Fanconi Cancer Foundation
Mental Health & Wellbeing Considerations for Individuals with Fanconi Anemia & Family Caregivers

---

Prepared for FCF by

Megan E. Voss, DNP, APRN &

Kathleen Bogart, PhD

31st July, 2024

Executive Summary

In 2022, Fanconi Cancer Foundation (FCF) funded a two-part study to examine the experience of individuals living with Fanconi Anemia (FA) and the prevalence of mental health symptoms in the community. The results of the ***Psychosocial Experiences of Adults with Fanconi Anemia: A Participatory Mixed-Methods Research Study*** are currently under peer review for publication in a manuscript titled *Mental Health in the First Generation of Adults with Fanconi Anemia.* Drs. Kathleen Bogart and Megan E. Voss, together with research team members Madeleine Limon and Sophia Fischer, and lived-experience advisory board members Jasmine Bennetsen, Sean Breininger, Anna Chou, and Dexter Sherrell have drafted the following considerations for the larger community of individuals with FA, their family caregivers, clinicians and scientists. These findings should be considered tentative until the study is published, when full results of the study will be shared.

Findings

- Adults with FA had significantly poorer health-related quality of life outcomes than the general population.
- Members of the FA community likely have high rates of Post-Traumatic Stress Disorder (PTSD), anxiety, and depression and additional screening and diagnostic assessment performed by licensed mental health professionals is warranted.
- Stigma was the most consistent risk factor for mental health symptoms, predicting anxiety, depression, and PTSD. Concern with death and dying and fatigue predicted both anxiety and depression. Therefore, addressing stigma, concern with death and dying, and fatigue should be prioritized in the FA community.
- The physical symptom fatigue was predictive of both anxiety and depression and low post-traumatic growth was predictive of PTSD.
- FA significantly impacts all aspects of an individual's life, as well as that of their caregivers.
- Significant systemic barriers exist that worsen risk factors and make seeking help challenging.

Recommendations

Screening
- Organizations that support FA families could support their mental health by developing a web-based screener that includes the tools discussed in this document to screen for mental health risk and protective factors. An individual report could be generated and shared with health and mental health professionals as a catalyst for referrals and resources in individuals' local communities.

Education
- FCF already plays a critical role in educating individuals facing FA and their family caregivers. Individuals can utilize these resources to enhance their quality of life. FCF and other organizations that support FA families may consider creating additional education materials based on the emerging evidence, discussed in this document, that suggests where resources are most needed. There may also be a need to increase awareness of resources and encourage community attendance and utilization of these resources.
- Organizations that support FA families could extend educational efforts to mental health professionals who are lacking the skills necessary to adequately care for the FA community.

Communication Tools
- Organizations that support FA families could create communication tools for individuals and caregivers to reduce the burden of seeking support in the form of a mental health storyboard and sample accommodation letters.

Support Ongoing Research
- Additional research is needed to determine the risk factors that affect younger members of the FA community as well as caregivers of all people with FA.
- Assessing the interventions recommended in this document through additional research can guide future recommendations.

Limitations

It is critical to recognize that this document is a preliminary outline of the needs of the community and possible solutions based on existing evidence. While much of this document is based on the existing quantitative evidence, the qualitative study outlines how challenging implementing evidence-based interventions can be in certain communities. Gaining access to competent mental health resources poses significant challenges for many members of the FA community and their caregivers.

Individuals also described how engaging in the FCF community was both helpful and distressing at times. This phenomenon needs to be better understood as many of these recommendations rely on FCF community engagement. The best way to navigate these conflicting realities is by continuing to engage with the community to determine the best way to support them.

Conclusion

The first study examining the prevalence of mental health symptoms, risks, and protective factors in individuals with FA revealed a significant need in the community. Individuals with FA place a high importance on quality of life, mental health, and wellbeing. While acknowledging that many gaps exist in understanding the experience of living with FA and access to providers who understand that complex experience, this document can serve as a starting point to providing much needed support. There is an urgent need to continue engaging experts in this area and the community to create innovative solutions to enhance the mental health and wellbeing of this entire community. **Members of the FA community have been clear that their quality of life is significantly jeopardized and without efforts to improve it, extending the quantity of their life may only result in more suffering.**

Detailed Report

Introduction

The results of the *Psychosocial Experiences of Adults with Fanconi Anemia: A Participatory Mixed-Methods Research Study* suggested that adults with FA had significantly poorer health-related quality of life outcomes than the general population. Specifically, individuals with FA in the study sample experienced higher rates of anxiety, depression, fatigue, sleep disturbance, and pain; they also experienced poorer cognitive function, physical function and social participation, when compared to the general population (Bogart, Voss, Limon, 2024).

When validated screening tools were administered to the study participants, results showed that 50% of individuals with FA in the sample screened positive for symptoms indicative of PTSD, 33% screened positive for anxiety, and 25% screened positive for depression (Bogart, Voss, Limon, 2024). While these estimates are not representative of rates of diagnoses, they indicate that members of the FA community likely have high rates of PTSD, anxiety, and depression and that additional screening and diagnostic assessment performed by licensed mental health professionals is warranted.

In the study sample, a known mental health disorder (at some point in their lifetime) was reported by 44% of participants; of those, half reported active mental health symptoms and 15% believed they were experiencing symptoms of a mental health disorder but had not been diagnosed by a mental health professional (Bogart, Voss, Limon, 2024).

In a second study, in-depth interviews with adults with FA were conducted. Findings from this study are being prepared for submission to a scientific journal. The study revealed five themes:

1. **FA is all-encompassing.** This means FA affects all areas of life and future planning.
2. **Mental health care barriers exist.** Barriers to accessing quality mental health care and competent providers are prevalent. Adults with FA felt that many mental health providers lacked knowledge about FA or chronic illness. There was also a desire for providers who share or understand other intersecting identities.

3. **Stigma and trauma take a toll on mental health.** Discrimination, body image issues, gender norm violations, isolation, and healthcare trauma can lead to mental health symptoms.
4. **Individuals with FA undergo a process of accepting the disease.** This involves factors like keeping occupied, developing optimism, being perseverant, appreciating the present moment, and valuing one's uniqueness.
5. **Connection is bittersweet.** Connecting with the FA community (FAmily) has many benefits but it can also involve grief. Individuals with intersecting minority identities didn't always feel welcomed or included in FA spaces.

Individuals with FA live all over the world and most often are not geographically close to an FA Center, therefore recommendations in this document rely on the partnership of primary care providers, oncology teams, and mental health professionals in local communities. Individuals with FA and their family caregivers can advocate for themselves by providing their local providers with this information. This information can also be made available through FCF's website and publicized to increase awareness.

Family Caregivers

The *Psychosocial Experiences of Adults with Fanconi Anemia* study unfortunately did not have the opportunity to capture the experiences of caregivers. While it is critical that subsequent research is done to gain a clearer understanding of the caregiver experience, the community can begin to act now to respond to the needs of caregivers based on anecdotal evidence as well as extrapolated experiences from general caregiver and cancer caregiver literature.

One study that examined the link between cancer survivor health and mental health and spousal caregiver mental health concluded that mental health outcomes of individuals and their caregivers are connected. Caregivers of individuals with higher amounts of physical health care needs were less likely to receive mental health support from a licensed mental health professional. Conversely, if the individual they were caring for was receiving mental health care, the caregiver was three times more likely to also be receiving mental health care (Litzelman, et al, 2021). Family-centered care that includes mental health care for both individuals and their family caregivers is important. Offering more mental health care to people living with rare diseases and cancer may help their family caregivers access mental health care. Finally, the study acknowledged that family caregivers are met with more barriers to accessing and participating in mental health

care due to limited resources of time, money, and energy (Litzelman, et al, 2021). A recent Cleveland Clinic survey found that 35% of caregivers report struggling with depression or anxiety, while 60% of caregivers reported feeling anxious at least one time per week (Cleveland Clinic, 2022). These figures are significantly higher than individuals who are not caregivers.

While there are no studies to date that have explored the incidence of mental health challenges in the FA caregiver community, the anecdotal evidence in the community suggests that caregivers are in need of mental health support just like the individuals they are caring for. Whenever possible, caregiver recommendations are included in this document alongside recommendations for individuals living with FA. However, as evidence in this area evolves, more specific and detailed recommendations should be created for caregivers.

Goals

The purpose of these recommendations is to standardize mental health screening, prevention, and treatment by making best practice recommendations for individuals living with FA, and when possible for their caregivers as well. Ultimately the vision is to improve quality of life and wellbeing for all in the FA community.

1) **Part 1: Screening Guidelines**
   The guidelines have been informed by the study findings. They will outline which individuals may be at highest risk due to their disease characteristics, treatment side effects, and individual disease trajectory. They will also outline protective factors to consider or encourage.

2) **Part 2: Evidence-Based Interventions**
   Many of the risk factors assessed in the study are modifiable with a variety of relatively easy to employ interventions. Part two of this document will cover high level strategies that may be employed by individuals, the FCF community, and local organizations and communities. Interventions are organized into three tiers based on the level of autonomy and intensity of the interventions.

Part 1: Screening Guidelines

Introduction

The following screening tools, most of which were used in the study, can provide a whole-person picture of risk and inform the need for preventative and treatment interventions in individuals in the FA community. The tools are divided into those that screen for mental health diagnoses and health-related quality of life factors, and tools that screen for risk and protective factors. In subsequent sections of this document, the relationship between mental health diagnoses and risk/protective factors will be described. Though little detail is known about caregiver mental health in the FA community, due to the low burden and high therapeutic potential, it is reasonable to also screen caregivers using the same tools as they apply to each individual caregiver.

Screening Protocol: Mental Health & Quality of Life

- ➔ Mental Health Diagnoses
    - ◆ Depression: PHQ-9
    - ◆ Anxiety: GAD-7
    - ◆ PTSD: PTSD Checklist for DSM-5
- ➔ Health-Related Quality of Life
    - ◆ Patient-Reported Outcomes Measurement Information System (PROMIS-29+2)
        - ● physical function
        - ● anxiety
        - ● depression
        - ● fatigue
        - ● sleep disturbance
        - ● ability to participate in social roles and activities
        - ● pain interference
        - ● cognitive function

Screening Protocol: Risk & Protective Factors
- ➔ Social Support
    - ◆ The 4-item PROMIS Companionship and Emotional support scales
- ➔ Post-Traumatic Growth

- ◆ The 4-item PROMIS Psychosocial Illness Impact-Positive form
  - ● Assesses positive psychosocial outcomes of illness
  - ● Acceptance
  - ● Benefit-finding
  - ● Meaning-making
  - ● Appreciation for relationships
- ➔ Stigma
  - ◆ Neuro-QOL 8-item stigma scale
- ➔ Disability Self-Efficacy
  - ◆ 6-item University of Washington Disability Self-Efficacy Scale
- ➔ Spirituality
  - ◆ Spirituality scale of the World Health Organization Quality of Life Instrument
- ➔ Concern with Death and Dying
  - ◆ Neuro-QOL 6-item Concern with Death and Dying

Predictive Factors

Once an individual is screened utilizing these tools, predictive factors can help health and mental health providers determine an individual's unique risk of developing a mental health diagnosis of anxiety, depression, or PTSD. The following lists provide predictive factors for each mental health diagnosis:

Predictors of Anxiety
- ➔ Fatigue
- ➔ Concern with death and dying
- ➔ Low self-efficacy
- ➔ Stigma

Predictors of Depression
- ➔ Fatigue
- ➔ Concern with death and dying
- ➔ Stigma

Predictors of PTSD
- ➔ Low post-traumatic growth
- ➔ Stigma

Screening Protocol: Timing & Frequency
Because this study revealed relatively high rates of PTSD, anxiety, and depression in individuals with FA who were unlikely to be in the immediate peri-transplant period, currently hospitalized, or undergoing other significant health stressor or crises, the recommendation is for all individuals with FA to be screened annually at minimum. The risk of screening is low. Access to screening tools can be provided to the community at no cost and with minimal time commitment. The potential benefits of annual or bi-annual screening are significant. Early detection and intervention can improve the quality of life of individuals living with FA and their family caregivers.

Although this study did not predict periods of increased risk of mental health disturbance in the trajectory of an individual's FA diagnosis, from what is known about FA, it is reasonable to consider the need for heightened awareness and increased screening vigilance during the following points in time. Due to the direct link between individual physical and mental health and caregiver mental health, it would be reasonable to consider caregivers at the same points in time.
- Significant life stressors
- Lengthy diagnostic process (including initial diagnosis & subsequent diagnoses)
- Misdiagnoses
- Acute health crises of individual or caregiver
- Extended periods of cancer or other health surveillance
- Uncertainty
- When an individual or caregiver experience feelings of lack of control
- When there is increased fear of disease progression
- Time period surrounding hematopoietic stem cell transplantation
- Annual or additional cancer screenings
- At the time of a cancer diagnosis
- When an individual is undergoing cancer treatments
- When a member of the FCF community is at the end of their life and in the subsequent bereavement period

Part 2: Summary of Evidence-Based Interventions to Support Mental Health & Wellbeing for Individuals with FA and their Family Caregivers

Introduction

Though there is an urgent need to better understand the incidence and risk factors related to mental health diagnoses in the FA community, there are evidence-based strategies that can be recommended in this phase of early discovery. This first comprehensive study of the community showed that psychosocial factors, managing physical symptoms, and employing existing mental health interventions can modify the risk of individuals with FA developing mental health diagnoses.

Taking a whole person approach to caring for individuals with FA and their caregivers is the most effective way to both modify risk and enhance overall wellbeing. The summary of evidence-based interventions is best situated in the context of a broad definition of wellbeing, such as the one depicted in the model below from the University of Minnesota. Wellbeing includes being happy and healthy in the broadest sense, not just physically, but mentally, emotionally, and spiritually healthy. In fact, a person can be experiencing significant physical health challenges, yet still experience high levels of wellbeing. Wellbeing is a state of being in balance or alignment. It includes feeling content, peaceful, connected to purpose, in harmony and safe (University of Minnesota, 2024).

In this model, there are six domains of wellbeing: health, purpose, relationships, community, security, and environment. Throughout this document the impact these domains have on mental health and wellbeing has been described. The strategies listed below can help individuals and caregivers enhance wellbeing by focusing on the various domains.

The Wellbeing Model

Earl E. Bakken Center for
SPIRITUALITY & HEALING

Mary Jo Kreitzer, PhD, RN, FAAN
© 2012 Regents of the University of Minnesota

Strategies

Strategies for improving mental health and overall wellbeing have been organized into three levels based on the level of autonomy and intensity of the interventions. Level one strategies are things individuals can do on their own. Level two strategies are things that can be done with the support of individuals' local community resources. Finally, level three strategies engage the healthcare system.

Level One Strategies: Things I can do on my own

The first level of supportive strategies are things that individuals can pursue on their own, with their family caregivers, and in their community. These strategies occur outside of the health care system and focus on improving the health and wellbeing of the whole person. Rather than focusing on mental health diagnoses that may come and go throughout a person's lifetime, these strategies focus on building lifelong resilience.

Wellbeing Strategies

Wellbeing can be enhanced by practicing mind/body skills that enhance resilience. Mind/body skills have been shown to improve sleep, help people manage stress, pain, and uncertainty, and even prepare for planned medical procedures. Below is a non-exhaustive list of wellbeing strategies that may be appropriate for individuals in the FA community.

- Mindfulness
- Guided Imagery
- Movement
- Spending Time in Nature
- Creative Arts Therapies (music, photography, culinary arts, etc)

Social Support

Community & relationships play an important role in enhancing wellbeing. As previously discussed, communities that can support intersectional identities can increase self-efficacy and reduce stigma. Community support and healthy relationships also create safe spaces to explore difficult thoughts and emotions, such as those brought up by disability and death.

Education

Education empowers people. Navigating a complex illness, such as FA, whether you yourself have the diagnosis or you are caring for someone who does, can be like navigating a foreign country when you don't speak the language. FCF and other organizations that support FA families already play a critical role in educating and empowering individuals facing FA and their family caregivers. Individuals can utilize these resources to enhance their quality of life. Organizations that support FA families may consider creating additional education materials based on the emerging evidence that suggests where resources are needed the most.

Level Two Strategies: Things I can do with the support of my local community

Level two strategies engage the help of community resources and licensed mental health professionals. Working with a local licensed mental health professional provides the opportunity to receive a full mental health diagnostic assessment. Sometimes mental health professionals focus on treating symptoms. An individual can experience mental health symptoms without meeting diagnostic criteria for any one diagnosis. For example, many individuals who have undergone extensive medical treatment have symptoms related to medical trauma, but they may or may not meet diagnostic criteria

for PTSD. Having a diagnosis doesn't always change the course of treatment, but it is a good idea for individuals who have been struggling with mental health symptoms for more than a few consecutive weeks to seek professional support. Individuals can be treated by mental health professionals whether or not they have a mental health diagnosis.

Psychotherapy

Psychotherapy is a broad category that describes many types of therapy that can support individuals or groups of individuals in processing their lived experiences. In this document, cognitive behavioral therapy is most often referred to due to its robust evidence-base. Other types of therapy may also be supportive. Choosing a licensed mental health therapist with whom an individual, couple, family, or group feels comfortable and safe is of the utmost importance. Licensed mental health therapists can diagnose and treat mental health concerns. There are general mental health therapists who may be helpful for processing a variety of mental and emotional experiences. The following list highlights some specialized roles and therapies that may be appropriate for individuals in the FA community.

Health psychologists have specialized training in working with individuals with significant health diagnoses. Some health psychologists may also have experience working with family caregivers as well. For individuals who are struggling with managing physical symptoms, such as sleep disturbances, chronic pain, or physical disability, a health psychologist may be especially helpful.

Trauma-focused therapies such as cognitive-based trauma therapies and eye movement desensitization and reprocessing (EMDR) can be effective at reducing the symptoms associated with trauma and PTSD.

CBT-i is a type of cognitive therapy that is designed to help treat insomnia. Evidence shows that it should be the first line treatment for insomnia and is more effective than medications at enhancing sleep.

Acceptance & Commitment Therapy (ACT) focus on helping individuals move forward in their lives by accepting that deep emotional responses to life stressors are appropriate and warranted. In ACT, the individual is encouraged to feel their emotions rather than denying or avoiding them.

This level of acceptance helps them to move forward with their lives despite the challenges they are facing.

Grief Therapy focuses on processing loss. Grief and loss can be defined broadly. The grieving process often begins long before a person dies. Both individuals with FA and their caregivers experience a great deal of grief and loss. In addition to thinking about losing a loved one in the future, individuals experience the loss of other community members, and loss of the life they planned for, including physical or cognitive abilities, fertility, career and other dreams. Grief therapy can walk an individual, family, or community through the complicated steps of the grieving process.

Integrative Therapies

Integrative therapies are a broad set of non-medical therapies that can help manage symptoms and support overall wellbeing. Many integrative therapies have a growing evidence base for helping people manage things like insomnia, pain, and anxiety. Some integrative therapies can be practiced at home, making them accessible. Those were discussed above in the 'Wellbeing Strategies' section. Other integrative therapies require a highly trained and/or licensed professional. While they can be effective, they are not always easily accessible or covered by health insurance. Talk to your health care team before engaging in a new integrative therapy. You can read more about connecting with safe and reputable integrative therapy providers in your community on the [Taking Charge of Your Survivorship](#) website.

Acupressure or acupuncture may be useful in managing pain, fatigue, anxiety, insomnia, and depression. There are some risks involved, so make sure to talk with your healthcare team before initiating acupuncture. Only a licensed acupuncturist or other licensed health care provider with additional training in acupuncture (such as a licensed chiropractor, naturopathic physician, nurse/nurse practitioner, or DO/MD) can provide acupuncture. You can read about [finding a reputable practitioner](#) and [safety considerations](#) on the Taking Charge of Your Survivorship website. You can also explore [acupressure for self-care](#) you can use yourself at home.

Chiropractic care may be useful in managing certain types of pain. There are some risks involved, so make sure to talk with your healthcare team before initiating chiropractic care. Only a licensed chiropractor can provide chiropractic care. You can read about the risks and benefits of chiropractic

care on the [NIH website.](#)

Massage Therapy may be useful in managing pain, fatigue, anxiety, and insomnia. There are some risks involved, so make sure to talk with your healthcare team before initiating massage therapy. Only a licensed (or in a few states, certified) massage therapist can provide massage therapy. You can read more about [finding a reputable practitioner](#) and [safety considerations](#) on the Taking Charge of Your Survivorship website.

Music Therapy may be useful in managing pain, fatigue, anxiety, and insomnia. Music therapy involves very little risk. Music therapists are highly trained and board-certified professionals who focus on therapeutic goals. You can [learn about music therapy](#) and get started with a [music therapist](#) on the Taking Charge of Your Survivorship website.

Yoga Therapy can be done working with a certified yoga therapist. [Yoga](#) can also be practiced at home. You can learn more about the risks and benefits of yoga on the [NIH website.](#)

Level Three Strategies: Things I can do with the support of my healthcare team

Finally, individuals in the FA community may need additional assessment, diagnostics, and pharmacological interventions (medications) to help them manage some of their mental health symptoms or physical health symptoms that are negatively impacting their mental health. Level three strategies require enlisting the support of licensed health care providers to diagnose and treat mental health and related symptoms.

Psychiatric (Medication) Management of Mental Health
Psychiatry is the health care specialty that focuses on using both medication and supportive psychotherapy to improve mental health. Psychiatric providers can assess, diagnose and treat mental health conditions. Psychiatric Mental Health Nurse Practitioners (PMHNP), Physician's Associates (PA), and Psychiatrists (MD) can all provide psychiatric services. Individuals can often receive an expedited referral for psychiatric care by talking to a primary care provider (PCP) or a provider on their oncology team. Sometimes PCPs and oncology team providers may also prescribe medications to help with mental health. If an individual has been receiving mental health medications from their PCP or oncology team and their symptoms are not well managed, they should request a referral to a provider who specializes in psychiatry.

Pain Management
Pain management is another health care specialty that may be useful to individuals with FA. Sometimes these health care providers are part of a palliative

care team. Pain management and palliative care providers specialize in improving an individual's quality of life through symptom management. Palliative care is not the same as hospice and is not reserved for end-of-life-care. Many individuals work with palliative care teams to improve their quality of life for many years while simultaneously pursuing curative treatment when possible.

Physical Function
Physical function can be addressed by many members of the health care team. PCPs or oncology team providers may be a good place to start. Sometimes improving physical function can be achieved by working with a physical (PT) or occupational (OT) therapist. More information on that can be found on [Taking Charge of Your Survivorship](#). There is also a medical specialty called Physical Medicine & Rehabilitation (PM&R). A referral to a provider who specializes in PM&R may be useful if physical function does not improve with PT or OT.

Sleep
Disturbed sleep, also known as insomnia, can be addressed by a number of health care providers if self-care strategies are not improving sleep satisfactorily. PCPs, oncology team members, psychiatry providers, and palliative care providers can all address insomnia. Additionally, there is a medical specialty called Sleep Medicine that may be appropriate for individuals with FA if other strategies are not working. Nearly any member of the healthcare team can provide a referral to sleep medicine.

Aligning Evidence-Based Interventions to Individual Needs & Risk Factors

In this next section, risk and protective factors will be broken down and outlined to help the individual, the FA community, and the larger structures individuals with FA live, work, and go to school in to better understand how to improve the quality of life and mental health & wellbeing outlook for a person living with FA.

Stigma predicted anxiety, depression, and PTSD, while concern with death & dying and fatigue predicted anxiety and depression. Therefore, addressing these topics should be prioritized in the FA community.

Table of Risk and Protective Factors

Risk/Protective Factors	Anxiety	Depression	PTSD
Stigma	X	X	X
Concern with Death & Dying	X	X	
Fatigue	X	X	
Disability Self-Efficacy	X		
Low Post-Traumatic Growth			X

Understanding Each Protective/Risk Factor

The following definitions describe each protective/risk factor from the study as well as other concerns reported by individuals with FA that appeared in the sample, but were less strongly correlated with mental health. There is a brief discussion on how it applies to individuals with FA, and in many cases, their caregivers as well. Each factor has a table that outlines high level strategies for mitigating risk and enhancing protective factors. In the tables below, strategies have been broken down into, 'What can I do?' which is a list of individual strategies for people with FA; 'How can FCF and other organizations that support FA families support me?' which is a list of strategies that could help individuals in the FA community minimize their risk factors; and finally, 'What can the community I live in do to support me?' which lists strategies and resources at the local community level.

Stigma

The study suggested that reducing stigma could impact an individual's risk of anxiety, depression, and PTSD. Impacting stigma involves individual understanding, community support, and structural changes. There are three types of stigma that impact individuals: public stigma, which involves how the public perceives and treats individuals with FA; self-stigma, refers to negative perceptions, thoughts, and beliefs an individual internalizes about themselves based on the way others perceive FA; and structural stigma, which extends to things like public policy and even discrimination (APA, 2024). Individuals with other minority identities in addition to FA often experience heightened stigma.

Reducing Stigma	
What can I do?	● Utilize cognitive behavioral therapies to process negative beliefs, feelings of shame, or other challenging mental & emotional states that have been internalized. ● Spend time in communities, such as the FCF community, that normalize and validate experiences had by a person with FA and their intersecting identities.
How can FCF and other organizations that support FA families support me?	● Raise public awareness on FA and the impact of caregiving. ● Continue to build community spaces and increase multiculturalism and intersectional inclusion. ● Support public policies that ensure non-discrimination practices for individuals with FA and family caregivers. ● Provide individuals with FA and their family caregivers with sample accommodation letters that can be used in school and employment settings.
What can the community I live in do to support me?	● Make accommodations for work & school that align with and go above and beyond non-discrimination policies, including the Americans with Disabilities Act, to protect an individual with FA or a caregiver.

Sleep Disturbances

The study suggested that sleep disturbances were prevalent in the FA community, even though they weren't statistically correlated with the mental health diagnoses that were screened for. Still, improving sleep could have significant positive impacts on mental health and quality of life for individuals with FA and their family caregivers as well.

Enhancing Sleep	
What can I do?	● Utilize cognitive behavioral therapies to process emotions and manage stress. Utilize CBT-i when appropriate. ● Follow sleep hygiene guidelines which can be

	found on the [Taking Charge of Your Survivorship](#) website.
	● Advocate for treatment of insomnia from your medical team when self-care strategies are not working. Your medical team may consider both non-pharmacological and pharmacological interventions.
How can FCF and other organizations that support FA families support me?	● Provide education on enhancing sleep through online webinars & at the FA Family and Adult Retreats.
What can the community I live in do to support me?	● Accommodate flexible work hours & time off. ● Understand when I need to set boundaries or cancel obligations last minute due to physical, mental, or emotional exhaustion.

Concern With Death & Dying

In the study, concern with death and dying was correlated with both depression and anxiety. It was measured by asking questions about how often a person thinks about death, worrying about their family coping with their death, anxiety related to dying, and thoughts about ending their own life (Neuro-QOL 6-item Concern with Death and Dying scale National Institute of Neurological Disorders and Stroke, 2015). It is important to normalize thoughts about death and dying for people with FA and to support them in making sense of and processing these thoughts. The goal is not to suppress, avoid, or shame a person for experiencing concern with death and dying. Instead, families and communities should create a safe space that invites exploration of these thoughts and feelings so that individuals are not left to face them alone. Although caregivers were not assessed in this study, it is reasonable to assume that they also need space and support to explore their own concern with death and dying.

Managing Concern with Death & Dying	
What can I do?	● Utilize grief therapy to process emotions and manage the stress of uncertainty ● Explore avenues of meaning-making. ● Engage in legacy work, projects, or art.

	● Explore resources on Taking Charge of Your Survivorship: 　○ Grief 　○ Meaning & Purpose
How can FCF and other organizations that support FA families support me?	● Provide education on grief & anticipatory grief. ● Continue to host grief-focused support groups for individuals and caregivers both online and at the FA Family and Adult Retreats. ● Ensure that FCF support groups and resources also include a focus on caregiver needs.
What can the community I live do to support me?	● Provide local grief support. ● Provide local resources that support spirituality and/or religion.

Fatigue

The study suggested that fatigue was prevalent in the FA community, even though it wasn't statistically correlated with the mental health diagnoses that were screened for. Fatigue, or feeling excessively tired, is a common physical symptom experienced by people with chronic disease and caregivers. Some causes of fatigue may be reduced in partnership with a health care provider. At other times, lifestyle modifications may be the best strategy for managing fatigue. Always talk with your healthcare team if you experience new or worsened fatigue. Fatigue can both be a cause of mental health diagnoses as well as a symptom of them.

Managing Fatigue	
What can I do?	● Focus on the physical and psychological building blocks for increasing energy: 　○ Sleep 　○ Nutrition 　○ Movement 　○ Managing Stress 　○ Processing Emotions
How can FCF and other organizations that support FA families support me?	● Provide education on managing fatigue of individuals and caregivers both online and at the FA Family and Adult Retreats.

What can the community I live in do to support me?	● Accommodate flexible work hours & time off. ● Understand when I need to set boundaries or cancel obligations last minute due to physical, mental, or emotional fatigue.

Disability Self-Efficacy

The study also showed that increasing disability self-efficacy may reduce an individual's risk of developing an anxiety diagnosis. Disability self-efficacy describes an individual's confidence in their ability to accomplish the things they need and want in life, including managing issues related to FA. It is impacted by the person's sense of self, their behaviors, their access to information, and the environment in which they live and work (Amtmann et al, 2012). In other words, when individuals with FA live and work in supportive environments, have access to appropriate health information, and are surrounded by communities that positively impact their belief in their own ability to reach their full potential, they are more likely to do so. It is likely that caregiver self-efficacy is equally important in the FA community. Caregivers need to feel like they can manage the challenges of caregiving effectively, surrounded by other individuals and communities that can support them.

Increasing Self-Efficacy	
What can I do?	● Utilize cognitive behavioral therapies to process negative self-beliefs and build confidence & self-esteem. ● Spend time in communities, such as the FCF community, that support confidence, self-esteem, and provide information on FA management. ● Spend time in caregiver communities, such as The Negative Space community or FCF sponsored caregiver events, that support confidence & self-esteem of caregivers.
How can FCF and other organizations that support FA families support me?	● Provide education to build individual's confidence in managing FA. ● Provide education to build family caregiver's confidence in managing FA and their caregiving role. ● Provide individuals with FA and their family caregivers with sample accommodation letters that

	can be used in school and employment settings.
What can the community I live in do to support me?	● Make accommodations that increase an individual's likelihood of achieving their goals.

Low Physical Function

Low physical function was reported by individuals with FA even though it wasn't statistically correlated with the mental health diagnoses that were screened for. Reduced physical function may show up in a variety of ways in an individual's life. The interventions to manage challenges with physical function will vary greatly depending on the type of physical function that's been impaired and the severity of impairment.

Enhancing Physical Function	
What can I do?	● Explore resources for physical activity on Taking Charge of Your Survivorship. ● Advocate for further evaluation & treatment from your medical team when self-care strategies are not working. ● Focus on nourishing your body. Explore resources on food & nutrition on Taking Charge of Your Survivorship.
How can FCF and other organizations that support FA families support me?	● Provide education on enhancing physical function and safe physical activity both online and at the FA Family and Adult Retreats. ● Provide caregivers with education and access to screenings related to the physical health risks associated with caregiving.
What can the community I live in do to support me?	● Provide accessible areas for moving your body, such as outdoor spaces & workout facilities with adaptive equipment.

Pain

The study suggested that pain was prevalent in the FA community, even though it wasn't statistically correlated with the mental health diagnoses that were screened for. Pain is a common experience of people with chronic illness. It can be acute (short-term) or chronic (long-term). It can be caused by a variety of underlying conditions, and sometimes the cause is not known. Pain impacts the physical body, the mind, and the spirit. A whole person approach to treating pain, especially chronic pain, is the best way to enhance quality of life.

Managing Pain	
What can I do?	- Explore resources for understanding pain on the Taking Charge of Your Survivorship website. - Explore mind-body practices that have proven beneficial for chronic pain including: - Acupressure - Clinical hypnosis - Meditation - Mindfulness - Tai Chi, QiGong, Yoga - Engage in cognitive behavioral therapy to help you manage the pain experience. - Advocate for further evaluation & treatment from your medical team when self-care strategies are not working.
How can FCF and other organizations that support FA families support me?	Provide education on living with and managing pain both online and at the FA Family and Adult Retreats.
What can the community I live in do to support me?	- Accommodate flexible work hours & time off. - Understand when I need to set boundaries or cancel obligations last minute due to physical, mental, or emotional pain.

Cancer Diagnosis

The FA community knows just how significant the impact of a cancer diagnosis is for someone living with FA and their family members. More research is needed to understand the impact of undergoing regular cancer screenings, diagnoses, and treatments on the mental health of individuals and their caregivers.

Coping with a Cancer Diagnosis	
What can I do?	● Utilize cognitive behavioral therapies to process new life stressors such as a cancer diagnosis. ● Incorporate mind/body strategies to manage the stress of a new diagnosis. ● Advocate for comprehensive care from the healthcare team that manages all physical symptoms, thereby improving mental and emotional wellbeing.
How can FCF and other organizations that support FA families support me?	● Provide education on managing the impact of a cancer diagnosis. ● Host support groups for individuals and caregivers managing a cancer diagnosis both online and at the FA Family and Adult Retreats. ● Ensure that FCF support groups and resources also include a focus on caregiver needs.
What can the community I live in do to support me?	● Accommodate flexible work hours & time off. ● Understand when I need to set boundaries or cancel obligations last minute due to physical, mental, or emotional pain or other symptoms.

Low Post-Traumatic Growth

Low post-traumatic growth was correlated with PTSD in the study. Post-Traumatic Growth can be described as the positive psychosocial outcomes of illness including acceptance of the illness, benefit-finding, meaning-making, and appreciation for relationships. Individuals who experienced low post-traumatic growth were more likely to have experienced PTSD. This is likely true for caregivers as well.

Increasing Post-Traumatic Growth	
What can I do?	- Utilize Acceptance & Commitment Therapy to process life stressors and encourage growth. - Engage in meaning-making activities like writing, art, or volunteer work. - Engage in supportive relationships. - Develop a formal gratitude practice. - Explore related resources on [Taking Charge of Your Survivorship.](#)
How can FCF and other organizations that support FA families support me?	- Provide education on post-traumatic growth. - Host support groups for individuals and caregivers to process their traumatic experiences. - Ensure that FCF support groups and resources also include a focus on caregiver needs.
What can the community I live in do to support me?	Provide a supportive sense of relationships & community.

Recommendations

The following recommendations can be acted on right away as ongoing research to better understand the needs of the community continues. Recommendations have been organized into four categories: screening, education, communication tools, and support for ongoing research.

Screening
FCF and other organizations that support FA families could support the FA community by developing a web-based screener that includes the tools described in this document. Individuals with FA and their family caregivers could screen themselves for mental health risk and protective factors. An individual report could be generated and shared with health and mental health professionals as a catalyst for referrals and resources in individuals' local communities.

Education
FCF already plays a critical role in educating and empowering individuals facing FA and their family caregivers. Individuals can utilize these resources to enhance

their quality of life. FCF and other organizations that support FA families may consider creating additional education materials based on the emerging evidence, discussed in this document, that suggests where resources are most needed. There may also be a need to increase awareness of FCF resources and encourage community attendance and utilization of these resources.

The qualitative findings revealed a significant need for clinician and provider education. FCF and other organizations that support FA families could play a role in training mental health professionals to be better equipped to work with rare diseases such as FA. Developing online educational resources for mental health professionals has the potential to decrease burden on individuals with FA and caregivers.

Communication Tools

Individuals with FA and their caregivers experience significant burdens related to caring for themselves. Whenever possible, the FCF and other organizations that support FA families should seek to create tools to reduce that burden. Two examples of communication tools that could reduce burden are the mental health storyboard and sample accommodation letters.

A mental health storyboard is a summary of physical, mental, and emotional challenges an individual faces. It could provide high level information about mental health in the FA community and direct mental health professionals to additional online resources. A tool like this could streamline the work an individual or caregiver must do to find a mental health professional who is competent and comfortable working with FA.

Sample accommodation letters could empower individuals with FA and caregivers to seek accommodations from educational and employment institutions that reduce burden on the individual and support their physical, mental, and emotional needs. Often, individuals and their healthcare professionals do not have the skills necessary for creating and advocating for effective accommodations. FCF and other organizations that support FA families could develop and provide sample accommodation letters for community members that could be adapted to fit their individual needs and used to advocate for themselves.

Support Ongoing Research

While there are many competing priorities for FA funding, the findings presented here support the need for ongoing research in the areas of mental health, wellbeing, and quality of life. **Members of the FA community have been clear that their quality of life is significantly jeopardized and without efforts to improve it, extending the quantity of their life may only result in more suffering.**

Additional research is needed to determine the risk factors that affect younger members of the FA community as well as caregivers. Assessing the interventions recommended in this document through additional research can guide future recommendations for improving mental health, wellbeing, and quality of life.

Limitations

It is critical to recognize that this document is a preliminary outline of the needs of the community and possible solutions based on existing evidence. The document was compiled relying on a great deal of clinical judgment and expertise to draw upon general evidence in the absence of evidence specific to the FA community. The authors acknowledge that much more research is needed to create clinical practice guidelines for mental health in the FA community.

While much of this document is based on the existing quantitative evidence, the qualitative study outlines how challenging implementing evidence-based interventions can be in certain communities. Individuals shared personal accounts of working with mental health professionals who were ill-equipped to work with someone with FA. In those instances, seeking help was more detrimental to their mental health and wellbeing. This challenge was even more significant for individuals with minority identities. Additionally, access to mental health care is an ongoing challenge in nearly all communities. Individuals also described how engaging in the FCF community was both helpful and distressing at times. This phenomenon needs to be better understood as many of these recommendations rely on FCF community engagement. The best way to navigate these conflicting realities is by continuing to engage with the community to determine the best way to support them.

Conclusion

The lived experiences of individuals with FA, as documented in the study and through the work of the lived experience advisory board, demonstrate that whole person health is of paramount importance. Individuals with FA place a high importance on quality of life, mental health, and wellbeing. While acknowledging that many gaps exist in understanding the experience of living with FA and access to providers who understand that complex experience, this document can serve as a starting point. It is the beginnings of a roadmap to supporting individuals with FA and their caregivers. There is an urgent need to train mental health professionals to better understand FA and the caregiving experience and to develop additional interventions to support the mental health and wellbeing of this community.

References

Amtmann, D., Bamer, A. M., Cook, K. F., Askew, R. L., Noonan, V. K., & Brockway, J. A. (2012). UW-SES: A new self-efficacy scale for people with disabilities. Archives of Physical Medicine and Rehabilitation. 93(10), 1757-1765. doi:10.1016/j.apmr.2012.05.001

American Psychiatric Association, Stigma, prejudice and discrimination against people with mental illness. Psychiatry.org - Stigma, Prejudice and Discrimination Against People with Mental Illness. (2024, March). https://www.psychiatry.org/patients-families/stigma-and-discrimination#:~:text=Researchers%20identify%20different%20types%20of,relative%20to%20other%20health%20care.

Bogart, K. R., Voss, M. E., & Limon, M. (2024). Mental Health in the First Generation of Adults with Fanconi Anemia. [Manuscript submitted for publication.] School of Psychological Science, Oregon State University and School of Nursing, University of Minnesota.

Cleveland Clinic. (2024, April 30). Importance of supporting family caregivers. https://newsroom.clevelandclinic.org/2023/02/15/importance-of-supporting-family-caregivers

Litzelman K, Choi H, Maher ME, Harnish A. Role of cancer survivor health and health service use in spouses' use of mental health-related care. Cancer. 2021 Apr 1;127(7):1146-1153. doi: 10.1002/cncr.33345. Epub 2020 Dec 8. PMID: 33290580.

National Institute of Neurological Disorders and Stroke. (2015). User Manual for the Quality of Life in Neurological Disorders (Neuro-QOL) Measures, Version 2.0. www.neuroqol.org

University of Minnesota, (2024). The wellbeing model. Taking Charge of Your Wellbeing. https://www.takingcharge.csh.umn.edu/wellbeing