Bloom Syndrome and Cancer: Knowledge is Power!

Bloom Syndrome Patient & Family Conference

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What We’re Going to Cover Today

- Cancer 101
- Diagnosis
- Treatment
- Prevention and risk reduction
- What YOU can do to make progress

Meet Our Cancer Warriors!

Interviewer: Lorne Yasbin

Cancer 101: Definitions

- What is cancer?
  - Group of related diseases where cells divide without stopping and sometimes spread to surrounding tissues

Cancer 101: Definitions

- What is a tumor?
  - A lump or growth of tissue. Tumors can be benign (not cancer) or malignant (cancer)

Cancer 101: Definitions

- How is cancer diagnosed and staged?
- Biopsy is a procedure where a small sample of tissue is taken and examined under a microscope
- Metastasis is when cancer cells spread from the original site to other parts of the body
- Stage is the extent of cancer in the body, including the size of the tumor and if it has spread. Stages range from 0 (very early) or IV (advanced)

Cancer 101: Definitions

- What are cancer treatment types?
- Chemotherapy are medicines to kill cancer cells or stop them from going
- Radiation therapy uses high-energy rays to kill cancer cells or shrink them
- Surgery is a procedure to remove the cancer from the body
- Immunotherapy is a treatment that harnesses the immune system to fight cancer
- Targeted therapy is a drug that targets specific molecules involved in growth and spread of cancer cells

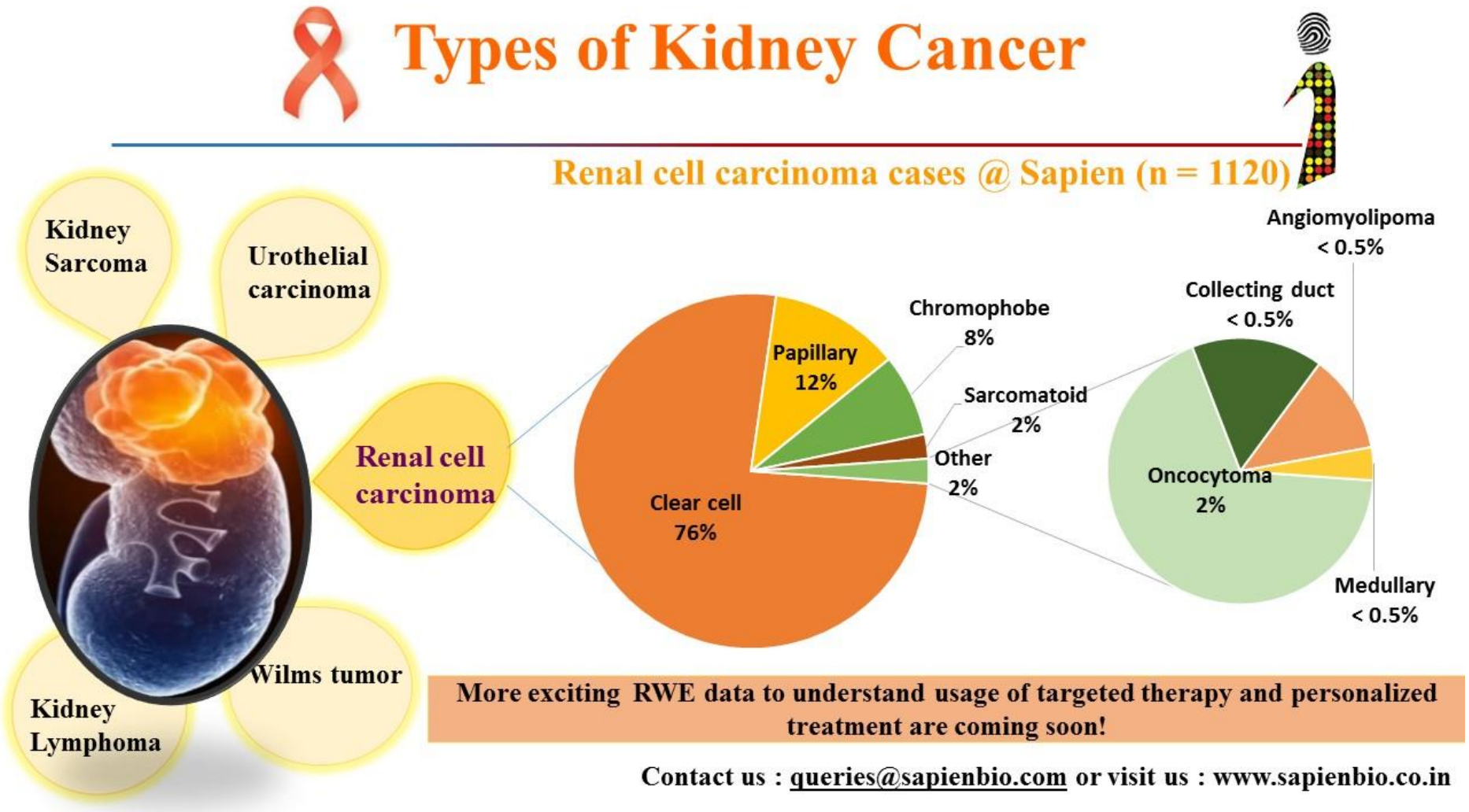
Cancer 101: Definitions

- Remission is when signs and symptoms of cancer are reduced or gone
- Recurrence is when cancer comes back after treatment

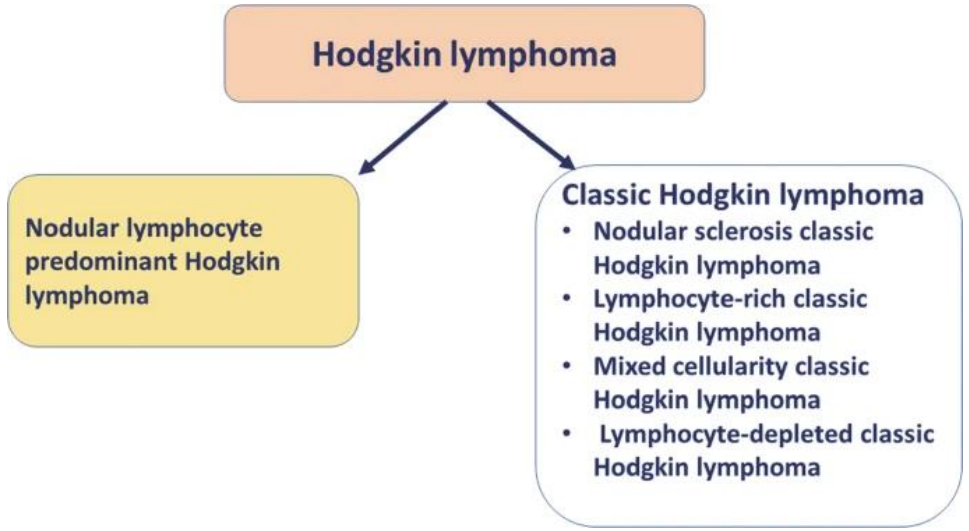
Introduction to cancer

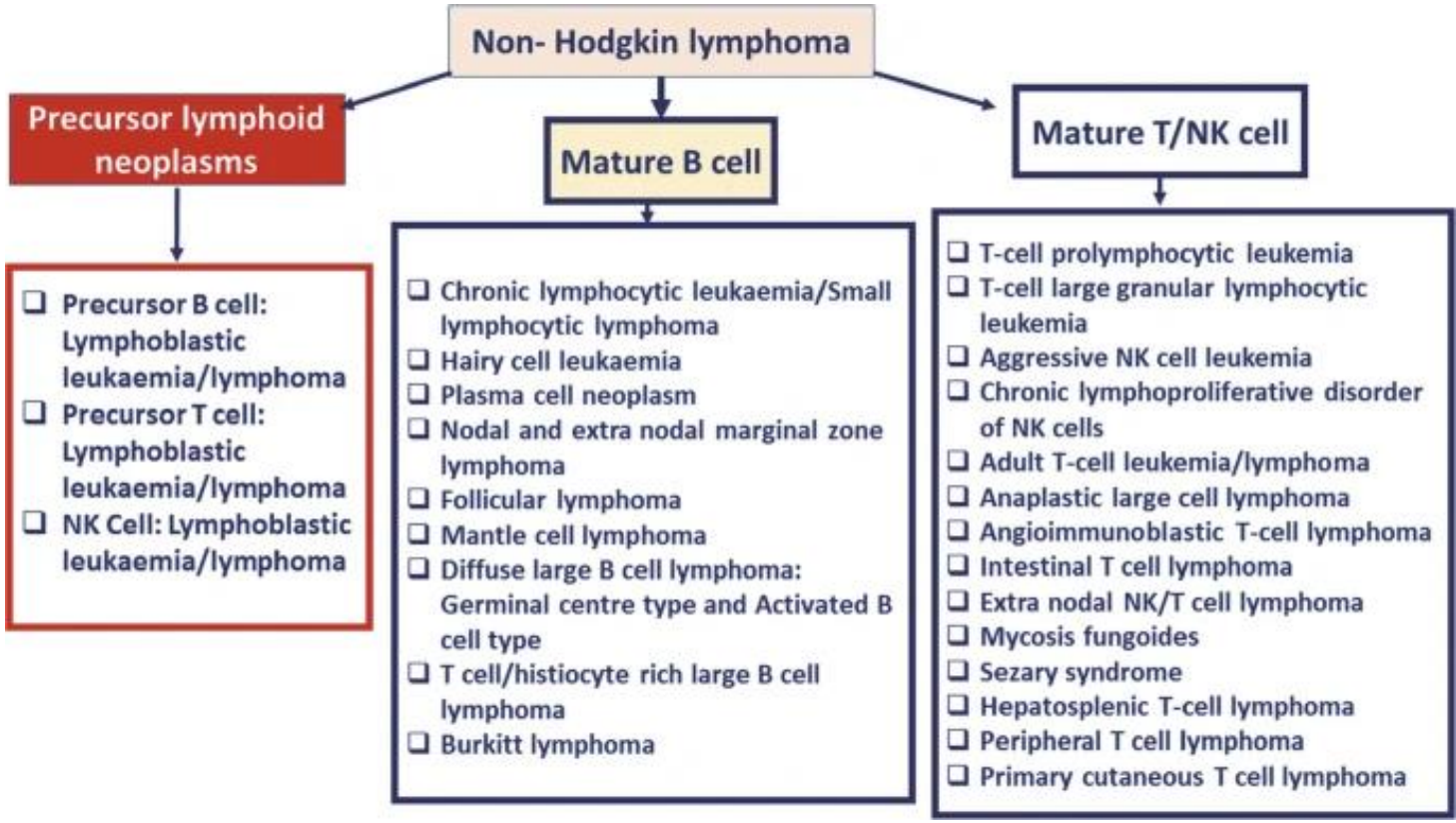
- Cancer can develop in virtually any part of the body
- Can behave differently, cause different symptoms, treated differently
- In lay language, most common cancers are referred to as the part of the body that it originates from
  - E.g. Breast cancer, colon cancer, lung cancer
  - More specific names depending on cell of origin
- Leukemia/lymphoma are blood and immune system cancers

Introduction to cancer



Introduction to cancer





Dey, P. (2021). Classification of Lymphoma, Different Markers and Approach. In: Diagnostic Flow Cytometry in Cytology. Springer, Singapore.

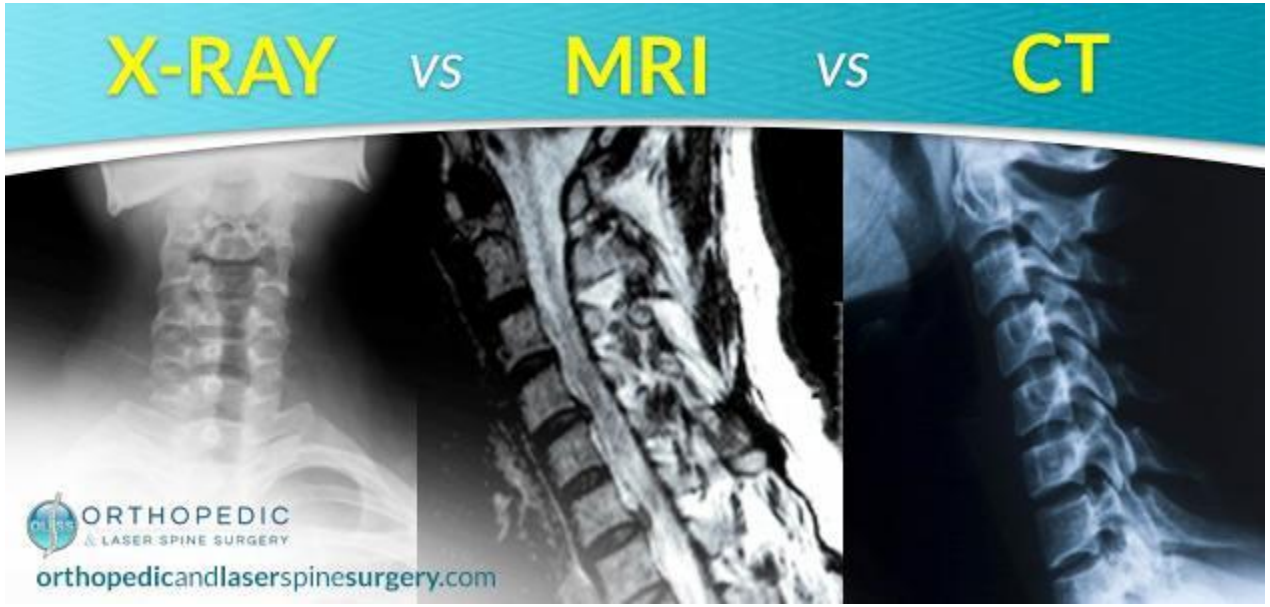
Cancer in Bloom syndrome

Cancer Diagnosis

- How is cancer usually diagnosed?
  - Common symptoms or through surveillance
    - Unexplained weight loss
    - Prolonged fever
    - Fatigue
    - Lumps or bumps
    - Pain
    - Unusual bleeding
- See a doctor and be a persistent advocate for yourself because early detection significantly improves chances of successful treatment

Cancer Diagnosis

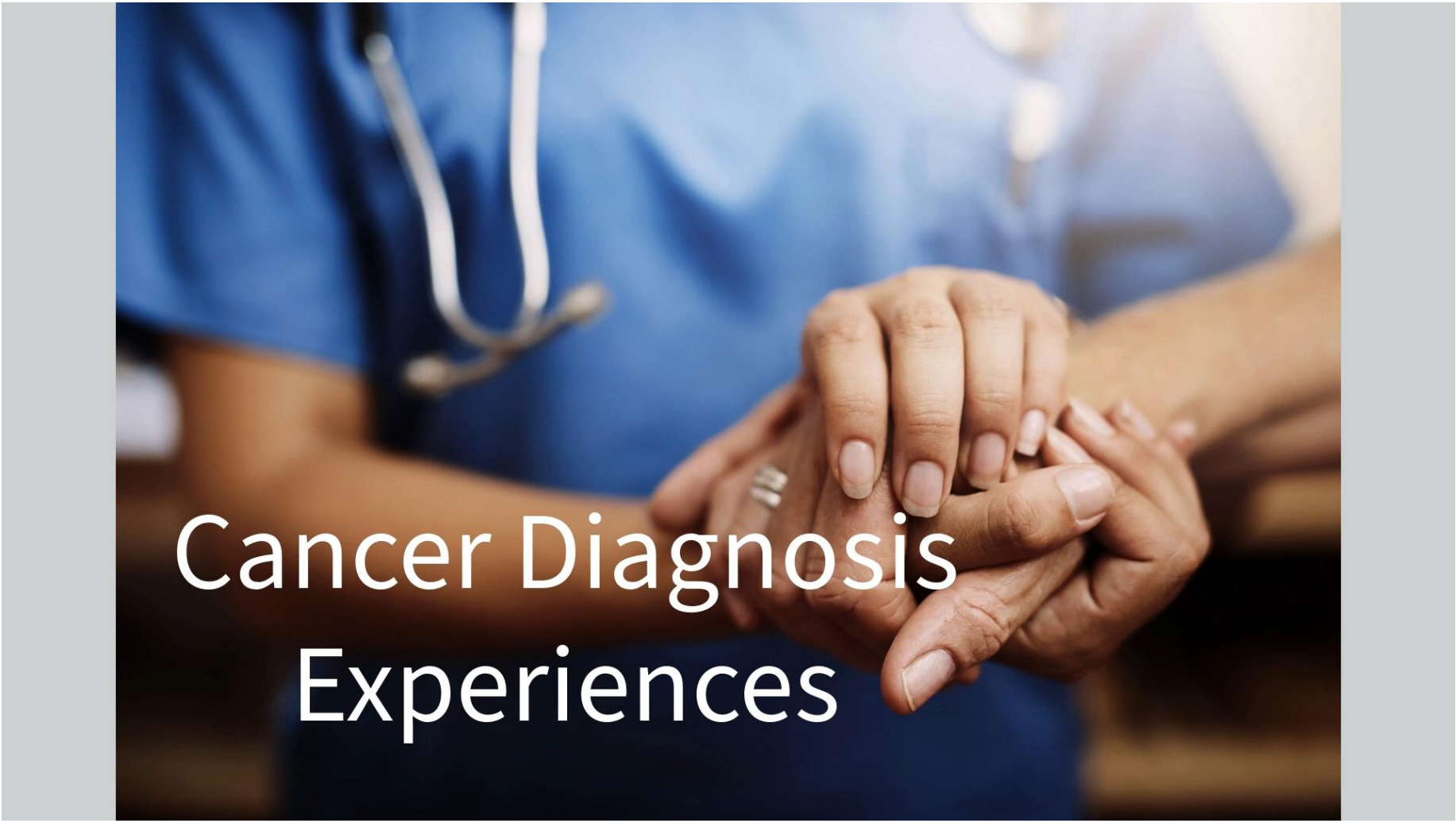
- Cancer can be diagnosed through a variety of methods
- Medical history, physical exam
- Imaging tests (radiology)



Cancer Diagnosis

- Procedures (lab and pathology medicine)
  - Biopsies
  - Surgeries
- Molecular and genomic tests (lab and pathology medicine)

Diagnosing Cancer in Bloom Syndrome



What’s known about cancer in Bloom syndrome?

- Two primary sources
  - Bloom Syndrome Registry
  - Published literature, usually case studies
  - Plus anecdotes from the community
- Limitations
  - Outdated information
  - Publication bias
- By coming together, we can start to overcome these limitations

What's known about cancer in Bloom syndrome?

- Information from 290 participants in the BSR, 1960-2021
- 155 (53%) participants developed 251 cancers

- The most common cancer is leukemia/lymphoma
- The most common solid cancers are colorectal, breast, and oropharyngeal

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Table 1 Malignancies reported among the 155 individuals in the Bloom Syndrome Registry

Malignancy Type	Frequency (% of Total Cases)	Median Age at Diagnosis, y	Mean Age at Diagnosis, y	Range, y
Total cases	251 (100%)			
Leukemia	41 (16.3%)	18	18	2-40
Acute myeloid	22	22	19	2-39
Acute lymphoblastic	13	15	18	4-40
Other (biphenotypic/ undefined)	6	16	18	4-39
Lymphoma	42 (16.7%)	23	23	4-49
Hodgkin	3	19	20	16-26
Non-Hodgkin	33	25	24	4-49
Burkitt	4	11	14	9-24
Other (mixed/undefined)	2	26	26	8-45
Oropharyngeal	28 (11.2%)	34	36	25-48
Tongue	10	39	40	30-48
Pharyngeal	7	32	34	30-45
Tonsillar	4	39.5	37.5	25-46
Other	7	31	30	25-34
Upper gastrointestinal	17 (6.8%)	31	32	15-48
Esophageal	5	39	37	25-48
Gastric	7	27	30	21-49
Pancreatic	4	34	35	28-44
Other	1	N/A	N/A	N/A
Colorectal	30 (12%)	36	35	16-49
Breast	29 (11.6%)	32	33	18-52
Ductal	12	32	33	26-42
Other	17	31	32	18-52
Genitourinary	14 (5.6%)	24.5	30	10-54
Cervical	5	22	22	19-23
Testicular	3	22	19	10-26
Other	6	41	42	33-54
Skin	29 (11.6%)	35	33	18-55
Basal cell	18	32	33	18-55
Squamous cell (uncategorized)	8	35	34	25-42
Undefined	3	35	34	25-42
Lung	4 (1.6%)	36.5	36	32-40
Wilms tumor	9 (3.6%)	3	4	1-11
All other	8 (3.2%)	N/A	N/A	N/A

N/A, not available

What’s known about cancer treatment in Bloom syndrome?

- Increased sensitivity to standard chemotherapy and radiation
- Few published reports
- Increased side effects and secondary cancers
- <= 50% of standard chemotherapy dosing
- Avoidance of alkylating agents (busulfan, melphalan, cyclophosphamide)
- Avoidance of ionizing radiation

What’s known about cancer treatment in Bloom syndrome?

- 18 → 22 published reports on cancer in patients with Bloom
  - 7 mention doses of chemotherapy
  - 5 mention doses of radiation
  - 4 mention surgery used
- Side effects
  - Prolonged bone marrow suppression- Fever, infection, bleeding
  - Gastrointestinal complications- Nausea, vomiting, mucositis, liver injury
  - Endocrine- High blood sugars
  - Strictures after radiation

Treating Cancer in Bloom Syndrome

Cancer screening in Bloom

- Cancer screening is challenging due to the different types of cancer
- Screening recommendations are based on the types of cancers that have been found in Bloom patients and the average ages that they have been found

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Cancer Surveillance Guidelines

2017 AACR recommendations	2018 AJMG recommendations
• CBCs every 3-4 months, starting 18y • Renal ultrasound at diagnosis/birth, every 3 months, through age 8y • Colonoscopy with fecal immunochemical testing every 6 months, starting 15y • Annual breast MRI, starting 18y	• Abdominal ultrasound every 3 mo, diagnosis to age 8y • Annual colonoscopy and FIT every 6 months, starting at age 10-12y • Whole body MRI every 1-2 years, starting age 12-13y • Annual breast MRI, starting age 18y

Practicing Cancer Surveillance

What to do if you receive a cancer diagnosis

- Questions to ask your oncologist
  - What type of cancer is it? (copy of pathology report)
  - How much experience do you have treating this type of cancer? How much experience do you have treating cancer in a patient with Bsyn?
  - Are there any targeted or personalized therapies available?
  - Should I get a second opinion?
  - What are the risks/benefits of each treatment option?
  - How can I manage potential side effects?
- Contact the Bloom Syndrome Registry
  - Put your healthcare team in touch with them as EARLY as possible.

What we still want to know

- Surveillance
  - What are effective screening strategies?
  - Can we detect abnormalities in blood sampled over time that signals development of cancer?
- Cancer diagnoses
  - Are cancers in patients with Bloom the same or different from patients without Bloom?
- Treatment
  - How does the treating oncologist select the best regimen/modifications?
  - Would patients with Bloom respond to immunotherapy?

How to get involved

- Register in the Bloom Syndrome Registry
  - Not sure if you’re registered? Contact the Registry.
- Let the Registry know any health updates
  - Annual questionnaire
  - Treatment roadmaps
  - Documentation of side effects
  - Outcomes
- Provide samples
  - Serial blood samples
  - Tumor samples - contact Registry as early as possible

Parting Words of Wisdom

Advice from Our Cancer Survivors

Thanks! Questions?