# “PUTTING ONE FOOT IN FRONT OF THE OTHER”: PARENTAL RESILIENCE DURING THE COVID-19 PANDEMIC

by

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This manuscript is dedicated to

My sister, Colleen, without whom the world would be a little less bright.

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## ABSTRACT

*Introduction:* During the COVID-19 pandemic, parents of disabled children faced a variety of challenges stemming from loss of services, medical concerns, caregiving stress, and changes to support systems. The purpose of this study was to explore the experiences of parents of children with Smith-Lemli-Opitz syndrome (SLOS), a genetic developmental disorder, during the COVID-19 pandemic, focusing on what factors impacted their ability to develop or maintain resilience. *Methods:* Eight mothers of children with Smith-Lemli-Opitz syndrome participated in this study. The participants filled out an online questionnaire and participated in an online interview. Those interviews were analyzed through thematic analysis. *Results:* Five major themes were identified: 1) Experiences with SLOS, 2) COVID-19 impacts, 3) Support and Services, 4) Parental and Caregiving Strain, and 5) Resilience. *Conclusion*: All of the mothers demonstrated resilience throughout the pandemic and throughout their experience parenting a child with SLOS. The findings from this study contribute to an understanding of what factors serve as protective factors for parents of children with developmental disabilities and provide insight into the experiences and needs of these women during the COVID-19 pandemic.



*Keywords: disability, COVID-19, resilience, adaptation*

# Chapter 1

# LITERATURE REVIEW

## Introduction

During the past two years, individuals and families across the country and throughout the word have experienced the effects of the COVID-19. From social distancing to enforced lockdowns, to sudden homeschooling and quarantines, the impacts of this pandemic have been far-reaching and striking in their intensity. COVID-19 has had a remarkable impact on almost every aspect of life, on every discipline in academia, and on everyone who lives in this society. Studies are coming out at an incredible rate, telling us just how much of an impact COVID-19 has had on a variety of populations, and the disabled community is just one of many vulnerable populations who face increased risk during this pandemic. Individuals with disabilities (Jeste et al., 2020; Chen & McNamara, 2020) and families of children with intellectual and developmental disabilities (IDDS) (Neece & Fenning, 2020; Redquest et al., 2020) face unique challenges during a disaster like this pandemic. Understanding the current and future needs of this population is paramount in identifying what resources need to be available to families during crises such as the COVID-19 pandemic. This study focused on the experiences of parents of children diagnosed with Smith-Lemli-Opitz Syndrome – a genetic developmental disability – during the COVID-19 pandemic in order to understand how they experienced or maintained resilience.

**Smith-Lemli-Opitz Syndrome**

Smith-Lemli-Opitz Syndrome (SLOS), or RSH syndrome, is a congenital, autosomal recessive developmental disability that results in an irregular metabolism of cholesterol due to a mutation and resulting "deficiency of the enzyme 7-dehydrocholesterol (7-DHC) reductase" (Nowaczyk & Wassif, 2020). It is named after the three doctors who identified the syndrome: Smith, Lemli, and Opitz. There is a wide spectrum within this disorder, with symptoms varying significantly depending on the severity of the disability. However, some common symptoms are as follows: behavioral problems, autistic symptoms and/or diagnosis, delayed developmental milestones, intellectual disabilities (Thurm et al., 2016), disturbed sleep (Zarowski et al., 2011), microcephaly (small head), "cleft palate, cardiac defects, underdeveloped external genitalia in males, postaxial polydactyly, and 2-3 syndactyly of the toes" (Nowaczyk & Wassif, 2020). Aggressive behavior is often present and, in one study, was found in approximately half of the participants (Thurm et al., 2016). Treatment varies significantly based on the child's needs and symptoms, but there is no cure. One relatively consistent treatment is the use of cholesterol supplementation, as it is, or should be, suggested for all children born with SLOS (Svoboda et al., 2012; Thurm et al., 2016). As of 2015, approximately 1 in 40,000 children in the US are born with SLOS, and approximately 1% of the population are carriers (Crest et al., 2015). To have a child with SLOS, both parents must be carriers. This syndrome is less common in those of African or Asian

descent (Wright et al., 2003); “both heterozygote advantage and founder effect have been suggested to explain the relatively higher incidence of SLOS among those of European descent” (Kelley & Hennekam, 2000, p. 322). And, due to the fact that genital defects are easier to recognize in males, females are not tested for SLOS at the same rate as males (Nowaczyk & Wassif, 2020).

**COVID-19 & Disability**

Individuals with disabilities and their families are a vulnerable, high-risk population during pandemics. While the full extent of the impact of COVID-19 on the disabled community is difficult to determine due to the fact that we have insufficient disability data (Reed, Meeks, & Swenor, 2020) and a limited understanding of the mortality risks for disabled people (Bosworth et al., 2021), there has still been a considerable impact. Armitage & Nellums (2020) discuss how people living with disabilities (PLWD) are at a higher risk for morbidity and mortality, have difficulty accessing health services despite their need for them, face discrimination and stigma, and have a greater need for services that are disrupted by social distancing and isolation. COVID-19 exacerbated many of these existing issues for PLWD.

The pandemic resulted in disruptions to service delivery for many disabled individuals all over the world. In Scotland, for example, learning disability services were disrupted (Middleton et al., 2020). Additionally, many individuals with significant behavioral conditions who reside in congregate settings were unable to completely isolate due to their caregiving requirements. Mills et al. (2020) state that within congregate settings, “isolating infected individuals with significant behavioral conditions was more

difficult, requiring frequent staff redirection to their room, and additional assistance with hand hygiene, cleaning and mask placement" (p. 495).

Concern over the welfare of disabled individuals during the pandemic resulted in stress and anxiety for parents and other caregivers as well. Parents of children with neurodevelopmental disabilities in Italy experienced symptoms of anxiety, depression, and stress that were significantly associated with their concerns for their child and the COVID-19 contagion (Grumi et al., 2021). Individuals who work with disabled people also faced increased stress during the pandemic, partially due to concern over the high risk of their clients (Lunsky et al., 2021).

Individuals with specific disabilities such as posttraumatic stress symptoms (PTSS) (Morabito, Bedford, & Schmidt, 2021), traumatic brain injuries (Morrow, Patel, & Duff, 2021), and individuals with IDD who experience significant behavioral conditions (Mills et al., 2020) are especially vulnerable to negative COVID-19 impacts, such as functional impairment or difficulty isolating. The following sections discuss vulnerability as it relates to disaster situations such as COVID-19, research specific to families of disabled children, and a brief overview of resilience, particularly within disability studies.

## Disabled Individuals & Families

### *Vulnerability*

Certain individuals and communities can be considered especially vulnerable to disaster situations, whether those are natural disasters or epidemics such as the current one. Bankoff (2019) states that vulnerability as a concept "emerged and gained validity

during the 1970s…[and] its chief proponents were practitioners and scholars highly motivated by concern with the plight of citizens" (p. 222). As mentioned before, the disabled community is a particularly vulnerable community during pandemics. Within the disasters discipline, research on vulnerabilities has been conducted based on factors such as location (Adger et al., 2005), population structure (Chen et al., 2014), the presence of urbanization (Joerin et al., 2014), and disability (Twigg, 2014), to name a few. Focusing on disability as it relates to vulnerability and the factors that impact vulnerability, is especially necessary when studying disabled individuals and the parents of children with developmental disabilities.

### *Research on Families of Disabled Children*

Research has been conducted on the experiences of families of children who have chronic conditions, are medically fragile (Patterson, Leanoard, & Titus, 1992), and are disabled. Understanding the experiences and needs of families of children with intellectual or developmental disabilities (IDDs) is critical. From the extensive literature on parents of children with IDD, and mothers in particular, research shows that parents of children with IDD have higher rates of depression (Manor-Binyamini, 2020; Singer, 2006) and struggle with their child's behavioral issues, such as hyperactivity, tantrums, and stereotypy (Muller et al., 2019). For families as a whole, there is evidence that the family quality of life is lower for families that include a child with an IDD (Brown et al., 2006). Research has thus been conducted on what factors prevent or mitigate these negative outcomes and how those factors impact an individual's or family's resilience.

Past research has addressed the needs of disabled children and their families as well, including community services. In a study of Canadian parents of children with IDDs, Hiebert-Murphy et al. (2008) discuss three important factors that impact families' entry into community services: identifying issues in development, the child's diagnosis, and "linkage" – or referrals – to community services. The parents in this sample clearly required more assistance from medical professionals, and particularly from the professionals involved in the diagnosis of their child. Hiebert-Murphy et al. (2008) state:

> Ongoing communication between providers of diagnostic services and community-based health and social services is needed so that families can be given current information about available services and/or referred to appropriate services. If such linkage does not occur at the point of diagnosis, some families will not receive support until they are in crisis and some children will lose the opportunity to benefit from early intervention programmes (p. 430).

Navigating support services for children with IDDs, and support services for the family as a whole, requires time and effort; for many families, it requires time they simply do not have. Dreyfus and Dowse (2020) make a compelling case for understanding what parents need and how they cope with the difficulties they encounter, warning that it is an error for "services and professionals [to] underestimate or discount parental expertise" (p. 20). These support services are a necessity for many families and can go a long way toward improving their family's – and their child's – quality of life. Access to these services may also serve as a protective factor when analyzing resilience (Olsson, 2008).

Research has already begun to emerge on families of children with IDDs during the pandemic. Embregts et al. (2021) conducted a qualitative study on the experiences of mothers caring for a child with an intellectual disability during the pandemic in the

Netherlands. Their study is, in fact, very similar to the one detailed in this paper. They interviewed five mothers of children with IDDs and found three themes:

> (1) We need to stay healthy, which centres on the mother's urge to protect their child's well-being; (2) We make it work, which provides insight into how the mothers were handling the drastic changes in their family; and (3) My child's and family's place in the world, which focuses on the mothers' experienced position in the world around them (Embregts et al., 2021, p. 760).

The findings of this study echo and support Embregts' findings, which suggests that the experiences of the mothers in this study are similar to those experienced by mothers of children with other disabilities.

**Tragedy narratives**. Recently, scholars have critiqued the accepted framework of research on families or parents of children with IDDs, sometimes called a "tragedy narrative", suggesting that a more positive emphasis is needed. This negative narrative persists throughout research in this discipline, highlighting the struggles and tragedies of raising or living with an individual with an IDD. Focusing solely on the negative side of experiences can produce a narrative of blame, with statistics and conclusions that make it seem as though the child's diagnosis is the sole contributor to those struggles. While the child's specific characteristics and diagnosis may contribute to parental stress, it is certainly not the only factor and should not be treated as such.

Researchers know that there are certainly challenges for parents or families of disabled individuals, but they have shown that those struggles can be attributed to a number of variables, such as socioeconomic status (Spencer, Blackburn, and Read, 2015, cited by Hastings, 2016), accessibility to services (Dreyfus & Dowse, 2020; Hiebert-Murphy et al., 2008), and concern over future care (Glidden & Jobe, 2007, cited by

Hastings, 2016). Hastings (2016) discusses the difference between a child's diagnosis and the other contributing factors mentioned, stating that "what is notable about most of [these] factors…is that they could be changed" (p. 175). The current, negative narrative contains the air of inevitability or the suggestion that having a child with an IDD automatically results in decreased psychological wellbeing. Understanding the positive experiences that parents report and their impact on family members requires another approach, such as a focus on resilience and protective factors – or, as Hastings (2016) puts it, factors that can change.

When studying resilience, it is therefore important to put as much emphasis on protective factors as risk factors, focusing on access to necessary services or accommodations in order to minimize some of the inevitable difficulties that comes with having a child with severe medical issues or disabilities. In other words, approaching research on parents of disabled children assuming that they are experiencing a tragedy is unhelpful. The tragedy is not the child; the tragedy is not having the resources to function or flourish with that child. An individual cannot adapt if they do not have the tools with which to do so.

**Resilience and Adaptation**

At its most basic, resilience can be defined as 'rebounding' or 'bouncing back' from something – a disaster, an event, trauma. This is overly simplistic, and scholars often add to this definition. There are a myriad of conceptualizations of resilience, and which conceptualization is best suited to each individual study depends on a variety of factors, including the area of research, the unit of analysis, and whether resilience will be

considered a process or an outcome. Uddin et al. (2020) explain the difference between these two views:

> In the process-oriented definition of resilience, recovery is primarily related to bouncing forward or moving to a different trajectory from the pre-disaster state (Manyena et al., 2011; Norris et al., 2008), while the outcome-oriented definition considers recovery as regaining the prior equilibrium (i.e. bounce-back) (Gilbert, 2010; Zhou et al., 2010) (p. 2).

There are scholars who do work with both definitions. For example, Fois and Forino (2014) view and discuss resilience – and particularly community resilience – as a process that "cannot be achieved in the short term [and] requires time and constant efforts by the community" (p. 724). Whether or not resilience is a process or an outcome could also depend on the type of disaster or event that a person or community experienced. Asking whether it is even possible for a person or group to 'bounce back' to the state they were in before the event in question is an important step in determining how best to conceptualize resilience. Are parents of children with Smith-Lemli-Opitz syndrome able to bounce back to their pre-COVID state, or will resilience look different for this population?

### *Overview of the Concept*

Since the 1980s, a growing body of research has focused on mediating factors related to stress, coping, adaptation, and resilience in parents of developmentally disabled children (Hassall & Rose, 2005; Hastings, 2016; Olsson, 2008; Patterson, 1991; Patterson, 2002), and researchers have become increasingly interested in the adaptation

and resilience of parents and families of children with IDDs (Olsson, 2008; Greef, 2013; Bristol, 1987; Patterson, 2002). Resilience itself is not a new concept or research interest, and many researchers cite Holling's work from 1973 as pivotal work in the area of resilience (Uddin et al., 2020; Kendra et al., 2018). The terms resilience and adaptation often appear together in literature. Some scholars argue that adaptation is simply another form or conceptualization of resilience, or "little more than a form of resilience in another guise" (Bankoff, 2019, p. 233-234), and it can be measured at an individual, family, or community level. Family adaptation can be defined as "the outcome of efforts to bring balance, harmony, coherence, and functioning to the demands and stress of daily life" (McCubbin et al., 1996 as cited in West et al., 2020, p. 154). West et al. (2020) explain that the interactions of a variety of factors, including family structures, vulnerabilities, and resources, can result in two separate outcomes: "well adapted" and "maladapted crisis" (p. 154). Individual level resilience can be traced back to psychology and studies on the resilience of children (Masten, Best, & Garmezy, 1990). The initial focus in these studies was on personal characteristics and experiences – i.e., what makes a person resilient? This has since evolved in the social sciences, where other theories – such as socio-ecological theory – have been applied to understand how one's social environment and circumstances impact their ability to be resilient (McConnell et al., 2013; Sage et al., 2014).

Researchers in many academic disciplines have conceptualized and developed the term resilience. Within criminology, for example, researchers analyze and discuss the resilience of survivors of intimate partner violence (Miller, 2018), protective factors

surrounding dating violence and adult intimate partner violence (Greenman & Matsuda, 2016), and even resilience in relation to counter-terrorism (Hardy, 2015). Disasters research also focuses heavily on resilience, having adapted the term from ecology, where a resilient ecosystem is "one that can absorb a short-term shock to the system, and then can, in a reasonable span of time, return to the state of the ecosystem before the shock" (Birkland, 2016, p. 117).

The concept of resilience is particularly prominent within disasters research. The COVID-19 pandemic has been classified as a disaster, and within the discipline there are recent publications on the resilience of care organizations (Orru et al., 2021), the impact of COVID-19 on community disaster resilience (Zu et al., 2020; Zhang et al., 2020), disaster management policies (Sengupta & Jha, 2021), the spread of misinformation during disasters and its impact on resilience during the COVID-19 pandemic (Barua et al., 2020), the impact of COVID on caregiving responsibilities and the increase of caregiver strain (Russell et al., 2020), and COVID response in healthcare and resilience planning (Keenan, 2020).

### *Resilience and Adaptation in Disability Studies*

Like other disciplines, there are multiple conceptualizations of resilience within disability studies. One definition is "positive family adaptation in response to, or despite, exposure to child behavior problems" (McConnell et al., 2013, p. 836), and another is "coping in difficult life situations" (Muir & Strnadova, 2014, p. 922). Researchers who apply resilience theory to this area of study are primarily interested in understanding what factors result in resilience for parents or family members of children with IDDs. In many

instances, researchers split these factors into protective and risk factors (Olsson, 2008; Patterson, 2002), viewing resilience "as a process where there are interactions between risks and protective factors relative to a specified outcome" (Patterson, 2002, p. 352). Patterson examined family resilience, using the family stress theory as an analysis tool, and focusing on protective factors and processes as well as the risk associated with having a child with chronic illnesses. They argue that "the key to understanding family resilience is the identification of protective factors and processes that moderate the relationship between a family's exposure to significant risk and their ability to show competence in accomplishing family functions" (p. 356). In another study conducted by Patterson (1991):

> nine family coping strategies or processes were identified that seem to be protective for these families: (a) balancing the illness with other family needs, (b) maintaining clear family boundaries, (c) developing communication competence, (d) attributing positive meanings to the situation, (e) maintaining family flexibility, (f) maintaining a commitment to the family as a unit, (g) engaging in active coping efforts, (h) maintaining social integration, and (i) developing collaborative relationships with professionals (Patterson, 2002, p. 357).

The ability to adapt to one's situation and cope with the stress that accompanies being a parent and caregiver to a disabled child is a protective factor when it comes to resilience. In other words, the ability to cope impacts one's ability to be resilient.

Olsson (2008) also focuses on protective and risk factors, suggesting that the reason we see such high rates of depression in mothers of children with IDDs, but also see evidence that mothers are doing quite well, is that there is a complex interplay between those risk and protective factors for each individual. Olsson (2008) discusses socio-ecological factors, such as financial situation and support, as both protective and

risk factors for families. The findings from McConnell et al. (2013), which included an analysis of socio-ecological factors, suggest that culturally relevant resources, such as financial resources and the level of social support, are a significant indicator of adaptation and, therefore, resilience in parents. They found that those with adequate socio-ecological resources adapt better than those without, despite stressors. Their analysis treats socio-ecological factors, such as financial hardship and social support, as mediating factors while also taking into account child behavior (McConnell et al., 2013). Bristol (1987) analyzed predictors of successful adaptation in families of autistic children, finding that social support, family resources, and coping patterns were related to adaptation.

The Family Adjustment and Adaptation Response Model is one method for understanding resilience which has been applied to families of disabled children. Figure 1 is an illustration from Patterson's article on family resilience (2002) and demonstrates the different protective and risk (demands) factors associated with adaptation and resilience.

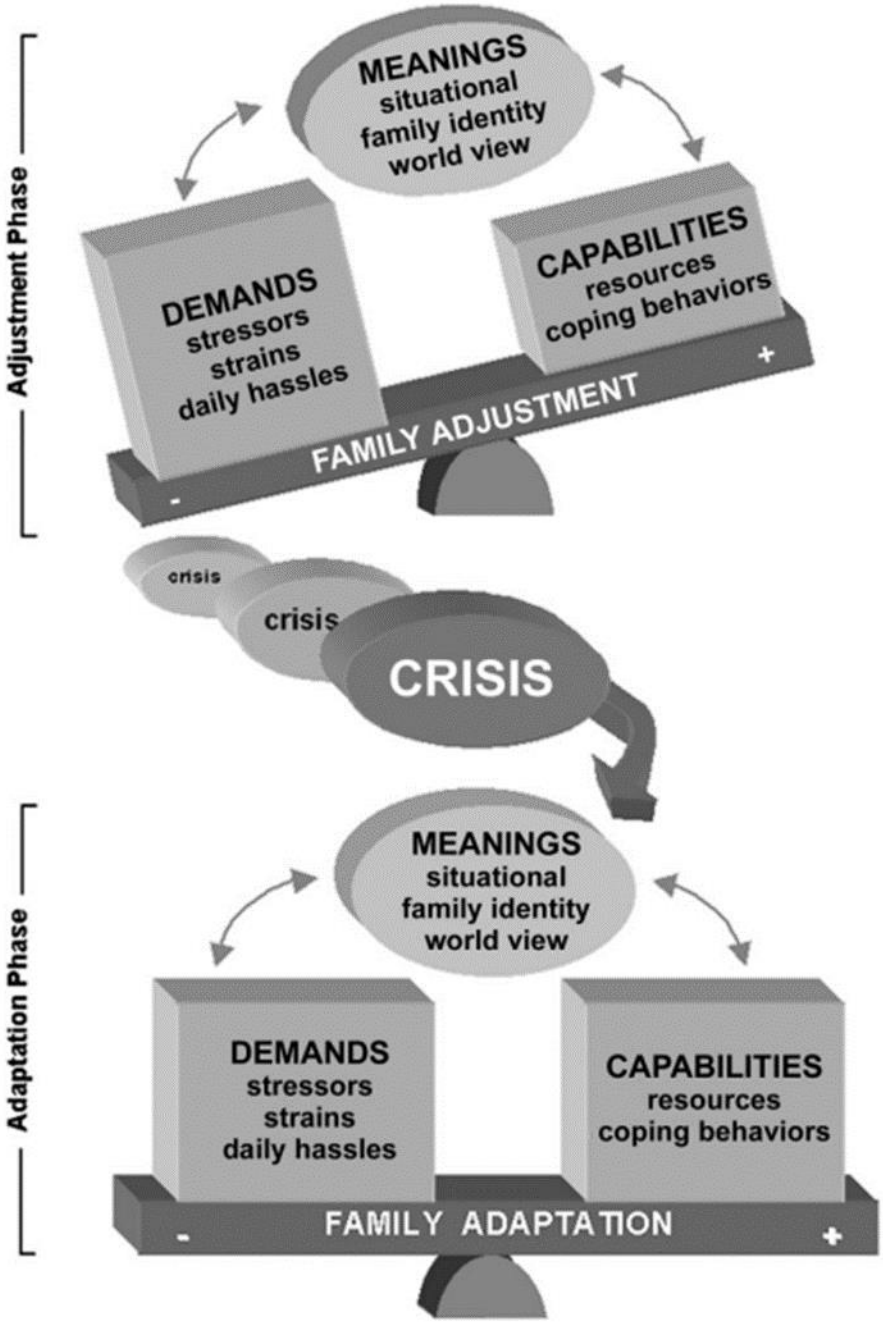



**Figure 1.** Family Adjustment and Adaptation Response Model

Note: From "Integrating Family Resilience and Family Stress Theory", Patterson, J.M. (2002), Journal of Marriage and Family, 64(2), 349-360.

Patterson suggests two possible outcomes based on the FAAR model: stable patterns of interacting (what could be referred to as a resilient response to the initial stressor or event) or crisis, "which is a period of significant disequilibrium and disorganization in a family" (p. 351) caused by the demands outweighing the capabilities. Adaptation to that crisis can result in resilience.

Quinlan et al. (2016) suggest that "being clear in advance as to why one wants to measure or assess resilience is the key to how the simplification should occur" (p. 685). The purpose of this study was to understand the experiences of a particular population (parents of children with Smith-Lemli-Opitz Syndrome) in order to assess if and how they maintained or developed resilience during the COVID-19 pandemic. For the purposes of this study, resilience will be conceptualized as 'a complex interaction between risk and protective factors that results in positive adaptation during or after stressful experiences'. This basic conceptualization was developed based on a review of the current conceptualizations of the term in the relevant literature, drawing primarily from the work of Olsson (2008) and Patterson (2002) and their theoretical models of resilience. And while this is a simplification of a complex concept, it is appropriate for this population and this context based on previous research on disabled individuals, their families and the experiences they have, particularly during disaster situations.

# Chapter 2

## PURPOSE OF STUDY AND RESEARCH QUESTION

The purpose of this study was to understand the experiences of parents of children with Smith-Lemli-Opitz Syndrome (SLOS) during the COVID-19 pandemic in order to understand what factors impacted their resilience, or lack thereof. The research question that guided the study is:

- How is the COVID-19 pandemic impacting parents of children with Smith-Lemli-Opitz Syndrome and their ability to develop or maintain resilience?

### Intellectual Merit and Broader Impact

The results of the study could potentially inform future research on resilience, Smith-Lemli-Opitz Syndrome (for which there is no published research in the social sciences), and disability studies. Information on the specific experiences of parents of children diagnosed with SLOS, particularly experiences surrounding healthcare and support during the pandemic, could be utilized for advocacy purposes. This study is also an important addition to disabilities literature, as it is the first social research conducted focusing on this particular syndrome.

The varying effects of the COVID-19 pandemic have been constantly discussed in reports and articles over the last year, both within and outside of academia. The studies and data available tell us that this pandemic is having a serious impact on disabled

individuals and their families (Jeste et al., 2020; Chen & McNamara, 2020; Neece & Fenning, 2020; Redquest et al., 2020, Embregts et al., 2021). Understanding the current needs of this population is paramount in identifying what resources need to be available to families during crises such as the COVID-19 pandemic. Embregts et al. (2021) argue that "research into the potential challenges and benefits parents have experienced in the current COVID-19 pandemic is vital in order to gain insight into how to support families at this difficult time" (p. 761). The purpose of this exploratory study was to understand those challenges and benefits through the lens of resilience.

**Relevance and Generalizability**

This study is an important addition to disabilities literature, as it is the first social research conducted focusing on this particular syndrome. As every syndrome is different, it is vital to understand and document the challenges and limitations – along with the positive experiences – that accompany specific diagnoses. This is particularly important when studying resilience, as it is impacted by both positive (protective) and negative (risk) factors (Olsson, 2008) and, as such, both should be taken into consideration. When considering both the fact that negative and positive factors can differ by diagnosis and the idea that "resilience...starts with the recognition of the individual variation in people's response to the *same experiences* [1]" (Olsson, 2008, p. 284), it is essential to look at resilience for each population, as opposed to looking at the resilience of all parents – or all parents of developmentally disabled children – as a whole. Parents of children with Down Syndrome, for example, will not necessarily have the same experiences as parents

[1] Emphasis is mine.

of children with SLOS (Olsson, 2008), and so they are not directly comparable. To study a group of parents and analyze the factors that either promote or inhibit resilience, I would argue that those parents must have relatively similar experiences surrounding their children and their child's diagnosis.

One downside to this method of studying and analyzing resilience is a lack of generalizability. While there are some experiences or factors that can be applied to a larger population, such as socioeconomic status and availability of necessary resources, other factors are more specific to a particular diagnosis. For example, because SLOS is a rarer syndrome, many doctors have not heard of it. One likely would not find that to be the case with other disabilities, such as autism[2], but they are both considered developmental disabilities. As such, certain findings and results of this study could be generalizable, while others could not be.

The reverse may also be true: other studies' results may not be applicable to this particular population for the same reason that these results may not be applicable to other populations. This only serves to strengthen the argument that research on this population is needed in order to understand and add to the literature on resilience and, specifically, the resilience of parents of children with developmental disabilities.

[2] This is not to say that most doctors know how to correctly treat or manage autism, rather that they will have heard of and/or have some knowledge of the disorder itself.

# Chapter 3

# METHODS

## Positionality Statement

Before discussing the methods utilized for this study or the findings, and in the spirit of self-reflexivity and transparency, I would like to acknowledge my particular position in regard to this research. I am a member of the Smith-Lemli-Opitz syndrome community, both as a sister to an individual with SLOS and a carrier of the gene. While this relationship to the sample population worked in my favor in regard to building rapport with participants, it is possible that it created unwanted bias as well. Furthermore, I am a white woman in a position of both educational and socioeconomic privilege, and I acknowledges that privilege can have an impact on research –particularly qualitative research. I have made a concerted effort to avoid any possible bias and represent the data as accurately as possible. This included researching information I have personal knowledge of to ensure that the information presented is accurate and ensuring that I avoided confirmation bias to the best of my ability by including and making note of discrepancies in responses or experiences, especially those that I do not have personal experience with

**Data Collection**

The population for this study was parents of children diagnosed with Smith-Lemli-Opitz Syndrome. The requirements for participation included being over the age of 18, residing in the United States, and the ability to speak English. To recruit participants, I identified two social media groups on Facebook where recruitment flyers could be distributed. As a family member of someone with SLOS, I was already familiar with, and a member of, one of those groups. The other group is a private group on Facebook and is restricted to parents of children with SLOS. Before distributing (posting) the recruitment flyer, I reached out to the members of the Smith-Lemli-Opitz/RSH Foundation requesting assistance with recruitment. After communicating with their Director of Media Relations & Communications, she agreed to post the recruitment flyer in the private group, and I posted the same recruitment flyer in the public group. The flyer was posted and/or reshared three times in the private group, at my request. Additionally, I contacted Dr. Elaine Tierney, M.D. at Kennedy Krieger Institute, and Dr. Forbes D. Porter, Ph.D., M.D. at the National Institute of Health to request assistance with recruitment. Both doctors are experts on SLOS, and both agreed to circulate the recruitment flyer if possible.

This study included both a quantitative and qualitative elements, and all items in the protocol were approved by the Institutional Review Board at the University of Delaware. A Qualtrics survey was used to collect demographic information, preliminary information on the participant's child/children, and for resilience and religiosity scales. Following the survey was a semi-structured interview, conducted via Zoom. The

recruitment flyer included my name and email address, and individuals interested in participating were instructed to send an email to the address listed.

**Consent and Protection of Information**

Upon receiving an email, I provided the interested individual with the consent form, which included a brief explanation of the study and what they would be asked to do should they consent to participate. If the individual consented to participate, they were asked to sign the consent form and scan it back me. Once this was complete, they were sent a link to the Qualtrics survey. When the questionnaire was completed, I reached out to schedule a Zoom interview. As information on both the questionnaire and interview were included in the first consent form, written consent was not requested prior to the interview portion, but verbal consent was obtained before the interviews began. Participants were encouraged to voice any questions or concerns they had prior to the beginning of the interview, or at any time during the interview if a question or concern should arise. The interviews were recorded and transcribed with the participants' consent, both written and verbal, and kept in a secure folder on my computer. The transcripts were then cleaned, the names of participants were replaced with pseudonyms, and the files were password protected and kept in a secure file. Only I had access to the transcripts and the recordings, which were deleted upon the completion of the transcription. All names used in this paper are pseudonyms, and certain demographic factors, such as the town/city the individual resides in, were excluded.

**Questionnaire and Interview**

The Qualtrics questionnaire included 45 questions on demographic information, the child/children's symptoms, basic information on the child's experience with doctors, the participant's religiosity, mental health, and stress. The questionnaire itself can be found in Appendix A. Two scales were included in the questionnaire: a resilience scale and a religiosity scale. For the quantitative measurement of resilience, found in the questionnaire, the Brief Resilience Scale (BRS) (Smith et al., 2008) was utilized. This scale was selected based on the fact that it has been studied and found useful for individual resilience (Smith et al., 2008), has been tested for internal consistency (a = 0.71, Fung, 2020), and the fact that it is shorter and was, therefore, more suited to the questionnaire for this study. This scale includes 6 items/questions - which can be found in the questionnaire in Appendix A - and puts an emphasis on stress and responses to stress, which is something the conceptualization I am utilizing in this study focuses on. The dependent variable in the quantitative portion of this study is resilience, as measured by this scale. An additional scale was included to measure religiosity, in light of the links between religiosity and resilience identified in prior research (Choi & Hastings, 2018; Peres et al., 2007): the Centrality of Religiosity Scale (CRS). The version utilized in this questionnaire is the CRSi-14, which is an interreligious version of the scale, allowing for the inclusion of non-Abrahamitic (Judaism, Christianity, Islam) religions (Huber & Huber, 2012), and it has 14 items/questions.

The interview was semi-structured, and the protocol can be found in Appendix B. Participants were asked about a variety of topics, including: their families, their

experiences with the syndrome, healthcare access, stress, and their experience during the COVID-19 pandemic. Interviews lasted between 34 and 105 minutes.

**Analysis**

Quantitative analysis for this study was simple, as in-depth statistical analysis would not be appropriate for this sample size. The resilience and religiosity scales were created in SPSS, as were the crosstabulations provided in the results section of this paper. The interviews were analyzed using thematic analysis. I utilized a basic form of thematic analysis used in the social sciences (Braun & Clarke, 2006), which is a type of analysis used on qualitative data to identify patterns and themes within the data. The process for this included: transcribing the interviews, reading through the transcripts, identifying patterns that emerged, creating codes based on those patterns, and grouping those codes into broad them

# Chapter 4

## RESULTS AND ANALYSIS

### Demographics

**Table 1.** Participants

| Participant[3] | Age | Race/Ethnicity | Number of Children | Child(ren) with SLOS[4] | Age of Child(ren) |
|---|---|---|---|---|---|
| Christina | 53 | White | 2 | Abby | 18 |
| Jessica | 54 | White | 3 | Isaac | 20 |
| Lisa | 47 | White | 1 | Bryan | 16 |
| Nicole | 42 | White | 3 | Morgan & Matthew | 14 & 6 |
| Julia | 37 | White | 3 | Harry | 4 |
| Lena | 30 | Hispanic | 2 | Evan | 4 |
| Natalie | 46 | White/Hispanic | 3 | Thomas | 13 |
| Felicia | 44 | Chicana | 2 | Daisy | 13 |

#### *Gender and Race*

There were eight participants who took part in this study. All of them identified as female and were the mothers of the children with SLOS. Their ages spanned from 30 to 54. Respondents were asked their race and ethnicity as an open-ended question to leave room for identities that may not match the standard options utilized in surveys. Out of the eight women, six identified as white or Caucasian. One of those women also identified as

[3] Pseudonyms.
[4] Also pseudonyms.

Hispanic. Another identified solely as Hispanic, so her race is unknown, and the last women identified as Chicana. The sample is overwhelmingly white, with three Hispanic/Chicana participants.

### *Marital Status*

Respondents were asked to provide their marital status in the questionnaire. Seven of the women selected 'married/domestic partnership/cohabitation', and one woman selected 'divorced'. Each woman discussed her relationship status in the interview portion of the study, and from that it was concluded that seven of the women are married to the father of their child with SLOS, and one woman is divorced but currently co-parenting with her ex-husband.

### *Education, Income, and Insurance*

**Table 2.** Educational Attainment

| Highest Level of Education | Frequency | Percent |
|---|---|---|
| Associate Degree | 2 | 37.5 |
| Bachelor's Degree | 2 | 25.0 |
| Master's Degree | 2 | 25.0 |
| Professional Degree | 1 | 12.5 |

The participants in this study were highly educated, with each of them having received a degree past high school. Only 1 participant did not respond to this question.

**Table 3.** Income

| Income | Frequency | Percent |
|---|---|---|
| $50,000-100,000 | 3 | 50.0 |
| $100,000-200,000 | 2 | 33.3 |
| $200,000+ | 1 | 16.7 |

The household income of this sample varied, with a range of $50,000 - $225,000. Two participants did not provide their household income. Seven of the eight participants (87.5%) reported having private insurance, and one participant reported having Medicaid as her insurance.

***Location***

The participants who took part in this study all live in the United States, spanning 7 different states, from every part of the country. In the interest of protecting confidentiality, the individual states will not be provided. The majority of respondents (75%) reported living in a suburban area, with one respondent living in an urban area and another respondent selecting ‘other’ and describing their environment as a ‘college town’.

## Quantitative Results

### *Resilience*

**Table 4.** Resilience Score

| Score | Frequency | Percent |
|---|---|---|
| 2.17 | 1 | 12.5 |
| 3.17 | 2 | 25.0 |
| 3.50 | 1 | 12.5 |
| 3.67 | 2 | 25.0 |
| 4.00 | 2 | 25.0 |

The scores in Table 4 were calculated utilizing the Brief Resilience Scale (BRS) (Smith et al., 2008). The possible scores range from 1 (low resilience) to 5 (high resilience). The participants in this study demonstrated both low, moderate, and high resilience (Table 5) based on their responses to 6 questions. Half of the participants had scores that indicate high resilience.

**Table 5.** Level of Resilience

| Level of Resilience | Frequency | Percent |
|---|---|---|
| Low resilience | 1 | 12.5 |
| Moderate resilience | 3 | 37.5 |
| High resilience | 4 | 50.0 |

When looking at resilience as it relates to other demographic characteristics, crosstabulation reveals that income could act as a protective factor (Table 6), but age does not appear to have any identifiable relationship to resilience in this sample (Table

7). More advanced statistical testing was not possible due to the sample size. Respondents whose income fell within the $50,000-100,000 range scored at every level of resilience (Table 6). The only individual with a low resilience score was in this group. Those with higher household incomes, in the $100,000-200,000 and $200,000+ ranges score moderately and highly on resilience.

**Table 6.** Income and Resilience

| | Low resilience | Moderate resilience | High resilience |
|---|---|---|---|
| $50,000-100,000 | 1 | 1 | 1 |
| $100,000-200,000 | 0 | 1 | 1 |
| $200,000+ | 0 | 0 | 1 |

There does appear to be a possible, positive correlation between income and resilience, with income acting a protective factor.

**Table 7.** Age and Resilience

| | Low resilience | Moderate resilience | High resilience |
|---|---|---|---|
| 30s | 1 | 0 | 1 |
| 40s | 0 | 2 | 2 |
| 50s | 0 | 1 | 1 |

While some research suggests a positive correlation between age and resilience (Portzky et al., 2010), other studies have demonstrated a lack of effect of age on factors related to resilience (i.e. depression and anxiety) (Bitsika et al., 2015). The mixed findings on age and resilience suggest that the relationship between these factors could be dependent on

the population being studied. As stated above, age and resilience do not appear to be related in this sample.

### *Religiosity*

Prior research has identified a relationship between resilience and religiosity (Choi & Hastings, 2018; Peres et al., 2007). In this sample, four respondents considered themselves religious. Of those four, three completed the questions in the survey surrounding religiosity. The religiosity scale (CRSi-14) scores range from 1 to 5, with lower scores meaning low religiosity and higher scores meaning higher religiosity.

**Table 8.** Religiosity and Resilience

| | Low resilience | Moderate resilience | High resilience |
|---|---|---|---|
| Religious | 0 | 1 | 0 |
| Highly religious | 0 | 0 | 2 |

Two respondents scored ‘highly religious’, with scores between 4.0 and 5.0, and one respondent scored ‘religious’, with a score between 2.1 and 3.9. Both individuals who scored highly on religiosity also scored highly on resilience.

## Interview Results

### *Coding*

The transcriptions of the 8 interviews were analyzed utilizing a thematic analysis approach (Braun & Clarke, 2006) to identify patterns and themes in the interviews. As the interviews were semi-structured, themes surrounding and relating to the specific questions emerged, as did themes that were not directly related to the questions.

The first step in this analysis was reading through the transcripts after they had been fully cleaned. After becoming more familiar with the data, the researcher created codes based on observed patterns. Specific codes were created, such as “mentioned school services” or “access to cholesterol”, which were then grouped into more specific themes. The process for creating the theme ‘support and services’ is detailed in Figure 2 below. The codes on the left were the initial codes for this theme, which were then condensed into ‘access to services and treatments’ and ‘social support and connection’. Those themes were merged due to the overlap between them throughout the interviews, creating one of the major themes discussed in this section.

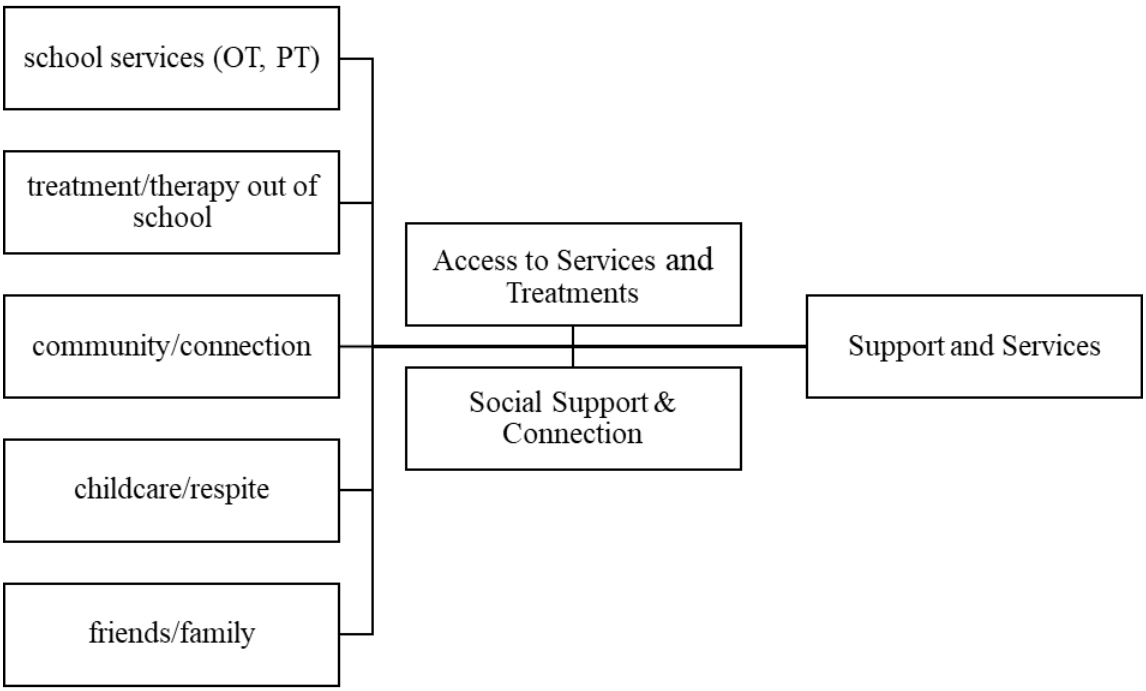



**Figure 2.** Thematic Coding Example

The broad themes that were identified through this analysis are experiences with SLOS, impacts of COVID-19, support and services, parental and caregiver strain, and resilience. Within these themes, protective and risk factors are identified and discussed.

### *Experiences with SLOS*

To understand how parents of children with SLOS develop or maintain resilience during a pandemic, and to have a more thorough understanding of their experiences during the pandemic, the first thing that must be analyzed is the experience of having a child with SLOS. As mentioned before, there is a wide spectrum of the severity of symptoms that accompany this diagnosis. Many aspects of this syndrome can create stress for parents, not only because of the physical or behavioral symptoms, but because of the difficulty in finding treatment or managing symptoms as well. Issues such as the diagnosis process and treatment, medical complications, inadequate or unstable insurance coverage, and the management of behavioral problems were discussed during the interviews, as were positive experiences and what Patterson (2002) refers to as the meaning making process.

**Diagnosis process and treatment**. Each of the mothers in this study clearly conveyed how the issues surrounding this syndrome and the treatment for it were often stressful for them and their children. Patterson (2002) argues that "the diagnosis of a child's disabling condition would be an example of an individual level [family] demand" (p. 350), which is a factor in adaptation in the Family Adjustment and Adaptation Response Model.

Getting a SLOS diagnosis is heavily dependent on the doctors you have working with you and their familiarity with genetic testing and genetic syndromes. This can be a long and complicated process, which creates stress and anxiety in many parents. Some mothers, like Lisa, received a diagnosis quickly, but others had to wait a long time. Nicole, who has two children with SLOS, lives in a state where she believes there is only one geneticist. This geneticist had to drive out to do the testing and Nicole subsequently struggled to get the diagnosis for her first child. Her second child, Matthew[5], was diagnosed quickly though. Christina, Jessica, and Lena all struggled with the diagnosis process, stating:

> …every three years or so we'd go in to see the geneticist and they'd draw blood and they'd do whatever…the most recent things that they were doing. [We went to] a neuro genetics clinic, which is very thorough, you know, you see psychiatrists, you see occupational therapists, you see neurologists, you see genetics, genetic counselors, you know, we were there the whole day. And they put everything together with respect to her conditions and they ran tests, every test that they could think of, or at least they told us they did. (Christina)

> It was almost six months from the time we first saw somebody till we got an actual diagnosis…the testing is expensive and insurance companies don't want to do it and – and there's not very much known about SLO. (Jessica)

> We didn't get [the diagnosis] until he was about 20 months. (Lena)

Even after seeing numerous doctors throughout Abby's life, her parents did not receive an SLOS diagnosis until Abby was 15. A diagnosis and subsequent understanding of the syndrome is necessary for appropriate medical treatment in many cases. Christina

[5] The participants described their children in detail, and while that aspect of the interview was not used in this analysis, brief descriptions of the children will be included in the footnotes. Matthew was described as a class clown and "the sweetest boy you'll ever meet" (Nicole).

expressed the belief that despite the lack of diagnosis, she believes that they did get the therapies that Abby needed, although she wonders if cholesterol supplementation (a common treatment suggested for SLOS) would have made a difference if they had been able to introduce it earlier.

Communication issues were also a source of stress regarding the diagnosis process. Some of the mothers did not understand what the doctors were testing for or why while others disliked the way their doctors talked to them about what was going on. One participant found out about her child's diagnosis, not from a doctor, but from a web portal for medical notifications:

> So I found out he had SLO just like reading about it, and not from like anyone like talking to me or explaining it to me, so I quick like googled 'what's Smith-Lemli-Opitz syndrome' and it's like when you just Google it, it's terrifying. I freaked out, like I called our pediatrician and she like left whatever she was doing to like talk to me on the phone, so I was like hysterical. Because the first thing it says is like the survival rate, you're just like, 'the – the what?!' (Julia)

She was not the only participant who experienced significant stress and anxiety from a lack of clarity on the syndrome at the time of diagnosis. Felicia was close to tears recalling the process:

> [The geneticist] said it looks like Smith-Lemli-Opitz syndrome…of course, I looked it up and that's when I was like, 'oh my God'…I get emotional thinking about it, um, but I thought 'she doesn't have it', but they called and said she did. (Felicia)

Communication surrounding the diagnosis and treatment of SLOS is especially important considering the amount of and severity of some of the medical issues that stem from the syndrome.

**Medical complications**. The physical symptoms of SLOS can be severe, and there are many medical complications that can arise. Several participants mentioned

surgeries their children needed, medical issues that are recurring (i.e. severe ear infections), and nutritional deficiencies that stem from the inability or disinclination to eat. The number of specialists that children with SLOS often have to see can be overwhelming for parents.

> He's seen [a] neurologist. Who does kidneys? Whatever kidney specialist is…the pulmonologist. Optometrist for his eyes. The feet, what is that – podiatrist? Podiatrist. I mean, you name it we've kind of seen a lot of specialists, we still see a gastroenterologist just because of his G tube, that's something we have to see him for and then also a dietitian because of the food intake. (Natalie)

These experiences are stressful for both the parents and the children, particularly when the procedure or symptom is severe, or the outcome is unpredictable. "You know, constantly being in and out of the hospital was like a little overwhelming too. So, I have like a lot of anxiety from it" (Lena) Hearing about other families' experiences with medical complications also creates stress and anxiety. Julia, for example, mentioned their hesitance to get their son Harry a feeding tube because "a little boy just passed away from complications with the feeding tube".

Two participants discussed the medical trauma that their children have experienced as a result of these complications, recurring doctors' visits, and procedures and connected this trauma to COVID:

> I think, just like the frequent COVID tests that they've had – he's had to have three, and this poor kid has had so much medical trauma…it's just been so traumatic that it's just not worth – it's not worth the trauma [to go get his cholesterol levels checked], especially if you're going to have to have a COVID test like every month or something. (Julia)

> Evan is already traumatized when it comes to doctors, so now like COVID test is something else to add to it. (Lena)

**Insurance**. All of the participants in this study had some type of insurance, with the majority having private insurance. Their children were all covered by at least one type of insurance, but often by multiple. Children with disabilities often have access to secondary insurances through the state, or through programs that are state specific[6]. Several participants described their experiences with those programs, detailing the process for qualifying or receiving those services.

While they had access to insurance, several participants had difficulty getting coverage for some of the treatments or equipment that their children needed. For example, cholesterol supplementation is not covered by all insurances. Nicole shared that they gave her daughter the cholesterol suspension using the recipe from the NHI, but "obviously [insurance] didn't cover that". Felicia also struggled with the insurance company denying coverage for the cholesterol, although she did eventually get it covered through a state insurance program. One participant described her fight with insurance to get coverage for cholesterol:

> So, insurance, like, denied that but then [state insurance program] paid for it, but then, at a certain point, there was – we got a change in insurance. So [state insurance program] said 'well, now we need a denial from your primary insurance' and I said 'okay, no problem, I'll get that', so we got the denial from our new insurance and then [state insurance program] goes 'yeah we're not going to pay for it either', and we were like 'wait, what?'…and we fought for over a year to get them to pay it the first time. And I just like, didn't have that in me anymore. (Jessica)

One participant applied for SSI, which was initially approved, but then they received a letter stating that SLOS doesn't qualify for SSI.

[6] In the interest of protecting confidentiality, the specific states and programs within those states are not provided.

**Behavioral problems**. Various therapies are available and recommended for children diagnosed with SLOS. Occupational therapy, physical therapy, speech therapy, feeding therapy, mental health services, and ABA therapy were all mentioned throughout the interviews. Some of these therapies address the behavioral problems that are often present in children with SLOS, such as aggression or screaming. Participants discussed these behavioral problems, often in a very matter-of-fact way: "So that's another thing about [Isaac[7]]. He goes on biting attacks. They're infrequent…but they're terrifying. And you don't always know when one is coming, that's the thing" (Jessica). Participants expressed some frustration surrounding access to certain therapies – ABA in particular.

Some of the mothers mentioned regressions in their child's behaviors. Where progress was being made before the pandemic, problem behaviors had reappeared during COVID-19. For example, Julia expressed frustration that her son's potty-training progress had stopped and his progress in feeding had halted. Nicole and Lena also reported a recurrence of specific behavioral problems during the pandemic, which were stressful and frustrating.

**Meaning making process**. Patterson (2002) discusses factors that promote adaptation and resilience in families, stating that "coping is viewed as part of family capabilities and…a component of protective factors associated with resiliency" (p. 357). One of the coping processes identified was the meaning making process, which impacts stress responses. A focus on positive experiences, positive characteristics of their

[7] Isaac was described as a "super perceptive and very bright" person who loves books and long walks (Jessica).

children, and viewing their situation or experiences as an opportunity for growth as a person or a learning experience contribute to this method of coping and impact adaptation. In other words, parents – in this case, mothers – cope with the negative experiences and challenges they face by altering their understanding and perception of their situation or life.

The mothers in this study did demonstrate this coping strategy, often mentioning the positive experiences they've had with their child or children, the way that their other children positively contribute to the family and the household, the impact their sibling's diagnosis has had on their other children, and the impact of having a child with a disability.

> Even though it is sometimes a harder existence, I feel like what you get kind of supersedes or overrides what you lose. (Jessica)

> He's very cheerful, and he's always smiling. So, he's really been just like a blessing, we say, because it's almost like our family's learning how to – we've never had any form of disability really in our family…it's kind of like taught them a little bit more about patience and understanding new things. (Lena)

> I think one of the silver linings with a family like ours is that the siblings develop a kind of empathy, compassion, and a different worldview – kind of a more mature view. (Jessica)

Some of the mothers expressed that without having a child with SLOS, they wouldn't have met others within the SLOS community, or the disability community as a whole, which they view as a positive experience as it often serves as an important support system for them. For example:

> I've made so many friendships…just the people I've met along the way, who have impacted his life and that I can see really care for him and love him. That's made a big difference in my life. (Lisa)

> I think one of the silver linings with a family like ours is that the siblings develop a kind of empathy, compassion, and a different worldview – kind of a more mature view. (Jessica)

The use of meaning making as a coping strategy was evident throughout the interviews, and it appears to be an important aspect of understanding their situation in a way that allows them to understand their lives in a positive way, which is likely beneficial to their mental health and contributes to their resilience.

### *Impacts of COVID-19*

Many of the issues and stressors all parents – not just parents of children with disabilities – face were exacerbated by the pandemic. The unique circumstances and issues that parents of children with SLOS deal with and their strategies for mitigating many of those issues were affected by COVID-19. Jessica stated in her interview that she and her husband "had the problem everyone across America had, but much more complex and challenging with a kid like Isaac". One of those complex issues surrounding the pandemic was explaining to their children why they were suddenly not allowed to do the things they enjoy or go out. "It's hard, because you can't explain to him what's going on, he doesn't understand why you can't go to school, you can't go horseback riding, you can't do anything" (Lisa).

The mothers that were interviewed discussed a wide variety of impacts that the COVID-19 pandemic had on them, their children, and their families. They also expressed varying degrees of concern surrounding the pandemic. The overwhelming majority of the

participants and their children were vaccinated[8], and many of them expressed concern about how COVID-19 would impact their children.

> I went back to work for a little bit last year for a suburban school district near here, but they just like had no COVID protocol and there – this is like before the vaccine and I was like, I can't take that risk, especially with like, not knowing how kids with SLO are affected. (Julia)

> We were very cautious, because we didn't know how bad it would affect him and when our pediatrician – plus Dr. Elias said that they were high risk, and I asked her specifically about Thomas and she looked over his charts, and yeah he is high risk, so we just stayed locked in. (Natalie)

> I kind of was like I don't want to go back anyways cause I don't want to catch this thing, you know? And I just didn't know how deadly it would be for us, especially for Daisy. (Felicia)

In some cases this was worsened by factors that increased the probability of their child contracting COVID-19, and some of the participants and their children did get COVID-19.

> We did, yes, we [got COVID] just this past May…and Bryan won't wear a mask…so that was hard, you know, taking him out. (Lisa)

> Harry[9] – it took Harry a long time to get used to a mask…there's one mask he will wear. (Julia)

There were varying levels of caution taken in regard to COVID-19 as well, with some mothers explaining how they completely isolated themselves and their families, and others took a less cautious approach. When asked if the pandemic impacted her support system at all, Nicole responded: "I wouldn't – maybe say it even improved it? You know, with my Bible study, like we're – we've gotten close and you know it's – we never wore masks, it was sort of like throw caution to the wind" (Nicole). But she also recognized

[8] Participants were not asked whether they were or were not vaccinated, but most of them offered that information on their own.
[9] Harry was described as a "quirky" kid who likes cars, garbage trucks, and bugs (Julia).

that her experience contracting COVID and having seen her entire family contract COVID had an impact on her perception of the pandemic, saying that she has a "flawed view of COVID".

The participants' concern for their children contributed to higher stress levels for some of the participants. Jessica talked about her worries, such as: her children and their schooling, her daughter who had just graduated from college, how the pandemic will affect them, their socialization, and their plans for the future. Some of them felt that one of the largest impacts the pandemic had on their family was the disruption of established routines or the creation of new routines:

> I think our typical day is like Groundhog day now, it's the same every day. It's like, and I know that's for everybody. And fortunately for autism that works. Yeah, they love a routine and we've kind of embraced that, because what other choice do you have? (Jessica)

> And it's hard to fill the time too because it's – it's – it's a routine every single Sunday we're [horseback riding] and – I mean, with everything, we have some routine. And we just lost it. Yeah that was hard. (Lisa)

For many of them, this disruption of routine occurred because of changing school experiences. Participants expressed frustration and dissatisfaction over their experiences with virtual schooling and virtual therapies, although it's worth noting that none of them expressed this frustration towards the individuals providing the services.

> [Therapy] is virtual and has been virtual since the start of the pandemic. Prior to that she would have had it in person, and I think it's much more effective in person, but it is what it is. (Christina)

> [Evan[10]'s] therapies and stuff, everything switched to virtual. Or it's just starting up now…that was kind of just the hard part, like a lot of his

[10] Evan was described as a kid who "has energy that'll last for days" (Lena), which the researcher can confirm. Evan was the only child with SLOS present for the interview, and he bounced around the entire time.

> therapies became online, like Zoom calls…for us, the challenge was just that he doesn't understand to sit still and participate…[so] we kind of just like opted out of a lot of those things" & "Like for a long time his like OT, his PT were virtual, so he wasn't getting the same benefits, and it was just frustrating for us like having to do distance learning and then on top of that, like we were the ones here doing like his therapies too. (Lena)
>
> [Abby][11] missed out on a lot for school last year, because it was virtual for so long…I think she missed out on the socialization of being in school…It was impossible to try and work at the same time that she was, you know, I was set up about six feet away from her to try and help, but that was – it was just a nightmare. (Christina)
>
> Daisy[12] made the transition [to a new school] during virtual, from sixth grade to seventh grade, which it really sucked to do it virtually. (Felicia)

Some of the women experienced financial impacts due to the pandemic, while others reported no significant impact on their financial situation. Access to unemployment, loans, the ability to work from home, stimulus money, child credit money, and hazard pay were some of the factors that protected them from experiencing financial hardship during the pandemic.

### *Support and Services*

The participants in this study had a variety of experiences with social support and access to services during the COVID-19 pandemic. Sufficient support and access to services operate as protective factors, while the lack of this support and difficulty accessing services as a risk factor.

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[11] Abby was described as a funny girl with a quick wit who has "always loved school, and…anything she can do to get to school, she will" (Christina).

[12] Daisy was described as a very social, very flirty girl who loves sound effects ("so if anyone's like coughing, sneezing, vomiting, she's loving it"), Elmo, and kittens (Felicia).

Access to support services was discussed in the interviews, and the mothers reported having access to the services their children needed, either through school or through private therapy. A recurring exception to this trend was limited access to ABA therapy, which often requires an autism diagnosis.

> So they call autism the million dollar diagnosis, because if they diagnose it that's maybe how much they're investing in this kid throughout his lifetime or her lifetime. So I know a lot of families with kids with SLO have had to get the autism diagnosis from Elaine Tierney or Kennedy Krieger, and then sometimes they don't accept that because it's out of state. (Jessica)
>
> ABA therapy – which I'm not a huge fan of ABA – but I liked the researcher who's running the lab a lot, and I think it would be a good fit for him, but yeah, it's like a two year waitlist. (Julia)
>
> We're on a waitlist for ABA and [his behavioral specialist] feels that that would really be like the best thing to help him as he gets older. (Lena)
>
> ABA has not been available to use simply for the fact that he doesn't have the autistic diagnosis. Even though the neurologist talked about that in her paperwork, we never got the official diagnosis, because we have to see some psychologists. And the insurance that we have right now doesn't cover it. (Natalie)

Some of the participants experienced a loss of support or inadequate support due to the pandemic:

> We definitely saw my parents a lot less. Because of their age, and we were just worried…yeah, it reduced our support system in terms of who we could reach out to. (Christina)
>
> So over COVID our nurse of five years got recruited away from us…and we've been trying to get another nurse and with COVID and the shortage, I mean, and they promised me too – 'you're going to have a nurse before school starts', because she left in April or May. But 'before school starts she'll have a nurse'. Nope" & "In the beginning, it was just, you couldn't get support, you couldn't get help because you don't want anybody in your house…so I fault nobody, but it was just no help. (Felicia)

Friends and family often provide support in both tangible and intangible ways. Almost all of the mothers in this study had more than one child, and many of them had children old enough to provide either babysitting or respite care for their sibling. Respite care and childcare was important for several of the mothers in this study. Jessica, for example, lives in a state that pays her to take care of her son, who is provided 90 hours of respite a quarter. Lisa had caregivers who helped her throughout the pandemic. Julia was paid to provide respite for her son. Felicia's parents provide respite and did so through the pandemic.

A healthy and functioning family dynamic also provides a type of support. Jessica described her philosophy surrounding family dynamic:

> I know that – that analogy with the airplane oxygen mask, you know, mom having to put that on herself so she can care for her kids and put one on the baby. And I feel like that has to – in a family, especially a family with this kind of dynamic – has to start with a couple. Like they have to be super strong or – or else you don't have the foundation for everyone else to feel secure…that's our goal in this house. (Jessica)

Jessica clearly makes an active effort to cultivate a healthy family dynamic, trying to ensure that her children are taken care of. Other mothers discussed positive experiences and relationships in their families, mentioning the support they receive from parents, their older children, and other family members, while others recounted a lack of support or understanding from their family.

> All three of the boys get along well. Brendan, our oldest, is 25, so there was that big age difference, so Brendan used to take care of Owen and Thomas[13] a lot, when I was still working, and Owen kind of stepped into that role…he's a helper. (Natalie)

[13] Thomas was described as a kid who loves cars, trucks, space shuttles, and swimming (Natalie).

> My dad struggles, sometimes, with Morgan's behavior…I would say that my mother-in-law and father-in-law struggled. And it was like they tried to fix it…like even now my mother-in-low is still trying to, like, 'Oh, she just needs Jesus'…and like this is not how this is working. (Nicole)

Interactions and connection to other parents with children with SLOS were also important for many of the participants.

> Before there was Facebook, there was a Yahoo support group. And that's how [my best friend and I] met. She understands what it's like. (Lisa)

> I'm a little jealous of the parents now who are just finding out because they do have that board and they can go on there and ask questions, and there are a lot of us who can [say] 'hey that's normal, that's okay'. (Natalie)

> Once we moved to the Facebook group, that was a real game changer because, you know, to have – to know so many families around the world, even if you've never met them, you know, they're there and that at least they get it, and that, like, that's invaluable, just to know like, some – you know, somebody in Romania, you know, knows with Smith-Lemli-Opitz syndrome is, and her kid's the same age as mine, and they do the same thing mine does. (Jessica)

The participants' experiences and attitudes toward support, both for them and for their families, suggest that this acts as a protective factor for them.

### *Parental and Caregiver Strain*

Parents and caregivers often face stressors and experience significant strain in their lives. Stress, particularly family stress, can impact resilience. According to Patterson (2002), "the daily caregiving demands for some chronic conditions can lead to physical and emotional exhaustion in parents, which may contribute to depression or other psychological symptoms" (p. 356), and the daily caregiving demands parents of children with SLOS face can be significant.

> Everything has to be with somebody, which is hard sometimes. Like I have to do everything with him or do everything for him" & "Every single

> day I do the exact same thing. I have to wake up, I have to get him ready, I have to do everything to help him, I have to help him use the toilet, I have to help him brush his teeth, I have to help him get dressed, I have to feed him breakfast. You know, it's just – it's just my life now. (Lisa)
>
> [Morgan[14]] doesn't understand kind of the severity of some of the things that she does. So that – it – that stresses me out, you know, because I feel like I'm constantly like helicopter parent. (Nicole)

Parents of disabled or medically fragile children can experience distress for a number of reasons, such as social isolation and the strain of daily caregiving tasks (Patterson, 2002). Caretaking needs were demanding for many of the participants in this study, particularly during the pandemic and virtual schooling. Jessica talked about the stress of doing therapy and exercises at home with her son and how this typically impacts mothers more than fathers, as mothers are often impacted more by issues surrounding their children. Julia shared how difficult it is to try to help her son participate in school and be more independent:

> I can't – I almost started crying when I got off the phone with her, I'm like I don't know, like I can't make him do any – like I can't like 'Okay, go like get yourself dressed now'…I don't know – I can't – what am I supposed to do? I can't like make him do – can't make him pee in the potty, because I – God knows, I would try, like I would if I could. (Julia)

Felicia's caregiving responsibilities make it difficult or impossible to schedule calls or meetings:

> They said, 'well, you can do an appeal [for more respite hours] and we can set up a Zoom meeting', and I'm like 'I don't have time, did you not hear me? I don't have a nurse'. You know, and they're like 'we can – when do you get a break?'. I'm like – Daisy's here all day. I said 'I don't get breaks'…don't add more stress to me. I'm trying to reduce it. (Felicia)

[14] Morgan was described as "a typical teenager" who loves music and plays guitar (Nicole).

**Responses to stress**. Some of the mothers described coping mechanisms or preemptive habits to avoid or deal with stress. Lisa runs, Jessica said she builds self-care into her daily routine, Nicole started taking anti-anxiety medication, and Lena has sought out therapy. Others, like Julia, shared that they struggle with stress and do not having coping mechanisms to help. Natalie expressed her difficulty with handling stressful situations, although she believes she recovers quickly from those situations:

> Um, shut down. Scream. And if I'm not screaming at someone that it's their fault, I'm shutting down" & "I just get drained. And once I'm drained, the stress just kind of follows, I mean it's just, I do what I can every day, I work out just to try to head that off because doctors tell you you should work out, it'll make you happy. I start out happy every day. By the end of the day, I'm a drained mess. (Natalie)

Overall, the participants expressed a variation in stress responses, both reporting positive coping methods and a lack thereof.

### *Resilience*

Respondents were asked at the end of the interview what the word resilient means to them and were subsequently asked if they consider themselves resilient. Overall, the women in this study do feel that they are resilient, or that they at least try to be. The following are their definitions and explanations of resilience:

> It is the ability to…bounce back from a situation, a negative situation. (Christina)

> I feel like the ability to like, be flexible and to, you know, to change course, if need be, and to – to…it's hard to put into words, really…the ability to, you know, like continue on in life when there – when something's – maybe the rug has been completely pulled out from under you or the, you know, - life has just done a 180, but you're still putting one foot in front of the other and carrying on. (Jessica)

> I think it means overcoming obstacles and challenges with a good attitude. Not letting a situation or an obstacle bring you down. (Lisa)

> Like, bounce back. Yeah – bouncing back. (Nicole)
>
> I guess, I don't know, I keep thinking like, bouncing back, like being able to bounce back from something. But I think, keep like – keep going, life if you're resilient, like you just do what you need to do and keep going. (Julia)
>
> Definitely like somebody who doesn't give up. (Lena)
>
> It means bouncing back. I haven't looked up the specific term, but I think that's – that's the way I always use it. Resilient, you can take stress and you can bounce right back, like you've grown from that moment. (Natalie)
>
> Being able to bounce back from bad stuff. (Felicia)

Their understanding of and experience of resilience fit within the most basic conceptualizations of resilience within the social sciences, which define resilience as bouncing back or recovering from an event or situation. More specifically, they fit into the conceptualization of resilience as a process (Uddin et al., 2020). The participants in this study emphasized this idea of bouncing back, not giving up, not being brought down – whether that's in relation to a situation, stress, or a challenge – and spoke about resilience as an action or a process. They spoke about being resilient and carrying on. After they had explained what the word means to them, participants were asked whether they think they themselves are resilient.

> Yes, yes…I think I'm a problem solver by nature. And when I see a problem, I try and figure out a way to fix it, as opposed to sitting and wallowing with the problem. (Christina, resilience score: 3.17 )
>
> Yes, I would. I mean – I strive to be…whatever happens in life, I'm always like, I don't know how I'll get through it, but I know I will. (Jessica, resilience score: 4.00)
>
> Oh, yeah I think so…I just think that trying not to let the worst of things get to you, and just trying to see the positive things makes you a resilient person. And I try to do that, and I try to do that with Bryan. (Lisa, resilience score: 3.50)

> Um, it's not the first thing that I would say about myself, it's not that I can't, but that I try not to let things get to me such that I would need to bounce back, if that makes sense. (Nicole, resilience score: 4.00)

> I don't think I have a choice. When you have kids, I think you kind of have to be. (Julia, resilience score: 2.17)

> I would hope so. I mean it's definitely felt like a never ending battle. So I would hope so. (Lena, resilience score: 3.67)

> I would say, yeah. I feel like I've grown. (Natalie, resilience score: 3.67)

> Somewhat. (Felicia, resilience score: 3.17)

Some of the participants mentioned their children being resilient as well. Lisa shared the following:

> Like I think I use [the word resilient] often about Bryan . He's very resilient. Because he'll just go through surgeries and things in life and challenges and he just always comes out happy. Like I say he's so resilient like, you put – put this obstacle in front of him, and you think it's going to bring him down or make him sad or, you know. And he just was like, 'it's okay', you know, 'what's next? I'll just deal with it'. And he's just – I think he's taught me a lot of like, to be that way too, you know what I mean? (Lisa)

She went on to describe how he has been resilient through medical complications and surgeries, coming out of the procedures happy and carrying on with life. Lisa also touched on his resilience through the pandemic, stating:

> I think all of this with COVID has been very challenging for everybody. And Bryan has been very resilient in all of this. And although he can't understand what's happening or why he couldn't go to school, or why we can't do anything…[he] showed how resilient he can be. (Lisa)

Similarly, when asked if she would call herself resilient, Felicia stated:

> Somewhat. I would say Daisy's very resilient. She can go from being very upset about something and then she can kind of let it go, you know, and yeah she has – she has the bounce back, she's great at it. I'm alright. She's good, she's great. (Felicia)

These mothers appear to view resilience as a process and as something that is continuous. Resilience, for this population, appears to be more of an action – something that they or their children are constantly doing or being, or even being "great at" (Felicia).

**Luck.** A recurring theme throughout these interviews was the feeling of luck, coincidence, or gratitude. This relates to resilience, as the things participants felt lucky about or grateful for could easily be considered protective factors. Often participants would express how they are lucky to have a certain service, or they are grateful that they receive respite, or how fortunate it was that a certain doctor was available or close enough for them to see:

> So it was just happenstance that we found this particular geneticist after 15 years, who had some experience with [SLOS] and just thought, you know, this looks really familiar. (Christina)

> We had to battle with our insurance company…for luck, one of the board members for that insurance company that we had was a doctor at [the hospital where Bryan was born]. (Lisa)

> I know we're pretty fortunate to have so [many services] available for him here. I don't think I've ever wished 'God I wish we had this or I wish we had that'. No, I don't think so. (Lisa)

This feeling of luck was present in discussions around the syndrome itself and their experiences with that, and throughout discussions about COVID-19 specifically. Participants also demonstrated an understanding of their situation in comparison to other families who have children with SLOS, often phrasing that understanding in terms of luck or gratitude. This was done in a general sense (i.e. luck surrounding lack of symptoms or medical problems), as well as their experiences with COVID-19.

> I mean, I guess I could say we're grateful, I mean [the SLOS conference] sort of made us think, you know, we are lucky compared to what other

> families are dealing with, especially families that have lost children to the disease. (Christina)
>
> I mean even like asking [if he'll live by himself], like a lot of parents and families with kids with SLOS, like that's not even something they think about because I mean their kids can't – like that – that – that was never even like a – like a possibility. We are lucky. (Julia)

Jessica expressed a clear understanding of the suffering other families are experiencing, citing various protective and risk factors in her explanation:

> I – I know this pandemic has been brutal for families of kids with disabilities, and I don't think people understand how – just how at risk these kids and families were in the first place, and you know, how challenging life already was, but I just really – it's scary to think about how much families are struggling. Emotionally, financially – and my family's been really lucky…I feel like it's brought us closer. And you know, I am really aware that many, many families have not had that experience and we haven't had to worry about finances and, you know, just – it's – there are, yeah, families struggling all over the place, then – and the food insecure families, who lost two meals a day at school, and you know that's a – that's a huge problem. (Jessica)

These findings are consistent with previous research on resilience, protective and risk factors, and COVID-19. The following chapter provides a discussion of these findings, the limitations of the study, and a conclusion.

# Chapter 5

## DISCUSSION

This study explored the experiences of parents of children with Smith-Lemli-Opitz syndrome during the COVID-19 pandemic. Interviews conducted with eight mothers revealed five broad themes: experiences with SLOS, impacts of COVID-19, support and services, parental and caregiver strain, and resilience. The findings from this study provide insight into the experiences and needs of this population during the COVID-19 pandemic and give us an understanding of resilience as it applies to this specific population. Resilience in this study was conceptualized as a complex interaction between risk and protective factors that results in positive adaptation during or after stressful experiences. The participants in this study identified both risk and protective factors for them and their families during the COVID-19 that had an impact of their ability to function, their mental health, and their experience and response to stress, including access to and availability of additional caregivers (i.e. nurses, family members, babysitters), financial stability, and the time and ability to cope with stressful situations. Furthermore, the findings of this study point to the need to conceptualize resilience as more of a process than an outcome for this population.

Participants reported both positive and negative experiences during the COVID-19 pandemic, citing struggles with routine disruption, access to services, additional strain

due to concerns over health, but also additional family time, bonding, and gratitude for additional time with their child at home. However, difficulties in parenting a child with SLOS were sometimes exacerbated by the pandemic, especially when there were communication barriers with their child or children.

Mothers reported different access to services and support based on a variety of factors, such as location (i.e. available state programs), financial situation, insurance, and the school their child attends. Those services and supports were interrupted by the pandemic. Social support was likewise impacted in some cases due to isolation and distancing requirements.

Parental and caregiver strain was impacted both by the symptoms and difficulties associated with SLOS and the change in routine and daily life caused by the pandemic. The necessity to take care of their child or children at home, to help with virtual learning and therapy, and to mitigate negative experiences their child or children may be experiencing due to frustration with or lack of understanding of the pandemic were all sources of stress for participants in this study. Additionally, some mothers had the added strain of continuing to work through the pandemic.

Results from this study align with similar research, especially with similar research conducted during and in relation to the pandemic. Embregts et al. (2021) published findings from one such study. They found that the mothers in their study experienced positive and negative impacts due to the pandemic, experienced a decrease in personal time and therefore an additional need for self-care, required and employed

different coping strategies, and felt significant fear for their children. All of these themes were echoed in the interviews conducted with mothers of children with SLOS.

All of the participants in this study demonstrated some level of resilience, and all of them considered themselves to be resilient, although some were more confident in their resilience than others. Protective and risk factors such as access to insurance, access to services, level of support, financial stability, and stress/strain were discussed throughout the interviews and appear to be incredibly relevant in the understanding and experience of resilience for this population. The participants' discussions and definitions of resilience point toward resilience being a process for this population, as opposed to an outcome. These women are continuously resilient – it is not a state of being that they can achieve. Stainton et al. (2018) argue that "resilience is a dynamic process by which individuals utilize protective factors and resources to their benefit" (p. 725). They suggest that research utilizing this conceptualization of resilience – what they call a multimodal model of resilience – could be useful from an intervention standpoint. Understanding resilience as a process, and not simply a characteristic or outcomes, also suggests that it can be learned. The findings from this study suggest that the mothers of children with Smith-Lemli-Opitz syndrome experiences resilience as a process, which in turn suggests that there is a possibility of interventions to teach others how to utilize the resources and protective factors that are available to them, particularly during a pandemic.

**Limitations**

There were several limitations to this study, the first being the small sample size. The target sample size going into this study was 15-20 individuals, and that target was

not reached. Having 8 participants limits the possible analyses and makes drawing conclusions difficult, particularly with quantitative data. One explanation for the low response rate could be the COVID-19 pandemic itself. Participants themselves had asked whether other parents were responding, stating that this is a very hectic and stressful time for a lot of them. It is possible that, had the recruitment period been longer, more participants would have volunteered their time and energy for these interviews.

All of the participants in this study were women. While this is consistent with previous research on parents and caregivers, it is a significant limitation. Gaining an understanding of the perspectives of fathers or other primary caregivers would be beneficial in the future.

The sample was also overwhelmingly white. While three women identified as Hispanic or Chicana, there were no participants of any other race. This is not wholly unexpected for this population, as the syndrome is more common in those with European ethnicity (Kelley & Hennekam, 2000) and less common in Asian or Black individuals (Wright et al., 2003), but it is still a limitation of this particular study. Future research should be conducted with this in mind, with efforts made to include a more diverse sample.

**Conclusion**

Despite the limitations of this study, the findings provide a deeper and more complex understanding of the experiences of parents of children with a developmental disorder during the COVID-19 pandemic. Research has shown that parents and families of children with IDDs face unique obstacles that can result in negative outcomes (Manor-

Binyamini, 2020; Singer, 2006), particularly during disaster situations such as a pandemic (Neece & Fenning, 2020; Redquest et al., 2020). This study's findings are consistent with past research, showing that parents of children with Smith-Lemli-Opitz syndrome face a variety of challenges in their day to day lives which were exacerbated by the pandemic, but they also report positive experiences with both the syndrome and their experiences during COVID-19. The insights provided in this study may prove useful for providers and medical professionals. Future research could be conducted specifically on the impact of stress on parents, focusing on detailing what factors contribute to stress in this population and what factors might mitigate or alleviate that stress. Resilience, as a simplified concept, is the ability to bounce back, to recover from stressful or traumatic situations, and research should focus on what factors promote resilience within specific populations while acknowledging that resilience may look very different in different contexts. The interaction between the protective and risk factors demonstrated by the participants' accounts of their experience during the pandemic resulted in resilience in the sense that the protective factors outweighed the risk factors or demands they experienced during the pandemic. This study clearly demonstrates the necessity and importance of those protective factors in the ongoing resilience of this population, particularly during a disaster situation like the COVID-19 pandemic.

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# Appendix

## Appendix A

## QUALTRICS QUESTIONNAIRE[15]

The following questionnaire was designed to provide insight into the experiences and needs of parents of children diagnosed with Smith-Lemli-Opitz syndrome. Please answer the questions truthfully to the best of your ability. There are no repercussions if you decide to skip certain questions or cease participation at any time. All personal identifying information (for example, your name) will be kept confidential.

The following questions are about yourself:

1. What is your name?
2. What is your age?
3. What is your gender?
4. What is your race/ethnicity?
5. Please select your marital status:
    a. Single, never married
    b. Married/Domestic Partnership/Cohabitation
    c. Widowed
    d. Divorced
    e. Separated
6. What type of insurance do you have? Select all that apply.
    a. Private insurance
    b. Medicaid
    c. Medicare
    d. Other:
7. How many children do you have?
8. What is the highest level of schooling you have completed? No schooling completed
    a. Grades 1 through 12

[15] Skip patterns were utilized in the Qualtrics survey.

b. 12th grade—no diploma
c. Regular high school diploma
d. GED or alternative credential
e. Some college credit
f. Associate's degree (for example: AA, AS)
g. Bachelor's degree (for example: BA. BS)
h. Master's degree (for example: MA, MS, MEng, MEd, MSW, MBA)
i. Professional degree beyond bachelor's degree (for example: MD, DDS, DVM, LLB, JD)
j. Doctoral degree (for example, PhD, EdD)

9. (If married or in a partnership) What is the highest level of schooling your partner has completed?
    a. No schooling completed
    b. Grades 1 through 11
    c. 12th grade—no diploma
    d. Regular high school diploma
    e. GED or alternative credential
    f. Some college credit
    g. Associate's degree (for example: AA, AS)
    h. Bachelor's degree (for example: BA. BS)
    i. Master's degree (for example: MA, MS, MEng, MEd, MSW, MBA)
    j. Professional degree beyond bachelor's degree (for example: MD, DDS, DVM, LLB, JD)
    k. Doctoral degree (for example, PhD, EdD)
10. What town/city do you live in?
11. How would you describe the area you live in?
    a. Urban
    b. Suburban
    c. Rural
    d. Other:
12. What is your family's yearly income? An estimate is fine.
13. Do you consider yourself religious?[16]
14. How often do you think about religious issues?[17]
    a. Several times a day
    b. Once a day

[16] If they answered 'no', the survey skipped to question 28.
[17] These questions are drawn from the Centrality of Religiosity Scale (CRSi-14) (Huber & Huber, 2012).

c. More than once a week
d. Once a week
e. GED or alternative credential
f. Some college credit
g. Associate's degree (for example: AA, AS)
h. Bachelor's degree (for example: BA. BS)
i. Master's degree (for example: MA, MS, MEng, MEd, MSW, MBA)
j. Professional degree beyond bachelor's degree (for example: MD, DDS, DVM, LLB, JD)
k. Doctoral degree (for example, PhD, EdD)

15. (If married or in a partnership) What is the highest level of schooling your partner has completed?
    a. No schooling completed
    b. Grades 1 through 11
    c. 12th grade—no diploma
    d. Regular high school diploma
    e. GED or alternative credential
    f. Some college credit
    g. Associate's degree (for example: AA, AS)
    h. Bachelor's degree (for example: BA. BS)
    i. Master's degree (for example: MA, MS, MEng, MEd, MSW, MBA)
    j. Professional degree beyond bachelor's degree (for example: MD, DDS, DVM, LLB, JD)
    k. Doctoral degree (for example, PhD, EdD)
16. What town/city do you live in?
17. How would you describe the area you live in?
    a. Urban
    b. Suburban
    c. Rural
    d. Other:
18. What is your family's yearly income? An estimate is fine.
19. Do you consider yourself religious?[18]
20. How often do you think about religious issues?[19]
    a. Several times a day
    b. Once a day

[18] If they answered 'no', the survey skipped to question 28.
[19] These questions are drawn from the Centrality of Religiosity Scale (CRSi-14) (Huber & Huber, 2012).

c. More than once a week
d. Once a week
e. One or three times a month
f. A few times a year
g. Less often
h. Never

21. To what extent do you believe that God or something divine exists?
    a. Very much so
    b. Quite a bit
    c. Moderately
    d. Not very much
    e. Not at all
22. How often do you take part in religious services?
    a. More than once a week
    b. Once a week
    c. One or three times a month
    d. A few times a year
    e. Less often
    f. Never
23. How often do you pray?
    a. Several times a day
    b. Once a day
    c. More than once a week
    d. Once a week
    e. One or three times a month
    f. A few times a year
    g. Less often
    h. Never
24. How often do you meditate?
    a. Several times a day
    b. Once a day
    c. More than once a week
    d. Once a week
    e. One or three times a month
    f. A few times a year
    g. Less often
    h. Never

25. How often do you experience situations in which you have the feeling that God or something divine allows for an intervention in your life?
    a. Several times a day
    b. Once a day
    c. More than once a week
    d. Once a week
    e. One or three times a month
    f. A few times a year
    g. Less often
    h. Never
26. How often do you experience situations in which you have the feeling that you are in one with all?
    a. Several times a day
    b. Once a day
    c. More than once a week
    d. Once a week
    e. One or three times a month
    f. A few times a year
    g. Less often
    h. Never
27. How interested are you in learning more about religious topics?
    a. Very much so
    b. Quite a bit
    c. Moderately
    d. Not very much
    e. Not at all
28. To what extent do you believe in an afterlife – e.g. immortality of the soul, resurrection of the dead or reincarnation?
    a. Very much so
    b. Quite a bit
    c. Moderately
    d. Not very much
    e. Not at all
29. How important is it to take part in religious services?
    a. Very much so
    b. Quite a bit
    c. Moderately
    d. Not very much

e. Not at all

30. How important is personal prayer for you?
    a. Very much so
    b. Quite a bit
    c. Moderately
    d. Not very much
    e. Not at all

31. How important is meditation for you?
    a. Very much so
    b. Quite a bit
    c. Moderately
    d. Not very much
    e. Not at all

32. How often do you experience situations in which you have the feeling that God or something divine lets something be communicated or revealed to you?
    a. Several times a day
    b. Once a day
    c. More than once a week
    d. Once a week
    e. One or three times a month
    f. A few times a year
    g. Less often
    h. Never

33. How often do you experience situations in which you have the feeling that you are touched by a divine power?
    a. Several times a day
    b. Once a day
    c. More than once a week
    d. Once a week
    e. One or three times a month
    f. A few times a year
    g. Less often
    h. Never

The following questions apply only to you child [or children] who are diagnosed with SLOS. Please keep this in mind when answering.

34. Please list your child's [children's] physical symptoms. Please be as detailed as possible:

35. Please list any behaviors your child exhibits that are characteristic of SLOS. Again, be as detailed as possible.
36. What is your child's living situation? (For example, do they live at home? Do they go between two or more homes? Do they live in a group home?)
37. Please rate your level of agreement with the following statements (strongly agree, agree, neutral, disagree, strongly disagree)
    a. My child's pediatrician has/had a good understanding of SLOS.
    b. My child's current doctor (if different from their pediatrician) has a good understanding of SLOS.
    c. My child's doctors have been willing to learn more about SLOS in order to treat my child.
38. Has your child seen any specialists? If so, please list which specialists.

The following questions are about you. Please answer only for yourself.

Please respond by marking/selecting one answer [strongly disagree, disagree, neutral, agree, strongly agree] for each statement.[20]

39. I tend to bounce back quickly after hard times.
40. I have a hard time making it through stressful events.
41. It does not take me long to recover from a stressful event.
42. It is hard for me to snap back when something bad happens.
43. I usually come through difficult times with little trouble.
44. I tend to take a long time to get over set-backs in my life.

45. How do you think your answers to the previous set of questions would have been different before the pandemic?
46. How many hours do you generally spend actively caring for your child/children a day?
47. How many hours did you generally spend actively caring for your child/children a day before the pandemic?
48. How would you rate your own mental health before COVID on a scale from 0 to 10, 0 being terrible and 10 being excellent?
49. How would you rate your own mental health now on a scale from 0 to 10, 0 being terrible and 10 being excellent?
50. How would you rate your level of stress before COVID on a scale from 0 to 10, 0 being terrible and 10 being excellent?

[20] Brief Resilience Scale (Smith et al., 2008)

51. How would you rate your level of stress now on a scale from 0 to 10, 0 being terrible and 10 being excellent?

## Appendix B

## INTERVIEW PROTOCOL

Introduction and brief, general explanation of why I am researching this.
First, I'm going to ask you some questions about your family members and your child.

1. Please tell me a little bit about the members of your immediate family.
    a. Who do you currently live with?
    b. Tell me a bit more about your child [with SLOS][21] (What is their personality like? What do they like to do?)
    c. What is the family dynamic like?
2. What types of activities does your child enjoy?
    a. Prompts[22]: What do they like about this activity? How long have they been doing this activity? Has this been disrupted by COVID?
3. Walk me through a normal day for you and for your family/child[23] before the pandemic.
    a. How has that changed since the pandemic?
4. Describe to me, in detail, some things you enjoy doing with your child?
5. What was the diagnosis process like?
    a. Prompt: Did you have to travel to see a specialist? Were you *able* to see a specialist?
6. Did your child's primary doctor or healthcare provider know about Smith-Lemli-Opitz syndrome?
    a. Prompt if yes: What did they know? Did you have to search for additional information elsewhere?
    b. Prompt if no: Did they make an effort to become familiar with it?
7. What has your experience with insurance coverage for your child been like?
    a. Prompt: What type of insurance do you have? Your child?
8. If your child exhibits any behavioral problems, describe them for me.
    a. What strategies has your family used to handle these issues?

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[21] "your child" and "child"/"children" were replaced with names in the actual interview

[22] prompts were not needed in every interview

[23] child if there are only 2 people in the home (i.e. mother and child with SLOS), family if there are more members of the household

    b. Do you have access to help and medication for these problems if it's needed?
    c. Have these problems changed since the beginning of the pandemic?
9. How does the rest of your/your partner's family behave around [child's name]?
    a. Prompt: Do they include [child's name] in family activities? Are they accommodating?
10. What kind of help do you have with childcare?
    a. Prompt: Do you feel that this help is sufficient? What kind of help do you need?
    b. Prompt: Has this changed since the start of the pandemic?

Now I'm going to ask some questions about you specifically.

11. To what extent and how has your child's diagnosis impacted your life?
    a. Prompt: Your career? Please provide an example.
12. Thinking back to before COVID began, to what extent are you able to participate in any activities outside your home? In the community?
    a. Prompt: What about now?
13. Describe to me how you typically respond to stressful situations.
    a. Prompt: Do you recover quickly from these situations?
    b. Has your response to stress changed since COVID began?
14. What do you do for yourself to cope with stress?
    a. Prompt: To what extent do you have support from friends and family?
15. Do you consider yourself religious?
    a. Prompt: To what extent do your religious beliefs and activities help you cope with stress?[24]

Next I'd like to discuss support and services for your child and your family.

16. What support services are available to your child, to the best of your knowledge?
    a. Prompt: How did you find out about them?
17. Does your child or family utilize support services?
    a. If yes: What services do you utilize?
    b. If no: Are there any services you would like to utilize? Are you facing any barriers to these services?
18. Are there any services you wish were available that are not?

[24] included if they responded on the questionnaire that they consider themselves religious

19. What do you think would have helped you adapt early on in your child's life?
    a. Prompt: More education on the syndrome? More services? Support?

Finally, I'd like to discuss the impact this pandemic has had on you and your family.

20. How has COVID impacted your life and your child/children's life?
    a. Prompt: How are you coping with stress now?
    b. Prompt: Has your child's access to healthcare changed during the pandemic? What about access to school/day programs/care? (If they have home aids, has that changed?)
    c. Prompt: How has your child responded to these changes?
21. To what extent and how has the pandemic impacted your support system?
22. Has the pandemic impacted your or your family's financial situation?
23. Describe to what extent you are still able to access the programs you need for your child and your family?
24. What does the word 'resilient' mean to you?
    a. Do you believe that you are resilient?
    b. If yes, what do you believe makes you resilient?

**Institutional Review Board**
210H Hullihen Hall
Newark, DE 19716
Phone: 302-831-2137
Fax: 302-831-2828

DATE: July 16, 2021

TO: Megan Wissert, B.A.
FROM: University of Delaware IRB

STUDY TITLE: [1765845-1] Parental Resilience: Adaptation During the COVID-19 Pandemic
SUBMISSION TYPE: New Project

ACTION: APPROVED
EFFECTIVE DATE: July 16, 2021
NEXT REPORT DUE: July 15, 2022

REVIEW TYPE: Expedited Review
REVIEW CATEGORY: Expedited review category # *(6,7)*

Thank you for your New Project submission to the University of Delaware Institutional Review Board (UD IRB). The UD IRB has reviewed and APPROVED the proposed research and submitted documents via Expedited Review in compliance with the pertinent federal regulations.

As the Principal Investigator for this study, you are responsible for, and agree that:

- All research must be conducted in accordance with the protocol and all other study forms as approved in this submission. Any revisions to the approved study procedures or documents must be reviewed and approved by the IRB prior to their implementation. Please use the UD amendment form to request the review of any changes to approved study procedures or documents.
- Informed consent is a process that must allow prospective participants sufficient opportunity to discuss and consider whether to participate. IRB-approved and stamped consent documents must be used when enrolling participants and a written copy shall be given to the person signing the informed consent form.
- Unanticipated problems, serious adverse events involving risk to participants, and all non-compliance issues must be reported to this office in a timely fashion according with the UD requirements for reportable events. All sponsor reporting requirements must also be followed.

The UD IRB REQUIRES the submission of a PROGRESS REPORT DUE ON July 15, 2022. A continuing review/progress report form must be submitted to the UD IRB at least 45 days prior to the due date to allow for the review of that report.

If you have any questions, please contact the UD IRB Office at (302) 831-2137 or via email at hsrb-research@udel.edu. Please include the study title and reference number in all correspondence with this office.

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