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"Doctors can read about it, they can know about it, but they've never lived with it": How parents use social media throughout the diagnostic odyssey

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Abstract

Parents of children with undiagnosed conditions struggle to obtain information about how to treat and support their children. It can be particularly challenging to find communities and other parents who share their experiences and can provide emotional and informational support. This study

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AUTHOR CONTRIBUTIONS

All authors contributed to the study design. CR, HKT, KEO, ND, JK, and JY developed the interview guide. ND conducted the interviews with participants. EB, KEO, ND, and JY performed the coding and data analysis. JAB and MTW contributed the funding for the project and critically revised the manuscript. ND and MH drafted the paper and all authors participated in revisions and approval of the final paper.

Conflict of Interest

Natalie Deuitch, Erika Beckman, Meghan Halley, Jennifer L. Young, Chloe M. Reuter, Jennefer Kohler, Jonathan A. Bernstein, Matthew T. Wheeler, Kelly E. Ormond & Holly K. Tabor have no conflicts of interest to report with respect to the manuscript.

COMPLIANCE WITH ETHICAL STANDARDS

Ethical approval

The Stanford University Institutional Review Board approved all aspects of this study (Protocol IRB-46950). All procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with the Helsinki Declaration of 1975, as revised in 2000. The clinical trial (NCT02450851) was approved by the Institutional Review Board of NHGRI (NIH Study Reference Number 15-HG-0130),

Human studies and informed consent

Informed consent was obtained from all individual participants included in the study.

Animal studies

No non-human animal studies were carried out by the authors for this article

Data Sharing and Data Accessibility

Data presented in this manuscript have not been publicly archived.

sought to characterize how parents use social media, both throughout the diagnostic odyssey and post-diagnosis, to meet their informational, social and emotional support needs. We conducted qualitative semi-structured interviews with 14 parents from the Stanford site of the Undiagnosed Diseases Network (UDN), including five whose children had received a diagnosis through study participation. Interview recordings were analyzed using inductive, team-based coding and thematic analysis based in grounded theory using Dedoose qualitative analysis software. Through this process we identified four key themes related to social media use. First, parents struggled to find the "right" community, often seeking out groups of similar patients based on symptoms or similar conditions. Second, though they found much valuable information through social media about caring for their child, they also struggled to interpret the relevance of the information to their own child's condition. Third, the social support and access to other patients' and families' lived experiences were described as both highly valued and emotionally challenging, particularly in the case of poor outcomes for similar families. Finally, parents expressed the need to balance concerns about their child's privacy with the value of transparency and data sharing for diagnosis. Our results suggest that the needs and experiences of undiagnosed patients and families differ from those with diagnosed diseases and highlight the need for support in best utilizing social media resources at different stages of the diagnostic odyssey.

Keywords

undiagnosed diseases; social media; diagnostic odyssey; rare diseases

INTRODUCTION

Globally, 200–400 million people live with a rare disease. A majority of these are pediatric onset diseases (Nguengang Wakap et al., 2020). Rare diseases are notoriously difficult to diagnose; children with rare diseases often undergo a multitude of tests and evaluations, sometimes over many years, in an attempt to find an underlying etiology for their condition (Mcconkie-Rosell et al., 2018; Pelentsov et al., 2015). The time in which children are "undiagnosed" is often referred to as the "diagnostic odyssey" (Pelentsov et al., 2015; Rosenthal et al., 2001). Despite such extended diagnostic odysseys, a sub-set of children will remain without a definitive diagnosis, leaving them and their families to navigate the healthcare system and complex medical decisions without a clear understanding of the underlying problem they are facing. For these children and their families, being undiagnosed can come with significant uncertainty and insufficient information to support clinical decision-making, and also limited social support from other patients and families who have undergone similar medical challenges (Germeni et al., 2018; Lewis et al., 2010; Lipinski et al., 2006; Pelentsov et al., 2015; Rosenthal et al., 2001; Skinner & Schaffer, 2006; Yanes et al., 2017).

Studies of patients and families coping with diagnosed rare diseases have suggested that, although social support from family and friends can be a valuable resource, there are unique benefits of connecting with people outside the family who share similar medical experiences or shared symptoms (Akre et al., 2018; DeHoff et al., 2016; Van Uden-Kraan et al., 2008). These benefits include: 1) learning about their child's future and prognosis through

connecting with older children in other families, 2) sharing and receiving information about interventions and management with other parents, and 3) finding emotional support from others who understand their current circumstances (DeHoff et al., 2016; Rosenthal et al., 2001; Van Uden-Kraan et al., 2008).

More recently, studies have demonstrated the ways in which patients and families with complex medical conditions and rare diseases have been able to source informational, social and emotional support through social media (Akre et al., 2018; Barton et al., 2019; Dhar et al., 2018; Geense et al., 2017; Gundersen, 2011; Van Uden-Kraan et al., 2008). Beyond facilitating the exchange of information and social support, studies of patients and families with complex medical conditions have found that participating in social media based support groups can enhance feelings of empowerment and disease acceptance, as well as overall well-being (Dhar et al., 2018; Van Uden-Kraan et al., 2008). Social media also brings the added benefit of providing instant, easily accessible support, thereby overcoming barriers to attending in-person support groups, such as geography, time, and mobility limitations. Remote access to support can be particularly valuable for patient populations with rare diseases, which, by default, contain only a very small number of individuals who are typically geographically distant (DeHoff et al., 2016). Parents have also reported that connecting with families with rare diseases online allows them to find information that may be more practical, specific, and applicable to their child's experience than information from a healthcare provider (HCP) (Barton et al., 2019; DeHoff et al., 2016; Geense et al., 2017; Van Uden-Kraan et al., 2008). While social media has been documented as a source of social support for families impacted by rare diseases, it is unclear how having, or not having, a diagnosis plays into a person's abililty to harness that benefit.

Social media-based support is not without its drawbacks. Concerns have been raised about privacy on social media in general populations as well as the impact it can have on mental health (Nicholas et al., 2020). In addition to finding that social media was highly empowering to individuals with complex medical conditions, studies such as Van Uden Kraaden *et al* have found disempowering features in social media based support groups such as uncertainty around quality of information and being confronted with negative sides of the disease (Van Uden-Kraan et al., 2008).

Little is known about the experiences of parents of children with an undiagnosed disease, and if they find social media to be informative and/or supportive, or if they have specific concerns related to using social media.The goal of this study was to explore the experiences of parents using social media throughout their child's diagnostic odyssey, including social media's role in providing both information and support.

METHODS

Participants

We recruited parents and primary caretakers of participants enrolled in the Stanford University clinical site of the Undiagnosed Diseases Network (UDN; https://undiagnosed.hms.harvard.edu/). The UDN is a national research collaboration supported by the National Institutes of Health (NIH). The goal of this network is to use technologies

like genomic sequencing to find diagnoses for patients who remain a medical mystery. However, it also offers the unique opportunity to follow parents throughout their diagnostic odyssey and assess how to better care for and support patients and families with rare and undiagnosed diseases. The UDN consists of 12 clinical sites, as well as a coordinating center and several scientific cores; it enrolls both adult and pediatric participants. To be evaluated by the UDN, an individual must not have received a unifying diagnosis, have at least one objective clinical finding, and have been thoroughly evaluated by other healthcare providers (Clinical Trial ID NCT02450851) (Reuter et al., 2018; Splinter et al., 2018).

Participants were included in the study if they were the parent or caretaker of a child enrolled on the Stanford UDN protocol. Parents were excluded if they were under 18 years old, did not use social media, were unable to complete an interview in English, or if the clinical team felt it was not an appropriate time for them to be contacted (e.g., recent loss of a family member or current medical crisis). Diagnostic status of the child was not used to determine study eligibility as we wanted to assess how receiving a diagnosis might impact social media use. Eligible participants (n=56) were identified by two members of both the UDN clinical team and the study team (CR, JK) and were sent a recruitment email about the study from the Stanford UDN staff, which included a screening questionnaire. This screening questionnaire confirmed inclusion/exclusion criteria and collected some basic information about the child's condition, diagnostic status and the participant's social media use. Recruitment occurred from September 2018 to February of 2019. The Stanford Institutional Review Board approved all aspects of this study.

Instrumentation

Demographics and social media use were collected through an online screening questionnaire and review of the patient's UDN records. A semi-structured interview guide (Supplementary file S1) was developed by a team that included genetic counselors involved with the UDN (JK, CR), genetic counselors with experience in qualitative research (ND, EB, KO) and bioethicists with experience in qualitative and social media research (HKT and JY). Questions were drawn from the existing literaute and from the experiences of the research team. They focused on 1) the experience of having a child with an undiagnosed medical condition, 2) how parents use social media to find information and support in relation to their child's condition, and 3) barriers to accessing social media resources for families with undiagnosed children. In the interview guide social media was defined as being any internet-based platform used to communicate with other individuals that a user may or may not know in person.

Procedures

Data were collected remotely between September 2018 and March 2019. After participant consent, interviews were conducted and audio-recorded using Zoom, a secure HIPAA compliant web conferencing application, by a single interviewer (ND). Participants had not previously interacted with the interviewer and were informed that the research was conducted in partial fulfillment of genetic counseling training. Interviews lasted between 35 and 75 minutes. After interview completion, parents received a $20 Amazon gift certificate for their participation.

Data Analysis

We conducted an exploratory analysis designed to identify emergent themes in the participants' narratives. Our approach drew on the concepts and methods of grounded theory to identify these themes (Strauss & Corbin, 1990). Interview recordings were transcribed verbatim and identifying information was removed. Transcripts were then uploaded to Dedoose qualitative analysis software version 8.1.8. for coding ("Dedoose Version 8.1.8, Web Application for Managing, Analyzing, and Presenting Qualitative and Mixed Method Research Data.," 2019).

The analytic team (ND, EB, JY, and KO) then reviewed a subset of the transcripts to develop a preliminary codebook. Intermediate and final codebooks were created through two rounds of coding of additional transcripts, and the analytic team adjudicated the codebook to consensus at each step. Two members of the analytic team (ND, EB) then independently coded all 14 transcripts with the final codebook and reviewed all coding to resolve discrepancies through consensus. The analytic team then reviewed all coded excerpts to create a list of themes and subthemes, which were then discussed and refined by the entire research team. Due to the challenges in identifying and interviewing parents of children with undiagnosed diseases, we interviewed the maximum number of parents available within the time frame for the study. At the completion of analysis, the research team found that all themes were consistently represented across interviews.

RESULTS

Sample characteristics

Twenty-one parents completed a demographic screening questionnaire (38% of those invited to participate). One parent was excluded because they did not use social media in any context. Fourteen parents (n = 13 mothers, n = 1 father) completed the interview. The remaining did not respond after three scheduling attempts. All interviews were conducted with just one parent. The majority of parents were of self-identified European race/ethnicity, with a range of educational backgrounds and household income brackets (Table I). Consistent with the overall diagnostic rate of the UDN, which is approximately 35%, one third of participating parents had children who had received a diagnosis (n = 5; 36%), while the remainder were still considered undiagnosed (n = 9; 64%) at the time of interview. All parents with diagnoses for their children had received them within five years of the interview date, although the specific amount of time varied by particpant. One child (included in the undiagnosed group) had just received a candidate diagnosis that had not yet been confirmed (n = 1; 7%). For families with a diagnosis, the time reported between symptom onset and receiving a diagnosis ranged from 1.5 to 13 years (mean = 6.57 years). For families without a diagnosis, the time spent between symptom onset and the date of interview ranged from 2 years to 14 years (mean = 7.36 years). When discussing social media use in general (not necessarily in relation to their child's medical condition) all parents reported using Facebook (n = 14; 100%), and all but one used more than one social media platform (n = 13; 93%). YouTube (n = 11; 79%) and Instagram (n = 9; 64%) were the next most frequently used platforms. Almost all (n = 13; 93%) parents belonged to some sort of social media group related to their child's symptoms, primarily on Facebook. Parents described using YouTube

for watching videos to gain informational support about their child's condition or caring for their child, and many used Instagram to follow other families with rare and undiagnosed diseases to learn about their stories.

Navigating social media as the parent of an undiagnosed child

Our analysis of participants' experiences using social media in relation to their child's condition resulted in four key themes: 1) parents' struggles to find the "right" community on social media; 2) the value and limits of medical information from social media; 3) the benefits and challenges of finding social support through social media; 4) balancing privacy and transparency on social media. Below we describe these themes in detail and provide exemplary quotes to illustrate their underlying meaning.

Theme 1: Parents struggle to find the "right" community on social media

Parents with undiagnosed children described the challenges of finding the "right" online community through social media, strategies they used for connecting, as well as the drawbacks of these strategies. In addition, parents of children who had recently received a diagnosis described how the diagnosis had changed their social media use.

Uniquely isolated—Parents described the experience of having an undiagnosed child as a unique form of isolation. While parents of children with undiagnosed diseases described wanting to connect with other individuals who they felt would understand what they were going through, they also described how the lack of a diagnosis for their child(ren) hindered their ability to find such groups. As one mother described:

> "It's not like I can go somewhere and say, 'My kid has some genetic condition,' and talk to families that have a certain diagnosis. It's just a whole bunch of different symptoms together, then I haven't been able to use anything, or talk to anybody that would know exactly what it is." (P14, mother, undiagnosed child)

Parents also attributed their difficulty in finding a group on social media to the heterogeneity inherent to the rare and undiagnosed diseases community. As one mother said:

> "I think the difficulty with undiagnosed cases is everyone's got something different. There's a reason they're undiagnosed. It's because they're going through something that nobody has really never seen before, for the most part. It might kind of look like one thing or kind of look like another, but at the end of the day, it's not either of them." (P11, mother, undiagnosed child)

Parents were asked if they used groups specifically oriented towards those with undiagnosed diseases. A minority of parents actively used such groups, citing that they were too broad and lacked information that was specific to their child's own experience.

Strategic Connecting—Without a diagnosis to help them identify specific groups, parents described using various elements of their child's condition (e.g., seizures, G-tubes) to find others who might have similar experiences. Granular searching allowed them to connect with others who could offer either general or targeted advice for managing shared symptoms. One mother described, "even if the kids had different diagnoses, it was actually

similar enough that you could kind of find some overlap or just like, “Hey, I’m trying to figure out how can I find this piece of equipment? What’s the deal with weighted blankets?” (P12, mother, undiagnosed child)

Parents described belonging to several groups, each focused on different conditions, symptoms or issues, to address specific aspects of their child’s condition. When asked about which groups they belonged to, some struggled to remember all of them because they had joined so many. One mother said, “I tried to make a list of groups, but it was too long, so I gave up.” (P14, mother, undiagnosed child)

Other parents described finding a “close-match” social media group for a diagnosis with not just one, but several symptoms similar to their child, to be somewhat useful. However, even in these relatively similar groups, they described sometimes feeling like outsiders. As one mother explained: “You know our kid’s got an autism diagnosis. They legitimately met the criteria for it. But I almost felt like an imposter in that community because it’s a bit of a stretch. But I needed to join some community – I wanted to leverage the knowledge in that community and the power of a group.” (P8, mother, undiagnosed child) However, being undiagnosed still limited the benefits even a close-match group could provide. As one mother described:

> “When you have a kid that does have a definitive diagnosis, you can look and be like, ‘Oh. Well a kid that’s like my kid should experience X, Y, and Z.’ But our kid does stuff that no other kid does. On social media, we can’t go to, say, the [epilepsy] support group, and ask a question because other kids aren’t going through the same thing.” (P6, mother, undiagnosed child)

Another mother who was very involved with a symptom-based support group summarized this sentiment, saying, “…sometimes I feel like I’m just not in the right “family”. I’m pretty sure that I’m not.” (P4, mother, undiagnosed child)

Diagnosis as social (media) belonging—Parents who had recently received a diagnosis for their child did describe how their ability to use social media changed after receiving a diagnosis. Five participants had received a diagnosis for their children through the UDN. Four of these parents were able to find social media groups for their child’s specific diagnosis. One of these families had two distinct diagnoses for their child and joined social media groups for both. Only the fifth parent, who had received an extremely rare genetic diagnosis for their child, had yet to find another family on social media with the same condition or any support group specific to this rare condition.

Three out of these parents said that finding social media communities was one of the most important consequences of the diagnosis. When parents described joining a diagnosis-specific group, they said they went from feeling like imposters to feeling like they really belonged and had a strong social connection and community. Even in the absence of changes in treatment – the diagnosis still came with the benefit of identifying social support. One mother said, “We knew it [the diagnosis] would be unlikely to find a cure. We’d still be doing things the same way if we had a diagnosis or not. But I think the biggest part was just

to have the support from other people with the same diagnosis." (P10, mother, diagnosed child)

Social media allowed for parents to connect for the first time with families whose children looked like theirs and shared their seemingly unique characteristics. As one mother described this experience, "I came across the group and all the kids in the group looked exactly like my child…When we found the group, I found all these kids that could pass as her twin brothers or sisters, and it was just kind of an eye-opener for us. I remember it really just took my breath away*."* (P9, mother, diagnosed child)

The feelings of isolation expressed by parents of undiagnosed children and the dramatically enhanced social support parents described as resulting from a diagnosis highlight the extent to which having – or not having – a diagnosis impacts the ability of families with rare diseases to access support and information through social media.

Theme 2: The value and limits of medical information from social media

Parents described how participation in social media communities filled gaps in information from healthcare providers, particularly in how to care for an undiagnosed child or a child with a rare condition. Some parents said they found the expertise of social media communities to be more reliable than that of local medical specialists, who might have had limited exposure or training to care for such conditions. By sharing questions, stories and photos, they were able to obtain timely and unique feedback from others with similar symptoms and conditions. As one parent described:

> "You see a geneticist, but he's never seen a kid that has something similar. You can't blame him, it's just a very rare disease. But on Facebook you get, 50, 100, 300 people with similar symptoms. My child has tapered fingers, and we're trying to figure out what this is a symptom of. So, I take a picture of his fingers, and I uploaded it to Facebook group and say, 'Okay, does anyone have these fingers?' Then I can get data that I don't think any doctor can get." (P4, mother, undiagnosed child)

Parents of children with and without a diagnosis also described social media communities as a more accessible source of information. Several parents discuss how social media was available wherever and whenever they needed it. Parents described using social media late at night, when doctors' offices might be closed, "I mean, it's there at 2 a.m. if you're up worried about stuff." (P9, mother, diagnosed child) or to get information during times of crisis such as when their child was in the hospital, "Well, it's much easier because we all have our apps on our phones. When I go to the hospital and waiting anxiously when he's getting a heart catheter I can just post right away, "Hey guys, here I am. This is what's happening. What should I expect?" (P13, mother, undiagnosed child)

Parents also described social media as a valuable source of unique, practical solutions for the day-to-day challenges of caring for their children that were rooted in others' experiences: "Parents have their tricks and their DIY MacGyvering, or just, like, good resources for stuff that the hospital can't recommend. You can patch together things from social media that the doctors can't tell you, because it's not an official or by-the-book kind of way." (P12,

mother, diagnosed child) This was especially true for alternative treatments that might not be shared by doctors. One father said, “They’ll [the group members’] point me to things like supplements that a doctor wouldn’t necessarily have. Because the docs will only point you to things that have been validated and are in the mainstream literature. You come across more advanced or alternative treatments on Facebook.” (P2, father, diagnosed children) In addition, four parents specifically and spontaneously mentioned using social media to find information about Cannabiodiol (CBD) as a therapy, namely for seizures, in their children. One mother said:

> “It’s been helpful medicine wise, what seizure meds people have tried. We’re using CBDs a lot with my daughters for seizures. Being able to talk to people about where they’re getting their CBDs and what dosage because medical marijuana is still new and all. These are things where you can’t get good information through the hospital.” (P7, mother, diagnosed child)

While parents saw social media as a valuable source of information, they also noted concerns and skepticism about such information. Many described the importance of taking information from social media with a “grain of salt”. They recognized that information might not be accurate, or even applicable to their child. As one father described, “ … you have to take those with a grain of salt. Because what works for their kid may not work for yours. You have to sift through that information, which can be time consuming and challenging. You know you don’t want to hurt your kid by trying some kind of crazy supplement or something that worked for one family in [some other location].” (P2, father, diagnosed children) This concern was exacerbated for parents of undiagnosed children who already found that social media groups did not fit their child’s condition.

Most parents articulated their preference to get medical information from providers, who they thought could provide more reliable information. Others felt, however, that their child’s situation was so critical they had to look to other sources, including social media, to fill in the gaps of what healthcare providers could give them.

> “We’re dealing with something where the medical world is unable to help us and we’re going into the world of integrative or alternative measures, which doctors don’t dare to talk about [sic]. These parents are dealing with this reality, so we’ll do everything. If medical can’t help, it will be alternative [sic]. It will be Chinese medicine, whatever it is…. We’re dealing with extremely rare, and extremely high-risk conditions here. All these parents eventually come to a point where we have to believe in miracles, in remedies that nobody would even consider serious. Because that’s what it is. Again and again, the medical world is unable to help us [sic].” (P13, mother, undiagnosed child)

Most parents had not discussed their social media use with their healthcare providers. Some parents thought that healthcare providers could help parents to find appropriate social media communities. Others felt that talking to providers would be embarrassing and a few referred to a negative stigma that providers associate with health information obtained online and through social media.

> "I feel like there's a stigma in researching medical issues online.... It's so hard not knowing answers and watching your kids go through this stuff. You're desperate for answers, so anything and all things should be considered if you think it might relate to your child. I just wished that we felt more open to sharing [with providers]." (P9, mother, diagnosed child)

Parents appeared to struggle to balance their desperation to find information to help their child and their desire for reliable evidence, ideally from a physician.

Theme 3: The benefits and challenges of finding social support through social media

In addition to practical and treatment information, parents also described the value of having information from the lived experiences of other members of social media groups, as a form of social support. As one mother commented, "Doctors have given me the knowledge, but the Facebook groups have given me other people's experiences, which is equally as valuable. Doctors can read about it, they can know about it, but they've never lived with it." (P9, mother, diagnosed child)

Parents also described drawing on the lived experiences of other children with similar symptoms as a potential source of information about their own child's future. One mother described seeing what another parent posted:

> "'This is what my kid's doing! She's taking steps.' And it's like, 'Okay, that's a potential future for our daughter.' She's going to take steps on her own, she's going to be able to use an iPad for communication... It pushes us to work harder on things. And to hope that that work will eventually lead to her doing things and progressing." (P10, mother, diagnosed child)

In addition, parents with undiagnosed children described social media as a source of hope for the future, and specifically that their child might eventually receive a diagnosis. Hope of finding a diagnosis was especially true when parents were able to connect with others who were on a similar diagnostic odyssey. As one mother said, "there have been a couple times where someone's posted 'oh my god, we got a diagnosis', and that's pretty incredible to think that someone who was in my shoes is actually not there anymore." (P6, mother, undiagnosed child) Through social media, parents were able to access unique and deeply personal information and connect with other families whose experiences felt directly relevant to their own very unusual medical issues.

While parents recognized the value of social media as a potential source of social support, they also described the emotional roller coaster that came with connecting with other families dealing with undiagnosed and/or rare diseases. As one mother said, "You get a lot of support, but then you're also face-to-face with some of the worst-case scenarios. It's a double-edged sword, I guess. It can be a great tool, but it can also be a heartbreaker at the same time." (P9, mother, diagnosed child)

Seven parents talked about the specific impact of posts about children dying, forcing them to consider their child's own potentially limited life expectancy. One mother said,

> "There are a lot of funerals posted on those groups and I mean, she's five. We didn't expect her to make it this far at one point. It's scary either way. If she lives to be 50 it's terrifying, because how am I going to be taking care of her and lifting her and all of that stuff is very scary." (P12, mother, undiagnosed child)

In addition, for families without a diagnosis, it was particularly difficult for them to know if the negative outcomes they saw posted by other families on Facebook would apply to their own child, leaving them with more uncertainty. Some described balancing the benefits of the information and support from the groups with the challenges of scary and/or challenging information, choosing to titrate how often to read information from the groups.

> "You read about all these things and you didn't know what to expect. Knowledge is power, but it's also very scary, especially if you don't know what you're dealing with. You also don't want to waste energy being scared of something that isn't relevant. But I don't want to miss something as well. You know, you have to read. You can't put your head in the ground and ignore it. Yeah, it's kind of a balance. You can't read every day." (P4, mother, undiagnosed child)

Recognizing the emotional toll social media could have, three parents described carefully curating what they shared on social media because they did not want to overly trouble other families in their disease-related social media groups. They were hesitant to share information about their child that might worry others. One mother said: "Part of what makes it hard for me to participate too much in these specialized groups is that my daughter is, for parents that are just getting a diagnosis for their kids with infantile spasm[s], my child is, in their mind, a worst-case scenario. I don't want to scare other parents." (P6, mother, diagnosed child)

Theme 4: Balancing privacy and transparency on social media

Parents expressed a range of concerns related to their family's privacy and sharing their children's information on social media. Some concerned parents described "lurking" in groups, or reading information others posted, but not sharing their own personal information, stories or questions. As one mother explained: "…we're pretty private, so I definitely would just – you know I reserve things that I talk about to people – that I've connected with – that I trust." (P1, mother, undiagnosed child).

The majority of parents reported taking note the privacy settings of the groups they joined. While more open or closed privacy settings did not seem to impact if they would join a group or not, parents were generally more comfortable posting in smaller, more targeted groups with tighter privacy settings.

A few parents expressed concern for the ways in which their posting of information might negatively affect their children in the future, in both personal and practical ways. One mother articulated specific risks related to insurance: "Once they become adults and how [sharing medical information] is going to affect them later in life, like with insurance. I don't know if they'll be able to ... they may not be able to, but life insurance policies or anything like that. I don't know how that would affect them." (P14, mother, undiagnosed children). Another mother wondered how her child might feel about her sharing in the future:

> "Well, he's only six right now, so he doesn't use social media, but I think if I were to put more information out there and really share his story or show day-to-day, he needs this, so we have to do this to help him, or he's unable to do this, then one day he would see that, and I wouldn't want him to feel insufficient because I had shared that information or struggles." (P11, mother, undiagnosed child)

On the other hand, parents also argued that the potential benefits of sharing their child's information on social media outweighed their privacy concerns. One mother explained how her desperation to find a diagnosis influenced what she was willing to post: "When it comes to [Child's] diagnosis, really again, I'm like it's everywhere. I told someone if I can just put his DNA on Google for everyone to look, if that gets me diagnosed, I don't care." (P4, mother, undiagnosed child). Another mother explained that her privacy concerns diminished as the gravity of her son's condition came into focus.

> "No, initially I was very careful about everything because I am all for privacy. Keep things, and I used to think oh my goodness, he's going to find out that I posted about him and he's 15 and he's going to hate me for that. Then I realized we're dealing really, I mean we're dealing with death. Anything I can share, actually, is in the hope of finding something to make his life better, extending it, making it more quality or maybe finding a cure." (P13, mother, undiagnosed child)

In addition, parents also described a certain degree of resignation to maintaining privacy, as well as an obligation to speak for their children when they could not do so for themselves. As one mother recounts:

> "...I made a decision in the very beginning to be just super, super open. I mean, her genetic stuff is all over out there in the world, I'm sure. It's all the studies and all the HIPAA forms I've signed, and the blog. But that was just my decision, and if ... I mean, I can see it might've been more awkward if she'd been more able to speak for herself, in a way. I don't know if I would've felt the same way, but I'm her voice." (P12, mother, diagnosed child)

Parents' discussions of privacy highlights the delicate balance that parents must find in deciding how to approach their social media activity. They express a strong need to protect their child's privacy and ability to access resources, such as insurance, but at times the gravity of their children's condition and the urgent need for information can outweigh those concerns.

DISCUSSION

This study provides an in-depth exploration of parents' experiences using social media throughout the diagnostic odyssey. Parents described social media as a source of information that doctors could not or would not provide, as a window into the lived experiences of other families struggling with the complex, rare conditions, and as a source of accessible social support support. However, this valuable resource was not without its drawbacks, as parents also struggled to cope with being exposed to the many challenges faced by other families, including the decline and death of other children. In addition, while some parents raised concerns about privacy when discussing their child's condition over social media, this

concern was tempered by the perceived value of sharing information as part of the search for a diagnosis.

Our results resonate with the existing literature on social media use among parents of children with diagnosed rare diseases, and highlights both empowering and disempowering features (Van Uden-Kraan et al., 2008). However, these studies have focused primarily on families who already have a diagnosis for their child (Barton et al., 2019; Gundersen, 2011; Jacobs et al., 2016; Pelentsov et al., 2015; Roche & Skinner, 2009). Barton *et al.* described social media use at different points during the genetic testing process, and identified that a lack of diagnosis can be barrier to social support. Our focus on families who remained without a diagnosis despite significant medical evaluation allowed us to build on the preliminary findings of Barton *et al.*

We highlight specific challenges faced by parents of children with undiagnosed diseases when using social media that are distinct from parents with diagnosed rare diseases. For example, when parents of undiagnosed children join social media groups, they strategically identify groups based on symptoms or “close-match” diagnoses, but still struggle to determine the relevance of the experiences they encounter on social media to their own situations. The lack of a unifying diagnosis may lead parents feel like outsiders or even imposters within existing rare disease communities. This is consistent with evidence that parents of undiagnosed children reported lower rates of social support when compared to parents of children with a diagnosed medical condition (Yanes et al., 2017). Given the well-documented importance of social support for mental health, and the high rates of anxiety and depression in this population (Lipinski et al., 2006; Mcconkie-Rosell et al., 2018), helping parents leverage social media to find support could be highly beneficial.

Creating social media communities designed to support the undiagnosed diseases is one way to better address the gaps in support. Organizations such as Syndromes Without a Name (SWAN) (SWAN; https://www.undiagnosed.org.uk/) and a handful of parent-initiated groups have done just that, by creating family-focused forums. Similarly, the UDN PEER group was specifically created for those enrolled in the UDN (UDN Peer; https://undiagnosed.hms.harvard.edu/peer/). However, the low participation in these groups among our participants suggests that while these groups could provide some a sense of comraderie in the experience of being on the diagnostic odyssey itself, the breadth and heterogeneity of undiagnosed conditions as a whole appeared to make them less valuable for tangible, practical support. While a person with a diagnosed disease may be able to find informational, social and emotional support in a one-stop group specific to their experience, groups focused on the experience of being undiagnosed may be more likely to be just one piece of a diverse portfolio of necessary support groups.

Key issues for parents in this study included the use of social media to find complimentary and alternative therapies for their child, as well as concerns surrounding privacy of their child’s medical information on social media. In both of these examples parents voiced a preference to be more private or to get medical advice from their healthcare providers, but it seems that the reality of being undiagnosed pushed some to utilize social media in potentially riskier ways out of perceived neccessity.

Our study indicates that some parents may be gathering extensive information on various strategies for care management and treatment of their children through social media. However, it also appears that some may not discuss the information they are gathering on social media with their physician or feel embarrassed to do so, including information on potential alternative therapies. Concerns about disclosure of alternative therapies have been noted with other complex and life-threatening diseases, such as cancer (Asadi-Pooya et al., 2019; Stub et al., 2021). However, parents struggling through an extended diagnostic odyssey may be especially frustrated and skeptical about the ability of evidence-based medicine, and its practitioners, to meet their child's needs and may more actively use social media to find such therapies. This highlights the importance of healthcare providers being open to engaging parents in a conversation about the information they are encountering through social media.

Parents sharing information about their children on social media is becoming more prevalent. Parental sharing of children's photos and personal information from a young age has been described using a new word – "sharenting" (Duggan et al., 2015; Keith & Steinberg, 2017; LaFrance, 2016; O'Keeffe et al., 2011). This creation of a child's digital identity well before the child is able to speak for him or herself has raised a number of ethical, legal and social concerns regarding children's right to privacy in the age of social media (Children's Online Privacy Protection Rule ("COPPA") | Federal Trade Commission, n.d.; Keith & Steinberg, 2017). While our results suggest that parents of children with undiagnosed diseases do consider their child's privacy in their social media activity, they also perceive a significant potential benefit to sharing their child's information – connecting with others with similar genetic variants and/or symptoms and potentially finding a diagnosis. For the undiagnosed community a heightened need for support may skew the balance of benefits to risks of sharing on social media, perhaps even more so than in parents with diagnosed rare diseases.

Practice implications and research recommendations

Helping families access informational, social and emotional support is an essential role for genetic counselors. Counselors working with families with rare and undiagnosed diseases can better care for their patients by discussing strategies for finding more valuable social media-based support (such as providing a list of helpful resources or search terms to find groups) and providing anticipatory guidance and context information they might come across in social media groups (such distressing posts about other children). Additionally, genetic counselors may help families weigh the benefits and risks of sharing personal medical information online and come to their own decision about what works best for their family.

When it comes to issues about use of alternative therapeutics, genetic counselors may be in a unique postion to serve as a bridge for families who are uneasy in sharing such information directly with their physicians. Genetic counselors should consider engaging parents in conversations about the information they are encountering through social media, and how they might be applying that information to the care of their own child, in a non-confrontational or judgmental manner. Such conversations would likely be beneficial

to most families – diagnosed or undiagnosed – however, it is particularly relevant for those struggling through an extended diagnostic odyssey. Providing an environment in which parents feel comfortable disclosing all supplements and alternative medications will encourage them to involve their doctors in conversations about alternative therapies, rather than leaving them to rely on social media for such information. This is essential not only for safe medication management, but also for fostering a supportive relationship with the child's family.

Future research should focus on understanding the core values and concerns of familes with undiagnosed diseases and how they specifically impact decision making around privacy and therapeutics (Courbier et al., 2019). Studies should also explore targeted interventions for helping families on the diagnostic odyssey to navigate social media platforms for informational, social and emotional support. More nuanced guidelines may be needed for clinicians when talking to their undiagnosed patients and families about data sharing to help maximize benefits and minimize risks.

Study limitations

This study has a number of limitations, including, but not limited to, a small sample size drawn from a population that is, by its very nature, heterogeneous. However, it is also a geographically dispersed and sometimes difficult-to-reach population, and families are often overwhelmed with complex medical needs and may not have time to participate in research. We have attempted to overcome the heterogeneity by focusing on the impact of caring for a child who has – or has had – an undiagnosed disease as a central focus. In addition, as our sample was drawn from existing UDN patients, it may not be representative of the broader population of parents of children throughout the diagnostic odyssey. Parents with children enrolled in the UDN may be particularly motivated compared to other parents of children with undiagnosed diseases who may not have sought out such a resource. Further research is needed to confirm the generalizability of our findings in the broader undiagnosed community.

CONCLUSIONS

Our findings suggest that parents of children with undiagnosed diseases use social media as a valuable source of information and social support. However, these parents also face unique challenges related to finding the right community and evaluating the relevance of information and others' lived experiences for their own child's condition. They express nuanced perspectives with regard to data sharing and privacy and use of alternative therapies. Our findings point to a gap in essential social support for many parents of children with undiagnosed conditions, and also significant informational needs that either are not or cannot be met by their healthcare team. Providers and national organizations working with rare disease communities should consider more targeted outreach and specific resources that may leverage social media platforms to patients and families living with undiagnosed rare disease and that are tailored to their unique information and social support needs.

Supplementary Material

Refer to Web version on PubMed Central for supplementary material.

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WHAT IS KNOWN ABOUT THIS TOPIC

Families of children with undiagnosed diseases are known to face unique challenges in managing their diseases and have overall lower coping.

WHAT IS NEW ABOUT THIS TOPIC

Social media can be a highly valuable tool for parents of children with undiagnosed diseases, as it allows them to find informational and social support outside what the medical system can provide. However, it can also present challenges to parents as they struggle to find connections, navigate posted information, and balance privacy concerns.

Table I:

Demographic Characteristics

Parent Gender (N=14)	**n (%)**
Male	1 (7%)
Female	13 (93%)
Age of Parent	
25–34	2 (14%)
35–44	8 (57%)
45–54	4 (29%)
Employment Status	
Working full-time outside of the home	3 (21%)
Working full-time from home	2 (14%)
Working part-time outside of the home	3 (21%)
Working part-time from home	1 (7%)
Full-time caretaker	4 (29%)
Full-time (from home and outside the home)	1 (7%)
Highest Degree of Education	
High school degree or equivalent (e.g. GED)	
Some college, no degree	2 (14%)
Associate or Bachelor's degree	6 (43%)
Master's degree	5 (36%)
Professional degree (PhD, MD)	1 (7%)
Household Income	
Less than $30,000	2 (14%)
$30,000 to $49,999	1 (7%)
$50,000 – $99,999	6 (43%)
$100,000 – $149,999	2 (14%)
$150,000 – $199,000	1 (7%)
$200,000 – $249,000	2 (14%)
Ethnicity	
Caucasian	8 (58%)
Hispanic/Latino	3 (21%)
Asian	2 (14%)
African American	1 (7%)
Number of Affected Children	
1	10 (71%)
2	4 (29%)
Diagnostic Status of Child (ren)	
Undiagnosed	8 (57%)
Diagnosis Confirmed	5 (36%)

Candidate Diagnosis	1 (7%)
Primary Symptom Type of Child(ren) (N=17)	**n (%)**
Neurologic	13 (76%)
Musculoskeletal	2 (12%)
Gastroenterology	1 (6%)
Hematology	1 (6%)
Pulmonology	1 (6%)
Age of Affected Child (n=17)	
Median	7.5 years old
Range	3–17 years old