Updates from the Bloom Syndrome Association & Registry

Mary Beth Campbell, Chair of Board of Directors, Bloom Syndrome Association

Nicole Kucine, Director, Bloom Syndrome Registry

Agenda

Bloom Syndrome Association

Who we are

Recent activities

What's ahead?

How YOU can get involved

Bloom Syndrome Registry

What it is

How it works with the BSA and others

How YOU can get involved

Questions & Answers / Discussion

Our mission is to connect, educate, and support the international Bloom syndrome community and stimulate research that leads to longer, healthier lives.

Our community, at a glance

Individuals with Blooms syndrome

72

Countries

18

Mailing list participants

400

Facebook members

297

Board Members & Volunteers

16

bloom syndrome association

Board of Directors

Join Us!

Mary Beth Campbell

Chair

Rode bike across the state of Iowa (471mi)

Tanja Obermeir

Jamaican by birth

Eileen Rosenthal

Held a drag queen bingo to raise funds for the BSA

Mary Treder

Vice-Chair

Has a road in Uganda named for her

Lorne Yasbin

Secretary

Once had to answer 'yes' to 'is there a doctor on this plane?'

Victoria Warren

Has traveled to 47 US States

Talon Webb

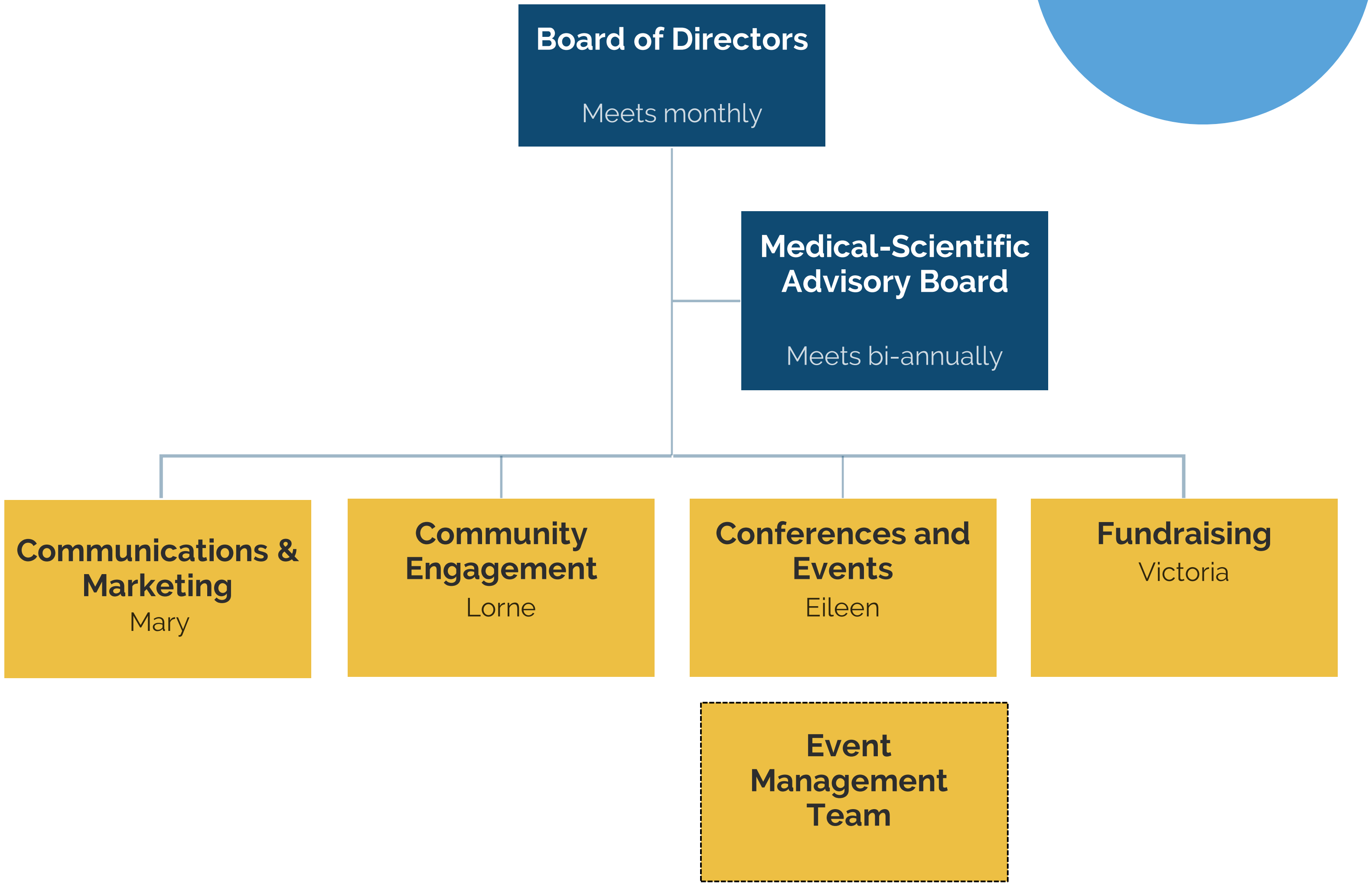
Treasurer

Has 34 nieces and nephews

Org Chart

Committees

Our committees and advisory groups help ensure the work of the BSA gets done



Board of Directors

Meets monthly

Medical-Scientific Advisory Board

Meets bi-annually

Communications & Marketing

Mary

Community Engagement

Lorne

Conferences and Events

Eileen

Event Management Team

Fundraising

Victoria

We need your help!

BSA programs/activities over the last two years

Refreshing our Look

- New logo
- Updated website

Stimulating Research

- iPSCs
- Research Roundtable
- Cancer Experiences

Forging Partnerships

RARE-X

FORCE

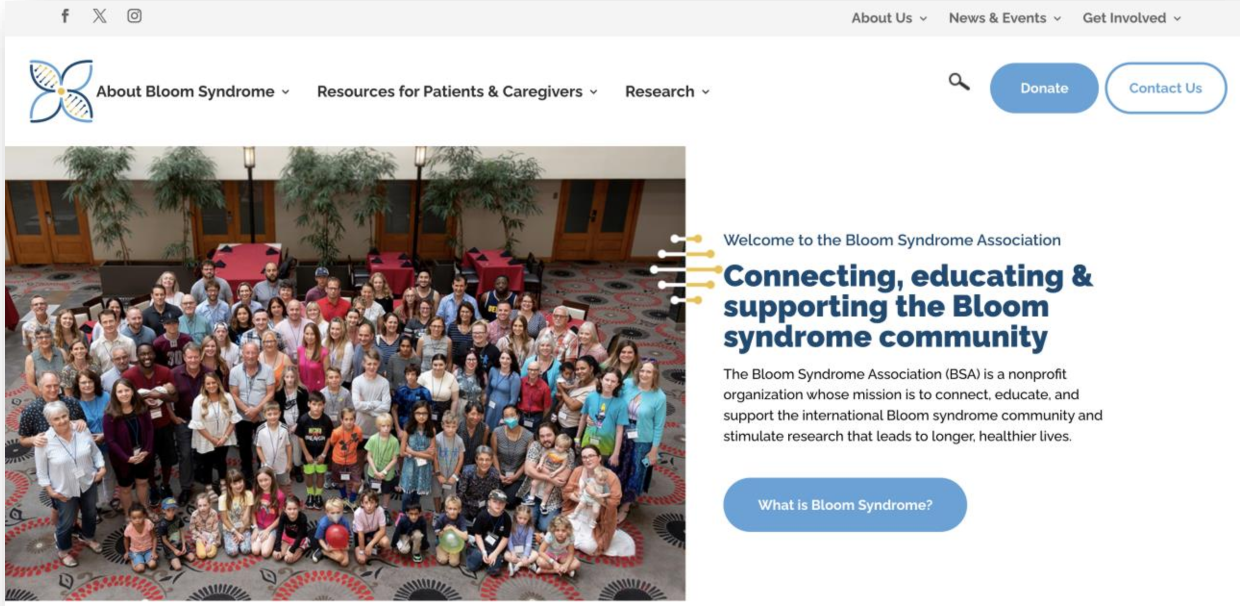
Creating a Sustainable Organization

- Virtual operations
- Fundraising

Building Bridges

- Conference
- FB group

The BSA website is a hub for all things Bloom syndrome



Welcome to the Bloom Syndrome Association

Connecting, educating & supporting the Bloom syndrome community

The Bloom Syndrome Association (BSA) is a nonprofit organization whose mission is to connect, educate, and support the international Bloom syndrome community and stimulate research that leads to longer, healthier lives.

https://www.bloomsyndromeassociation.org

- Handbook
- Health supervision recommendations
- Information on how to sign up for Registry, RARE-X
- Ongoing research studies
- Videos, and past webinars
- Streamlined donation page

Stimulating research…and more to come!

- **iPSC lines**

  3 lines currently being programmed from volunteer samples…thanks to your support!

  Will be released to researchers following successful reprogramming

- **Connecting community to emerging & ongoing research studies**
  - Cancer organoids study
  - Registry
  - RARE-X

- **Research Roundtable**

  First ever BSA-sponsored think tank for clinicians and researchers

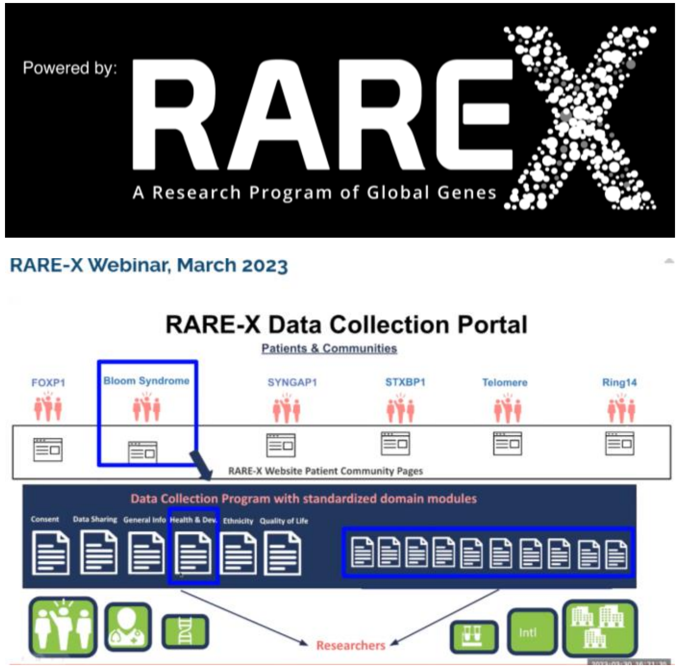
- **Patient-focused Research Roadmap**

  Solicited feedback from community on research priorities, discussed yesterday at RR

  More to be discussed tomorrow!

Forging Partnerships

with organizations that will help us meet our mission



Powered by: RAREX

A Research Program of Global Genes

RARE-X Webinar, March 2023

RARE-X Data Collection Portal

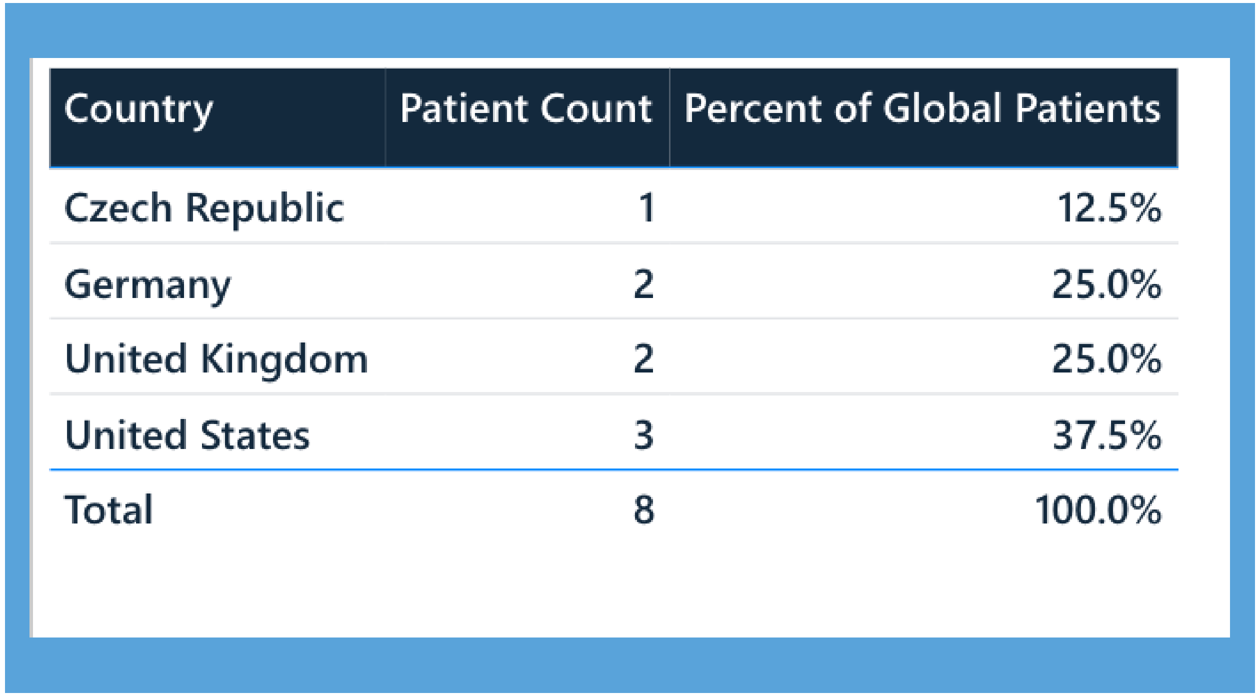
June 27, 2024

THE BLOOM SYNDROME ASSOCIATION

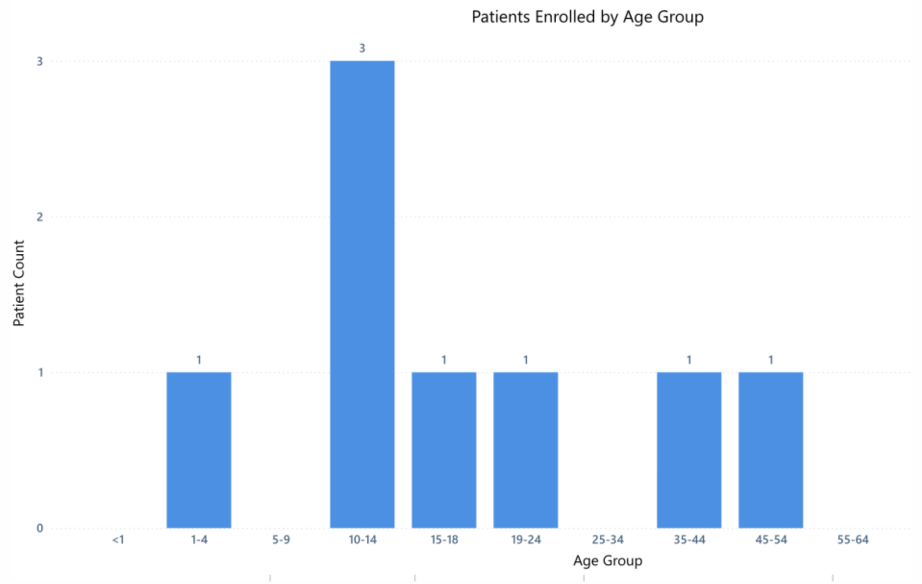
Posted in: Research , Genetic Testing , Hereditary Cancer - General , Featured Partner
Tags: Hereditary Cancer , Hereditary Cancer Research , Patient Advocacy , Inherited Mutations , Genetics Research , Medical Decisions , Coping With Risk

FORCE | 25 years

Facing Hereditary Cancer EMPOWERED



Country	Patient Count	Percent of Global Patients
Czech Republic	1	12.5%
Germany	2	25.0%
United Kingdom	2	25.0%
United States	3	37.5%
Total	8	100.0%



RARE-X...

is easy to participate in!

helps us connect to other rare diseases!

SIGN UP TODAY! SET A REMINDER TO UPDATE ON YOUR/YOUR CHILD'S BIRTHDAY, AND AS NEW SYMPTOMS DEVELOP!

Where we are going

Some of our upcoming priorities….

- Cancer treatment: what to do when cancer strikes
- Patient-focused research roadmap: a new era in Bloom syndrome
- Updating the Patient & Family Handbook
- Reaching more patients, in more languages

What do you want to see?

No level of involvement is too small!

The Bloom Syndrome Registry

Partnering with the Bloom Syndrome Community

Nicole Kucine, MD, MS
Director, Bloom Syndrome Registry

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Let’s talk about…

- What *is* the Bloom syndrome registry (BSR)?
- How the registry works with:
  - The BSA
  - The Bloom syndrome research community
- How YOU can be involved in the Bloom syndrome registry

What is the Bloom Syndrome Registry (BSR)?

- The BSR is a long-term natural history study of Bloom syndrome
- There are 2 key components to the BSR:

Sample Database

Deidentified samples:

- Blood samples
- Tissue samples
- Tumor samples

Information Database

Secure database
Demographic information
Clinical information:

- Genetic testing
- Symptoms
- Outcomes

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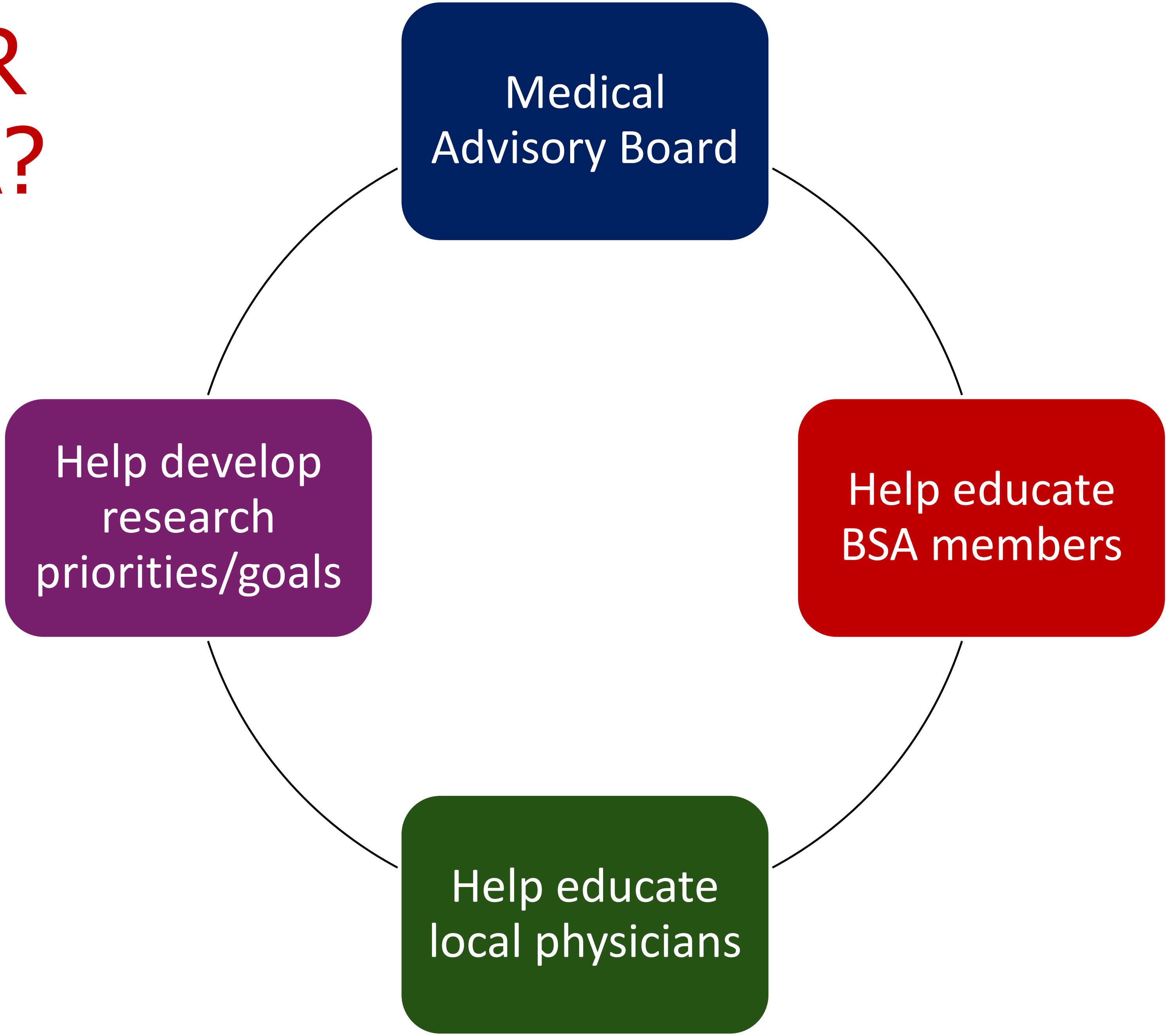
Who is currently in the registry?

- 311 enrolled since inception
- Just over 50% of subjects are male
- 156 individuals with cancer reported
  - A number of people are lost to follow-up so likely higher than this

How does the BSR work with the BSA?



Medical Advisory Board

Help educate BSA members

Help educate local physicians

Help develop research priorities/goals

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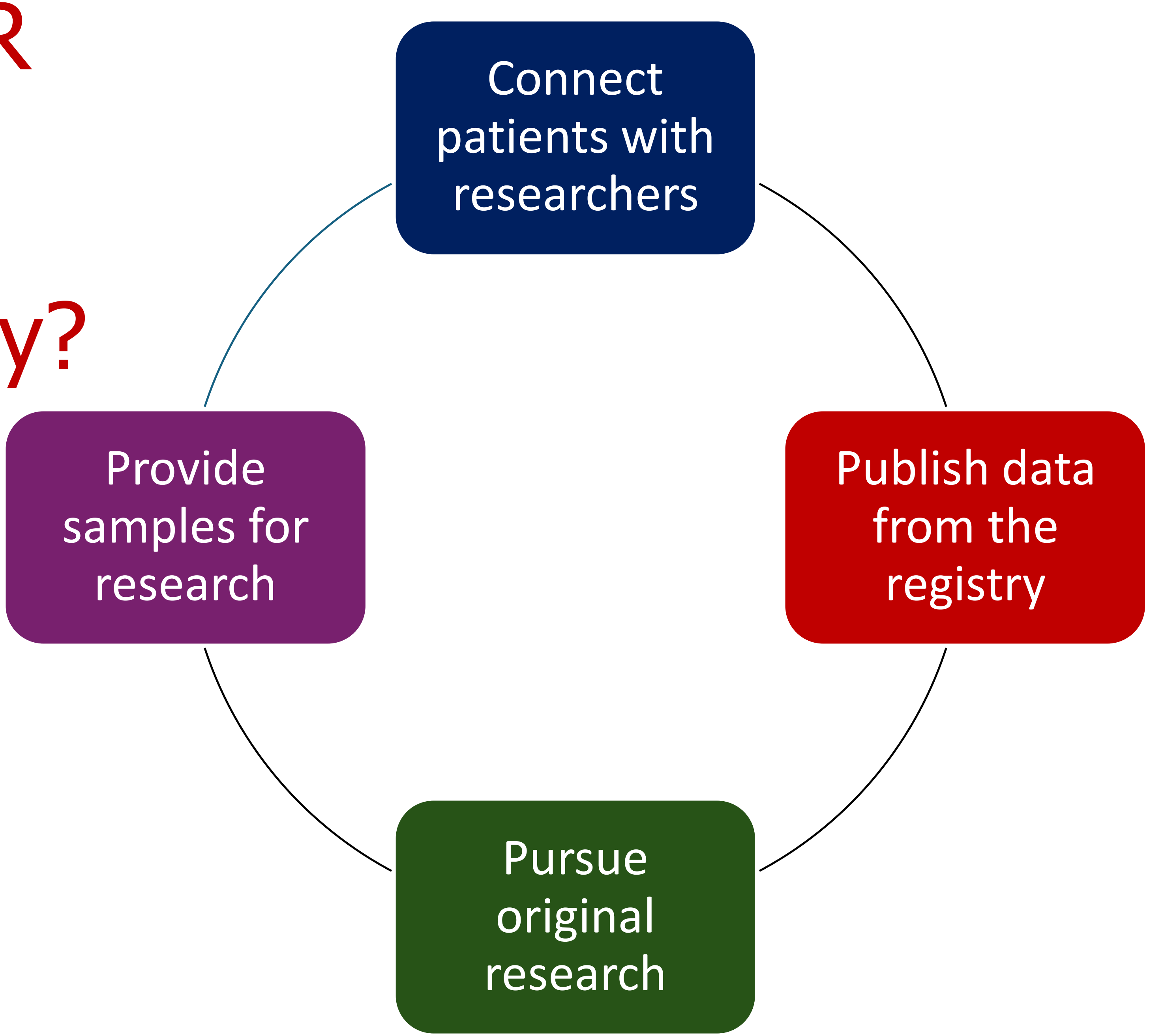
How does the BSR work with the broader Bloom research community?



Connect patients with researchers

Publish data from the registry

Pursue original research

Provide samples for research

BSR research collaborations

- Vivian Chang - UCLA
- Nathan Ellis - University of Arizona
- Stefan Meyer - Manchester University
- Additional collaborators at
  - Weill Cornell Medicine
  - Dana Farber Cancer Center/Boston Children's Hospital
  - NY Genome Center

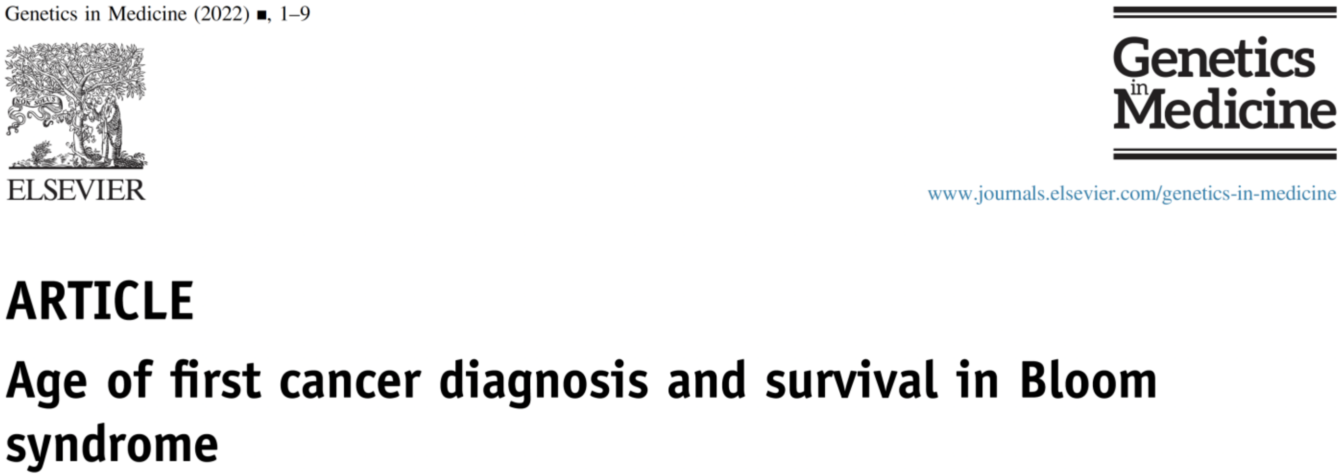
  and more!

Publications from the BSR

AJMG AMERICAN JOURNAL OF medical genetics PART A

ORIGINAL ARTICLE

Health supervision for people with Bloom syndrome



Genetics in Medicine (2022) ■, 1–9

ELSEVIER

Genetics in Medicine

www.journals.elsevier.com/genetics-in-medicine

ARTICLE

Age of first cancer diagnosis and survival in Bloom syndrome

What is involved in enrolling in the BSR?

- Sign consent/assent form
  - Currently only have forms in English

- Provide initial clinical information
  - Send updates with new labs, tests, changes in symptoms

- Share samples when getting routine blood or tissue biopsies
  - Can connect us with your doctors

- Can participate as much or as little as you are comfortable!

Sign consent/assent form

- Can be done in person or via zoom with e-signed documents
- Review consent form in depth
- Important issues to discuss include:
  - Confidentiality
  - Risks/benefits
  - Current and future research opportunities
- Children aged 7-17 sign an “assent” form
  - Very simplified but ensures they are willing to participate

Provide initial clinical information

- Complete an intake form
- Things that would be very helpful
  - Connect us with the diagnosing physician
  - Provide genetic testing report
  - Provide any SCE testing report
  - Provide any additional relevant reports/clinical notes/labs

Share samples when getting blood or tissue biopsies

- If you are going for routine surveillance testing
  - Blood
  - Colonoscopy

- If you are having unexpected tissue biopsies
  - Masses found on exam or imaging

Can participate as much or as little as you are comfortable!

- There are multiple elements of the consent that are optional
- Do not have to provide photos
- Do not have to allow additional genetic testing
- The more the better for research, but we are happy for all you are willing to share!

A note about working with outside researchers…

- All samples collected/used for the registry will be **deidentified**

- What might get shared when we work with other researchers?
  - Bloom diagnosis
  - Genetic Bloom mutations
  - Gender
  - Age at time sample was collected

- What is *NOT* shared with other researchers?
  - Name, address, date of birth, etc
  - Identifiers are not shared and don’t need to be
  - If an outside researcher wants to connect with you, we would ask you first before making that connection

What can the BSR *not* do?

- Cannot conduct all Bloom syndrome research on our own
  - Need different kinds of resources & expertise for different kinds of research studies
  - <u>This is why collaboration is crucial and best for the community</u>

- Cannot give specific medical advice for you or your child
  - The BSR team cannot & should not replace your own doctors
  - We can partner with them to share our knowledge and experience, and connect them with relevant specialists

What’s the bottom line?

- The Bloom syndrome registry has been working to further our understanding of Bloom syndrome for many years
- Data from the Bloom syndrome registry has helped contribute to practice guidelines and educates physicians and families
- Only through working with the Bloom syndrome patient and research communities can the registry be most effective
- Our goal is for **all** patients with Bloom syndrome to be enrolled on the registry

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Thank you!

- Please come chat if you are interested in enrolling in the registry!
- If you have previously enrolled, please come fill out an updated consent form!
- Teamwork is the best way to make progress!

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