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"I would love to talk to someone that actually understands": Psychosocial experiences of adults with Fanconi anemia

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Abstract

Fanconi Anemia (FA) is a rare disease characterized by bone marrow failure and increased cancer susceptibility. Although the adult FA population is growing due to medical advancements, there has been little research on their psychosocial experiences. Participants (*n* = 18) completed semi-structured interviews about mental health. Additionally, participants had the option of participating in Photovoice, taking photos that represented their experiences. Conventional qualitative content analysis identified five themes, including "FA is all-encompassing," Stigma and trauma take a toll on mental health, Connection is bittersweet, Mental healthcare barriers, and "A process of accepting the disease." Findings indicate a need to address mental health barriers including a lack of mental health providers knowledgeable about rare disorders and FA. Social support and connections with the FA community –key protective factors– should be made more accessible to minorities and individuals outside of Westernized countries.

Keywords

cancer predisposition, fanconi anemia, participatory research, rare disease, rare disorder

Fanconi Anemia (FA) is a rare, autosomal recessive disorder of the DNA repair pathway (Peake and Noguchi, 2022). Disordered DNA affects every cell in the body and leads most individuals with FA to have bone marrow failure and a high susceptibility to cancers (Peake and Noguchi, 2022). FA can also cause issues such as skeletal anomalies, kidney problems, gastrointestinal difficulties, and infertility (Peake and Noguchi, 2022). Although previously thought to be a pediatric disease, medical advancements in allogeneic hematopoietic stem cell transplants to address bone marrow failure have enabled many individuals with FA to live into adulthood for the first time (Peake and Noguchi, 2022).

In the United States, a rare disease (RD) is defined as a condition that affects less than 200,000 people, with an estimated 30 million Americans living with a RD (National Institutes

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of Health, 2020). RDs present unique challenges beyond the experience of common chronic diseases, including difficulty accessing information, treatment, psychosocial support, and coping with stigma and uncertainty (Bryson et al., 2021). Research has found that individuals with RDs experience lower health-related quality of life compared to US norms and compared to people with common chronic conditions (Bogart et al., 2022).

Psychosocial research on FA and related conditions

Research on the psychosocial experiences of people with FA, especially adults, is limited. In one study, Kearney et al. (2012) conducted a chart review of children and adults with FA receiving transplants referred to psychiatry consultation (Kearney et al., 2012). Of these, 50% were diagnosed with mood disorders, 46% were diagnosed with adjustment disorders, and 23% with anxiety disorders. These findings indicate that children and adults with FA are at high risk for psychological distress during this difficult transplantation period. However, approximately two-thirds of the sample were children, so little is known about mental health for the larger FA adult population during other developmental and treatment stages.

Notably, the first research focused on mental health of adults with FA was conducted by Amy Frohnmayer, who herself had FA. Her research focused on emerging adults was described in an unpublished master's thesis (Frohnmayer, 2010) and a published conference abstract (Frohnmayer, 2016). She identified psychosocial challenges across four domains: physical, medical, cognitive perceptual, and social. Frohnmayer found that individuals with FA use a combination of problem-focused and emotion-focused coping strategies. Active coping and acceptance were the most commonly used coping strategies. Problem-focused coping strategies were found to be positively associated with well-being, and avoidant, emotion-focused strategies like behavioral disengagement self-distraction, and denial, were found to be negatively associated with well-being. These findings echo broader work on chronic illness, finding that active, problem-focused coping can be more effective in promoting adjustment than passive or avoidant coping strategies (Stanton and Hoch, 2025). Frohnmayer also found that embracing FA as an identity and community comes with both positives and negatives, specifically that embracing FA can foster meaning and connection, but it can also distance individuals from a normal lifestyle and emphasize uncertainty about the future. This project continues Frohnmayer's work through further qualitative research on psychosocial experiences of adults with FA.

Recently, Bogart et al. (2025) conducted a large-scale quantitative study of mental health in 102 adults with FA. When compared to population norms, adults with FA had significantly poorer health-related quality of life. Results showed 50% of participants had probable PTSD, 33% had probable anxiety, and 25% had probable depression. Hierarchical regressions indicated protective and risk factors, respectively, for anxiety, depression, and PTSD included disability self-efficacy, post-traumatic growth, stigma, fatigue, and concern with death and dying. These findings indicate alarming levels of mental health issues in adults with FA and a need to understand their lived experiences more deeply.

Qualitative studies of people with other inherited cancer disorders suggest a complex relationship with one's body—people may become hypervigilant or emotionally detach from their bodies (Werner-Lin et al., 2022). Health crises, however, can abruptly recenter their physical being in their lives (Werner-Lin et al., 2022). The stress of constant bodily vigilance and frequent screenings is compounded by the grief experienced within FAmily and community circles (Forbes Shepherd et al., 2021; Wilsnack et al., 2021). The physical alterations resulting from cancer or its treatments can be distressing and stigmatizing, leading to shifts in one's body image and self-perception (Forbes Shepherd et al., 2021; Werner-Lin et al., 2022).

Minority status is known to result in mental and physical health disparities (Lick et al., 2013), and it could intensify the challenges, stigma, and healthcare access issues that those with inherited cancer predisposition disorders like FA face. Although many inherited cancer predisposition conditions—including FA—occur roughly equally across racial and ethnic groups, previous studies of inherited cancer predisposition conditions have been majority White and middle to high socioeconomic status (Werner-Lin et al., 2022). An important unanswered question is the role of minority status on mental health in heritable cancer predisposition disorders.

Building on the limited previous literature on adults with FA and related disorders, the current project aims to qualitatively explore the psychosocial experiences of adults with FA and intersections with race, ethnicity, nationality, sexuality, and gender. In addition, we examine barriers and facilitators to mental health.

Method

Participatory engagement

This project employed a participatory research approach (Balcazar et al., 1998; Bogart and Uyeda, 2009). An advisory board consisting of five adults with FA convened approximately quarterly to co-create the study aims and design and pilot interview questions. Additionally, the board facilitated recruitment via community networks and assisted in interpreting and disseminating findings. Efforts were made to ensure diversity in board membership, encompassing various dimensions such as age, disease stage, race/ethnicity, gender, and sexuality.

Participants

Participants were recruited from a pool of individuals from our previous quantitative survey who agreed to be contacted for future research (Bogart et al., 2025). Purposive sampling was used to maximize diversity of the sample. Specifically, participants who had reported being a sexual, gender, or racial minority were invited to participate first. As a strategy to ensure data quality and weed out fraudulent responses—a growing problem in qualitative research, especially when payment is offered (Lawlor et al., 2021; Schneider et al., 2024)—at the start of the interview we assessed the alignment of participants' reported demographic information and history of FA with data collected from the survey. If answers to more than two of these questions were inconsistent, participants were excluded (n = 8).

Included participants (n = 18, M age = 29) were 50% White, 28% Black, 11% Hispanic or Latinx, and 1% Indigenous. The majority of participants, 61%, were female, 33% were male, and 1% were nonbinary. Regarding sexuality, 61% of participants were heterosexual, 22% were bisexual, 6% were gay, 6% were asexual, and 6% were lesbian. In terms of nationality, 50% were from the United States, 11% from Canada, and 5.5% each from Colombia, Kenya, Nigeria, Norway, Spain, and Switzerland.

Procedure

This study was approved by the Oregon State University Institutional Review Board and participants provided informed consent, including consent to publish Photovoice pictures. To elicit deep and varied data from this understudied population, we borrowed from several qualitative interviewing techniques. Semi-structured interview questions focused on psychosocial experiences and mental health treatment-seeking. Additionally, modified from the Life Story Interview (Atkinson, 1998), we asked participants about a high point in their life with FA, a low point in their life with FA, and a turning point in their life with FA. Similar modifications have been used in recent disability research to prompt rich responses (Adler et al., 2022). Additionally, participants had the option of participating in Photovoice, a participatory method in which participants represent their experiences through photography (Hergenrather and Rhodes, 2007; Wang and Burris, 1997). Participants were invited to take pictures the week before their scheduled interview that represented answers to the following questions: "What does FA mean to you?," "What emotions and experiences stand out to you?," "What does FA mean for your mental health?," and "What support do you need most?." During the interview, participants were asked to describe and interpret their photos. The 30–60 minute interviews were conducted by one of two interviewers—the second and third authors–via Zoomin English ($n = 17$) or Spanish (n – 1). Interviews were offered in Spanish as well as English because consultations with the advisory board and the Fanconi Cancer Foundation, the largest FA organization, indicated that there was a relatively large and underserved Spanish-speaking population. Participants were compensated for their time with a $100 digital gift card.

Data analysis

All participants were given pseudonyms. The interviews were transcribed first by Zoom's auto-transcription software and manually checked by the interviewers. A bilingual research assistant translated the Spanish transcript to English, which was then checked by another bilingual research assistant. We employed conventional qualitative content analysis (Hsieh and Shannon, 2005), a method appropriate for health research in poorly understood topics. The entire interview was analyzed holistically, including answers to the semi-structured interviews, discussions of Photovoice, and life story interview questions.[1]

Using an inductive approach, each coder (the second and third authors) and the first author independently read each transcript and noted initial ideas. These study team members then met to discuss initial ideas, combining these into an initial list of codes. Next, in an iterative process, the coders independently coded several transcripts at a time, meeting with the first author to discuss revisions to the coding scheme and repeating until all transcripts had been coded twice. When coding disagreements arose, the coders and first author discussed until agreement was reached. The study team members then discussed how the codes clustered together into themes.

The themes and codes, along with exemplar quotes, were communicated to the Advisory Board and all participants using a structured member checking approach (Birt et al., 2016) and feedback was incorporated into results.

Positionality

One interviewer/coder identifies as a White, heterosexual, able-bodied, cisgender woman. The other interviewer/coder identifies as an Asian, mixed race, queer, chronically ill, disabled, cisgender woman. The first author identifies as a White, heterosexual, and cisgender woman with a RD. The last author identifies as a White, heterosexual, able-bodied, cisgender woman.

Results

Overall, 30 codes were identified and five themes were generated, which are discussed below in turn.

Theme 1: "FA is all encompassing"

"FA is a ticking time bomb." This code describes the unpredictability of having FA. Interviewees discussed feelings of uncertainty surrounding FA in their lives, such as not knowing whether or when they would need a transplant, anxiety around cancer surveillance, and/or the possibility of having cancer. One said, "it's kind of hard to accept it and know what's gonna happen, 'cause you have no idea." Another participant

discussed the inevitability of illness. "I'm feeling that someday or sometime I'm going to get very sick. Because that is what happens with this disease."

FA affects goals. FA altered many participants' life goals, including those related to careers and relationships. A participant shared frustrations when thinking about the future, "Like sometimes I don't know what plans to make 'cause it's like what's the point?'" One participant discussed being unable to return to work after the COVID-19 pandemic because her doctors deemed it unsafe. "I ended up using all my medical or all my vacation time, all of my sick time. And basically I had to quit my job. And now I'm on disability." A couple participants discussed having to change career prospects due to FA. "I wanted to be a doctor. A hematologist. My doctor told me, 'Why are you studying that? You, you can't study medicine because you're going to be exposed to many viruses, bacteria, and you can't be in a hospital because of my white [blood] cells.' And so I was very sad. . . I used to cry about it a lot." Another recounted, "I had big dreams. I wanted to serve in the military as a pilot, but due to my condition, I ended up changing my career. . . I could not serve."

FAmily planning/fertility. Experiences around FAmily planning ranged from concerns about infertility due to FA or treatments to fears of dying while raising children. One participant said, "I was told I didn't have enough eggs to freeze, that it would not be worth the money. . . I had like my dream of being a mom kind of taken away from me." One participant described her ambivalence about her fertility. "When I was young, I was like, 'That sucks, I won't have kids.' But. . .I'm not gonna be having kids in the next couple of years, anyway. . .I guess as I get older it's just kind of one of those things that looms over you, you know."

Male participants also experienced challenges with FAmily planning. One participant said he feels like he is not raising his children well because of his FA and subsequently, his partner does more of the labor. "Yeah, I feel like I'm not doing so [child-rearing], because my partner is the one doing most of the things for us." One participant mentioned that he felt his FAmily was delayed, saying, "I started my FAmily a bit late because of [FA]." Another participant described discussing adoption with his partner because it wouldn't be possible for them to have a biological child. "I had to go through a lot of counseling to accept the fact. Because it is the wish of any human being to have their own biological offspring. So this is an issue that affected me and I needed a lot of support and counseling."

"Professional patient." Due to the little research on FA and few healthcare providers with expertise, combined with the need for complicated, sometimes self-administered treatments, participants felt they had to become experts on FA. They described feeling responsible for researching FA online. One participant described researching FA online every day. "I Googled it [FA], and I was like, 'Oh, my God!' You know I kind of didn't understand much. . . I was like, 'What does this mean, the only thing I know is doctor and blood.' And then like I'd read the same Internet pages every day for years." Another participant started researching FA to advocate for herself. "That first year that I was diagnosed I read everything I could about Fanconi Anemia. I'm talking like every paper under PubMed in the past 50 years that I could get my hands on!"

Lauren shared a photo of medical tubing and needles to illustrate her demanding treatment regimen (Supplemental Figure 1). She described:

> I do infusions of plasma like three times a week from home . . . I have issues where it doesn't pierce the skin so, and it'll keep happening. And it can be like 20 times of trying to pierce the

> needle, and it doesn't wanna go in. I took a picture, I was like, this is a frustrating thing that other people don't have to deal with that it's like, 'Oh, I don't want to be ill, and I don't want to have to do this.' But I do it for myself, 'cause I'm like if I don't do it I start getting chest infections and the consequences are worse.

FA symptoms shape life. Interviewees described how physical symptoms from FA disrupt their daily lives, saying, "I'm tired, exhausted, I want to rest from this, but you know you can't because it's a disease that stays with you." When describing complicated medication management, Lucia shared a photo of medications she splayed out with Baby Yoda placed in the middle (Supplemental Figure 2). She described:

> They are elements that are always with someone. There's always one pill here, one pill there. There also [are] the needles because I have to prick myself every day. They form a part of you, like your companions, but they are also a little complicated because, now this one, another one now, now later it will be changing the dose. And well in the middle I put a Grogu, that is Baby Yoda. . . For me it's like, well I know that it's a really fun person. But he's there, he's not active and I have a little, a point of calm in the storm of pills and medication that there are that you take every day.

Supplemental Figure 3 is a photo from Lauren of her bedside table cluttered with candies, soda, a camera, and medications. She described working every day that month doing freelance photography. She described:

> A lot of time I don't have any time to go and get proper food and like make myself meals. . . . there's other parts of my life that sometimes take like a back step where I can't do as well in some parts 'cause I'm focusing on others. . . I feel kind of frustrated at myself 'cause I'm like, I should be cooking proper meals.

Ana shared a picture her son took of her sleeping, as fatigue was one of her prominent symptoms (Supplemental Figure 4). "People know this about me, that if I'm not working or preparing my classes, I'm asleep."

Theme 2: Stigma and trauma take a toll on mental health

Healthcare Trauma. Interviewees described trauma from frequent interactions with the healthcare system, including misdiagnosis, treatments, and screenings, all of which carried the threat of mortality. "I would say the health experiences I've been through have been a form of health trauma. . . going through chemo at a young age, and all those feelings you go through with getting the diagnosis of a severe health condition that you have to manage." This category also includes being invalidated or rejected by doctors. " . . .You find yourself with more of these problems with doctors who don't want to follow protocol. Doctors that want to discharge you. You will find yourself dealing with a problem for years and they do not listen to you."

One participant discussed the lack of confidentiality from healthcare providers they experienced as a minor during their transplant:

> I definitely wasn't honest with them [healthcare providers] 'cause. . . I was like, 'Well, if I tell anything to this nurse about how I feel, she's just gonna go tell my mom straight away.' I knew that. . . anything that comes out of my mouth, it goes to everyone. So I was like, 'Well, nothing's confidential now.' And I definitely needed that [confidentiality].
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> Another participant discussed the trauma from their transplant:
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> The anxiety, I think, which triggered with transplant itself. . . just knowing that I may not make it through transplant because then it was still very experimental. My parents [said] that I was a guinea pig because I was. I went through transplant without radiation. . . and then there were two other patients who went through the protocol and did not make it. I knew the two people after me because we were in the hospital together.

Isolation. Interviewees experienced social isolation due to FA, such as feeling different from others, that no one understands their situation, and overall loneliness. Individuals also discussed physical isolation during transplant or longer and more intense isolation due to their immunity status during COVID-19. One participant said, “I talked about the loneliness, you know, during [the] pandemic. You know, I probably went months without talking to another human being in person other than like the person drawing my blood at the hospital.” Participants discussed the social isolation they felt returning to school after transplant, “Not a lot of people to relate to. And then I felt kind of just in a different spot mentally than my peers.”

Body image and gender norms. Participants talked about the stigma they experienced toward observable aspects of FA, such as not fitting social norms of height, weight, femininity, and masculinity. Interviewees also discussed looking young and being underestimated or being bullied for having hand differences. One participant said, “How people see my appearance, the way I look. I don’t feel comfortable with that.” Another said, “Having FA you have so many things happening to your body, like I can have my physical appearance changing. So sometimes. . . I stigmatize myself.”

Discrimination. Interviewees discussed experiences of bullying and discrimination. One stated, “I was definitely bullied for, like not having hair was the main thing.” Having additional minority social identities, such as race, gender, and sexuality, intensified stigma already experienced due to FA. Another participant described facing discrimination in a healthcare setting, “My spouse [and I] belong to different races. . . But the first [inaudible] we are referred to, at first they thought, because we went, the two of us. They thought. . . my spouse was the one who was supposed to be receiving the treatment. So at first the reception was a bit warm. But when they noted that I was the one [who was] supposed to receive the treatment I noticed the reception kind of changed. We started having some cold treatment. Which, with my condition, I was not comfortable. And I just decided to forgo the process and seek another alternative.” A participant observed that “Mostly here in [African country], people have different cultural perspectives. So when they see me, and the way I look they see as if you know maybe it’s [FA’s] a curse, or something, or like sort from my ancestral lineage.”

Mental health symptoms and diagnoses. Often citing the challenges described in this theme, participants recounted mental health symptoms or diagnoses including depression and anxiety. Periods during which these were elevated included times of isolation, discrimination, life transitions, and while undergoing transplants.

Theme 3: Connection is bittersweet

Connecting with FAmily. Most interviewees talked about the value of spending time with the FA community (sometimes called FAmily) due to their shared understanding of FA and the opportunity to learn from others’ experiences. “To be able to connect with people, talk with people that understand you because they have the same thing as you. Well, it helps tremendously, because you don’t feel alone.” Several participants went to great lengths to connect with others, including traveling internationally or learning English (to speak with the predominantly English-speaking FA community).

Giving back to FAmily. Many interviewees found meaning and joy in giving back to FAmily. Examples include having careers in FA or chronic illness care, participating in research studies, volunteering, and supporting friends with FA. One interviewee shared, “Ever since my transplant I admired my nurses. . . They’re the reason why I became a nurse. . . To be a nurse in oncology has just made it even better because I'm able to give back."

FA community can be inaccessible. Interviewees discussed how FA communities can be inaccessible as they are primarily located in Western, English-speaking countries. "It's just weird being a minority in White spaces in general, it's not really specific to FA. But like I have noticed little slight microaggression things at those meetings before. . . I don't want to call anything out or anything but yeah, it's a little bit different. That's just kinda being a minority in the U.S. though. . . " A participant shared, "I'm lesbian and I think that definitely has kinda come up because I feel like within the FA community there has been like the few people I've met. . . You'll find out that they're actually really homophobic." Two participants who live in Africa noted they have not met anyone with FA in their respective countries.

Grief. Contending with illnesses and deaths in the FA community means people with FA experience grief in many forms, such as from the death of loved ones, their lives before FA, or from having a life-limiting disorder. A participant explained, "It's been difficult, because a lot of the friends I've made have passed away, and it kinda makes me not want to continue going to the [FA] meetings sometimes, 'cause it's just really upsetting. So sometimes I can handle going to those meetings, and sometimes I haven't been able to for a little while now."

Collective support. Interviewees discussed their families encouraging them to seek mental health support or finding them a provider. For instance, "My FAmily actually did all of that, you know, with the aid of my brother. . . He arranged my wait on the therapist for me." When speaking about the many local community members who have supported her, one participant said, "Just like the amount of people that think of me when I don't even know it. It just kinda like breaks me down 'cause it just makes me feel so special."

Ana shared a picture of her dining with her son and dog (Supplemental Figure 5), which she does every day. Ana was unsure if she would be able to have a child, and shared, "I feel blessed because I'm able to have time with my son. It's a gift: I am very pleased with whatever time I have with him."

Inappropriate support. Although many participants benefited from social support, there were also times when well-meaning individuals provided inappropriate or overbearing support. Examples include people offering unhelpful advice, being overprotective, or trying to convert them to religion. In one instance, a participant shared that, "I thought I was getting a bone marrow transplant. I was like, 'Okay, yeah, all in. Let's do it.' And she [participant's mom] was like, 'Oh, no, no, like. We need to get seven different opinions. And let's try other things first.' And I'm like, well, this is the reality of the situation."

FA affects romantic relationships. FA has complex effects on romantic relationships as individuals may face fears of entering a relationship, their partner being seen as their caregiver, or tolerating harmful relationships due to a lack of perceived alternatives. A participant shared, "I've had two ex-partners that used the disease as a form of manipulation. . . 'You have a disease, who is going to love you?' " Fortunately, some interviewees found loving, supportive relationships. Regarding his wife, one participant said, "Having my wife by my side always, it's a big help. It's a big tick in my fight with FA."

Living for others. Some individuals reported making an intentional decision to continue screening or treatment even when they felt like giving up, or avoiding conversations about death, as they did not want to cause their loved ones grief. A participant explained how growing up, “I knew that when I asked some questions about my condition, you know my dad would start crying. My mom would get all sad. . . So I was like, ‘Well, I’m not gonna share how I feel because this just upsets people.’ ”

Theme 4: Mental healthcare barriers

Providers lack knowledge. Participants often felt they had to educate their providers as many did not understand FA. One participant shared, “I would love to talk to someone that actually understands it because I’ve never done that.” Interviewees desired mental health providers with knowledge about FA, chronic illness, RDs, and/or cancer. “I would like someone [a mental health provider] who has experience with people that have had multiple medical problems and issues.”

Providers avoid FA. Interviewees who had previously seen a mental health provider discussed some providers’ unpreparedness or unwillingness to discuss FA or related topics. One participant described how their mental health providers did not know how to approach treatment, saying, “There are a lot of health professionals that are kind of scared of me when they hear my whole history, and a lot of them just don’t know how to deal.”

Provider concordance. Interviewees desired a provider with shared social identities, including race, gender, sexuality, and/or health status. A participant of color said, “So I think in the future I might look for someone that’s a little bit older, and probably preferably [a racial] minority. But that’s not, you know, I wouldn’t dismiss someone for not being, but it’s just like a different level of understanding.” Another participant of color stated that he “ . . .needed [a psychiatrist] who’d be open-minded. Who wouldn’t discriminate [against] me based on my race.”

Mental healthcare access. Interviewees shared experiences of learning to navigate mental healthcare services, insurance, traveling long distances for care, or long waiting lists. This category also includes an overall lack of providers, appointment delays, or rushed visits. Indeed, one participant noted, “There was [a] lack of mental services in my locality. So I had to move.”

Financial barriers. Participants discussed instances where financial issues kept them from seeking a mental health provider or continuing with care. Examples include losing insurance coverage or paying out of pocket to see a different mental health provider. One participant said, “Cost of accessing counseling services. It’s quite high to be honest. So that. . . has been the major hiccup I’ve faced trying to seek mental health treatment.”

Theme 5: “A process of accepting the disease”

Keeping occupied. Interviewees coped by spending time in nature and the outdoors, listening to music, spending time with pets, playing video games, watching or playing sports, and engaging in creative activities. One participant shared how, “Yoga really helps me and also like focusing on some things that I like to do, kind of like gets my mind off of things. I like to cook and bake and do art projects.”

I’m a fighter. Participants felt the low points in their life proved they could overcome challenges. One participant stated, “I feel like if I didn’t have FA then I wouldn’t be so resilient to lots of things my friends find like, really big issues in life.” Another participant said of surviving transplant, “ . . .made me proud of myself because in that moment I felt, ‘Okay, I like passed this very difficult moment of my life and I somehow made it.’ And that made me feel good about myself. . . ” Another said, “If it ever gets bad again I'm strong enough, and I can make it if it comes down to it because I've done it once."

Lucia shared a photo of a painted bowl she made in a class she teaches about mindfulness (Supplemental Figure 6). She described that the book in the center represents the trauma she experienced during her education. She always loved learning and wanted to succeed in school, but she was bullied by students and instructors:

> I missed a lot of class because of course, because you have a disease and you have to be continuously going to the hospital. But you also miss it because you don't want to go. To be seen as a weirdo. I had to learn to live with chronic pain. A pain that, it's a continuous pain every day, that isn't easy. You have to learn how to live with it, nobody was going to teach me how to live with it. I spent two years isolated at home when I had the transplant and then after, when I returned to high school, but nobody was going to explain to me how to relate with other people. . . developing into a trauma in which I could not enter a classroom or take exams.

Through therapy and mindfulness practices, she was able to move beyond this trauma and start learning again:

> I've always liked learning. . . I've come here because of all of the rejection that I've received throughout my life. I haven't stopped myself from being able to follow the path that I wanted to go down. The idea is I can transform a little of this pain as well into expressing it and getting it out. Also to transform it into something beautiful.

Optimism. Many interviewees strived to feel hopeful in their daily lives, such as by saying, "No matter what life throws at you, you can find the end of the tunnel. . . Just take it one day at a time."

Appreciate the current moment. Interviewees talked about being grateful for life, practicing mindfulness, and enjoying the present moment. Participants discussed living every day like there's no tomorrow, which occasionally involved taking risks that doctors may not approve of, like drinking alcohol. A participant shared, "Simple things like someone coming to see you at home, and I don't know, you drink a tea, you have a glass of water, and that's, just sometimes that's wonderful."

Supplemental Figure 7 is a photo of a singing meditation bowl shared by Lucia. She described:

> It is a sound that helps me a lot to find peace or calm in the many complicated or difficult moments that there are. . . A lot of times with a disease you get lost along the way. You let yourself be carried away by the pain, by the fear, by the uncertainty, by a lot of feelings that are completely normal but a lot of the time, I think that they are difficult as well. I think that sometimes they're so difficult that they drag us down and they make us forget where we are, who we are. And the fact of using these elements reminds you that you are here, that you are alive, that it's normal to have these fears, it's normal to have uncertainty, what's happening is normal. But it also reminds you that there are many times that you don't care about those fears, that reality. That inside of that uncertainty you can also find good things.

Acceptance. Interviewees talked about how they embraced who they are and what they cannot control, such as their mortality. One participant said, "It is a process of accepting the disease, from which you will learn to love yourself more. You will learn to respect yourself and also to see life in a different way."

Uniqueness. Some participants felt FA made them unique or gave them unique opportunities, such as traveling, learning English to communicate with the FA community, being sought after by researchers, and feeling mature beyond their years. One participant shared, "I see life a lot differently than a lot of my peers do and I savor moments a lot more than my peers do, too. I think it's made me grow up really quickly from a young age."

Supplemental Figure 8 is a photo of Ana teaching English to her university class. She lives in a primarily Spanish-speaking country, and she learned English so she could communicate with the FA community. "They say I'm the best teacher they have." She was bullied when she was younger about her deep voice, which was caused by FA medications. Now, as a university instructor, "My voice, which was a problem to me at that time when I was younger, now is a blessing. My tone is like very serious. So my students are like, 'Okay, she's talking, silence,' because it's very strong. I never thought that I would be teaching, never, ever, and I feel proud. I'm proud of myself."

Effective mental healthcare. Interviewees highly valued access to mental health therapy and/or psychiatric medications. For instance, "I decided to book some appointments with a therapist, which enabled me to like. . . It was actually helping me out as well as my FAmily."

Life story interview summary. Here, we provide a descriptive count of codes in response to each life story interview question. When asked about a low point in their lives with FA, seven participants described discrimination, five described fertility issues, and three described the transplant period. When asked about a high point, five described giving back to FAmily, four described connecting with FAmily, four described collective support, four described surviving transplant, and four described uniqueness. When asked about a turning point, four participants described the transplant period, three described acceptance, two described connecting with FAmily, and one described grief.

Discussion

This qualitative study explored the psychosocial experiences of adults living with FA. The five overarching themes include, "FA is all encompassing," Stigma and trauma take a toll on mental health, Mental healthcare barriers, Connection is bittersweet, and "A process of accepting the disease." This study adds to the small body of literature regarding the psychosocial aspects of living with FA and reveals crucial gaps in mental healthcare and support for adults with FA.

As the science of treating FA evolves, so do the psychosocial challenges that people with FA experience. Past research has found that individuals with FA and their families had perceptions of ambiguity due to conflicting scientific evidence about transplant (Hamilton et al., 2013). While those guidelines have become clearer as the science of transplant has evolved, families continue to face other sources of ambiguity. Throughout interviews participants expressed many ways in which "FA is all encompassing," including uncertainty about when a transplant would be needed, the ever-present threat of cancer, and what career and life goals to set for themselves. To mitigate uncertainty, those with FA become "professional patients," researching their disease as a way to gain control.

Stigma and discrimination were notable challenges for adults with FA and were described as the most common low point in participants' lives. Participants reported experiencing bullying and discrimination due to visible differences associated with their condition, such as small stature, thumb abnormalities, and hair loss. Additionally, they described feelings of ostracization stemming from life experiences markedly different from those of their peers. The previous quantitative study identified stigma as a strong predictor of anxiety, depression, and PTSD among adults with FA (Bogart et al., 2025). Although stigma is a well-documented challenge in broader RD research (Bogart et al., 2022; Munro et al., 2022), these findings underscore its critical yet poorly recognized impact on adults with FA.

Participants indicated that stigma and healthcare trauma take a toll on their mental health. In our previous quantitative study, half of participants screened positive for PTSD (Bogart et al., 2025). Healthcare trauma has been found to occur in a high proportion of hematopoietic stem cell transplant patients, with estimates of PTSD ranging from 3% to 28% (Janicsák et al., 2021). Moreover, high cancer surveillance has been associated with increased distress and lower quality of life in certain hereditary cancer risk populations (Gopie et al., 2012).

Participants experienced significant barriers to mental healthcare. People with RDs and disabilities desire providers with disability cultural competency and familiarity with the challenges of RDs, yet mental healthcare providers receive little training on these topics (Kole et al., 2010; Wang et al., 2024; Whittle et al., 2018). Minoritized individuals, including those with disabilities, also desire mental health providers with concordant identities, as they can enhance trust and rapport (Eken et al., 2021; Moore et al., 2023). Unfortunately, mental health providers with disabilities and other minority identities are underrepresented in the field, and may face complex decisions about whether to be "out" about their conditions (Battalova et al., 2020).

Because people of color with FA exist at the intersection of being both of color and having a disability, they have a higher need for mental healthcare that is both racially and FA competent. This means that if they can access mental health services, they may have to settle for a provider who is either racially competent or FA competent, which does not serve the whole individual.

Research on coping with chronic illness generally finds benefits to active, problem-focused coping, which was echoed in Frohnmayer's (2010) study as well as ours. People with FA often played a very active role in their medical care, following rigorous treatment regimens to the point where they felt like "professional patients." Although exhausting and all-encompassing at times, these self-management and self-advocacy skills also build disease self-efficacy, which was found to be a protective against mental health symptoms in our quantitative study (Bogart et al., 2025). However, when a stressor is uncontrollable, such as many aspects of FA, approach-based emotion coping can also be beneficial (Stanton and Hoch, 2025). Participants described a journey toward active acceptance and post-traumatic growth. We also found that participants desired to appreciate the current moment in a way that was not directly connected to FA. Frohnmayer noted that when reflecting on one's overall experience of FA, a common response was that it made individuals stronger, a protective aspect of post-traumatic growth participants in our qualitative and quantitative studies exhibited as well.

A rich literature points to the benefits of patient communities and support groups when coping with disabilities and chronic illness (Bardon et al., 2022), yet in the FA community, this "connection is bittersweet." When asked about their high points living with FA, individuals frequently highlighted meeting others with FA for the first time or helping others with FA. In Ashtari and Taylor's (2023) research on support groups for those with RDs, participants expressed the sense of community from online support groups helped with their mental health. In our study, however, participants also mentioned the need to take time away from FA support groups due to grief. Frohnmayer (2010) noted that for many individuals who attend FA meetings, feelings of grief are intertwined with individuals' experiences with FA. Participants with additional minority identities felt the FA community was inaccessible in multiple ways, such as being located in predominantly Western, English-speaking countries and primarily composed of people who were White, cisgender, heterosexual, and higher socioeconomic status. Despite these challenges, participants emphasized that the FA community provides valuable solidarity and resources that individuals with FA cannot access elsewhere.

Limitations and strengths

Despite efforts to recruit participants through diverse channels, most were already connected with the FA community. We can speculate that individuals who are not connected with this community may experience even greater challenges in mental health support. Our study had several strengths. A participatory approach ensured that the study reflected the experiences and needs of adults with FA. Our diverse advisory board and intentional sampling allowed us to recruit a far more diverse group of participants than is conventional in RD research.

Future directions

A multi-level and whole person approach to improving the mental health of adults with FA is needed. Their needs are not always well defined or captured by existing mental health diagnoses. A tiered approach that includes a solid foundation of wellbeing strategies such as social support, mindfulness, self-expression, empowerment and education can be useful (Voss et al., 2023). Building on that foundation is the need for community-based support such as connection with community organizations, peer support, mental health providers, and other allied health practitioners providing whole person health (music therapist, physical therapist, etc.). Finally, healthcare teams have a responsibility to attend to the needs of the whole person by offering expertise and specialty referrals as appropriate to enhance quality of life, such as pain management/palliative care, psychiatry, sleep medicine, etc. There is also a need for RD or FA-specific training to prepare mental healthcare providers.

Together, community-based services, comprehensive whole person care, and competent mental health care can begin to address the complex challenges described by this and other RD groups. Mental health and wellbeing are individual and multidimensional, therefore, they can only be addressed with tiers of interventions designed to meet those unique needs.

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Author contributions

KRB: conceptualization, data curation, formal analysis, funding acquisition, methodology, project administration, supervision, writing-original draft preparation, writing-review and editing. ML: conceptualization, data curation, formal analysis, investigation, writing-original draft preparation, writing-review and editing. SF: data curation, formal analysis, investigation, writing-original draft preparation, writing-review and editing. MV: conceptualization, funding acquisition, methodology, writing-review and editing.

Data sharing statement

The data presented in this article are not readily available due to their sensitive nature; participant responses may contain details that could identify them. Reasonable requests to access de identified data should be directed to Kathleen.bogart@oregonstate.edu

Declaration of conflicting interest

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Ethics approval

This study was approved by The Oregon State University Institutional Review Board Health Research Ethics Committee (approval no. HE-2023-316).

Informed consent

Participants provided written informed consent to participate.

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Notes

1. Traditionally, life story interviews are analyzed separately using an individualized narrative identity approach; which was beyond the scope of this paper.

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