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Mental health in the first generation of adults with Fanconi anemia

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ABSTRACT
Fanconi anemia (FA) is a rare genetic disorder involving bone marrow failure and cancer predisposition. Due to medical advances, people with FA are now living to adulthood and unmet mental health concerns are emerging. We aimed to explore mental health outcomes and protective and risk factors for the first time using a participatory research approach. Participants included 102 adults with FA from diverse national, ethnic, and racial backgrounds who completed validated measures of anxiety symptoms, depression symptoms, post-traumatic stress disorder (PTSD) symptoms, health-related quality of life, FA characteristics, personal factors, and social factors. When compared to population norms, adults with FA had significantly poorer health-related quality of life on all domains measured, including anxiety symptoms, depression symptoms, fatigue, physical function, sleep disturbance, social participation, pain, and cognitive function. Results showed 50% of participants had probable PTSD, 33% had probable anxiety, and 25% had probable depression. Hierarchical regressions indicating protective/risk factors for symptoms of anxiety, depression, and PTSD included stigma, disability self-efficacy, fatigue, concern with death and dying, and post-traumatic growth. Adults are at risk for symptoms of PTSD, anxiety, and depression. Just as people with FA should receive regular cancer screenings, they should also be screened for mental health symptoms.

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Fanconi anemia; rare disease; cancer predisposition; participatory research

Rare diseases and disorders (RD) constitute a public health priority (Schieppati et al., 2008). Only 5% of RDs have an effective treatment, and relatedly, research finds people with RD experience poor health-related quality of life (K. R. Bogart et al., 2022). A critical gap is that RD research has primarily taken a biomedical, rather than a biopsychosocial approach, such as the International Classification of Functioning, Disability, and Health (ICF; World Health Organization, 2002). It is imperative to integrate research on medical aspects of the disorder with *personal factors* such as demographics and psychological factors, and *social/environmental factors* like stigma and social support.

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Fanconi anemia is a prime example of a RD for which psychosocial research remains virtually nonexistent. Fanconi anemia was named for what was once its most consequential and deadly sign, pediatric anemia from bone marrow failure (Fanconi, 1964). Medical advancements enabled allogeneic hematopoietic stem cell transplants, curing bone marrow failure and allowing many individuals with FA to live beyond childhood for the first time (Peake & Noguchi, 2022). Yet, as individuals reached adulthood, a new challenge was discovered: an extremely heightened risk of aggressive cancer. It is now understood that FA is an autosomal recessive disorder of DNA repair (Peake & Noguchi, 2022). Adding to its complexity, few cancer treatment options exist because traditional chemotherapy and radiation can cause increased side effects including additional cancer in this population (Peake & Noguchi, 2022).

FA is slightly more common in males than females, and is found in all ethnic and racial groups, but at a higher rate in Ashkenazi Jews, sub-Saharan Black people, and Spanish Gitanos (Tipping et al., 2001). Some individuals are born with visible physical manifestations, including short stature, small craniofacial structure, thumb and arm abnormalities, or café au lait spots. FA or related treatments can also limit fertility. Men may be particularly stigmatized for small stature, while women may be stigmatized for fertility issues. Similarly, café au lait spots may be particularly salient on people with darker skin tones. Systemic bias in the healthcare system may amplify challenges for people of color and other minorities.

There is a dearth of research on the psychosocial experiences of adults with FA. More is known about the experiences of parents (Hamilton et al., 2013; Haude et al., 2017; Zierhut & Bartels, 2012), siblings (Hutson & Alter, 2007), and children with FA (Kearney et al., 2012). The only previous study examining mental health in FA focused on the peritransplant period and primarily sampled children (Kearney et al., 2012). Of 32 children and adults with FA receiving stem cell transplantation, 22 were referred for psychiatric consultation (Kearney et al., 2012). Of those, 50% were diagnosed with mood disorders, 46% were diagnosed with adjustment disorders, and 23% with anxiety disorders, highlighting the significant risk of psychological distress during this critical phase.

To our knowledge, the late Amy Frohnmayer Winn, who had FA herself, conducted the only research focused on mental health in adults with FA, as found in an unpublished masters thesis (Frohnmayer, 2010) and a published conference abstract (Frohnmayer, 2016). In a qualitative study, Frohnmayer (2010) interviewed 18 adults with FA and identified challenges including stigmatized visible anomalies, fear of death and dying, uncertainty about the future, and feeling isolated. Coping strategies included the following themes: positive reframing, resilience, social support, connecting with the FA community, and active engagement with medical management (Frohnmayer, 2010). In a second study, Frohnmayer (2010) conducted a quantitative survey on wellbeing among 96 adults with FA. The coping strategies that correlated positively with hope and wellbeing were spirituality, positive reframing, and active coping. Thus, our project incorporates key factors from Frohnmayer's work and continues her legacy by studying their role in mental health.

People with RDs like FA are likely at high risk of anxiety, depression, and PTSD due to stress and potentially traumatic events including diagnosis or misdiagnosis, acute health crises, treatments such as bone marrow transplants, and cancer (Langhinrichsen-Rohling et al., 2021). In a study of adults with the cancer predisposition RD Li-Fraumeni syndrome, the majority had clinically significant levels of distress and low mental health-related quality of life (Kiermeier et al., 2024). The DSM-5 criteria for PTSD includes 'life-threatening illness or debilitating medical condition' as a potential traumatic event (American Psychiatric Association, 2013). As described by the Enduring Somatic Threat Model, cancer and other life-threatening health conditions can be uniquely traumatic because they involve an internal, protracted existential threat (Edmondson, 2014).

Approximately two-thirds of people with RDs feel they do not get adequate social and psychological support (Limb et al., 2010). Emotional support signals that the person is understood and valued, while companionship support provides a way to cope with exclusion and stigma by increasing a sense of belonging (Bambina, 2007). A large study of adults with diverse RDs identified companionship and emotional support as key predictors of satisfaction with life (Bryson & K. R. Bogart, 2020). We will investigate social support as a protective mental health factor.

Adults with FA now outnumber children (Fanconi Anemia Research Fund, n.d.), and there is an urgent need to understand their mental health experiences. Given the dearth of existing research, we conducted a community-engaged, large-scale, comprehensive study to answer the following questions:

Research question 1: How common are symptoms of anxiety, depression, and PTSD in adults with FA?

Research question 2: What FA characteristics, personal factors, and social factors are protective and risk factors for mental health symptoms?

Method

Participatory engagement

This project followed a participatory research approach (Balcazar et al., 1998; L. M. Bogart & Uyeda, 2009). We assembled a diverse Lived Experience Advisory Board of five adults with FA. The advisory board met approximately quarterly, identifying research goals, key constructs, survey accessibility issues, and assisting with recruitment and interpretation of findings.

Participants

Participants were recruited via email and social media channels of the Fanconi Cancer Foundation (FCF), their international partner organizations, FA centers of excellence, and our advisory board's social networks. The study was based in the United States (US) but collected an international sample. Inclusion criteria were 18 years or older, having a diagnosis of FA, and ability to communicate in English or Spanish. Based on Frohnmayer's (2010) previous research and discussions with FCF and clinical partners, a target of 100 participants was selected as a feasible goal.

Online research presents concerns about inattentive responses, bots, and fraudulent responses, especially when a financial incentive is offered (Simone, 2019; Storozuk et al., 2020). We employed recommended strategies to maintain data quality (Lawlor et al., 2021; Storozuk et al., 2020). In total, 187 participants completed the survey. Individuals were excluded if they had two or more of the following data integrity issues: took less than 15 minutes to complete the survey, failed attention checks (there were three in total), failed reCAPTCHA, provided inconsistent responses to questions about demographics, phone number provided did not match reported country of residence, responded with an exact duplicate response to open-ended question, and reported receiving treatment, such as bone marrow transplant, at a medical center that does not offer that treatment ($n = 85$).

Procedure

This study was ruled as exempt by the first and third authors' Institutional Review Board (IRB-2022–1517) and the second author's Institutional Review Board (Study00017238), respectively. Participants provided informed consent and completed a 40-minute survey containing the measures described below. For accessibility, participants could request the survey in a variety of formats: online via Qualtrics ($n = 101$), mailed paper copy ($n = 1$), phone interview ($n = 0$), English ($n = 101$), Spanish ($n = 1$)). Data collection occurred between October 2022 and April 2023. Participants received a $40 digital gift card. Survey instruments are described below.

Measures

Health-related quality of life

Health-related quality of life is a multidimensional construct representing the impact of health status on wellbeing, and was measured with the Patient-Reported Outcomes Measurement Information System (PROMIS-29 + 2) (Cella et al., 2010; Health Measures, n.d.). This instrument includes domains of physical function (current sample Cronbach's $\alpha = 0.87$), anxiety symptoms ($\alpha = 0.93$), depression symptoms ($\alpha = 0.91$), fatigue ($\alpha = 0.90$), sleep disturbance ($\alpha = 0.82$), ability to participate in social roles and activities ($\alpha = 0.89$), pain interference ($\alpha = 0.94$), and cognitive function ($\alpha = 0.75$). Higher numbers indicate greater amounts of the domain. Scales were scored using the published T-scores calibrated such that a mean of 50 and a standard deviation of 10 is representative of the U.S. general population (Cella et al., 2010). PROMIS means for people with one or more common chronic diseases have also been published for the following scales: anxiety, depression, and fatigue in the US (Rothrock et al., 2010). Probable cases of moderate to severe anxiety and depression were determined using published cut-points of > 65, which have been validated in cancer populations (Cella et al., 2014).

Social support

Emotional support refers to connections with others who provide empathic concern, while companionship support refers to connections with others who provide a sense of belonging. These constructs were measured with the 4-item PROMIS Emotional support ($\alpha = 0.93$) and Companionship ($\alpha = 0.89$) forms (Health Measures, n.d.). Scales were scored using the published T-scores calibrated such that a mean of 50 and a standard deviation of 10 is representative of the U.S. general population.

Post-traumatic growth

Post traumatic growth includes positive psychosocial outcomes of illness, such as acceptance, benefit-finding, meaning-making, and appreciation for relationships. This construct was measured with the 4-item PROMIS Psychosocial Illness Impact-Positive form (α = 0.77; Health Measures, n.d.). This scale asks respondents to indicate how true statements were before and since their illness. Discussions with our advisory board indicated that this was not appropriate for an FA sample due to its congenital yet dynamic nature. Instead, we asked participants to consider their experience with FA overall. Responses were summed and higher numbers indicated more positive illness impact.

Stigma

Stigma includes negative judgments, discrimination, and shame, and was measured with the Neuro-QOL 8-item stigma scale (α = 0.88; National Institute of Neurological Disorders and Stroke, 2015). The scale has been validated for use among individuals with a range of chronic health conditions and rare cancers. This scale was scored using published T scores calibrated such that a mean of 50 and standard deviation of 10 are representative of the norms for populations with common chronic illnesses (National Institute of Neurological Disorders and Stroke, 2015). Higher scores indicate greater perceptions of disability-related stigma.

Concern with death and dying

People with life-limiting illnesses may experience worry and preoccupation with death and dying; these were assessed using the Neuro-QOL 6-item Concern with Death and Dying scale (α = 0.92) (Carlozzi et al., 2016; National Institute of Neurological Disorders and Stroke, 2015). This scale was developed for use in Huntington's disease and has been recommended for use in other rare terminal illnesses. Responses were summed, and higher scores indicate greater concern with death and dying.

PTSD

PTSD includes re-experiencing, avoidance, negative cognitions and mood, and arousal related to a traumatic event. The PTSD Checklist for DSM-5 (PCL-5) was used (α = 0.95; Blevins et al., 2015). This self-report measure corresponds with the DSM-5 criteria to assess the presence and severity of PTSD over the past month. This measure has been used in oncology populations (Jung et al., 2021). Following the scoring guide, a cutoff score of 31 or higher was used to indicate a provisional diagnosis.

Disability self-efficacy

Disability self-efficacy is confidence in managing disability-related issues that arise and was measured with the 6-item University of Washington Disability Self-Efficacy Scale. This instrument shows good psychometric properties (Amtmann et al., 2012). Scales were scored using the published T-scores calibrated such that a mean of 50 and a standard deviation of 10 are normed to a sample of people with chronic conditions. Higher numbers mean greater disability self-efficacy (α = 0.90).

Spirituality

Spirituality includes beliefs about religion and/or a connection to something greater than oneself. This construct was measured using the Spirituality scale of the World Health Organization Quality of Life instrument ($\alpha = 0.89$; World Health Organization, 2012). This cross-cultural measure was developed to assess individuals' self-perceptions of their life in the context of the culture and value systems in which they live. It was developed in 15 cultural settings and has since been tested in many more. Items were summed, with higher numbers indicating greater spirituality.

Demographics

We assessed gender, sexual orientation, country of residence, age, education level, race/ethnicity, and income.

FA symptoms and characteristics

Age of diagnosis, year of diagnosis, bone marrow transplant status, and cancer status were measured. Participants were asked about existing mental health diagnoses or symptoms and psychiatric medication use.

Data analysis

In order to account for multiple exploratory analyses, we used the Benjamini and Hochberg false discovery rate control approach (Glickman et al., 2014). This resulted in a critical p-value of .025, and we report tests with p-values less than this threshold value as significant.

To address research question 1 about the frequency of symptoms of anxiety, depression, and PTSD in adults with FA, frequencies of participants exceeding published cutoffs indicating caseness were reported. Participant data were also compared to published norms when available using one-sample t tests. (See Measures section for description of cutoffs and norms.)

Research question 2 examined risk and protective factors for mental health outcomes. First, we conducted bivariate correlations between outcome measures (i.e. anxiety, depression, PTSD) and all variables. Only variables that were significantly correlated with outcomes were then included in regressions with each respective outcome. Hierarchical multiple regressions were conducted following the ICF biopsychosocial framework (World Health Organization, 2001). The first step represented the health condition (FA symptoms), the second step included personal factors, and the third step added social factors. The addition of each step allowed us to test whether personal and social factors predicted distress over and above biological factors (which have been more thoroughly studied to date).

Results

The final sample totaled 102 participants, and Table 1 shows participant characteristics. Participants were, on average, 26.86 (SD = 5.46) years old, and the majority were male (60%). Most participants (71%) lived in the US, with 6% living in Canada, 3% living in the UK, and 20% living in other countries.

Table 1. Participant characteristics.

Characteristic	Frequency (%)	Mean (SD
Age		26.86 (5.46)
Age diagnosed		12.03 (7.10)
Country		
United States	71	
Canada	6	
UK	3	
Other	20	
Race/ethnicity		
African, Black, or Caribbean	34	
Asian	5	
White	40	
Hispanic or Latino/a/x	13	
Middle Eastern	1	
Indigenous	1	
Mixed race or multiple ethnic groups	3	
Prefer not to answer	6	
Gender identity		
Female	37	
Male	60	
Transman	1	
Prefer not to answer	2	
Sexual orientation		
Asexual	3	
Bisexual	6	
Gay	5	
Heterosexual or straight	78	
Lesbian	5	
Queer	1	
Other	1	
Prefer not to answer	1	
Gross income in U.S. $		
under $10,000	12	
$10,000–25,000	11	
$25,001–40,000	6	
$40,001–55,000	24	
$55,001–70,000	25	
$70,001–85,000	8	
$85,001–100,000	5	
$100,001 and above	9	
Education		
Did not complete high school or equivalent	3	
Completed high school or equivalent	29	
Completed community college or vocational school	20	
Completed bachelor's degree or equivalent	40	
Completed graduate degree/postgraduate degree	8	
Received bone marrow transplant	47	
Diagnosed with any type of cancer		
No	54	
Yes, 1 time	36	
Yes, 2–5 times	10	
Yes, 6–10 times	1	
Diagnosed with mental health disorder		
No	42	
Yes, I was diagnosed in the past, but I do not have symptoms	22	
Yes, I was diagnosed and I currently have symptoms	22	
I wasn't diagnosed but I currently have symptoms	15	

(*Continued*)

Table 1. (Continued).

Characteristic	Frequency (%)	Mean (SD
Current psychiatric medication use		
antidepressant medication	33	
anti-anxiety medication	25	
sleep medication	20	

Note. Numbers may not sum to 100 due to rounding. Other countries each represented less than 2% of the sample and included countries such as Ghana, Italy, the Netherlands, and South Africa.

Table 2. Health-related quality of life and psychosocial factors compared to published norms.

Variable	Current sample M (SD)	M difference from population norm	M difference from chronic illness sample
Anxiety	58.51 (10.04)	8.51*	6.55*
Depression	57.15 (9.63)	7.15*	5.19*
Physical function	43.71 (8.03)	– 6.29*	– 1.79
Fatigue	53.81 (8.54)	3.81*	1.28
Sleep	52.34 (7.32)	2.34*	NA
Pain	57.85 (7.96)	7.85*	4.11*
Social participation	47.58 (7.32)	– 2.42*	NA
Cognition	48.05 (6.93)	– 1.95*	NA
Emotional support	48.14 (6.88)	– 1.86*	NA
Companionship support	47.82 (7.25)	– 2.18*	NA
Stigma	58.63 (5.23)	NA	8.63*
Disability Self-Efficacy	45.18 (.04)	NA	– 4.82*

Note. NA indicates that published comparison data were not available. * denotes $p < .025$.

The first research question concerned levels of anxiety, depression, and post-traumatic stress disorder symptoms in adults with FA. Using published cut-points, 32.4% of participants screened positive for anxiety, 25.5% screened positive for depression, and 50% screened positive for PTSD.

We also compared health-related quality of life and psychosocial factors to published means when available, as shown in Table 2. Compared to population norms, adults with FA had poorer scores on all health-related quality of life and psychosocial factors, including anxiety, depression, fatigue, physical function, sleep disturbance, social participation, pain, cognitive function, emotional support, and companionship support. When comparison means for samples with common disorders were available, adults with FA fared comparatively poorer in domains of pain, anxiety, depression, disability self-efficacy, and stigma, but they did not differ in fatigue or physical function.[1]

The second research question asked what FA characteristics, personal factors, and social factors are protective and risk factors for mental health disorders. Bivariate correlations with all outcome measures (i.e. anxiety, depression, PTSD) are shown in supplemental material.

As shown in Table 3, in the first step of the model predicting anxiety symptoms, significant FA characteristics included sleep disturbance and fatigue. When personal factors were added in the second step, significant predictors included fatigue, concern with death and dying, and low disability self-efficacy. In the final step adding social

Table 3. Hierarchical linear regression predicting anxiety.

	Step 1			Step 2			Step 3		
Predictor variable	*b*	*SE*	*β*	*b*	*SE*	*β*	*b*	*SE*	*β*
FA factors									
Cognition	.19	.13	.13	.08	.12	.06	.20	.13	.14
Pain	.09	.16	.07	.08	.15	.07	.06	.15	.05
Sleep	.34	.13	.25*	.19	.12	.13	.15	.12	.11
Fatigue	.43	.12	.38*	.38	.11	.33*	.40	.11	.35*
Physical function	− .32	.16	− .25	− .03	.15	− .02	.01	.14	.01
Cancer status	1.21	1.23	.08	.08	1.10	.01	− .72	1.12	− .05
Personal factors									
White racial identity				− 2.23	1.72	− .11	− 1.85	1.64	− .09
Black racial identity				.60	1.70	.03	− .02	1.62	.00
Concern with death				.46	.15	.28*	.34	.15	.21*
Disability self-efficacy				− .30	.13	− .24*	− .28	.12	− .22*
Post-traumatic growth				.09	.10	.08	.15	.09	.12
Social factors									
Social participation							.10	.15	.07
Emotional support							− .12	.11	− .09
Stigma							.60	.18	.31*
Adjusted R^2			.54*			.65*			0.69*

Note. White racial identity was dummy coded such that 0 = not White and 1 = White. Black racial identity was dummy coded such that 0 = not Black and 1 = Black. * denotes $p < .025$.

Table 4. Hierarchical linear regression predicting depression.

	Step 1			Step 2			Step 3		
Predictor variable	*b*	*SE*	*β*	*b*	*SE*	*β*	*b*	*SE*	*β*
FA factors									
Cognition	.18	.11	.13	.07	.09	.05	.18	.10	.13
Pain	.09	.14	.07	.01	.12	.01	.00	.11	.00
Sleep	.29	.11	.22*	.12	.09	.09	.08	.09	.06
Fatigue	.43	.10	.40*	.37	.08	.35*	.38	.08	.36*
Physical function	− .35	.13	− .30*	− .13	.12	− .11	− .08	.11	− .06
Cancer status	2.10	1.02	.15	1.39	.85	.10	.53	.83	.04
Personal factors									
White racial identity				− 1.82	1.36	− .10	− 1.37	1.23	− .07
Black racial identity				1.53	1.32	.08	.96	1.22	.05
Spirituality				.22	.17	.08	.16	.16	.06
Concern with death				.43	.12	.28*	.32	.11	.21*
Disability self-efficacy				− .20	.10	− .17	− .19	.09	− .16
Post-traumatic growth				− .32	.21	− .11	− .19	.20	− .06
Social factors									
Social participation							.07	.11	.05
Emotional support							− .15	.10	− .11
Companionship support							.06	.09	.04
Stigma							.57	.13	.31*
Adjusted R^2			.65*			.77*			.82*

Note. White racial identity was dummy coded such that 0 = not White and 1 = White. Black racial identity was dummy coded such that 0 = not Black and 1 = Black. * denotes $p < .025$.

factors, significant predictors included fatigue, low disability self-efficacy, concern with death and dying, and stigma. Each step explained a significant increase in variance, and the final model explained 69% of the variance in anxiety.

The first step of the model predicting depression (shown in Table 4) indicated that significant FA characteristics included sleep disturbance, fatigue, and low physical function. When personal factors were added in the second step, fatigue and concern with death

Table 5. Hierarchical linear regression predicting PTSD.

	Step 1			Step 2			Step 3		
Predictor variable	*b*	*SE*	*β*	*b*	*SE*	*β*	*b*	*SE*	*β*
FA factors									
Cognition	−.08	.18	−.04	−.06	.16	−.03	−.02	.18	−.01
Pain	.58	.24	.30*	.34	.23	.18	.27	.22	.14
Sleep	.60	.19	.28*	.34	.18	.16	.24	.17	.11
Fatigue	.14	.17	.08	−.03	.16	−.02	−.10	.16	−.06
Physical function	−.39	.25	−.20	−.41	.25	−.20	−.21	.25	−.10
Cancer status	4.24	1.78	.18*	4.45	1.59	.19*	3.51	1.61	.15
Transplant status	−3.07	2.31	−.10	−3.43	2.40	−.11	−3.56	2.33	−.12
Personal factors									
White racial identity				−.93	2.48	−.03	−.95	2.35	−.03
Black racial identity				−2.81	2.45	−.09	−2.78	2.36	−.09
Sexual minority				.82	2.64	.02	.97	2.56	.03
Income				.07	.48	.01	.16	.50	.02
Spirituality				.05	.38	.01	−.02	.37	.00
Concern with death				.52	.22	.20*	.48	.22	.19
Disability self-efficacy				−.03	.17	−.02	.02	.17	.01
Post-traumatic growth				−1.73	.38	−.36*	−1.37	.38	−.28*
Social factors									
Social participation							−.19	.21	−.09
Emotional support							.15	.18	.07
Companionship support							−.16	.19	−.08
Stigma							.77	.25	.26*
Adjusted R^2			.64*			.74*			.77*

Note. White racial identity was dummy coded such that 0 = not White and 1 = White. Black racial identity was dummy coded such that 0 = not Black and 1 = Black. Sexual minority was coded such that 0 = heterosexual and 1 = sexual minority. * denotes $p < .025$.

were significant predictors. In the final step adding social factors, predictors included fatigue, concern with death and dying, and stigma. Each step explained a significant increase in variance, and the final model explained 82% of the variance in depression.

In the first step of the model predicting PTSD symptoms (see Table 5), significant FA characteristics included pain, sleep disturbance, and cancer diagnosis. When personal factors were added in the next step, significant predictors included cancer diagnosis, concern with death and dying, and low post-traumatic growth. The final model including social factors indicated that low post-traumatic growth and high stigma were significant predictors. Each step explained a significant increase in variance, and the final model explained 83% of the variance in PTSD.

Discussion

In the first large-scale study of the mental health of adults with FA, we assessed symptoms of anxiety, depression, and PTSD, and we explored biopsychosocial protective and risk factors. In our sample, 44% of participants reported having been diagnosed with a mental health disorder. Of those, 22% of those were currently experiencing symptoms. Another 15% believed they were experiencing symptoms of a mental health disorder but had not been diagnosed, suggesting unmet need. Using validated screening tools, we found remarkably high levels of probable PTSD, anxiety, and depression. The only previous study examining occurrence of mental health conditions in FA focused on a particularly precarious time, the peritransplant period, and primarily sampled children (Kearney et al., 2012). It is noteworthy that our community-based sample was very unlikely to be in peritransplant period or hospitalized at the time of study, yet mental health disorders were similarly elevated.

Several studies focusing on individuals with RDs have identified elevated levels of anxiety and depression (K. R. Bogart & Irvin, 2017; K. R. Bogart et al., 2022; van der Kloot et al., 2010; Wiener et al., 2012; Yang et al., 2018). Living with FA or other RDs can involve significant stressors such as a lengthy diagnostic process, misdiagnoses, acute health crises, extended periods of watchful waiting, uncertainty, lack of control, and fear of disease progression. Although PTSD has seldom been addressed in prior literature – with a few notable exceptions (van der Kloot et al., 2010; Wiener et al., 2012; Yang et al., 2018) – the alarmingly frequent occurrence in our sample suggests that this is a critical area for both research and intervention. Our findings indicate that providers should screen regularly for anxiety, depression, and PTSD and offer referrals to mental health providers. Echoing Frohnmayer's conclusion in 2010, greater awareness of mental health issues among providers and interventions to enhance wellbeing are still acutely needed.

Emphasizing the importance of a biopsychosocial approach to managing this condition, we found that a combination of FA symptoms, personal factors, and social factors significantly impact mental health outcomes. Stigma was the only consistently significant predictor for all three mental health conditions. Additionally, disability self-efficacy, concern with death and dying, and post-traumatic growth were notable predictors for symptoms of anxiety, depression, and PTSD, respectively.

While biomedical interventions for FA are crucial, psychosocial factors are in urgent need of study and intervention because they are more strongly associated with mental health. Fortunately, psychosocial factors may be malleable, and existing interventions could be modified to focus on specific FA challenges. For instance, reducing stigma could involve modifying structural barriers, supporting policies that ensure non-discrimination towards individuals with disabilities and health conditions, and fostering social connections within and beyond the FA community. Regarding disability self-efficacy, patient education plays a crucial role in building confidence in managing one's condition. Patient support organizations can provide opportunities for education and social support, such as in the form of conferences, which have been shown to reduce stigma and improve self-efficacy in other RD populations (K. R. Bogart & Hemmesch, 2016). Interventions that promote meaning-making and legacy can support concerns about death and dying and post-traumatic growth. In-person or phone-based CBT has shown promise for people with cancer diagnoses undergoing radiation or hematopoietic stem cell transplantation for blood cancers (Doolittle & DuHamel, 2015; Kangas et al., 2013).

Limitations and strengths

As a cross-sectional study, determining directionality and causality of findings is not possible. Our sample cannot be considered representative of the population of people with FA due to the nature of convenience sampling. Consequently, individuals with limited access to diagnosis, medical treatment, or FA organizations would be less likely to participate in our study.

Despite these limitations, our research has several strengths. A participatory approach was crucial to ensure the study reflected the experiences of adults with FA. Relatedly, our deep connections with stakeholders and an advisory board enabled us to recruit a more diverse sample than is typical in RD research, including racial, ethnic, and sexual minorities.

Future directions

Future directions include interviews to deeply explore the perceived barriers and facilitators around mental health in adults with FA. Ultimately, we aim to develop mental health guidelines for adults with FA and their healthcare team. This could include information alerting the healthcare team to common psychosocial issues and appropriate screening tools. Adults with FA could also use this as a resource to bring to their providers as a self-advocacy tool.

Note

1. The majority of the sample was from the US and the norms and comparison means are based on US samples. As a stricter test, we repeated these analyses on the US sample only ($n = 72$). The same pattern of results emerged, yet cognitive function and emotional support were no longer significantly different from norms, perhaps due to lower power. The pattern of results and significance did not change when comparing the US FA sample against comparison means of people with common chronic conditions in the US.

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Data availability statement

Data would be provided upon request.

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