Original Article

The role of primary care in rare disorders: A qualitative study of parents and patients managing complex vascular anomalies

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Abstract

Background: Patients with complex rare disorders often require the care of multiple specialists. Effective coordination between primary and specialty care is needed to ensure patients receive high-quality care. Previous research has documented the importance of primary care clinicians providing referrals to specialty care and the importance of specialists in helping patients reach a diagnosis. However, little is known about primary care clinicians' roles in the ongoing care of patients with rare disorders. In the current study, we explored the role of primary care clinicians in the care of rare and complex vascular anomalies. **Materials and Methods:** Data were collected using semi-structured qualitative interviews with 34 parents and 25 adult patients recruited from advocacy groups for patients with complex vascular anomalies participated. We asked participants about their diagnosis, care experiences, and communication with clinicians. We used thematic analysis to identify themes illustrating the roles of primary care clinicians. **Results:** PCC roles were characterized by four behaviors. *Supporting* behaviors included learning more about vascular anomalies and asking participants about the care they received from specialists. *Facilitating* included providing referrals, ordering tests, and engaging in problem-solving. *Interfering* included failing to provide referrals or help participants coordinate care, ordering incorrect tests, or making inappropriate recommendations. *Disregarding* included focusing narrowly on primary care needs and not showing concern about the vascular anomaly. **Conclusions:** The results reveal opportunities to improve primary care for patients with vascular anomalies. Disregarding and interfering behaviors furthered the division between primary and specialty care for patients with vascular anomalies and prevented patients from receiving comprehensive primary care. Supporting and facilitating behaviors convey genuine interest in the care of the vascular anomaly and a commitment to helping the patient and parent.

Keywords: Care coordination, primary care, rare disorders, vascular anomalies

Introduction

Approximately 30 million Americans are affected by rare disorders.[1] With the average time to an accurate diagnosis being seven to eight years, patients with rare disorders often face a long, difficult diagnosis journey.[2] Patients often rely on clinicians with specialized disease-specific knowledge to help them reach the end of their "diagnostic odyssey."[3] Even after a diagnosis, many patients struggle to coordinate complex multi-system care from multiple sub-specialists.[3] The focus on specialty care in rare disease research means that primary care clinicians' roles in managing rare disorders are often overlooked,[4,5] despite the fact that primary care serves as most patients' entry point into the healthcare system.[6] In the current study, we examine the role

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of primary care and family medicine physicians' in the care of patients with rare disorders.

In the context of rare disorders, high-quality primary care can help prevent delayed diagnoses.[5] Care coordination is an essential role of primary care clinicians.[6,7] For individuals with chronic illnesses, effective coordination between primary and specialty care is associated with higher quality and more efficient care.[8] However, primary care clinicians seldom manage patients with rare disorders (accounting for less than 2% of all visits between 2010 and 2014).[9] Continuous primary care can reduce the risk of emergency room visits, hospitalizations, and readmissions,[10] so identifying ways to optimize the role of primary care in the care of patients with rare disorders is essential.

Rationale

VAs are a broad spectrum of vascular tumors and malformations that range from simple "birthmarks" to rare and life-threatening conditions.[11] Complex VAs can be invasive and cause pain, ulceration, and organ and musculoskeletal dysfunction.[12] VA patients and their caregivers struggle with stigmatization and uncertainty.[4-7] The treatment of complex VAs requires the care of multiple specialties and is frequently unfamiliar to primary care physicians.[13] Consequently, many patients with VAs struggle to receive a diagnosis[14] or receive incorrect diagnoses leading to unnecessary or harmful treatments.[15] Like patients with other rare disorders,[16] VA patients and their caregivers commonly encounter clinicians who do not have the knowledge needed to diagnose or treat their condition or are dismissive or unwilling to investigate symptoms further.[17] Even when patients do access expert care, they struggle to with local clinicians who are unable to manage routine care between visits to specialized VA centers that are clustered in urban areas.[17] Furthermore, specialized care for VAs is typically located at pediatric institutions; the lack of VA care in adult medicine means adults with VAs face challenges transitioning to adult medicine and maintaining comprehensive care.[18]

Research has documented the importance of primary care clinicians (PCCs) providing timely referrals to specialty care for patients with VAs and the importance of specialists in helping patients make decisions and manage uncertainty.[19-21] However, little is known about the role PCCs play in ongoing care. Thus, we sought to answer the research question: How do patients and caregivers describe the role of PCCs in the care for VAs?

Materials and Methods

We conducted semi-structured interviews with adult patients and parents of children with complex VAs. The consolidated criteria for reporting qualitative research (COREQ) checklist was used during study development and reporting.[22] The Washington University institutional review board approved the study. The procedures were in accordance with the ethical standards of the IRB and with the Helsinki Declaration of 1975, as revised in 2000.

Recruitment and participants

We recruited participants through four patient support groups: Klippel–Trenaunay Support Group, CLOVES Syndrome Community, Project FAVA, and Lymphangiomatosis and Gorham's Disease Alliance (LGDA). Caregivers were eligible if they were the caregiver of a child (<18 years) with a VA. Adult patients were eligible to participate if they were diagnosed with VA and were between the ages of 18 and 39, based on the National Cancer Institute designation of young adults.[23] Inclusion criteria also included speaking English and being a resident of the United States. Participants received a $40 Amazon gift card.

Data collection

After providing verbal consent, participants completed an online survey to self-report age, race, ethnicity, income, education level, relationship status, insurance status, and VA diagnosis. We conducted semi-structured interviews via telephone or videoconferencing software. The interview guide asked participants about the diagnosis process, accessing care, and communicating with clinicians. Interviews were audio-recorded and professionally transcribed.

Data analysis

We used thematic analysis to characterize the primary care experiences of participants. Two authors (AK and CB) descriptively coded five transcripts to generate preliminary codes. During the first coding meeting, we organized the codes into categories (i.e., roles of primary care clinicians) refined through iterative cycles of independent coding and consensus meetings. We independently applied the final codebook to all 58 transcripts, reviewed each other's coding and resolved disagreements through discussion. The categories represent 100% agreement. Coding was completed using Dedoose qualitative analysis software.

Results

We interviewed 34 caregivers and 25 young adult patients. Participants were predominantly White, female, and had college or professional degrees [see Tables 1 and 2]. All caregivers identified as parents; their ages ranged from 21 to 54 years (Median = 42 years) and they cared for children with VAs ranging from infancy to 16 years (Median = 10 years). Adult patient participants' ages ranged from 18 to 39 years (Median = 29.5 years). Interview lengths ranged from 25 to 80 minutes.

The roles of primary care clinicians

We identified four categories of behaviors of primary care clinicians described by parents and adult patients with VAs: supporting, facilitating, interfering, and disregarding. See Table 3 for a summary of these clinician behaviors.

Supporting. Supporting behaviors were described by 5/25 patients and 13/34 parents. Supporting behaviors included demonstrating a commitment to learning more about VAs.

Table 1: Full sample demographics

Characteristic	Parents (*n*=34)		Adult Patients (*n*=25)*	
	n	%	*n*	%
Age in Years	Mean=41.3	Range=2-54	Mean=29.2	Range=18-39
Gender				
Woman	28	82%	21	84%
Man	6	18%	2	8%
Non-binary/third gender	--	--	1	4%
Race/Ethnicity†				
Asian	1	3%	1	4%
Black or African American	1	3%	2	4%
Hispanic, Latin, or Spanish	6	18%	2	8%
White	30	88%	22	88%
Education				
High School or Equivalent	--	--	3	12%
Some College	11	32%	3	12%
College Degree	10	29%	13	52%
Graduate/Professional Degree	13	38%	5	20%
Household Income‡				
$24,999 or less	1	3%	2	8%
$25,000 - $49,999	4	12%	2	8%
$50,000 - $74,999	3	9%	2	8%
$75,000 - $99,999	7	21%	2	8%
$100,000 or greater	15	44%	13	52%
Relationship Status				
Married or Living as Married	28	82%	10	40%
Never Married	3	9%	13	52%
Divorced/Separated	3	9%	1	4%
Child's Age in years	Mean=9.4	Range ≤1-16	N/A	
Child's Gender				
Boy	14	41%	N/A	
Girl	20	59%	N/A	

*One participant did not complete the demographic survey. †Not mutually exclusive. ‡Four parents and three patients did not report income

Table 2: Diagnosis characteristics

Diagnosis	Child Diagnosis *n*=34 (Parent-reported)		Patient Diagnosis *n*=25 (Self-reported)	
	n	%	*n*	%
Type of Vascular Anomaly[a]				
Lymphatic Malformation	18	53%	14	58%
Venous Malformation	12	35%	16	67%
Capillary Malformation	8	24%	5	21%
Arteriovenous Malformation[c]	5	15%	1	4%
Hemangioma[c]	3	9%	2	8%
Unsure of Definitive Diagnosis	3	9%	1	4%
Associated Syndromes or Disorders[b]				
PIK3CA-Related Overgrowth Spectrum	23	68%	18	72%
CLOVES Syndrome	8	24%	4	17%
Fibro-Adipose Vascular Anomaly	8	24%	5	21%
Klippel–Trenaunay Syndrome	2	6%	4	17%
Macrocephaly–Capillary Malformation	1	3%	--	--
PTEN Hamartoma Syndrome	1	3%	--	--
Kaposiform Lymphangiomatosis	3	9%	--	--
Gorham Stout Disease	2	6%	--	--
Generalized Lymphangiomatosis	4	12%	5	21%
Central Conducting Lymphatic Anomaly	1	3%	2	8%

[a]Not mutually exclusive. [b]Many of these disorders are often grouped in the larger diagnostic category of "PIK3CA-Related Overgrowth Spectrum (PROS)." However, many patients self-reported their disorders by these historic terms, rather than as PROS. These percentages reflect patients self-reporting. [c]Participants self-reported arteriovenous malformations and hemangiomas, however, these diagnoses are often incorrectly applied, and the associated disorders seldom include these lesions

Table 3: Primary care clinician role and frequencies across interviews

Behavior	Description	Frequency*
Supporting	Being committed to learning more about VAs through research or asking participants about the care they (or their child) received from specialists and how the VA affected overall health. Showing genuine interest in the patient and parent and being compassionate regarding care for VAs.	18/59 (30.5%)
Facilitating	Helping patients by providing referrals and ordering tests and engaging in problem-solving. Providing primary care that includes care of the VA symptoms or taking on the role of advocate or quarterback of the care team.	33/59 (55.9%)
Interfering	Failing to provide referrals for second opinions or help participants coordinate care. Providing incorrect diagnoses, ordering incorrect tests, or making inappropriate treatment recommendations. Failing to validate or follow-through on patient or parent requests.	13/59 (22.0%)
Disregarding	Focusing narrowly on routine primary care needs. Not asking about or showing concern about the VA or associated symptoms. Overlooking symptoms of the rare condition or dismissing them as "just a virus." Deferring all responsibility for care to specialists.	22/59 (37.3%)

*Behavior categories are not mutually exclusive. Participants often described multiple primary care clinicians throughout one interview

Clinicians conveyed support by doing research or asking about the care the patient received from other clinicians. One patient recalled, "The nurse practitioner, three primary care physicians before, she was amazing. She asked questions. You could tell that she had done some research, that she was trying." [Patient 7] Supporting PCCs were interested in understanding how VAs affect patients' overall health, showed genuine interest, and were compassionate regarding care for VAs. A parent described how her child's primary care doctor adjusted his routine care schedule to accommodate the added care needs: "His pediatrician likes to see him almost every two months instead of the normal wait period, just to keep an extra eye on him." [Parent 42]

Some participants acknowledged that VA care was beyond the PCC's level of expertise, yet appreciated the PCC's support and willingness to learn. However, some participants were frustrated with constantly having to educate PCCs:

"Every time I've gone [to a PCC], they're just more fascinated and learning about my disease instead of having knowledge about helping it, which is uncomfortable, as the patient, because everything is still coming from me. [..] They'll tell me, "I've never heard of this disease before. What's the name of it? How do you spell it? I wanna look it up and learn more about it." [..] At this point, I'm used to it, so I'll go along with it, but it doesn't necessarily make me the most comfortable with getting treatment, obviously". [Patient 6]

Facilitating. Facilitating behaviors, described by 14/25 patients and 19/34 parents, included providing referrals, ordering tests, and engaging in problem-solving with participants. For most patients, a referral from a PCC was the first step toward an accurate diagnosis. Referrals from PCCs allowed patients to access additional specialists or seek second opinions: "We've got a good pediatric doctor who has given us a lot of referrals." [Parent 31] For adult patients, finding a PCC who will facilitate referrals to a pediatric specialist is important. One patient explained,

"I've actually never been to a pediatrician before, so I was not in any system that would even warrant me being seen at the children's hospital, so I had to wait until that could be opened up for someone of my age for someone to even start treating and seeing me at the children's hospital" [Patient 2].

Participants described *facilitating* as providing more instrumental than emotional support: "I really just go to [my PCC] so I can get referrals." [Patient 17]. This was especially true for participants who did not have a longstanding relationship with their PCC. Some participants did not expect their PCC to do anything more than coordinate care:

"I wouldn't necessarily expect just a regular family pediatrician to get super involved with that or to know more about it, other than doing research maybe suggesting or recommending places to go to for treatment or to get second opinions or things like that. I would not expect them to all of a sudden take an interest and dive deep into the condition itself or counsels or treatments or anything like that necessarily". [Parent 104]

For others, however, facilitating also involved advocacy. One patient explained, "I have really good conversations with my family doctor, just talking about the need to do this. He validated, like, 'Hey, let's do something about it, I'm putting the referral in right now.' Listening to my concerns, caring about how that affects me personally." [Patient 114]

Interfering. Interfering behaviors, described by 5/25 patients and 8/34 parents, included behaviors (or a lack thereof) that obstructed their ability to receive a timely diagnosis and access or maintain care. Often, interfering included being reluctant to provide referrals or coordinate care by communicating with specialists. A patient recalled,

"None of them could do anything. I didn't know there was the possibility of trying a different specialty when we knew what it—even when we knew what it was, and they knew that there was a vascular component, none of them had the gumption to stand up and say, 'Oh, maybe somebody who specializes in vascular things would be better than me.' Nobody took that chance to help me when they could have just admitted they don't know anything. I understand it's hard to admit when you're wrong, or you don't know something, but you should want what's

best for the patient. Ethically, that's your responsibility as the doctor." [Patient 100]

Many participants described instances when PCCs provided incorrect diagnoses or missed symptoms indicating a vascular component: "I feel like with my PCP and this—and her just wanting to label what's going on with my left arm as a cyst, that just drives me crazy 'cause it's not a skin thing. It's not a cyst. It's just not. I've had cysts before. This is not a cyst." [Patient 17]

For other patients, interfering included ordering incorrect laboratory tests or making inappropriate treatment recommendations:

"[Our PCC] would try to give us things that we could do at home to help open up her airway, like give her steam showers, run humidifiers, put drops of eucalyptus into your—things like that, that she was just like, whatever you can do to help open up her airway, but there was no opening her airway, there was a gene mutation that was occurring that just kept closing it". [Parent 113]

Participants often described interfering clinicians who did not validate or follow through on patient or parent requests for assistance or information.

Disregarding. Disregarding behaviors, described by 9/25 patients and 13/34 parents, included being narrowly focused on routine primary care needs or showing a lack of compassion. Many participants recalled clinicians who did not show concern about the VA or associated symptoms; in some cases, the clinician ignored the VA altogether—failing to bring it up during routine visits even when it was included on the patient's chart: "I can't communicate with our pediatrician on this issue because it doesn't go anywhere. They lack knowledge on it. Like I said, I don't feel like—she's getting all this information, but I don't feel like she's reading it." [Parent 20]. Disregarding also included overlooking symptoms or dismissing them as "just a virus." Multiple participants reported bringing up the symptoms with multiple PCCs and being frustrated that "nobody seemed concerned."

Disregarding often included deferring all responsibility for care (ordering tests, calling in prescriptions, making decisions, etc.) to the specialist:

"I wish that our pediatrician would've said, "I'm with you. I don't notice a change. Let's see if we can find something else out." Instead, she just always fell back on, "Well, what does the dermatologist say? What does the dermatologist say?" I don't really—I don't blame her for that. Because she is aware that she does not have the training to distinguish the difference between a hemangioma and a venous malformation". [Parent 112]

Participants found disregarding especially frustrating with urgent care needs: "Since it was during the holiday, I went to my PCP. My PCP is like, 'This is not normal. Contact your vascular anomaly doctor. My vascular anomaly doctor was out of town. I had to wait till after Thanksgiving to get an answer [laughter].'" [Patient 7]

Disregarding caused many participants to intentionally maintain separation between primary and specialty care: "To be honest, I don't know that [my PCC] should have a role. Because she knows nothing about it. It's sad. She would be able to give me zero advice. I only trust the vascular anomalies clinic." [Parent 112] Some participants seemed to accept the limited role of PCCs: "It would've been nice to have—if they had had some more knowledge and involvement, but I just don't think they knew enough about it to know how to help." [Patient 12]

Discussion

This study investigated the role of primary care clinicians (PCCs) in caring for vascular anomalies (VAs). Parents and adult patients with VAs described four central behaviors enacted by PCCs: supporting, facilitating, interfering, and disregarding.

Our results suggest that many patients with VAs would benefit from PCCs who are actively engaged in their care. Participants in our study frequently encountered PCCs who behaved in a way that was interfering (failing to provide referrals or coordinate care, ordering incorrect tests, or suggesting inappropriate treatments) or disregarding (dismissing or overlooking care related to VAs or VA symptoms). These behaviors caused frustration and increased burden. They often prompted participants to avoid these clinicians altogether. These results suggest that PCCs who do not assist with care coordination, provide whole-person care that includes consideration of the VA, or become educated about the rare condition are furthering the divide between primary and specialty care for patients with VAs. Therefore, PCCs should incorporate supporting and facilitating behaviors described in our study.

Participants' appreciation for facilitating behaviors, including providing referrals, ordering tests, and acting as the quarterback of the care team, illustrates how much patients with VAs rely on effective care coordination to access and maintain care. Indeed, previous research suggests that patients benefit from having a PCC who is highly involved in coordinating care and transferring information to other clinicians.[24] However, our results suggest that relying solely on facilitating behaviors may provide patients and parents with informational and tangible support (i.e., sharing information, providing resources, and assisting with tasks)[25] but not important emotional support. Supporting behaviors are also needed to provide the psychosocial support patients and parents need when navigating care for a rare disorder.

Our results show the benefits of having a supportive PCC who is genuinely interested in learning more about the VA to provide comprehensive whole-person primary care.[26] PCCs should strive to become knowledgeable about the rare disorders they encounter, which involves relying on the expertise of the patients

and parents who become experts on the condition. However, the results of our study confirm previous research that constantly having to educate clinicians can be a burden.[2] This is particularly true for adults with VAs who routinely encounter clinicians in adult medicine who do not have the knowledge or expertise needed to care for VAs.[18] Therefore, PCCs should also make an effort to do their own research to demonstrate their commitment to the patient and family.

Overall, our results suggest that PCCs should focus on supporting and facilitating behaviors and avoid interfering and disregarding behaviors when caring for patients with rare disorders. These results reflect the recommendations for PCCs that were proposed in a proposed model of primary care in rare disorders: 1.) recognize deviations of common patterns of disorders that may indicate a low-prevalence disorder or need for referral, 2.) provide comprehensive care that includes both disease-specific and routine primary care conditions, 3.) become knowledgeable about the rare disorders you encounter, 4.) empower patients and families and support their emotional and social needs, and 5.) be an advocate for the patient and help them navigate social services, healthcare systems, and complex medical information.[5] Our findings extend this framework to define PCC roles that may either improve or complicate care for patients with VAs. Of course, we acknowledge that additional time spent coordinating care may increase the burden and stress for already-overworked primary care clinicians and staff. Thus, future work should examine system-level barriers that may be obstacles for PCCs.[27]

Our study is not without limitations. Participants were recruited from advocacy groups, so our sample does not include the potentially unique perspectives of patients recruited from specialized medical centers. Our recruitment may have also affected the diversity of our sample. Our participants were predominately female, White, and well-educated, which is consistent with individuals who are most likely to participate in online support groups, yet does not reflect the experiences of families of all socioeconomic statuses, genders, or racial/ethnic identities.

Conclusion

Our interviews with patients and parents revealed the four most common roles of primary care clinicians in the care for patients with vascular anomalies. Disregarding and interfering behaviors further reinforced the separation of primary and specialty care by focusing narrowly on routine primary care, dismissing patients' and parents' concerns about the VA, and failing to coordinate care. Conversely, supporting and facilitating behaviors conveyed genuine interest in the care of the VA and commitment to the patient and family by coordinating care, becoming educated about the condition, and providing comprehensive care that included the VA. Future research should examine clinicians' perspectives to build a comprehensive framework of the role of primary care in the care of VAs.

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Ethical approval

All research activities were reviewed and approved by the Washington University Institutional Review Board.

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Conflicts of interest

There are no conflicts of interest.

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