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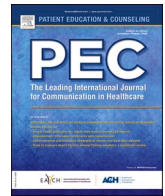
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Navigating care for rare diseases: Caregiver and patient advice for families and clinicians managing care for vascular malformations

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ABSTRACT

Objective: Families affected by rare diseases face many challenges finding adequate care and often report poor communication with clinicians. In the current study, we explore patient and caregiver advice for families and clinicians in the context of complex vascular malformations (VMs), a condition that is frequently misunderstood and misdiagnosed.

Methods: We performed semi-structured interviews with 21 adult patients with complex VMs and 24 caregivers of children with VMs. We analyzed the transcripts using thematic analysis.

Results: Participants advised patients and caregivers to advocate for care, address mental and emotional well-being, seek social support, and promote self-management and self-care. Participants advised clinicians to show care and concern, show commitment, empower and validate, communicate information clearly, address mental/emotional well-being, acknowledge the broad impact of disease and treatment, acknowledge your limitations, work as a team, and commit to learning.

Conclusion: Participants' advice revealed challenges related to family-centered communication and patient and caregiver quality of life and demonstrated the importance of self-advocacy and social support.

Practice implications: The result of this study can help newly-diagnosed families overcome challenges related to care and communication. Clinicians can also use the results to support families by offering them our accompanying handout to validate families' experiences and relay this advice.

1. Introduction

Approximately 30 million people in the United States live with a rare disease [1]. Patients with rare diseases face many challenges including delayed diagnoses, lack of specialized and coordinated care, and limited treatments [2]. Patients and caregivers report poor communication with clinicians due to their limited knowledge of rare diseases [3,4]. Communication with inexperienced clinicians can result in unmet information needs, misunderstandings, and inadequate support [3]. Consequently, patients often lose trust in the health care system and experience feelings of fear, frustration, and dismissal. Caregivers of children with rare diseases also report being dissatisfied with the support they receive from clinicians and frustrated by the lack of knowledge about their child's condition [4]. Ongoing communication challenges between clinicians and patients/caregivers can affect access to care,

quality of medical decisions, patient/caregiver self-efficacy, and, ultimately, physical and mental health outcomes [5].

Vascular anomalies are one group of rare diseases affecting infants, children, and adults that are often misdiagnosed and mistreated [6]. Vascular anomalies include vascular tumors and malformations that result from irregular development of blood or lymphatic vessels [7]. While some vascular anomalies are uncomplicated, or even resolve on their own, others are more complicated and invasive [6]. In the current study, we focus on the experiences of patients and caregivers managing complex vascular malformations (VMs). Complex VMs can be progressive and, if left untreated, result in pain, disfigurement, and organ and musculoskeletal dysfunction [8]. Some complex VMs are also associated with rare syndromes characterized by limb, bone, soft tissue, and lipomatous overgrowth. Consequently, patients with complex VMs should receive prompt referrals for evaluation from a multidisciplinary team of

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specialists [9]. However, due to the rarity of the condition and the lack of knowledge about vascular anomalies among clinicians, patients often struggle to find credible clinicians and get the care they need [10,11].

When patients and caregivers do not receive support from medical professionals, they often turn to other patients and caregivers living with the same condition for support and advice [12,13]. Advice refers to messages that offer guidance and, in the context of illnesses, are often considered a form of social support that helps facilitate coping and uncertainty management [14,15]. Exploring advice within particular health contexts can reveal gaps in the system of care for a particular condition, i.e. VMs, and challenges in the “communication work” involved in managing medical conditions [15]. Previous research has not explored advice in the context of complex VMs. Additionally, few empirical studies have integrated the advice of patients and caregivers. Therefore, the goal of the current study is to explore patient and caregiver advice for families and clinicians caring for complex VMs.

2. Methods

We conducted semi-structured interviews with adult patients and caregivers of children with complex VMs. The consolidated criteria for reporting qualitative research (COREQ) checklist was used during study development and reporting [16]. The Washington University institutional review board approved the study.

2.1. Recruitment

We recruited participants through two support groups for complex VMs: K-T Support Group and CLOVES Syndrome Community. These groups provide support for patients and families with complicated vascular anomalies associated with overgrowth syndromes [17]. Each group advertised via email, social media, and websites. Two authors (BAS, AMK) also hosted a webinar to introduce the study. Caregivers were eligible if they had a child (< 18 years) with a VM. Adult patients were eligible to participate if they were diagnosed with VM and were between the ages of 18–39, based on the National Cancer Institute designation of young adults [18]. While we purposively sampled for diversity in terms of sex, race, and socioeconomic status, recruitment was affected by the diversity of the organizations’ memberships.

2.2. Data collection

After providing verbal consent, participants completed an online survey to self-report age, race, ethnicity, income, education level, relationship status, and insurance status. All participants were asked to indicate their (or their child’s) diagnosis, the severity, and the extent to which the illness interferes with the individual’s life. Given that some participants may still be seeking an expert diagnosis, we included a list of diagnoses consistent with the International Society for the Study of Vascular Anomalies classification of vascular anomalies and related syndromes [17], and also provided an open-ended response option for participants to self-describe their condition. Two authors (BAS, AMK) conducted one-on-one semi-structured interviews via telephone or videoconferencing software. The interview guide asked participants about getting a diagnosis, accessing care, and communicating with clinicians. After asking participants to reflect on their experiences, we asked them to give advice to other patients, caregivers, and clinicians: 1.) Imagine you are talking to another patient/caregiver who is trying to get medical care for their/their child’s vascular anomaly. What advice would you give them? 2.) What advice would you give to doctors who care for patients with vascular anomalies? All participants received a \$40 Amazon gift card. Interviews were audio-recorded and professionally transcribed.

Table 1
Participant demographics.

	Caregiver (n = 24)		Adult Patients (n = 21)	
	n	(%)	n	(%)
Gender				
Woman	21	(87.5%)	19	(90.5%)
Man	3	(12.5%)	1	(4.8%)
Non-binary/third gender	–	–	1	(4.8%)
Race^a				
White	22	(91.7%)	19	(90.5%)
Black or African American	1	(4.2%)	2	(9.5%)
Asian	–	–	2	(9.5%)
Ethnicity				
Hispanic or Latin Origin	4	(16.7%)	2	(9.5%)
Education				
High School Degree (or equivalent)	–	–	2	(9.5%)
Some College	9	(37.5%)	4	(19.0%)
College Degree	8	(33.3%)	10	(47.6%)
Graduate or Professional Degree	7	(29.2%)	5	(23.8%)
Household Income^b				
\$24,999 or less	1	(4.2%)	2	(9.5%)
\$25,000 - \$49,999	3	(12.5%)	1	(4.8%)
\$50,000 - \$74,999	1	(4.2%)	2	(9.5%)
\$75,000 - \$99,999	6	(25.0%)	2	(9.5%)
\$100,000 or greater	11	(45.8%)	11	(52.4%)
Relationship Status				
Married or Living as Married	20	(83.3%)	8	(38.1%)
Never Married	2	(8.3%)	12	(57.1%)
Divorced or Separated	2	(8.3%)	1	(4.8%)
Child’s Gender^c				
Boy	11	(45.8%)	–	–
Girl	12	(50.0%)	–	–

^a Race and ethnicity responses were missing for one caregiver.
^b Income responses were missing from three adult patients.
^c Missing data from 1 caregiver for child’s demographic data.

Table 2
VA diagnoses.

	Caregivers’ Children ^a		Adult Patient	
	n	(%)	n	(%)
Self-Reported Diagnosis^b				
<i>Vascular Anomaly Diagnosis</i>				
Lymphatic Malformation	10	(41.7%)	11	(52.4%)
Venous Malformation	10	(41.7%)	15	(71.4%)
Fibro-Adipose Vascular Anomaly (FAVA)	9	(37.5%)	8	(38.1%)
Capillary Malformation or Port-Wine Stain ^c	8	(33.3%)	1	(4.8%)
Arteriovenous Malformation	4	(16.7%)	1	(4.8%)
Hemangioma	3	(12.5%)	2	(9.5%)
Central Conducting Lymphatic Anomaly	–	–	1	(4.8%)
Kaposiform Lymphangiomatosis (KLA)	1	(4.2%)	–	–
Lymphatic vascular malformation	1	(4.2%)	–	–
<i>Symptom</i>				
Limb Differences	1	(4.2%)	–	–
Macrodactyly	1	(4.2%)	–	–
Macrocephaly-Capillary Malformation (MCM)	1	(4.2%)	–	–
<i>Associated Syndromes or Disorders^b</i>				
CLOVES Syndrome	9	(37.5%)	4	(19.0%)
Klippel-Trenaunay (K-T) Syndrome	2	(8.3%)	7	(33.3%)

^a Missing data from 1 caregiver for child’s diagnosis.
^b Categories not mutually exclusive.
^c Self-reported categories that do not align with the ISSVA classification were included to represent participants’ current knowledge of their condition.

2.3. Data analysis

We analyzed survey data using SPSS 28.0 and analyzed qualitative data using thematic analysis to identify themes in participants’ advice [19]. All coding was completed using Dedoose qualitative analysis

Table 3
Summary of themes and concepts.

	Quote	Concept
Advice for Patients/Families		
Advocate for Care	<i>“Always advocate for your child, and never stop with just one doctor.”</i>	Advocacy
Address mental and emotional well-being	<i>“It is okay to talk about it and acknowledge that it’s something that you’re living with every day.”</i>	Emotional well-being
Seek social support	<i>“Join and find your community [...] it really makes a huge difference. [...] If we didn’t have social media, we would [...] definitely be in different place ‘cause it’s so easy to be in a silo and feel like, ‘oh, nobody understands, and nobody’s gonna help him.’”</i>	Social well-being
Promote self-management and self-care	<i>“Recognize that there are options available for pain relief and that it’s important, take care of yourself, practice self-care.”</i>	Social/physical functioning
Advice for Clinicians		
Show care and concern	<i>“Actively show that you care about the patient’s well-being, that you care about their life.”</i>	
Demonstrate commitment	<i>“It’s really important to not make your patient feel like a burden, but to—with every step of the way, show your patient that you are actively invested [...]. That has made a world of difference to me, having doctors that will repetitively and routinely reach out to me, even if I’m having a complication that doesn’t fall within their specialty.”</i>	Fostering relationships
Empower and validate	<i>“Emphasize that the parent and the caregiver are the quarterbacks, and the support system is there to work with them as a team.”</i>	
Communicate information clearly	<i>“You’re dealing with children and their parents, and they might be experts in a particular field, but it’s—if it’s not medicine, slow down, speak as plainly as possible, and repeat yourself a few times.”</i>	Exchanging information
Address mental and emotional well-being	<i>“I wish they knew the emotional toll that it takes. Obviously, doctors are very focused on the physical aspect of the condition.”</i>	
Acknowledge broad impact of disease and treatment	<i>“I would like them to honor that the diagnosis or the lack of a diagnosis impacts more than the patient. The whole family is involved here. I would like them to just know the impact, the ripple effects.”</i>	Responding to emotions
Acknowledge your limitations	<i>“Admit when you don’t know all the things you need to know. Find somebody else who does. Don’t take it personally.”</i>	
Work as a team	<i>“Get connected to the teams that see a larger number of patients [...] and work with them so like that, we have more knowledge out there throughout the country. [...] then we as parents wouldn’t have to travel so much for care.”</i>	Managing uncertainty
Commit to learning	<i>“Please keep up with the reading. If you have a patient who comes to you with [a] syndrome or just</i>	

Table 3 (continued)

Quote	Concept
<i>vascular malformations, please take a little time to read up on it and consider how that condition might factor into the overall treatments.”</i>	



Fig. 1. Advice to families.

software. Two authors (BAS; AMK) read all transcripts to familiarize themselves with the data and then descriptively coded five transcripts (10%) to generate preliminary codes. During the first coding meeting, we organized the codes into categories refined through iterative cycles of independent coding and consensus meetings. We reached saturation after 10 transcripts when no new categories were identified. A third coder (CB) joined to apply the final codebook to all 45 transcripts by independently coding transcripts, reviewing the other’s coding, marking disagreements, and resolving disagreements through discussion. The final categories represent 100% agreement among coders.

3. Results

We completed 45 interviews with caregivers (n = 24) and adult patients (n = 21) representing multiple VM diagnoses and symptoms (see [Tables 1 and 2](#)). The majority of the participants identified as women. All caregivers identified as parents caring for children from infancy to 16 years old (M=10 years). On a scale of 1–10, caregivers reported a high disease severity (M=7.0; SD=2.4) and interference (M=7.3; SD=2.2). The average age of adult patient participants was 20 years (Range: 18–30 years). On a scale of 1–10, adult patients reported a high disease severity (M=6.3; SD=1.8) and interference (M=7.4; SD=1.8). All caregivers (n = 24; 100%) and all but one adult patient (n = 20; 95.2%) reported that their (or their child’s) health care is covered by insurance. This sample is consistent with those individuals who participate most often in patient and family support groups.

Overall, participants shared advice for families and advice for clinicians related to advocacy, well-being, and patient-centered care (see [Table 3](#)).

3.1. Advice for families

Participants’ advice for caregivers and patients revealed four themes ([Fig. 1](#)) that highlight the importance of self-advocacy [[20](#)], while also attending to well-being [[21](#)].

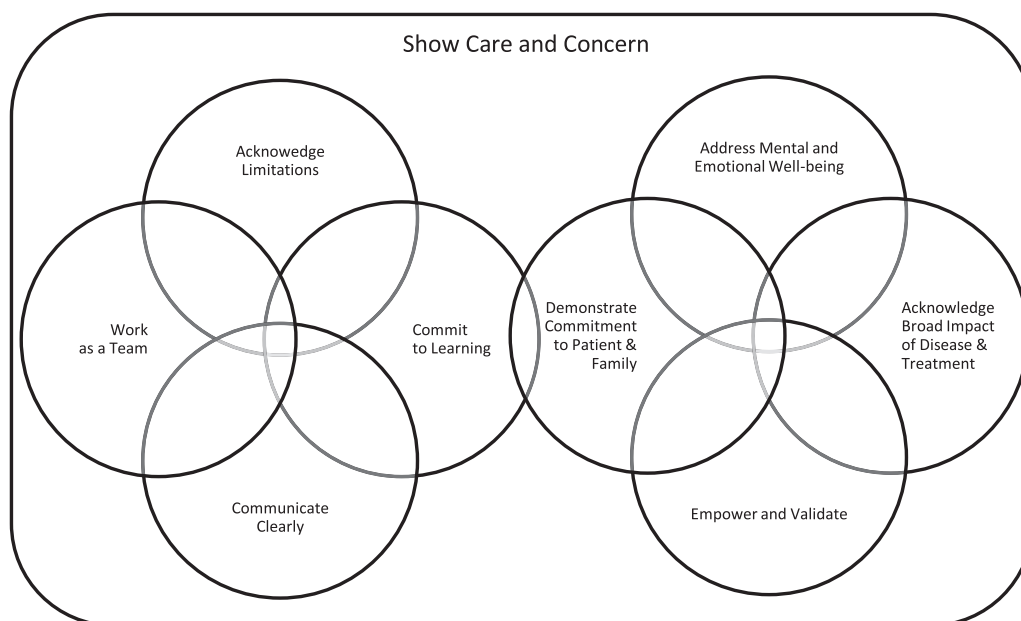


Fig. 2. Advice to clinicians.

3.1.1. Advocate for care

Participants advised others to advocate for care by obtaining needed information, staying organized, coordinating visits, asking for second opinions, and seeking expert clinicians. Caregivers recommended being assertive and perseverant to get children optimal care:

Always advocate for your child, and never stop with just one doctor. [...] It is really scary in the beginning but once you have a doctor who is willing to help you and is willing to set up a game plan, then that's when you know you're on the right path. (P42)

Caregivers emphasized the need to “not give up,” “keep pushing,” “be the squeaky wheel,” and “not worry about upsetting people.”

Patients offered similar advice, even recommending patients “be annoying” to get the care they need. For one participant, the regret of not self-advocating shaped their advice:

I wish I had been a better advocate for myself because you're the one that's gonna care about it the most. (P47)

3.1.2. Address mental and emotional well-being

Participants advised others to attend to their mental and emotional needs. Caregivers cautioned about the potential for stigma and encouraged caregivers to be a source of unconditional support for their children: “Make sure that your kid is loved, appreciated, taken care of, you—there is no better person on this planet to do that than you.” (P43). Patients offered strategies such as seeking therapy and emotional support:

It is okay to talk about it and acknowledge that it's something that you're living with every day. I think I was always afraid other people have it worse or that I would be seen as a negative person or a complainer by bringing it up or talking about how much it affected me. (P21).

Patients also emphasized the importance of finding things that bring them joy and provide distraction or redirection: “Go out, exercise, and when you're able to, go out and have fun with your friends. It doesn't have to change your life.” (P2).

3.1.3. Seek social support

Participants recommended connecting with others for both emotional and instrumental support. Primarily, they described the

benefits of relying on the expertise and experiences of other families:

Talking with the parents [in these groups], you're gonna be able to put yourself in a position where you know exactly where to go and pinpoint, versus relying on a doctor that doesn't understand the diagnosis and is just going through medical books and research papers to figure out what the diagnosis is and how to handle it. (P44)

A patient shared similar advice and highlighted the benefits of connecting with a community:

I realized there were so many other people like me who had information to share. It felt like a huge weight off of my shoulders. (P26)

3.1.4. Promote self-management and self-care

Participants encouraged others to focus on self-management. Caregivers recommended involving children in care early. One caregiver explained:

I've had to learn to let her blossom a little bit and let her experience some things on her own. She had to pack her pillbox and take it with her. Having to teach her how to be responsible for her medication care [...] and there's a lot of times as she gets older she'll be like, “I know mom, I know what I gotta do.” (P24)

Patients highlighted the importance of quality of life, self-care, and prioritizing treatments for their symptoms. One patient advised, “Recognize that there are options available for pain relief and that it's important, take care of yourself, practice self-care. Recognize that you're a person who deserves relief and that there are options out there.” (P30) Other patients emphasized the value of “learning to live” with the condition rather than surrendering to pain or other symptoms: “I think that if somebody would've just told me like, ‘Hey, this is what you have. Don't let it stop your life. You have to keep doing the things you love.’ [...] I would've accepted my diagnosis better.” (P35).

3.2. Advice to clinicians

Advice to clinicians represented nine themes (Fig. 2) addressing multiple elements of patient-centered communication [22,23].

3.2.1. Show care and concern

Participants advised clinicians to demonstrate empathy, kindness, and patience, and to help families feel less alone. Caregivers urged clinicians to “always be caring and nice to the caregivers as well as their own patients.” (P42) A caregiver explained, “Understand where we’re coming from and the roles and the responsibilities that we have.” (P39). One patient explained the importance of clinicians showing genuine care and concern for patients with rare diseases:

Treat us like people, not like zebras, not like unicorns. [...] Don’t come in with a look of shock on your face when you see something. I don’t like feeling like a science experiment. (P7)

3.2.2. Demonstrate commitment

Participants advised clinicians to show they are willing to take the time to provide (or find) quality care. Demonstrating commitment included ensuring families can easily contact the medical team, following up regularly, advocating for the family, and not making families feel like a burden. One patient explained the positive impact of committed physicians:

It’s really important to not make your patient feel like a burden, but to—with every step of the way, show your patient that you are actively invested in their life and their well-being, [...] That has made a world of difference to me and having doctors that will repetitively and routinely reach out to me, even if I’m having a complication that doesn’t fall within their specialty. (P8)

Caregivers and patients specifically advised primary care physicians to take the time to follow up with specialists and do research on VMs.

3.2.3. Empower and validate

Participants advised clinicians to empower and validate families by trusting the family’s insights, engaging caregivers and patients in care, validating their accounts of pain or discomfort, and empowering them to make decisions and advocate for care. Caregivers recommended acknowledging caregivers’ expertise: “Emphasize that the parent and the caregiver are the quarterbacks, and the support system is there to work with them as a team.” (P28) Similarly, one patient summarized,

[S]aying that you understand their wants, even though you don’t understand their experience, is very reassuring. That helps people know that you’re really trying to do what’s in their best interests. (P26)

Often, participants felt dismissed if their symptoms were not considered severe. One patient instructed, “just because you don’t think it’s the worst case you’ve seen, it’s still really scary for us [...] don’t try to diminish my experience because it’s not the worst case.” (P15).

3.2.4. Address mental and emotional well-being

Participants advised clinicians to pay attention to mental and emotional needs and attend to non-physical care needs that affect quality of life. Caregivers advised clinicians to acknowledge their emotional distress, “I wish they knew what it really is. What it’s like to have people just stare at your child all the time or what it’s like to hear negative or sly comments from other parents or strangers.” (P42).

Similarly, one patient reflected, “I wish they knew the emotional toll that it takes. Obviously, doctors are very focused on the physical aspect of the condition.” (P26) This patient explained the benefit of doctors asking about their emotional well-being:

I have had doctors say, “Okay. How does this affect not just you physically, but how does it affect you emotionally?” I think that if doctors had asked me that sooner, they would’ve had a better idea of what I was going through. (P26)

Another patient reflected, “It meant so much for someone to even just say, “This is really hard. This sucks for you. I’m really sorry.”” (P21).

3.2.5. Acknowledge the broad impact of disease and treatments

Participants advised clinicians to acknowledge how VMs affect many aspects of the patient’s and caregiver’s life. Caregivers urged clinicians to recognize how the child’s condition affects factors such as family functioning and financial well-being: “I would like them to honor that the diagnosis or the lack of a diagnosis impacts more than the patient. The whole family is involved here.” (P31) A patient elaborated on a similar desire for clinicians to acknowledge the frustrations of trying to seek care for a rare condition:

I wish they would know how frustrating it is to not have answers and to go through a lot of treatments that doctors don’t really know if they’re going to help or not and then end up in the same place where you were before without any relief really. (P12)

3.2.6. Acknowledge your limitations

Participants advised clinicians to admit when they do not know something and to be willing to reach out to experts and refer when appropriate (i.e., “Be honest. If you don’t know, you don’t know.” P14). One caregiver summarized,

I wish they knew that it’s okay. It’s okay for you to say “I don’t have any experience with this. I need help trying to find out this diagnosis,” and I don’t think that some doctors are willing to do that. (P33)

Participants described physicians who were reluctant to ask for help or appeared offended when families asked for second opinions. One patient asserted, “Admit when you don’t know all the things you need to know. Find somebody else who does. Don’t take it personally.” (P25) Another patient reflected, “I would rather them be honest instead of pretending or just dismissing it.” (P23).

3.2.7. Commit to learning

Participants urged clinicians to learn about VMs, stay up-to-date with guidelines and recommendations, and monitor research updates (i.e., “I think it would be helpful if doctors would be more educated.” P37). One caregiver summarized: “Keep increasing that knowledge and do that research and continue to educate yourself the best way that they can to keep our kids safe.” (P39).

For non-specialists, caregivers and patients advised clinicians to gain at least a basic understanding of VMs. One patient suggested, “If you have a patient who comes to you with [VM] syndrome or just vascular malformations, please take a little time to read up on it and consider how that condition might factor into the overall treatments.” (P30).

3.2.8. Work as a team

Participants advised clinicians to ensure all members of the patient’s care team have a shared understanding of the patient’s diagnosis and care needs. Participants also encouraged clinicians to broaden the network of specialists across the country. One patient explained,

My advice is that they really try to broaden their network across other specialties and other practices, ‘cause it seems like there’s a few really great doctors in each of like five or six big cities, but I think it’s probably hard if you’re not already part of their network to even really know how to get into their network. (P2)

Another caregiver explained the importance of physician collaboration, explaining that vascular anomaly care is often lacking the same collaboration found in other specialties:

[W]ork together more, for the sake of the kids; you don’t need to reinvent the wheel. A lot of this stuff each person has so much incredible information and so much knowledge, that might be their own skillset, but, wow, when we’re together, how much stronger that is, and how much more beneficial it is for the patients. (P48)

Participants described the importance of referrals, but often

experienced challenges getting referrals from doctors inexperienced with VMs. One caregiver stated: “I wish they knew other people to refer to. I really think that’s the biggest thing that I want out of doctors is that if you’re gonna tell us to do something, have the next thing to do available.” (P45).

3.2.9. Communicate information clearly

Participants advised clinicians to ensure families understand information about their diagnosis and treatments. Participants recommended using plain language, providing accurate information, and having useful handouts. Patients often wanted accurate *and* honest information. One patient suggested, “don’t sugarcoat it too much, because in the long run, that really does end up hurting the kid more than anything.” (P35).

Participants’ advice reflected the strong desire for doctors to spend time explaining the patient’s condition:

Even in doing some research, I still don’t know how all this works. I don’t know which things affect other things, and I’m gonna have to ask a lot of questions in that way. The more information that you can provide to me in a digestible way—give me examples. Tell me what another person’s experience has been. (P29)

However, one caregiver described the potential for information overload:

It’s overwhelming. You’re not seeing one physician. You’re seeing 10 physicians or more. I’m just not getting information from you. I’m getting so much information. A lot of times, you’re not the only appointment of my day. I have three to four hours of appointments back-to-back. It’s a lot of information. It’s mentally draining. (P20)

Participants offered many suggestions to improve the exchange of information. One caregiver suggested, “slow down, speak as plainly as possible, and repeat yourself a few times.” (P36) Another caregiver explained, “Not everyone is a doctor, as well; you need to know how to describe things in laymen’s terms and not overwhelm the caregiver.” (P42).

4. Discussion and conclusion

4.1. Discussion

In this study, we examined advice related to managing care for complex vascular malformations (VMs). This study integrates the perspectives of patients and caregivers, which creates a comprehensive framework of advice. While our sample represents a group of patients and families managing complex vascular anomalies with high disease severity, these findings may also apply to other families with rare diseases facing similar challenges [3,4]. Overall, participants’ advice revealed many challenges related to health-related quality of life and unmet support and communication needs.

Participants’ advice suggested that living with, and seeking care for, VMs affects patients’ and caregivers’ health-related quality of life, or “an individual’s ability to function and his or her perceived well-being in physical, mental and social domains of life.” (p. 350) [21]. This finding is consistent with previous research suggesting that rare diseases affect patient and caregiver well-being and supports calls for clinicians to provide family-centered support resources and services [3,4]. Indeed, participants expressed a strong desire for clinicians to demonstrate multiple elements of patient-centered communication: fostering healing relationships, responding to emotions, exchanging information, and managing uncertainty [22,23]. Fulfilling these communication functions affects important outcomes such as access to care, diagnosis timing and accuracy, self-care and self-efficacy, and health-related quality of life [22].

Our results also suggest that patient-centered communication when caring for patients with rare diseases involves committing to learning about the condition. This need for committed learning was especially

true for patients who live far from specialists and rely on the care of local physicians who are inexperienced or unfamiliar with the condition. However, given the rarity of VMs, participants emphasized the importance of clinicians openly admitting when they are uncertain and providing referrals to expert clinicians. Helping patients navigate the health care system is an essential aspect of clinician advocacy and patient-centered communication [22]. Our findings confirm that this advocacy is particularly important for patients with rare diseases, who often experience poor care coordination and struggle to navigate a health care system that is highly fragmented [24].

Given the effect of a VM diagnosis on quality of life and patient-centered care, participants’ advice revealed the importance of self-advocacy and social support. Participants emphasized three core components of self-advocacy: increased illness education, increased assertiveness, and mindful nonadherence (or a willingness to disagree with physicians’ recommendations) [20]. Indeed, this finding confirms that patients and their caregivers often become experts on VMs and take on many of the responsibilities of care coordination. This expertise is also why participants strongly advised others to seek support from other families. Connecting with other families can help minimize feelings of loneliness or isolation, and can also provide valuable information regarding diagnosis, treatment, and credible clinicians. This finding is consistent with previous research suggesting that families affected by rare diseases rely on disease-specific social support groups for emotional and informational support [13].

The contributions of our study are not without limitation. Our sample was relatively homogenous in terms of gender, race, ethnicity, and income level. Similarly, our sample primarily represented complicated vascular anomalies given our recruitment from support groups for vascular overgrowth syndromes and the relatively high disease severity reported by participants. Future researchers should seek to recruit diverse patient and caregiver samples to represent the breadth of experiences of all VM families. Our results also represent the perspectives of patients and caregivers who participate in support groups. Therefore, our findings may not reflect the experiences of individuals who are still trying to find a diagnosis, are struggling to get the support they need, or do not find benefit in VM support groups. However, given the challenges of recruiting participants in rare disease communities, these support groups offer a valuable resource for researching the experiences of patients with rare diseases. Additionally, this recruitment strategy allowed us to engage participants who have been unable to access multidisciplinary, expert care.

4.2. Conclusion

This study highlights advice for patients and caregivers seeking care for complex VMs and clinicians providing care for patients with VMs. Participants’ advice revealed challenges related to family-centered communication and patient and caregiver quality of life and revealed the importance of self-advocacy and social support. This advice can help families overcome these challenges and help clinicians improve care and communication experiences for families navigating care for VM. The advice given from this study could also be applied to other rare diseases and families navigating medically unexplained physical symptoms. Finally, the study highlights the benefits of seeking advice from individuals navigating the healthcare system to identify ways to improve partnerships between clinicians and patients/caregivers.

4.3. Practice implications

Our study offers implications for clinicians and advocacy groups supporting families affected by complex VMs. Families would benefit from an early intervention to minimize the negative impacts of a VM diagnosis on quality of life. The results also suggest that clinicians caring for patients with rare diseases should foster trusting relationships and acknowledge the toll navigating fragmented care can have on patient

Advice for Patients and Caregivers



Advocate for Care & Information

- Be assertive
- Stay informed
- Seek opinions from multiple doctors



Address Emotional & Mental Health

- Show unconditional love
- Talk about your emotions
- Do things that spark joy



Seek Social Support

- Connect with vascular anomaly families
- Find support groups online
- Seek advice from expert parents/patients



Promote Self-Management

- Empower children to participate in care
- Practice wellness and self-care
- Manage pain and symptoms

A vascular anomaly diagnosis can affect more than just physical health. This advice can help patients and caregivers connect with other families and improve patient and caregiver physical, mental, and social well-being!

and caregiver well-being. Clinicians should also admit when they are uncertain about how to care for VMs and help families find expert care. These patient-centered behaviors can help validate and empower patients; they can also help minimize the burden of advocacy. Lastly, clinicians might use these results to encourage families that they are not alone and provide resources for addressing emotional, social, and physical well-being. We have created an example of a potential handout that might validate families' experiences and relay this advice (Fig. 3).

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CRediT authorship contribution statement

Anna Kerr: Conceptualization, Methodology, Investigation, Formal analysis, Writing – original draft. **Christine Bereitschaft:** Formal analysis, Data curation, Writing – review & editing. **Kayla Duty:** Writing – review & editing. **Bryan Sisk:** Conceptualization, Methodology, Investigation, Data curation, Formal analysis, Supervision, Funding acquisition.

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Disclosure

The authors have no competing interests to disclose.

Appendix A. Supporting information

Supplementary data associated with this article can be found in the online version at [doi:10.1016/j.pec.2022.11.011](https://doi.org/10.1016/j.pec.2022.11.011).

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