

Advancements in Research

Towards a Patient-Focused Agenda

August 4, 2024

Research Roundtable Organizers, represented by:

Candice Bourke, Chris Brünger, Mary Beth Campbell,
Nathan Ellis, Anne-Marie & Henri van den Hurk



72

ACTIVE BLOOM SYNDROME PATIENTS

18

COUNTRIES

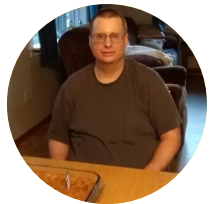
5+

CANCER DIAGNOSES IN PAST TWO YEARS



JASON

If we rely solely on "stimulating research" in the ways we have so far, we will be too late for many families



PHILIP

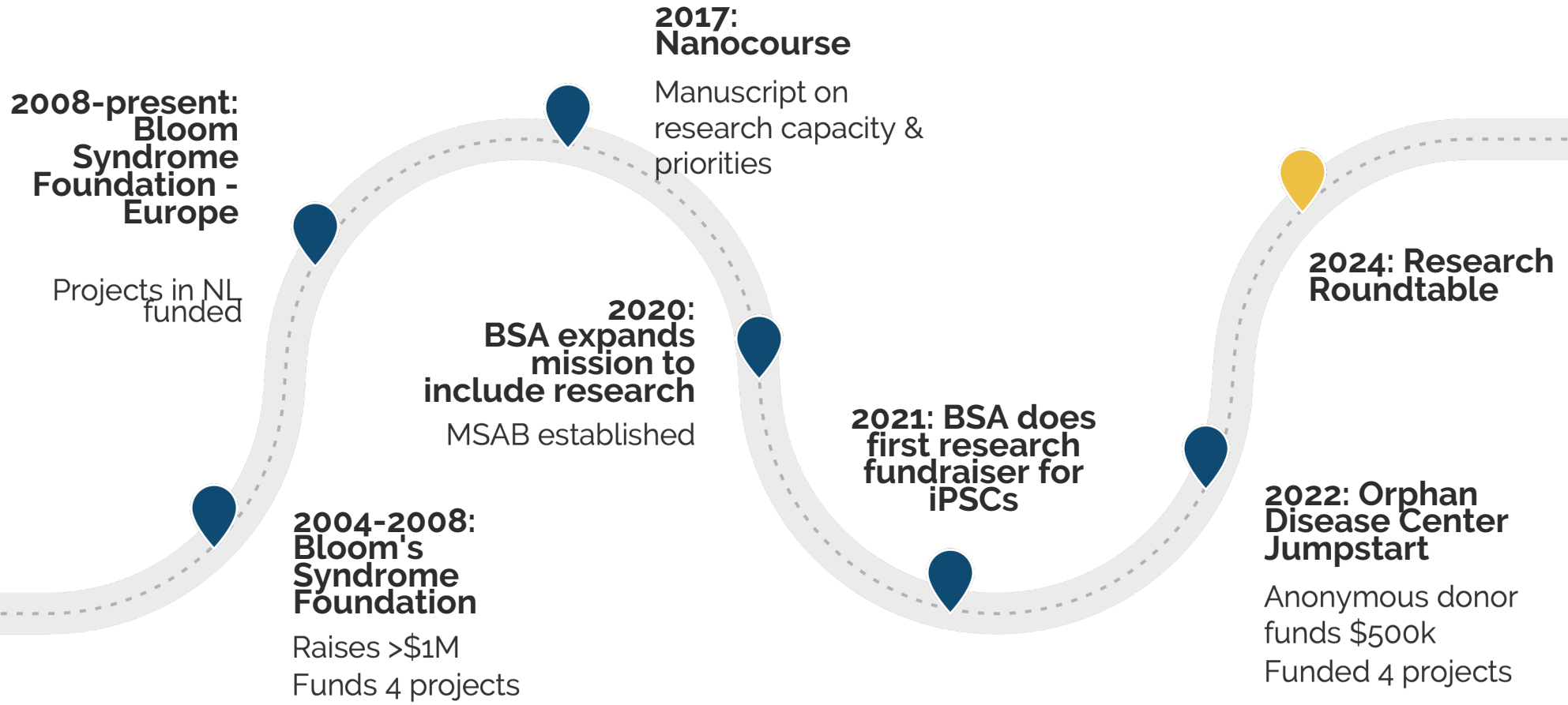


BONNIE



KAITLYN

Patient-Led Research Activities: A Brief History



Summary of Research Roundtable

Friday, August 2,
8:30am-4pm

15 participants:
clinicians,
researchers, patient
advocates, from 5
countries

Discussion of key
Bsyn research
priorities and
existing gaps

Some take-aways of key priorities & ideas:

1. **Need for DATA & INFORMATION & SAMPLE sharing, across borders: Bsyn too rare for any country, let alone one institution to do alone**
 - Cancer surveillance trial would require multi-national effort, complexities due to multi-cancers
2. **Cancer treatments & experiences:**
 - Need better understanding & data
 - International tumor board(s)
3. **How to facilitate data, information and samples to Registry: Missing link from patient to Registry? Hurdles?**
4. **Need for new, additional researchers & collaborators with expertise in specific areas (Bsyn Centers of Excellence?)**

Most important topics to address?

1. Reducing risks of cancers through standard surveillance
2. Detecting and treating other symptoms of Bsyn (e.g. diabetes, immunodeficiency)
3. Developing cancer treatments that are more effective, less toxic
4. Understanding the cause of, and treatment of, dermatological symptoms?
5. Finding a cure for Bsyn
6. Finding more individuals with Bsyn
7. **OTHER?**

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

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

**We want to
be "SECURE"**

1. Be Safe

1.1 Identify and address key concerns:

Cancer Risks
Other Risks

1.2 Evaluate the effect of sunlight on the development of mutations and cancer in BSyn

1.3 Evaluate the effect of ionizing radiation (e.g. x-rays) on the development of mutations and cancer in BSyn

1.4 Evaluate the effect of anti-cancer drugs on the development of mutations and cancer in BSyn

1.5 Conduct a clinical surveillance study using established methods

1.6 Conduct a clinical surveillance study using new methods

2. Effective Treatments

2.1. Perform a repurposing screen for efficacy in preventing/lowering SCEs (sister chromatid exchanges) and/or restoring function phenotypically (e.g. increasing lifespan, size, etc.) in BSyn models

2.2 Screen ACDs (anti-cancer drugs) /radiation / immunotherapies for appropriate efficacy dosage in the treatment of Bsyn cancers

2.3 Find a cure for Bsyn

3. Communicate/Collaborate with Stakeholders

**3.1 Within Bsyn
community**

**3.2 With the Bsyn
clinician/researcher
community**

**3.3 With other RECQ
disorder communities**

**3.4 With other cancer
predisposition
communities**

**3.5 With other ultra-
rare disease
communities**

**3.6 With regulators,
politicians, industry,
and media**

4. Understand Bsyn

4.1 Conduct research into the underlying mechanisms of BSyn to find effective treatments for it, and its various manifestations.

4.1.1 Understand role of BLM in aging

4.1.2 Understand role of BLM in immune function

4.1.3 Understand role of BLM in dermatology

4.1.4. Understand role of BLM in endocrinology

4.2 Foster understanding of BSyn as an important model of cancer development in general

5. Recruit

5.1 More individuals with BSyn through publicity, early diagnosis, dialogue with relevant societies

5.2 More researchers

5.3 More expert & speciality clinicians

5.4 More interdisciplinary teams

5.5. More friends, volunteers, and donors who support us

6. Engage

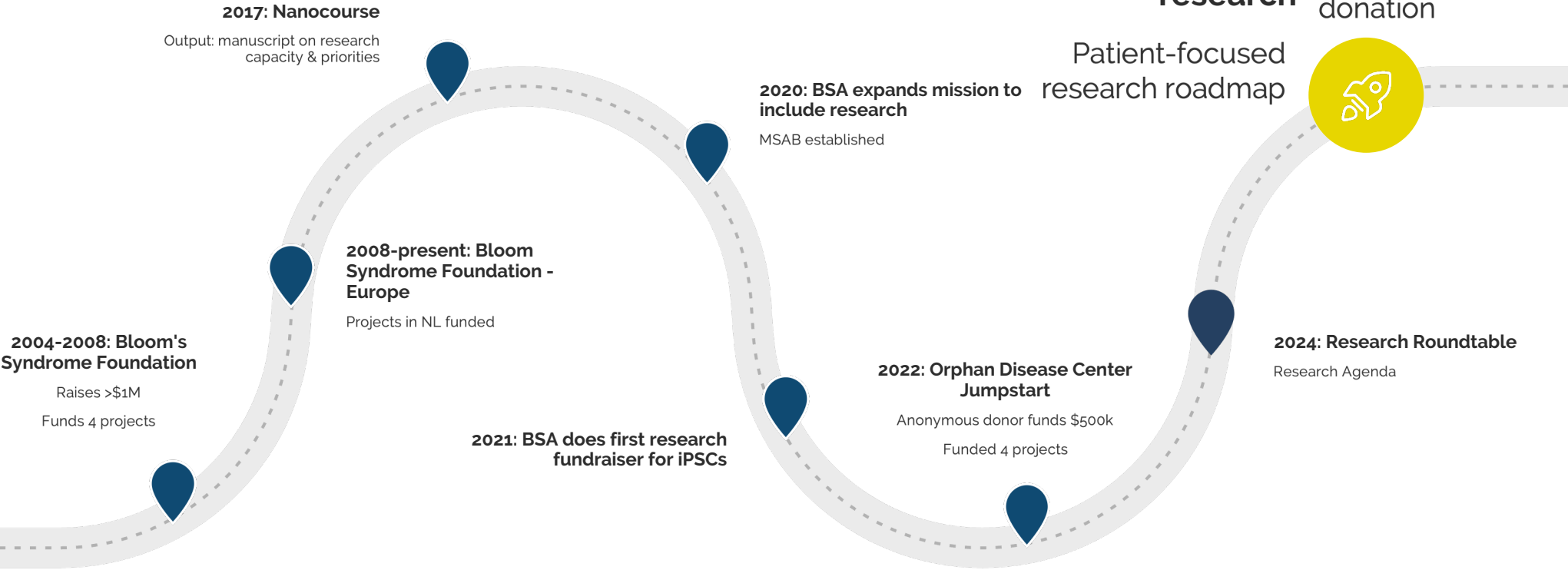
6.1 Actively engage in BSyn research through discussion, study participation, provision of data and samples

6.2 Actively engage in BSyn research through fundraising activities

6.3 Actively engage in BSyn research through evaluation of study outcomes and setting priorities

6.4 Actively engage in BSyn research by documenting impact of outcomes on one's own life

Patient-Led Research Activities: The Future



Funding Policy/Principles



Patient-driven

Projects need to be directly tied to identified priorities from the community, as articulated in patient-centered research roadmap



Community-building

Investigators need to be willing to collaborate, to share results/reagents with other researchers, and to provide updates to the patient & family community



Gap-filling

Projects would not likely be funded by more traditional funding mechanisms. Can include:

- Seed funding for preliminary data/validation of approach
- Literature reviews/white papers
- More directed activities
- Tool/reagent development



Independently-reviewed

Projects will be reviewed by unbiased technical experts to ensure scientific merit and feasibility

High-Level Funding Process



Call for Proposals: Sept 2024

1. Priorities identified, RFA drafted
2. Request for Applications announced by Orphan Disease Center (ODC)

- ODC solicits peer review feedback on applications
- BSA MSAB may also review and provide feedback

Review process

Recommendation

- "Research Executive" reviews proposals and expert ratings, makes funding rec. based on technical reviews & funding principles

- BSA Board of Directors makes ultimate funding decision
- BSF may choose to fund in conjunction or separately

Funding decision & project start: Jan 2025

Examples of Two Initial Projects Aligned with Roadmap

Cancer Treatment

Richarda deVoer *et al.*: Analysis of mutational signatures of Bsyn tumors

Possible outcomes: *clues* as to whether targeted therapies or immunotherapy may be particularly useful in Bsyn cancers

Cancer Surveillance

Vivian Chang *et al.*: validate use of novel, minimally invasive cancer surveillance methods

Possible outcomes: an understanding of promise or pitfalls of “liquid biopsies” in Bsyn

We cannot do this without you...

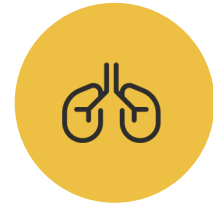
We will need **EVERYONE's** support



Funding



Data



Samples

Global Fundraising Will Be Coordinated



www.bloomsyndromeassociation.org

Funding:

Vivian Chang et al: *Cancer Surveillance in BSyn*



The BSA is a 501(c)3 and donations may be tax-deductible.

Tax ID: 800813615



www.bloomsyndrome.eu

Funding:

Ricarda de Voer et al: *Finding clues for future treatments of cancer like Immunotherapie*

NL79 RABO 0124853153

Stichting Bloom Syndrome
Nistelrode (The Netherlands)
Swift/BIC: RABONL2U

The BSF has ANBI status and donations may be tax-deductible

Fundraising Efforts: A New Era

Multi-year plan for achieving new levels in funding, to include:

- **Million Dollar Bike Ride (Jun 25)**
- **Bsyn Gala Ball (25/26)**
- **1:1 matching donations**
- **International funding opportunity repository**



Research Coordinators: Optimizing Data Flow & Sample Flow

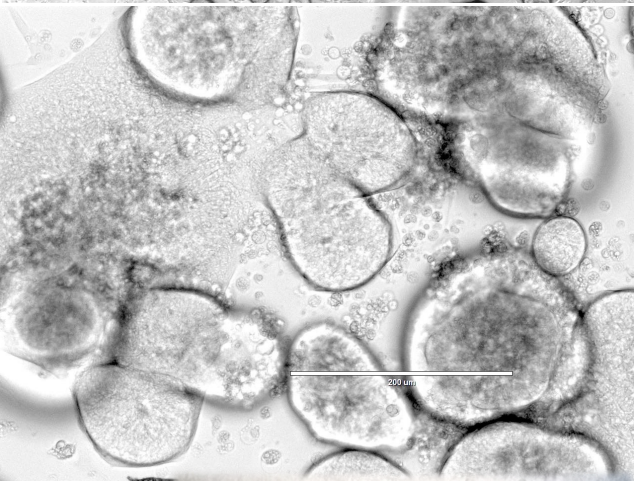
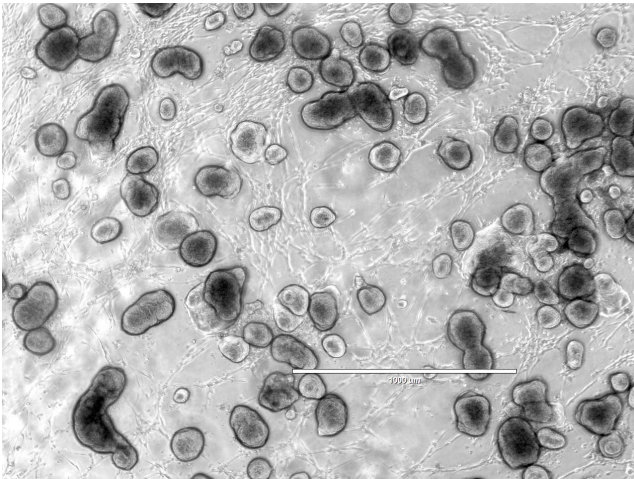
Australia: Candice Bourke

Japan: Chris Brünger

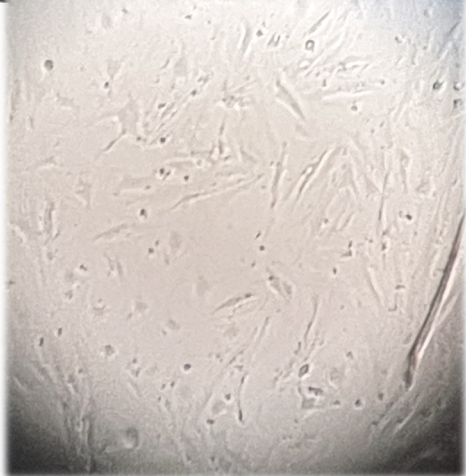
UK/EU: Ramsay Bowden

USA: Susan Zaslav (interim)

Other?



Colon mesenchymal cells from BS



Example: goals & aims of organoid project

Make tissue-like cell culture from normal cells and tumor cells from persons with Bsyn

Use the cell culture models to learn about toxicities to chemotherapeutic drugs from the normal cells and....

...to learn about the sensitivities to those drugs using the tumor cells.

Challenges...

Discussion

Around providing data & samples?

**Who would like to be involved in
our efforts?**

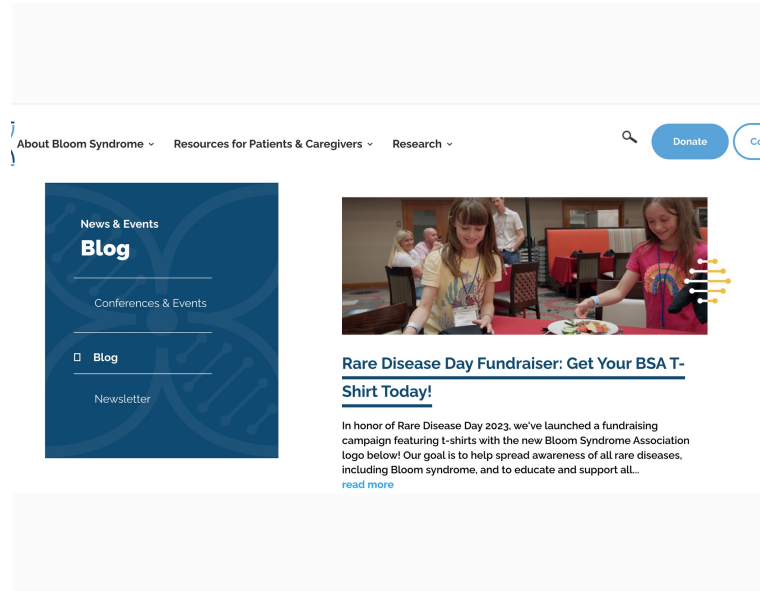
What's Next

How to keep up with what's going on



Webinars

Funded researchers and new project areas will be presented to the community



Blog updates

On BSA website:
bloomsyndromeassociation.org/news-events/blog/

Push to mailing list and FB page



Fundraising campaign

Updates on our progress
Requests for support