; AD Dick: methodology, investigation, resources, writing – review & editing; JS Rahi: methodology, investigation, resources, writing – review & editing; AL Solebo: conceptualization, data curation, formal analysis, funding acquisition, investigation, methodology, project administration, resources, supervision, validation, visualization, writing – original draft, writing – review & editing.

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## Ethical conduct of research

Ethics approval for this work was granted by the Health Research Authority London – London – Bloomsbury Research Ethics Committee, REC reference 20/LO/0661. Written consent was granted by participants or the parents/legal guardians of participants, including consent for use of anonymized responses. The researchers have followed the principles outlined in the Declaration of Helsinki for all human or animal research.

## Data availability statement

The participants of this study did not give written consent for their data to be shared publicly, so due to the sensitive nature of the research supporting data is not available.

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