

The Rare Covid Experience: Systemic and Health Factors, Coping, and Support Among  
Adults With Rare Diseases During Covid-19

by  
Annelise V. Hartinger

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## AN ABSTRACT OF THE THESIS OF

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Abstract approved: \_\_\_\_\_

Kathleen Bogart

Objective: There are currently 6,000-7,000 recognized rare disorders, affecting approximately 1 in 10 Americans. While each rare disease is unique, the experience of having a rare condition is relatively common. It is this common experience of having a rare disease during the Covid-19 pandemic that we aim to represent with this present study.

Method: We used an inductive qualitative analysis technique to analyze the responses of 9 open-ended questions from our 759 participants, representing 231 unique rare diseases.

Results: At the end of our analysis, we grouped responses into 20 codes, which were further consolidated into 3 themes: systemic and health factors, coping, and support. One of the most common experiences of the rare disease community during Covid-19, according to our study, is reduced access to healthcare. Our results also show a clear link between support and coping; those who have the necessary support could cope better during the pandemic, and vice versa. Finally, many participants hoped that Covid-19 would highlight inequalities, especially among the rare disease community, both in intangible ways such as greater understanding, and more tangible ways such as policies aimed to improve the quality of life of the rare disease community.

Keywords: rare disease, Covid-19, healthcare, support, coping

Corresponding e-mail address: [annelisehartinger@gmail.com](mailto:annelisehartinger@gmail.com)

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APPROVED:

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Kathleen Bogart, Mentor, representing Psychology

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Linda Richards, Committee Member, representing History

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Brooke Bryson, Committee Member, representing Psychology

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Toni Doolen, Dean, Oregon State University Honors College

I understand that my project will become part of the permanent collection of Oregon State University, Honors College. My signature below authorizes release of my project to any reader upon request.

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Annelise V. Hartinger, Author

## **The Rare Covid Experience: Systemic and Health Factors, Coping, and Support Among Adults With Rare Diseases During Covid-19**

A rare disease is a condition that affects a very small minority of individuals. In the US; a rare disorder was defined in the Orphan Drug Act of 1983 by Congress as a condition that affects fewer than 200,000 people in the country (NIH, 2021). However, different countries have varying definitions of a rare disease, like in Europe a rare disease is defined as a condition that affects 1 person per 2000 (Orphanet, 2012). For this study, we are following the US definition of a rare disease because with definitions similar to the European one, a disease can be rare or common depending on the population size you are comparing it to.

While an individual rare disease is uncommon, the prevalence of rare conditions as a whole is not. Currently, there are six to seven thousand recognized rare disorders, but as new diseases are discovered and added to medical literature, this number continues to rise (Orphanet, 2012). In America, there are 25-30 million people living with a rare condition (NIH, 2021). Each rare disease presents its own unique challenges, but members of the rare disease community face many similar problems that arise from having a condition that is not well understood.

With this current study, we aim to use an inductive qualitative analysis to document the experiences adults with one or more rare disorders had during the early months of the Covid-19 pandemic. We predicted that people with rare disorders would experience unique challenges related to access to healthcare and treatment, social support, and coping. We also hope to use the responses from our participants not only to spread awareness of their unique Covid-19 experiences, but to help inform future decisions and policies to help mitigate the challenges of living with a rare disease, especially during a pandemic.

## **Rare Diseases**

Previous literature has well established that those with rare diseases face unique trials beyond problems directly arising from their condition. Those with rare conditions have a much lower health-related quality of life compared to the able-bodied population, but this finding remains true when compared to individuals with common disorders as well (Bogart & Irvin, 2017). Some of the major causes of this decreased quality of life are difficulty getting a diagnosis, limited information on their condition, a lack of treatment options, limited psychosocial support, stigma, etc. (Bryson et al., 2019). Many changes are needed to begin closing the gap in the difference in quality of life between individuals with rare diseases and able-bodied people, including better awareness, social support, access to reliable information, improved access to medical care, etc.

Most rare diseases range from severe to very severe (Eurordis, 2005). They are also often degenerative or life-threatening. Additionally, rare diseases are incredibly difficult to manage with a lack of reliable information, knowledge amongst medical professionals and treatment options. The pain from rare diseases is further aggravated by psychosocial despair, often impacting the patients as well as their family. They are also much more likely to be misdiagnosed because of the rarity of their condition. A survey done by Eurordis (2005) found that 40% of Ehlers Danlos syndrome patients were misdiagnosed, some of them even receiving unnecessary surgery or psychological treatment as a result of their incorrect diagnosis. In the United States, the average individual with a rare condition has to wait 9 years for a correct diagnosis (Bogart & Irvin, 2017).

Outside the medical field, stigma and a lack of support, amongst other factors, further complicate the lives of those with rare conditions (Bogart et al., 2012). Often, the amount of support someone with a rare disease has access to can have the largest impact on their quality of life. Companionship and emotional support were shown to have some of the largest impacts of satisfaction with life amongst the rare disease community (Bryson & Bogart, 2020).

With this study, we aim to expand on previous rare disease literature by focusing on rare diseases in a pandemic situation. We expect that many of the challenges already attributed to rare conditions will be further exacerbated by the Covid-19 pandemic and its effects on society.

## **Covid-19**

While previous research demonstrates many of the unique problems attributed to rare conditions, very few have examined the impact Covid-19 has had on the rare disease community. Many of the studies published about disability and Covid-19 either focus on common conditions or specific rare diseases.

The studies concerning common conditions cover a wide variety of topics such as how telehealth highlights and aggravates barriers to healthcare for the disabled community (Annaswamy et al., 2020) to the trauma Covid-19 has had on disabled people, specifically from healthcare access, ableism in healthcare, and isolation (Lund et al., 2020). Rationing in healthcare, namely ventilators and ICU beds, is, in particular, highlighted as a concern of the disabled community (Andrews et al., 2020). Specifically, this research suggests that ableism,

perceived quality of life, and attitudes towards disabled individuals can influence rationing decisions unfairly.

The studies that have been published about rare disease tend to address multiple implications, similar to this one, but focus on a single condition or situation. One study focuses on the experience of individuals with rare connective tissue disorders, and argues that Covid demonstrates the vulnerability of these patients and steps that should be taken to help protect them in the future (Talarico et al., 2021). Another study focused on the impact Covid-19 has had on patients with rare diseases in Hong Kong, finding that most of them have had trouble accessing services, had their mental health negatively impacted, and had a change in financial status and more (Chung, 2020).

To our knowledge, the most relevant study to our current research was an internet based survey from the National Organization for Rare Disorders. This study was conducted in June 2020, and had 833 participants (71% individuals with a rare condition, and the rest being family members or advocates). This study in particular highlights the major healthcare access issues that arose throughout the rare disease community, as well as employment/financial concerns, and increased risk of Covid (NORD, 2020). They found that 79% of their participants had a medical appointment canceled because of the pandemic, and 62% were concerned about medical supply shortages. 37% of them had been impacted by loss of income, and their study provided qualitative data that the lack of information about Covid-19 affecting rare disorders prevents them from feeling safe. All of these results are major themes in this study as well. However, this study does not address coping or support during the pandemic. And it is gaps like this one that we are aiming to fill in the literature with this study.

## Method

### Participants

Participants were recruited from previous rare disease study participant contact lists and Coordination of Rare Diseases at Sanford. Recruitment information was also shared via email lists and social media links. There were a total of 759 participants. Participants were adults with at least one rare condition, however 212 (27.93%) of our participants had 2 or more rare disorders. 231 rare disorders were represented among our participants, the most common being spinocerebellar ataxia (10.3%), idiopathic hypersomnia (6.4%), Ehlers Danlos syndrome (5.7%), mast cell activation syndrome (5.5%), and narcolepsy (3.7%). The faculty mentor confirmed self-reported disorders were rare according to the NIH definition using their Genetic and Rare Disorder Information Center database.

The average age of our participants is 52.03 years old (SD = 15.39). 571 of our participants identify as female (75.23%), 183 identify as male (24.11%), and 4 reported “other” when asked to identify their gender (0.005%). In regard to race, 686 (90.38%) of our participants identified as White, 18 (0.024%) as Hispanic or Latino/a, 16 (0.021%) as Asian, 10 (0.013%) as American Indian or Alaskan Native, 3 (0.004%) as Black or African American, 1 (0.001%) as Native Hawaiian or Pacific Islander, and 18 (0.024%) as other. The median income among our participants was greater than \$90,001, with 37.7% of our participants falling into this range.

Most of our participants, 649 (85.51%), lived in America when participating in our study. However, 110 (14.49%) of the participants resided in a variety of other countries: 42 (0.055%) were from Canada, 23 (0.03%) from the United Kingdom, 18 (0.024%) from Australia, 5 (0.007%) from Spain, 4 (0.005%) from Germany, 4 (0.005%) from Ireland, 3 (0.004%) from the

Netherlands, 2 (0.003%) from Switzerland, and 1 (0.001%) from each Belgium, France, India, Italy, Mexico, New Zealand, Norway, Portugal, and South Africa.

## **Procedure**

Participants followed a link to the survey administration website Qualtrics, an encrypted, password-protected platform. Data collection occurred between May 6, 2020 and continued for 10 weeks, ending on July 15, 2020. If it was difficult for a participant to enter responses, they were permitted to dictate their responses to another person to enter for them ( $n = 19$  did so). The survey contained a mixture of closed-ended questions regarding quality of life and open-ended questions. The current paper reports on responses to 9 open-ended questions concerning the impact of the COVID-19 pandemic on life with a rare disorder, healthcare access, difficulties experienced during the pandemic, silver linings, coping, and cultural or societal changes that participants hoped would emerge.

## **Analysis**

Thematic analysis, a flexible qualitative analysis approach, was used (Braun & Clarke, 2006). An inductive, data-driven approach was selected due to the lack of existing knowledge and continually developing nature of the pandemic. First, the faculty advisor and second research assistant reviewed all survey responses independently, drafted potential code lists, and discussed until a combined coding scheme was developed. Next, the two research assistants used the coding scheme to code all participant responses. The coders met with the faculty advisor to discuss, resolve coding differences, and revise the coding scheme. Next, they re-coded all survey responses with the refined coding scheme. The final coding scheme contained 20 codes and 3 themes.

## Results

The theme systemic and health factors included codes that were a result of, or hoped for change in an organized system (i.e. healthcare, public health, work places, etc.). The theme coping included codes that reflect how participants felt during and managed the pandemic or activities or distractions that were used to help them carry on during COVID-19. The support theme included codes where participants mentioned any kind of support (emotional, institutional, instrumental, etc.) that helped them or that they were lacking during the pandemic. 2,099 of our 4,707 (44.6%) codes from responses fell into the systemic and health factors theme, 964 (20.5%) of our codes belong to the coping theme, and 1,044 (22.18%) codes make up the support theme. In the following list of themes, I have listed the codes from most prevalent to least within each theme.

### **Theme 1: Systemic and Health Factors**

#### *Healthcare Negative Effect*

This code was our most prevalent, being present in 645 responses, and is defined as a negative change or expected change in healthcare as a result of the pandemic. This includes institutional practices, insurance, pharmaceutical, mental health, etc. Participants often reported inadequate access to healthcare, including canceled appointments and/or treatments, telehealth, and medication shortages.

One participant reports not only having “less access to “non-essential” care like pain management”, but they are “scared about ending up in the hospital from Covid-19, because hospitals are actually kind of a dangerous place for me to be with my rare disease, like having staff enter my hospital room wearing fragrance (which causes anaphylaxis).” This

response demonstrates the rippling effect lacking healthcare access can have on the lives of adults with rare disorders. Not only does this participant lack the access they require for their rare disease, but it is causing them distress, another code discussed later on.

Another participant reported, “It is almost impossible to see my Dr. in person. We have lost supplemental health benefits, so I am not filling all prescriptions.” Not only were many participants unable to access medical providers, but many were also unable to access their medications and treatments. This participant was forced to forgo their prescriptions because of insurance, but others couldn’t access necessary medicine because of medication shortages, supply chain issues, and pandemic restrictions.

### *Values*

This code is defined as hoping for greater societal changes, including individuals or societal treatment of others, change in the government, or change in environmental practices. Participants often focused on greater acceptance and/or understanding of individuals with rare disorders across domains, including society as a whole, on an individual level, or government institutions. These participants also desired greater acceptance of all people across domains and hoped that this pandemic would highlight the need/benefits of protecting the Earth.

Some participants focused on their rare disorder or symptoms when hoping for a change, such as one participant who said that they hope the pandemic “makes people more caring about immunocompromised people”, but many focused on the needs of the rare disease community as a whole, such as a participant who said “I think the specific needs of those with rare disorders will be highlighted and better understood”. Responses such as these

demonstrate the fierce desire many adults with rare diseases have for others to understand them and their unique struggles, and the hope they have placed on the pandemic to spark such a change.

Others focused on values as a whole, unrelated to rare diseases. These responses largely focused on appreciating others, appreciating simplicity, or appreciating the environment. One participant desired “a change in values: more appreciation of friends and family and creativity and simplicity; less attention on material success.”

### ***Public Health***

This code is made up of responses that discuss public health, either current measures or predictions/desires for future measures. This includes following or improving current public health measures, improving public sanitation, planning or preparing for future pandemics, virus research, using masks in non-pandemic times, better personal hygiene, etc. Participants often desired these changes in public health to have an immediate positive impact on the rare disease community, such as increased sanitation making it safer for immunocompromised people in public. Others also hoped for better future public health measures because they felt the one for this pandemic did not adequately protect them or their community.

One of the most common public health responses we saw in our study was a desire for the acceptance of mask wearing to extend beyond the pandemic. For many, this made them feel more accepted and safer, especially if they were immunocompromised. One participant said that they “would like to see people keep distancing and wearing masks during flu season.” Others simply enjoyed wearing the masks, often to hide a difference, such

as one participant who said “I do think a lot about the irony of having to wear a mask. I often wish I could put a bag over my face so people would not see my [facial] paralysis, and now, I can use a mask without people finding it strange I do.”

Another one of the common responses we had in this code was a criticism of people not following current public health measures. One participant said that they have seen “a renewed sense of selflessness. I am disheartened and disgusted at the number of people who refuse to wear a mask.” The people who didn’t follow public health measures also added even more distress for people with rare diseases. A participant said that they “worry for my family and others who do not practice safe distancing or wear a mask to protect others or their families.”

### ***Work/Finance***

Responses in this code include a change in the participants’ work or responsibilities, accessibility, unemployment, engaging in work as coping, or change in income, savings, expenses, etc. This code in particular had a large mix of positive and negative responses. The responses gathered during the pandemic demonstrate how much a rare disease can impact work and how work, in turn, can affect their rare disease.

Some participants greatly benefited from changes in work during the pandemic, such as work from home policies and better accommodations. Many found working or attending school from home allowed them to better address their needs and symptoms. One participant, whose rare disease is a form of sleep disorder, said, “I find myself happier and more productive. My school moving courses completely online allowed me to take a course that I

otherwise wouldn't have been able to take because of my sleep disorder making it difficult to drive and requiring naps.”

Others suffered, either from losing employment/income, or symptoms/stress interfering with their job. As one participant said, “Working from home has been exhausting and hard to set boundaries between work/personal time. This has increased my stress level tremendously.” Another states that they “just returned from Italy where I had medical treatment. I spent all of my savings on my treatment and went into debt. I came home to zero income [because of the pandemic]. I cannot travel to my doctors and therapists. The pandemic has taught me to live one day at a time and to ask for help”

### ***Healthcare Positive Effect***

This code is defined as a positive change or expected change in healthcare as a result of the pandemic. This includes institutional practices, insurance, pharmaceuticals, mental health, etc. Most of the responses in this code hoped for better healthcare options or practices in the future. Other responses in this code result from better accessibility, usually through telehealth, and using healthcare as coping.

One of our most common responses for this group is a desire for universal healthcare. As one participant clearly sums up, “health care is a human right and should be available to everyone.” Many participants hope the pandemic “will shine a light on the inequality in healthcare in this country and will bring about universal healthcare.”

While telehealth was very detrimental for some people, seen in the healthcare negative effect code, for others telehealth was largely beneficial. One participant explains that they “think the positive that came out of this is the easier access to doctors that are far

away. I usually have to drive 360 miles for most of my healthcare, but those doctors have finally started doing telemedicine which has helped me greatly.” Another participant thinks that having telehealth as an option in the future would be very useful because it would be “possible (to use telehealth) in the future if too sick to attend in person.”

### *Symptoms*

Responses in this code include mention of changes or potential changes in symptoms of the rare disease due to COVID risk or restrictions. This code also includes participants mentioning an increased risk of getting COVID or severity of COVID due to the rare disease. Very rarely did the pandemic itself cause an increase in symptoms, unless the participant actually had Covid, but the stress, impacts on healthcare access, and other life impacts it had, did play a major role for our participants.

The limited access to healthcare in particular was one of the largest causes for increased symptoms in our study. One of our participants had to “take a break from the medication I take daily which I need that to stay awake throughout the day” which in turn “meant I have had to give into my illness for this period and my quality of life is not as good.” Additionally, the new challenges of the pandemic also caused an increase of symptoms in our participants. As one participant stated, “It is harder for me having my kids not able to go to school. Having them home all day and having to do homeschooling has caused my pain level to increase without a break. That has been my biggest challenge.” Another said that they “can't go to regular physical therapy for pain control, so I'm in more pain.”

## *Supplies*

The least prevalent code in this theme is described as a change in accessing or ordering supplies, including food, housing, water, etc. This code also includes hopes for change in supply chains in the future. Many participants often struggled to get the supplies they needed, either due to shortages or lack of accessibility, making many go without or rely on others to obtain supplies for them.

Some of our participants were able to benefit from pick-up or delivery systems, but many struggled to get the supplies they needed. One of the participants mentioned that one of their biggest challenges was that “medications/medical supplies are harder to find or much more expensive.” Another participant said that “my usual use of grocery delivery and curbside pickup went haywire, and was unreliable until recently. Shopping for groceries became extremely difficult due to the number of people suddenly accessing it.” One participant who struggled to fill their prescription said “I take hydroxychloroquine and had a hard time finding a pharmacy that had my medication when I needed a refill.”

## **Theme 2: Coping**

### *Distress*

This code is simply defined as participants expressing negative emotions within their responses. The most common types of distress were stress, worry, frustration, and anxiety. These negative emotions not only made living through a pandemic more challenging for our participants, but have several negative effects throughout their lives.

The lack of reliable information, especially about their rare disorder, caused many participants distress. One participant reported a “fear for my well-being given the lack of consistent information, or information on how COVID may affect [rare disorder] patients.” Another said, “it's caused a great deal of anxiety over possible infection and death.”

This stress has had many negative effects for our participants, especially for their health. One said that “the stress from this caused multiple ulcer flares.” Another reported the effects of the pandemic “caused greater stress which causes trickle down physical issues and sleep disorders.” Another one states that “the worst thing is the Covid-19 pandemic has caused stress.... and stress is NOT good for my rare disorder... causing falls, and injury.”

### ***Resilience***

This code was used when a participant’s response included their ability to adapt to challenges, including using gratitude, acceptance, meaning making, focusing on the positive, tenacity, self-advocacy, meditation, determination, etc. Self-care, positive thinking, and determination were some of the most common threads that are a part of this code.

Focusing on positive distractions or information was a common way people stayed resilient. One participant represents this method with their focus on “finding positive news and useful information. Thinking through processes and battling negative thoughts.”

Another frequent response in our data was for participants to acknowledge the bad, but not let it hold them down. One participant said that they “keep on moving forward. It is what it is. No need to dwell on it” Another focuses on “being the type of person that won't give up easily. My life has never been an easy one, so I guess I am used to challenges.”

### ***Physical Activity/Nature***

This code was used when a participant described a change in their physical activity, or access to nature, as a result of the pandemic. This included more negative responses like canceled physical therapy appointments, not feeling safe going outside, and gyms being closed. However, this code also included positive responses about using nature and/or physical activity as a way to cope.

The participants who lack proper access to physical activity or nature often suffered as a result. One participant “did not have [the] opportunity to regularly walk which has impacted my mobility”. Another had their sport practices canceled so “Self care and coping [through] sport was not available leading to anxiety and upset.”

Those who were able to utilize physical activity and nature often were able to cope much better. One participant was able to spend more time outside as a result of the pandemic and said “my husband and I built a hydroponic greenhouse from scrap materials and started a garden. It has been good for physical activity, and it's relaxing.” Another is “keeping a positive outlook, getting outside and walking more, doing yard work with my family.”

### ***Keeping Occupied***

A participant’s response was coded as keeping occupied when they described keeping themselves busy during the pandemic with entertainment, and/or hobbies. This form of coping was usually based on the idea of distracting one’s self from the pandemic with things they enjoy.

One participant is “keeping as busy as I can, doing online courses, doing crafts, too much online shopping (so I have something to look forward to).” Another likes to “watch a lot of mindless TV, preferably comedies, read silly books, and sleep a lot.” One of our other participants distracts themselves by helping others; they are “Keeping busy sewing and making quilts for others.”

### ***Routine***

This code was used whenever a participant set themselves a schedule and/or goals for themselves. This was usually the result of trying to stay productive while at home all of the time, trying to pick up new habits, or trying to utilize a schedule to help manage the symptoms of their condition.

Working from home allowed many of our participants more control over their schedules and could accommodate the needs of their condition more effectively. One participant said that they were “able to work from home some days allowing me to sleep in and nap during lunch or after the work day was over, which ended earlier.”

Others used a routine to help them manage life during a pandemic. Another participant is “Starting and trying to stick to new habits like taking vitamins with my daily medication, using the Nintendo Ring-fit game (a computer game that uses physical activity as an in-game mechanic) to prevent myself from being too sedentary.”

### ***Slower Pace***

This code is defined as having or desiring a slower or more relaxing pace to life for themselves or others. Individuals who mentioned a slower pace in their responses either liked

that Covid got everyone to slow down, or hoped Covid would encourage people to slow down with their lives, which mainly included a better work/life balance.

One of our participants predicted that “people will appreciate simple life and nature and true relationship connections more I think.” Another participant noticed that “the slowing down and staying home has re-engaged families in spending time together, appreciating what we have and maybe even stopped careless spending” and hoped this would continue post pandemic.

### **Theme 3: Support**

#### ***Family***

A participant’s response was coded as family when they experienced a change in support, distraction, companionship, stress, or time spent with family. For this code, we defined family as blood relations and partners/spouses. This code was our second most prevalent code, which demonstrates the large positive, or negative, effect family had during the pandemic.

In our study, the largest amount of support came from various family members, usually spouses. One participant stated that “having a solid marriage and a husband who is my rock has been a steadying force. We have ridden out other crises together and have only come out stronger.” Another participant details how their family has been supporting them, “friends and family have been essential to coping with COVID, for 6 weeks I moved in with a family member that was also isolating, and had no visitors and no trips outside the home. Family members brought our groceries and other items.”

As helpful as family could be, they could also be harmful as well. One participant, who is immunocompromised, thought they had Covid and their “family members made fun of me for taking it seriously.” They also had trouble getting their family to “understand my reasons for self-isolating.” However, those without family support often fared worse. One participant said “I have no family, no support system, - I am in constant pain, struggle to pay for daycare and general bills on my income alone”

### ***Companionship Support***

This code is defined as a change in a sense of belonging and shared enjoyment with others. This includes changing relationships with colleagues, friends, boyfriend/girlfriend, healthcare practitioners, others with rare disease, neighbors, church, pets, etc. Most commonly, this code was used to refer to the support or isolation from friends.

Sometimes the support they received was simple and tangible. One participant relied on their “boyfriend has helped me run errands.” However, most of the time the support was more emotional, especially to avoid feeling isolated. One participant noted that their “friends have also really been there for me when I need them.” Another has “videoconferenced some friends and played games which [have] been fun. Also, I am a part of some support groups on Facebook. They have held some "fellowship" Zoom calls, which has been refreshing.”

However, without this support, many individuals began to feel isolated and lonely. One participant has “been isolating [for] a few month(s), rarely seeing my friends. I feel abandoned by my doctors and am scared that my health will decrease with nobody noticing because I don't get my check ups.” Another has been feeling “Loneliness due to how people

have responded to being required to wear a mask in my state. It feels like no one cares if me or people like me are alive.”

### ***Informational Support***

This code represents a change in availability or reliability of information. This also includes participants changing the amount of news and/or information they are exposed to, or wanting greater importance placed on science, research, or healthcare. News dieting, which is limiting the amount of news you consume, was a very common practice among our participants.

One of our participants was using information as a way to cope. They said that they are “finding positive news and useful information. Thinking through processes and battling negative thoughts.” Many hoped that the pandemic would be used to further research on rare diseases. One participant said they “hope we will learn how to keep those of us with rare diseases healthy through things like this.”

While some people felt supported by available information, many felt let down by it. One participant was especially anxious because of the “lack of solid information about my true level of risk.” Another participant stated that for their coping they “unfortunately, -use a lot of avoidance techniques. I stay off of social media and rarely visit my support group websites. I go to health authority pages for updates and avoid looking at news articles or watching the news.”

## *Emotional Support*

A response was classified within this code if the participant experienced a change in feeling esteemed, or accepted, or believed that others may be able to understand the experience of having a rare disease better as a result of the pandemic (also known as experiential empathy).

Something as simple as feeling “well supported and encouraged” would fall within this code. However, most of these codes related to an increased understanding for the rare disease community. One participant said that it “feels like the rest of the world experienced what it’s like to be chronically ill, and having that understanding from others has been nice. Seeing people recognize that what we go through every day is trauma has been validating and makes me feel less like screaming than I normally do.” Another said that “hopefully, folks will have a better understanding of what it feels like to be isolated and unable to access our world; a reflection of what many of us experience already.”

One of my favorite quotes is from a participant who hoped that Covid finally let others walk a mile in their shoes. They said, “I think people, in general, are gaining a better understanding through the pandemic of what people with underlying illnesses face on a daily basis. Before this, so many people didn't understand why I'm late frequently, or why I can't work, or why I don't "just do [certain things] with my children", as others may do much more easily. I think the COVID phenomenon builds understanding for populations that previously didn't understand how much people like us struggle on so many levels, and therefore may build more empathy and compassion moving forward.”

### ***Instrumental Support***

This code was used when a participant experienced a change in availability of tangible support for activities of daily living, including housekeeping, caregiving, home maintenance, governmental support, etc. For the majority of our responses in this code, it was a loss of support.

One of our participants was frustrated at “not being able to access the help I already had to perform daily tasks.” Another was struggling to accept that they were “depending on others to help me.”

However, a few participants did have better support during the pandemic. One participant in particular is “quite happy that many stores have started offering curbside pick up. I have a great deal of trouble shopping at garden centers because they are so warm, so having a curbside pick up has actually encouraged me to shop more at the garden center. I also have started using the curbside at the grocery store and will probably continue to do this once the pandemic is over.”

### ***Helping Others***

This code is our smallest code and included responses where the participant described providing any kind of support to others. Many of our participants helped others by working with their rare disease support group or organization. One participant spent a lot of their free time “leading an organization focused on my disease.” Another, also with a rare disease group, “continued significant volunteer responsibilities, including leading state wide Zoom meetings, trainings, political advocacy, media interviews.” Others just aimed to fill a need they saw around them, like one participant who spent their time “making activated charcoal

filtered masks for my family and friends, and sharing of scientific information about the spreading and how to avoid it.”

### **No Noteworthy Issues**

This code, while being our second most prevalent at 600 responses, is not included in any of our themes. A response received this code when a participant answered a prompt, but their response indicated they did not experience the challenge mentioned in the prompt, or they felt the challenges they did experience were not the result of their rare disease or were no different from the average person.

For example, when asked about what barriers they faced managing their health and wellbeing during the pandemic, one participant said “My condition has been treated with medication and so for the past five years I have lived a full and healthy life with minimal side effects of the condition.” Because this participant had reliable access to medications that managed their rare disorder, their experience during the pandemic was similar to someone without a rare disease.

In response to a being asked how the pandemic has impacted their life with a rare disorder, another participant said, “Covid-19 hasn’t really changed things - it just seems to make it more difficult to surround myself with positive people.” This participant acknowledged that Covid-19 has had impacts on their life, in their case in regard to companionship and emotional support, but they also identify that these changes are not the result of their rare disorder.

Since our participants cover such a wide variety of conditions, it means they also cover a wide variety of experiences. Some rare diseases have mild symptoms, do not impact

their daily life, do not require regular medical monitoring/intervention, do not impact their immune responses, and so many other factors that can affect the way they experienced the pandemic. As such, we expected that not all of our participants would be impacted by the pandemic as a result of their rare disorder.

## **Discussion**

This study is an inductive qualitative analysis that explores the experiences of adults with rare disorders during the Covid-19 pandemic. As such, this study contributes to the psychosocial literature for both rare disease and Covid-19, and offers valuable information on making improvements for the rare disorder community, both in and outside of a pandemic. Our study demonstrates that systematic and health factors, coping, and support, or the lack thereof, played major roles in the experiences of adults with rare diseases early on in the pandemic. The use of narratives from our participants provide evidence and context to support previous research about the unique challenges the rare community faces, such as studies like Bogart & Irvin (2017), and Eurordis (2005), as well as the new challenges this pandemic caused them. While the pandemic caused obstacles for nearly everyone, rare disease or not, our study supports the additional challenged adults with rare diseases faced, particularly with healthcare access, distress, and a lack of support.

By far, the largest concern that was brought up by our participants was reduced access to healthcare. This took many forms from canceled appointments/treatments, issues getting prescriptions and medical supplies, unreliable information about the pandemic, especially how it affected their particular rare disease, and so much more. These results align very closely with the results of the NORD 2020 study. They also found that healthcare access was

a major concern for the rare disease community, especially with canceled/delayed appointments and medical supply shortages. The similarity of our results to NORD further support the extensive effect the Covid-19 pandemic has had on healthcare for the rare disease community and how this disruption caused waves of impact throughout our participants' lives affecting their conditions, mental health, work, basic life tasks and more.

One clear connection that arose from our study is that participants who were lacking support had a harder time coping. During the pandemic, most of our participants derived their support from family and friends. Other support methods that our participants had previously relied on were often disrupted because of the pandemic. Those with sufficient familial/friendship support often did not experience the same stress, uncertainty, feelings of isolation, inability to complete daily tasks, etc. as those who did not. Having this additional security allowed participants to better cope with many of the added stressors of the pandemic and, overall, have a better quality of life during this time frame. These findings are supported by previous studies that also demonstrate better outcomes in the rare disease community as a result of support, such as Bryson et al. (2020) which found that emotional and companionship support increase satisfaction with life amongst individuals with rare conditions.

While the majority of our participants' responses address challenges or negative experiences, there were some positive experiences or expectations that arose as well. Work from home policies especially seemed to help many of our participants, who often reported feeling less stigma and being able to manage their condition more effectively when able to work from home. Participants also hoped that the current pandemic would improve public health measures, both in and out of pandemics. There was also a great desire for societal

changes. Our participants wanted more understanding and accepting of differences, both from rare disease and otherwise, they wanted society to be more accepting and accommodating of the unique needs of rare diseases, and they wanted better government policies/institutions to recognize or address the unique challenges this community faces. Our participants hoped that Covid-19 would highlight current inequities and inspire the changes they so deeply desire.

The aspects of our studies concerning coping, support, and silver linings are largely missing in the current academic discourse. Most rare disease studies from the pandemic tend to focus on the healthcare and economic impacts of rare disease instead. For example, Chowdhury et al. (2021), a literature review about how rare disease research was being conducted and how the respective conditions were being managed, found similar results regarding the disruption of healthcare access for individuals with rare disorders. As such, it is our conclusions in these areas that make the biggest impact in filling current literature gaps.

In regard to future research, we would recommend further investigation into the link between support and coping, and how we could address support needs of the rare disease community in the future. In this study, the connection became clear from participant quotes that directly cite family or companionship support as the cause for their ability to cope, or people who point out their lack of support as to why they can't cope. We did not ask any direct questions about the relationship between support and coping, so this connection has a lot of room to be further flushed out. Additionally, our study does not address a way to meet the support needs of this community. With proper support in place, individuals with rare disease would likely fare much better in unprecedented and normal times.

## **Strengths and Limitations**

The major limitation of this study is the demographics. Among our participants, people who identify as white and female who are upper-middle aged, and make more than \$90,001 are overrepresented. Since our participant demographic likely varies so much from the demographics of the rare disease community, there could be several unforeseen impacts on our results. Mainly that the over representation of these identities could skew our results towards their average experience, away from the average experience of the rare disease community. Participants who identify as white or upper-middle class likely had greater access to privilege and resources compared to the true population of the rare disease community, which means the experiences represented in our study may be more positive than the reality.

Another aspect of our research is both a strength and limitation. Our data was collected from May 6th 2020 to July 15th 2020. Since our data comes from a small window of time early in the Covid-19 pandemic, it would be impossible to replicate our findings when accounting for all of the developments that have occurred since then. However, our data also represents a moment in time when some of the greatest uncertainty of the pandemic occurred. The effects of this uncertainty is seen throughout our data and offers a rather comprehensive snapshot of arguably the most difficult time from the pandemic.

One of the major strengths of our study is that the researchers of this study also have rare disorders. While formulating, coding, and analyzing this study, our own unique experiences as individuals with rare diseases during a pandemic allowed us to zero in on specific issues often overlooked by other researchers. This study is done by researchers with

rare disorders, of participants with rare disorders, in order to better our community and help close some of the gaps in rare disease literature.

## **Conclusion**

Participants were greatly impacted by the Covid-19 pandemic. As noted in previous literature, those with rare disease often face unique challenges, and this proved to be especially true during Covid. Our participants noted inequalities and problems with systemic and health practices that only add to the challenges of having a rare disease. They also responded how they were unable to cope or what tools they employed to be able to cope with their unique challenges. They also demonstrated the importance of having support through such unprecedented times, and the consequences that result from not having it.

Our study demonstrates how the current pandemic both exacerbated existing problems as well as added new challenges for this rare community. Proper healthcare access, especially in regard to diagnosis and treatment, has always been difficult for the rare disease community, but became nearly impossible during the pandemic. Necessary support has also been a very present challenge in this community, but now that support affected their ability to cope with the trauma and stress of a pandemic as well.

Our study also shows how many of our participants are hopeful that Covid-19 can inspire the changes they so desperately want. They anticipate that the inequities highlighted by the pandemic, especially those affecting individuals with rare diseases, will finally be addressed and greater respect and understanding will be had for everyone. They also hope that emphasizing the flaws in public health and healthcare systems, will prompt the changes needed to improve these systems for everyone.

**Figure 1**

*Graph with the Prevalence of Codes*

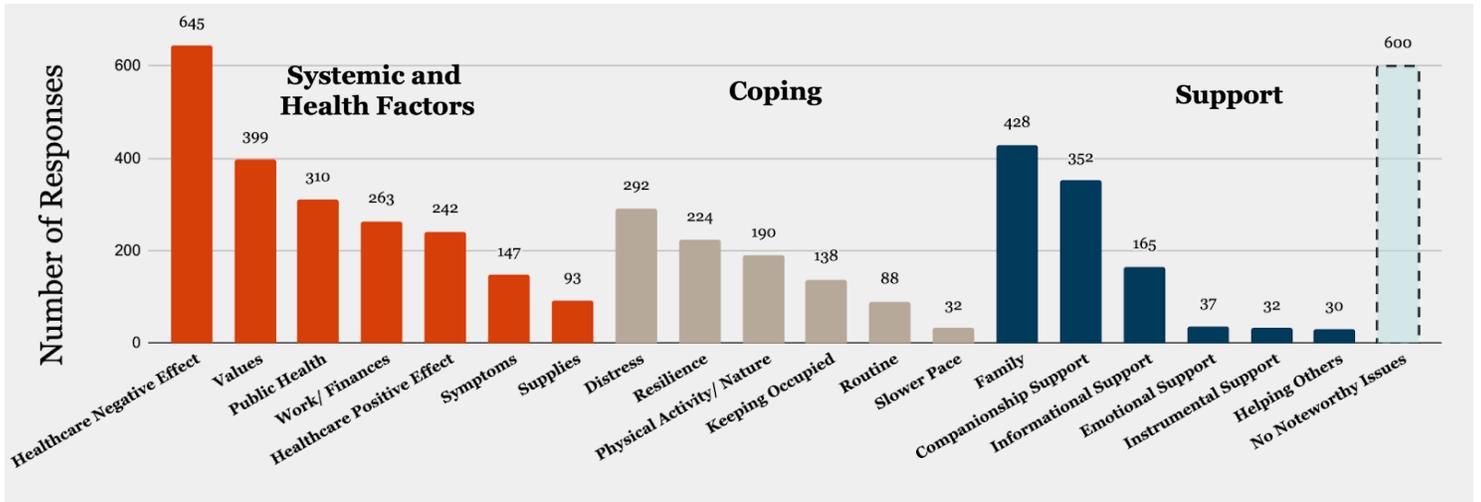
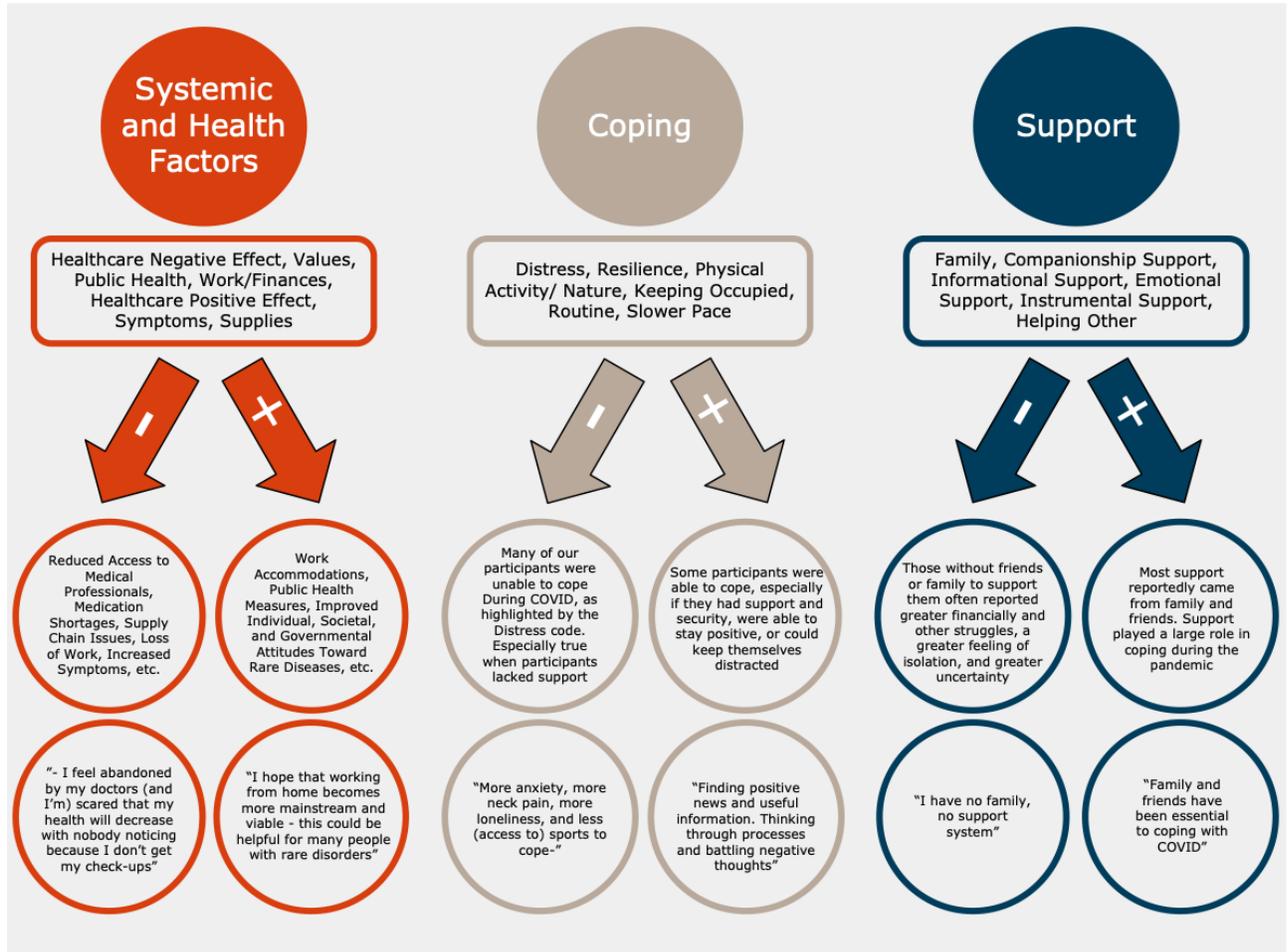


Figure 2

Theme Map



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