



Life Beyond the Diagnosis: Wellbeing & Community in Bloom Syndrome

Bloom Zoom

October 25, 2025

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Bloom Zoom

Agenda (P.T.)

- **Welcome & Logistics (12:00-12:05)**
- **Background (12:05-12:20)**
- **Integrating Mental Health into Research via the IBSR (12:20-12:25)**
- **Building Community & Peer Support (12:25-12:35)**
- **Discussion & Community Input (12:35-12:55)**
- **Closing (12:55-13:00) + Chat**



12:00-12:05

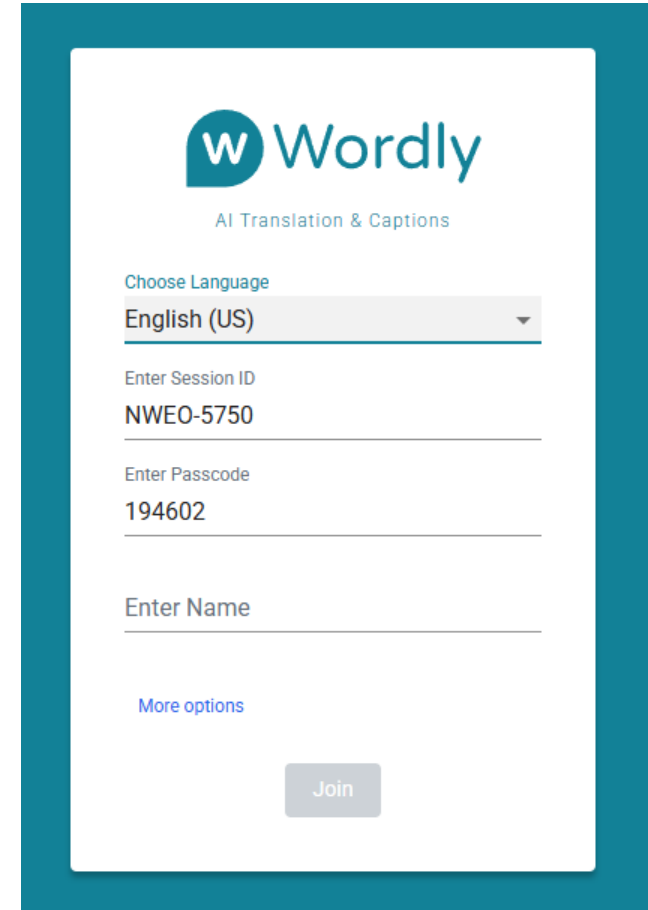
Welcome & Logistics

kalos ilthate hush kelibsiz
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Wordly Translation Tips for Today's Meeting

Wordly translation has been activated for this meeting. If you would like to use translation and have not already opened it, here is the link: <https://join.wordly.ai/join/NWEO-5750?key=194602>

- When you select the link, you will see an invitation that looks like this:
- Select your preferred language, enter your name, and select Join.
- You'll see a Wordly welcome screen – this is your live translation stream. Keep the window/tab open during the meeting so you can glance at the live translation when needed.
- The best browsers to use are Google Chrome or Microsoft Edge.
- There will be no audio translation offered today.
- If you run into issues, please let us know through the meeting chat.

A screenshot of the Wordly AI Translation & Captions join screen. The screen has a white background with a teal border. At the top is the Wordly logo (a teal circle with a white 'w') and the text 'Wordly AI Translation & Captions'. Below this is a 'Choose Language' dropdown menu with 'English (US)' selected. Underneath is a text input field for 'Enter Session ID' containing 'NWEO-5750'. Below that is a text input field for 'Enter Passcode' containing '194602'. At the bottom is a text input field for 'Enter Name'. A 'More options' link is located below the name field. A 'Join' button is at the bottom right.

Welcome! Today's Logistics



Please Mute

In order to avoid cross-talk or distractions for others, please stay muted unless asking a question.



Questions

Raise your Zoom hand or put a note in the Chat.

For any anonymous questions, direct them in the chat to Mary Treder and she will ask them anonymously.



Post-Meeting Dissemination

This meeting will NOT be recorded due to sensitive nature of discussion. Slides and a high level summary will be put on the BSA website.



Tech Issues

Wordly: chat Chris Micka or Susan Zaslaw

Non-Wordly: If you have non-Wordly tech issues, please contact Mary Treder via chat or via FB messenger



Active Participation

We encourage you to ask questions or provide feedback, during this session and after!



12:05-12:20

Background

Goals for Today's Bloom Zoom

- Share how the topic of this Bloom Zoom came about
- Share what we know and what we're learning about mental health in DNA repair disorders, including Bloom syndrome
- Introduce upcoming BSA research and community initiatives
- Invite open discussion and ideas

Taking Care as We Talk about Hard Things

- *Today's conversation will include themes that may feel emotional, such as experiences with illness, uncertainty, and loss, as well as reflections on resilience, connection, and hope.*
- Take care of yourself in any way that feels right:
 - *Step away or turn off your camera for a few minutes if you need to.*
 - *Reach out to a friend, family member, or another member of the Bloom community.*
 - *Remember: you are never alone here.*
- If you need additional support:
 - 988 Suicide & Crisis Lifeline (US): Call or text 988 (24/7, free, confidential)
 - Crisis Text Line: Text HOME to 741741 (US & Canada)
 - International Helplines: findahelpline.com for global numbers
 - E.g. 0800-113 in NL, 116-123 in UK, etc.
- BSA Community Resources:
 - Email info@bloomsyndromeassociation.org if you'd like to connect privately after this session or if you'd like to get involved.

Understanding Impact + Providing Support for Mental Health is a Key BSA Research Council Deliverable

Planning and Initiation

2. Complete RC training on its role and tasks, and be fully functioning.
4. Update the Patient-Centered Research Strategy & Action Plan.

Outreach

5. Provide updates to the community via webinars, blog posts, and more.
6. Design and implement a fundraising plan.
7. Enroll 300 BSyn persons globally in the IBSR through outreach & intake.

Registry

9. Review options and make a recommendation on Registry platform(s).
10. Put the augmented registry in place & complete the initial phase of the New Census of BSyn.
11. Complete Natural History Study data collection, analysis, and reporting.

Cancer

12. Establish a biobank network, sample collection and distribution system.
13. Collect and evaluate data on cancer diagnosis and treatment regimens.
14. Establish a Tumor Board for evaluation and guidance on active BSyn cancer cases.
15. Design and initiate a BSyn cancer surveillance study.

Resources

16. Update the BSyn Patient & Family Handbook.
17. Compile information on BSyn health maintenance.
18. Define scope of psycho-social impact of BSyn and efficient support measures.
19. Establish BSyn Centers of Excellence

Note: Deliverables 1, 3 and 8 are complete.

The Mental Health Gap in Rare Diseases, including Bloom Syndrome

“Mental health is a state of mental well-being that enables people to cope with the stresses of life, realize their abilities, learn and work well, and contribute to their community. It has intrinsic and instrumental value and is a basic human right.”

- World Health Organization

- Mental health and wellness is a vital aspect of quality of life, but is often overlooked in rare diseases.
- Very few studies have been done on mental health and well-being in rare diseases.
- Of studies that have been done, common themes of isolation, anxiety, depression, uncertainty, but also resilience.
- Lack of prioritization of mental health during rare disease diagnosis and care may exacerbate patients' overall health outcomes.

Example: UK Study (2022)

RESEARCH

Open Access

Mental health care for rare disease in the UK
– recommendations from a quantitative survey
and multi-stakeholder workshop



Rosa Spencer-Tansley, Nick Meade, Farhana Ali, Amy Simpson and Amy Hunter*

- Survey of 1,231 rare disease patients and 564 caregivers
- >90% felt worried/anxious
- 36% of patients and 19% of caregivers had suicidal thoughts
- Challenges that negatively impact mental health:
 - Limited knowledge of disease by healthcare providers (88%) and not being taken seriously by them (80%) or being treated as a medical curiosity (50%)
 - Lack of information about the condition (76%), finding worrying information online (56%)
 - Feeling socially isolated (76%)
 - Almost half have *never* been asked about mental health by their healthcare providers, and only 8% are often or always asked

Insights from Fanconi Anemia




<https://news.cci.fsu.edu/cci-news/cci-alumni-news/fisher-family-fanconi-anemia-inspire-character-in-comm-alumnas-novel/>

- About Fanconi Anemia:
 - Causative genes are involved in DNA repair
 - Cancer predisposition (and bone marrow failure) syndrome
 - Individuals have short stature, microcephaly
 - Range of other potential health issues
- 80-90% require a bone marrow transplant before early adulthood
- Cancers are common (up to 76% by age 45) doi: 10.1182/blood-2002-05-1498

Insights from Fanconi Anemia (2025)

Mental health in the first generation of adults with Fanconi anemia

Kathleen R. Bogart ^a, Megan E. Voss^b and Madeleine Limon^a

^aSchool of Psychological Science, Oregon State University, Corvallis, OR, USA; ^bSchool of Nursing, University of Minnesota, Minneapolis, MN, USA

- 102 adults with FA surveyed
- Adults with FA had significantly poorer health-related quality of life on all domains measured, including anxiety symptoms, depression symptoms, fatigue, social participation and others.
- Results showed 50% of participants had probable PTSD, 33% had probable anxiety, and 25% had probable depression.
- “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.”
- Recommendation from paper: “Just as people with FA should receive regular cancer screenings, they should also be screened for mental health symptoms.”

Insights from Fanconi Anemia (2025)

Interviews with 18 adults with FA:



<https://fanconi.org/supporting-mental-health-and-wellbeing-for-individuals-with-fa-and-caregivers-key-insights-and-recommendations/>

- 1. Living with FA is a full-time job:** Managing FA is a constant, all-encompassing task that requires ongoing attention and effort.
- 2. Struggling to find and access mental health care:** Many adults with FA experience difficulties in finding mental health professionals who understand their unique needs, let alone their chronic illness.
- 3. Facing stigma and isolation:** 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. Grief, loss, and community connections:** Connecting with the FA community has many benefits, but it can also involve grief when friends encounter serious medical problems.
- 5. Finding ways to cope and stay optimistic:** Despite the challenges, many individuals with FA use various strategies to cope, such as staying occupied, cultivating optimism, and embracing their uniqueness.

Insights from Fanconi Anemia (2025)

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

Things I can do on my own

- Wellbeing strategies (mindfulness, movement, etc.)
- Social support
- Education about the disease to be empowered

Things I can do with the support of my local community

- Psychotherapy (CBT, ACT, grief therapy, etc.)
- Integrative therapies (non medical therapies that can help support symptoms)

Things I can do with the support of my healthcare team

- Psychiatric management, including medication
- Pain management
- Physical function (e.g. PT or OT)
- Sleep

What Do We Know About Mental Health in Bloom Syndrome? Not a lot...

Qualitative themes from discussions at Patient & Family Conferences, Bloom Zooms:

- *Fear of cancer and loss*
- *Social isolation, especially for adolescents and adults*
- *Caregiver emotional strain and anxiety*
- *Desire for connection and practical advice*

Survey (6 respondents):

- 5/6 say the biggest emotional or mental health challenges they face is *anxiety about health and future*

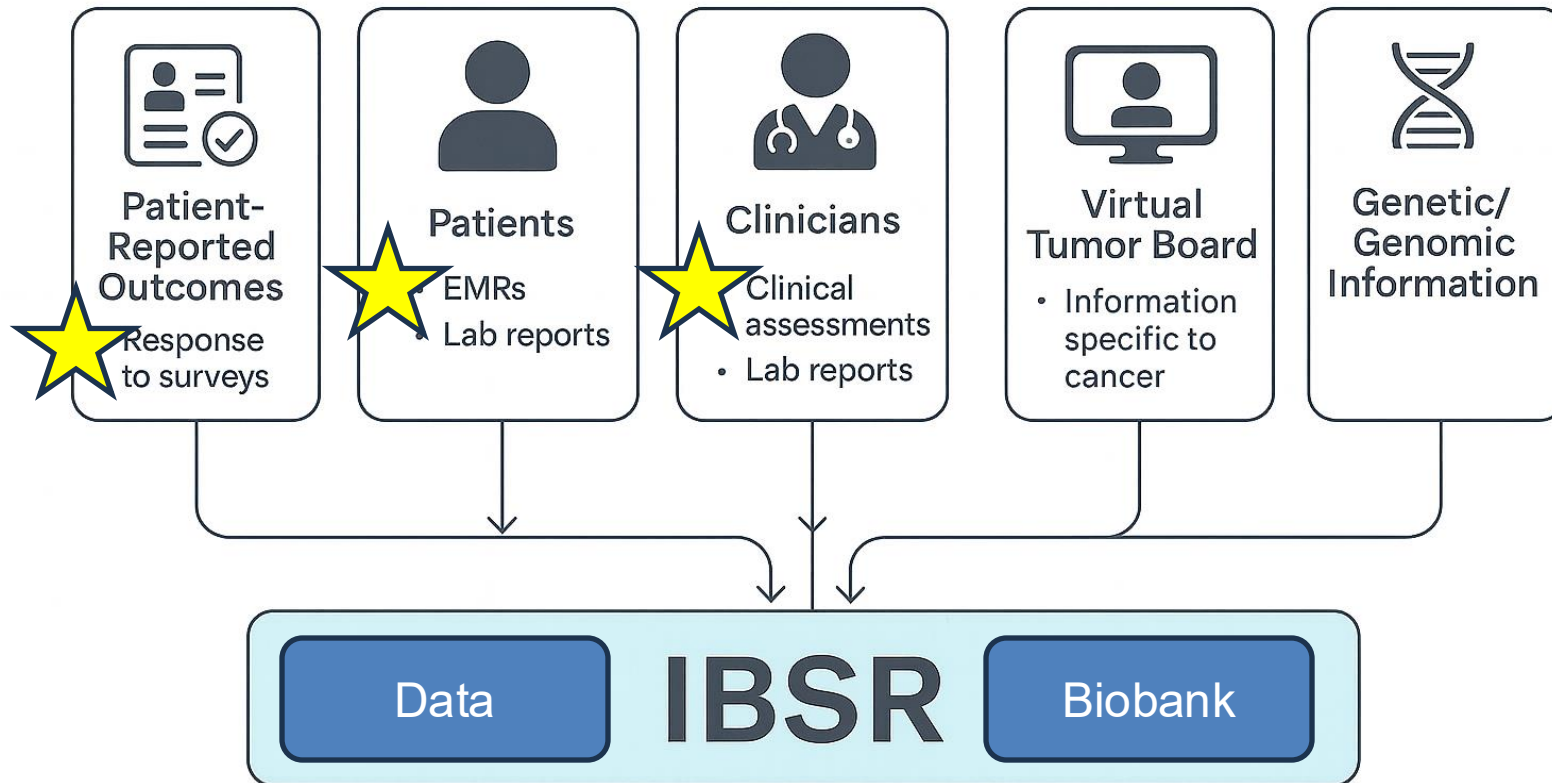
What words come to mind when you think of Bloom syndrome and well-being?



12:20-12:25

Integrating Mental Health into Research

IBSR: Redefining the Face of Bloom Syndrome



- BSA is launching the International Bloom Syndrome Registry (IBSR) before the end of the year
- Build the evidence base for clinical care and quality of life improvements
- Completely private and secure; data de-identified and only available for research purposes
- More information to be given in Dec webinar

Mental Health & Quality of Life Data in IBSR

- IBSR will include validated participant/caregiver surveys that include quality of life
- IBSR will also contain open fields for lived experiences
- Plans for future: targeted surveys on short stature, fatigue, stigma, resilience
- Will be the first time QoL in Bloom syndrome is documented

Global02	In general, would you say your quality of life is:	5 = Excellent
		4 = Very good
		3 = Good
		2 = Fair
		1 = Poor
Global03	In general, how would you rate your physical health?	5 = Excellent
		4 = Very good
		3 = Good
		2 = Fair
		1 = Poor
Global04	In general, how would you rate your mental health, including your mood and your ability to think?	5 = Excellent
		4 = Very good
		3 = Good
		2 = Fair
		1 = Poor
Global05	In general, how would you rate your satisfaction with your social activities and relationships?	5 = Excellent
		4 = Very good
		3 = Good
		2 = Fair
		1 = Poor

Why Does It Matter to Collect Research-Quality Information?

- Data can support holistic care guidelines, including mental health support
- May encourage clinicians to screen for psychosocial needs and provide support as healthcare team
- Enables evidence-based advocacy, funding, and clinical studies



12:25-12:35

Building Community & Peer Support

What Our Community Has Asked For



Ron Squire session at 2024
Beyond Borders conference

- Peer connections by age/life stage (teens, young adults, parents, siblings) and life events (cancer, bereaved)
- Safe online spaces for sharing emotions and advice (tips & tricks)
- Access to professional support: counseling, webinars/workshops, mental-health resources

Some Resources that Exist Today



Bloom Syndrome Community

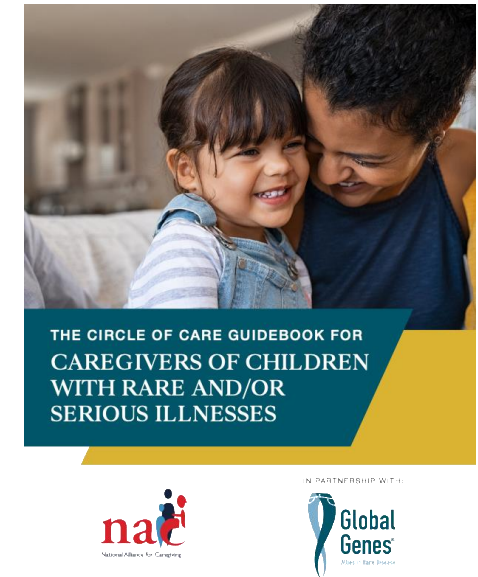
Private group · 342 members



Community Resources

Patient & Family Handbook

Personalized Guidance › Connect with a Peer Navigator volunteer for confidential 1:1 support by phone or email and receive a free, expert-reviewed resource guide tailored to you.	Support Meetings & Partner Events › Review our calendar to connect with others and obtain support and vital resources.	Private Facebook Group › In this group, you can connect with others who are affected by hereditary cancer. Let's unite and support each other.
Message Boards › Our message boards are available 24/7 and are the easiest way to reach out and connect with others who are facing the challenges of hereditary cancer.	Helpline › Peer volunteers return calls within 48 hours and provide nonjudgmental support and resources in English and Spanish. A Genetic Counselor helpline is also available.	Find Healthcare Providers › Find medical experts in cancer genetics, from genetic counselors and oncologists to surgeons, fertility experts, and menopause specialists.



<https://www.bloomsyndromeassociation.org/>

<https://www.facingourrisk.org/support>

<https://globalgenes.org/wp-content/uploads/Rare-Caregivers-Guidebook.pdf>

Planned BSA Community Programs & Initiatives

- **Peer Support Circles** (pilot Q4 2025/Q1 2026):
 - Structured small group discussions led by trained peer facilitators
 - Virtual, global, privacy-respecting model
- **Updated Patient & Family Handbook** (Q4 2025): Section on mental health
- **Health Maintenance Strategies** (Q1 2026): Focus on integrative health
- **Expert-led Webinars** (TBD): coping skills, stress management, navigating uncertainty
- **Blossoming Hope 2026 Conference** (Aug 2026): “Well-Being Track”
 - Building off session and discussions at Beyond Borders conference



12:35-12:55

Discussion & Community Input

Discussion

- Do the mental health challenges described match your experience?
- What helps you or your family feel connected?
- What kind of peer support or mental-health resources would be most helpful?
- How can the IBSR capture what matters most to you?
- How can we make mental-health support accessible globally?
- What would you want included in the “well-being” track at the Blossoming Hope conference?



12:55-13:00

Conclusion + Chat

Thank You!

- Thank you for attending!
- Slides will be placed on the BSA's website and we will have a blog post about this session week of November 2
- Contact info@bloomsyndromeassociation.org with any questions or requests.
- If you are able to, feel free to stay on for a continued chat!

Backup



The BSA's mission is
to connect, educate,
and support the
international Bloom
syndrome
community and
*stimulate research
that leads to longer,
healthier lives.*

S: We want to be Safe

E: We want Effective Treatments

C: We want to Collaborate & Communicate with stakeholders

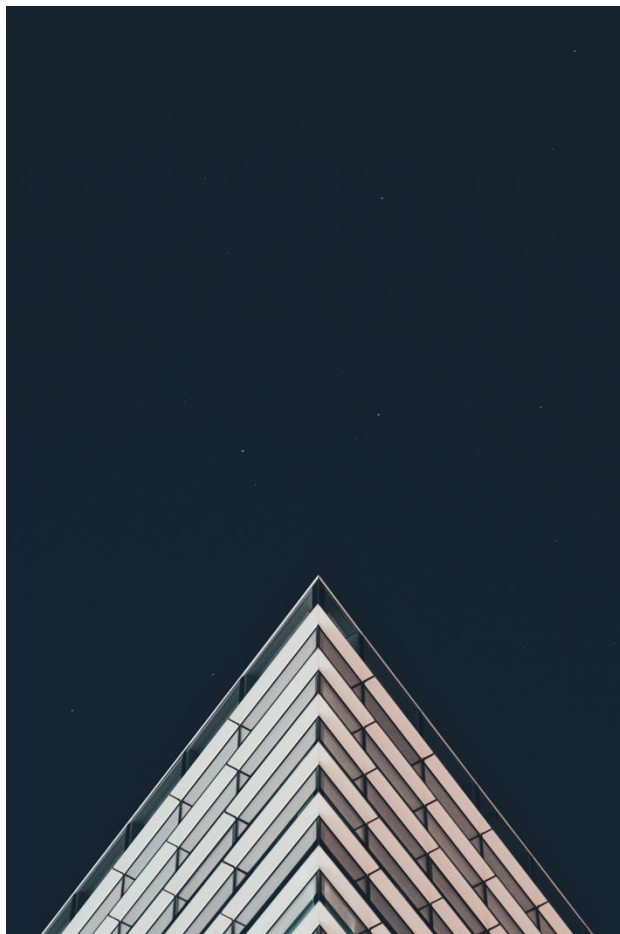
U: We want to Understand

R: We want to Recruit more patients, researchers and others who can help us

E: We want to be actively Engaged in BSyn research and fundraising

**We want to
be "SECURE"**

Emerging Vision of Strategy



**Better Care
Guidelines**

Treatments

Cure

Cell models

**Animal
models**

Assays

**Disease
Biology**

Centers of Excellence

Virtual Tumor Board

IBSR

**Natural History
Study**

**Biobank
Network**

Organization

Research Council

RC is a new group designed to advise and guide the BSA's research activities

BoD & ED ensures all RC activities are aligned and in sync with committees

