Cancer in Bloom Syndrome: What Do We Know From Registry Data?

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Key Points

- Bloom Syndrome Registry information
- Types of cancer diagnosed
- Age when first cancer diagnosed
- Survival

Bloom Syndrome Registry

Demographic	Number of patients (%)
Total Members	284 (100%)
Sex Male Female	 148 (52%) 136 (48%)
Number living	150 (53%)
Number who developed cancer Hematological cancer Solid tumor	147 (52%) 68 (24%) 99 (35%)
Genotype Homozygous Compound Heterozygous Only 1 allele detected (Affected)	 104 (37%) 56 (20%) 9 (3%)

Cancers seen in the BSR

As of March 2019, 147 (52%) registry participants had developed 235 cancers

Of the 235 cancers, 34% were hematologic, and 66% were solid tumors

Cancer Type	Frequency (% of total cases)	Median age of diagnosis (yrs)	Mean age of diagnosis (yrs)	Range (yrs)
Leukemia	40 (17%)	18	18	2-40
Lymphoma	39 (17%)	21	22	4-49
Colorectal	29 (12%)	36	35	16-49
Skin	25 (11%)	35	32	18-53
Breast	24 (10%)	26	29	10-47
Head and neck	24 (10%)	36	37	26-48
Stomach/esophagus	15 (6%)	32	33	15-48
Genitourinary	15 (6%)	22	21	19-23
Wilms tumor	8 (3%)	3	3	1-8
Lung	4 (2%)	37	36	32-40
All others	12 (5%)			

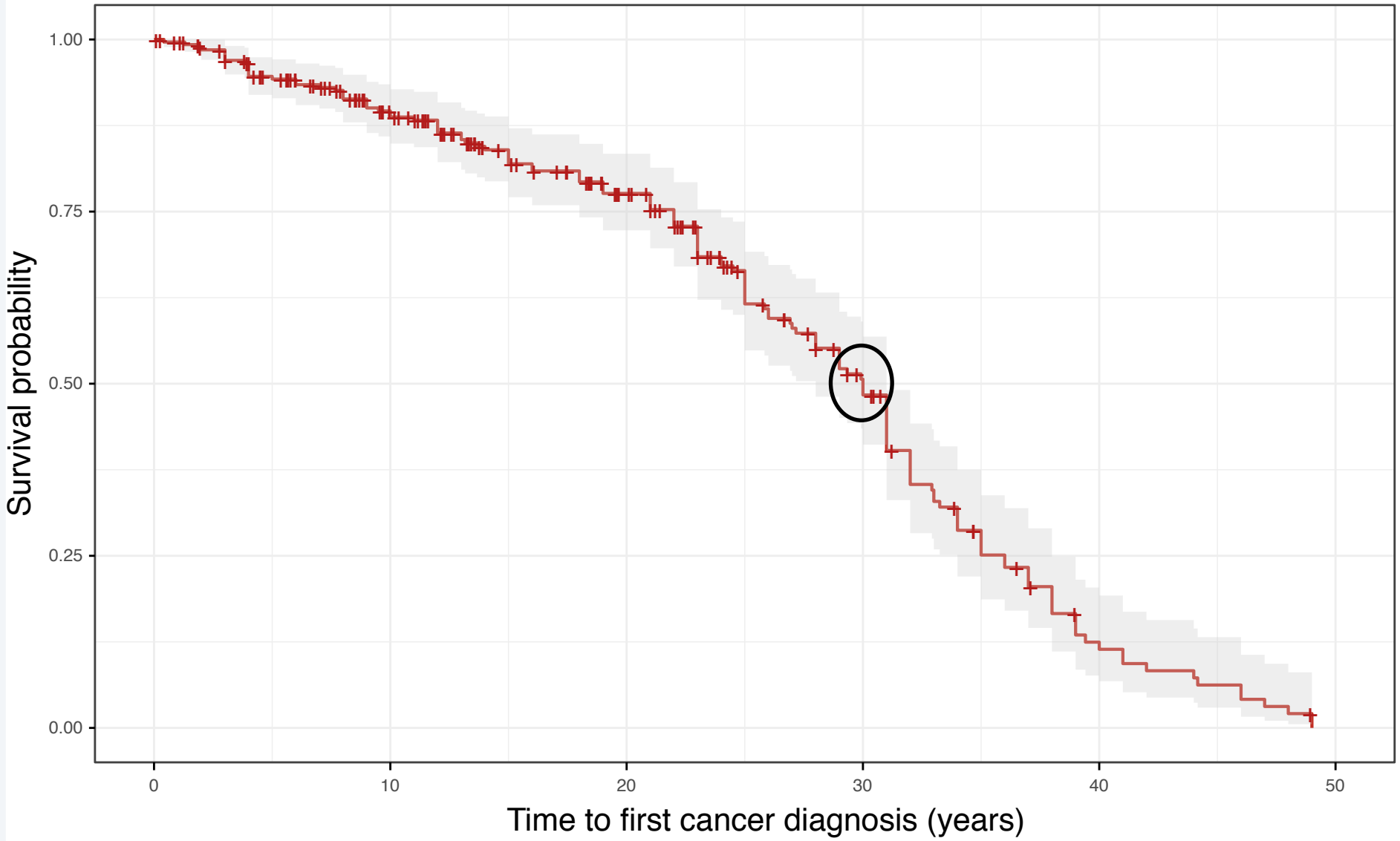
Who is being included

All registry participants

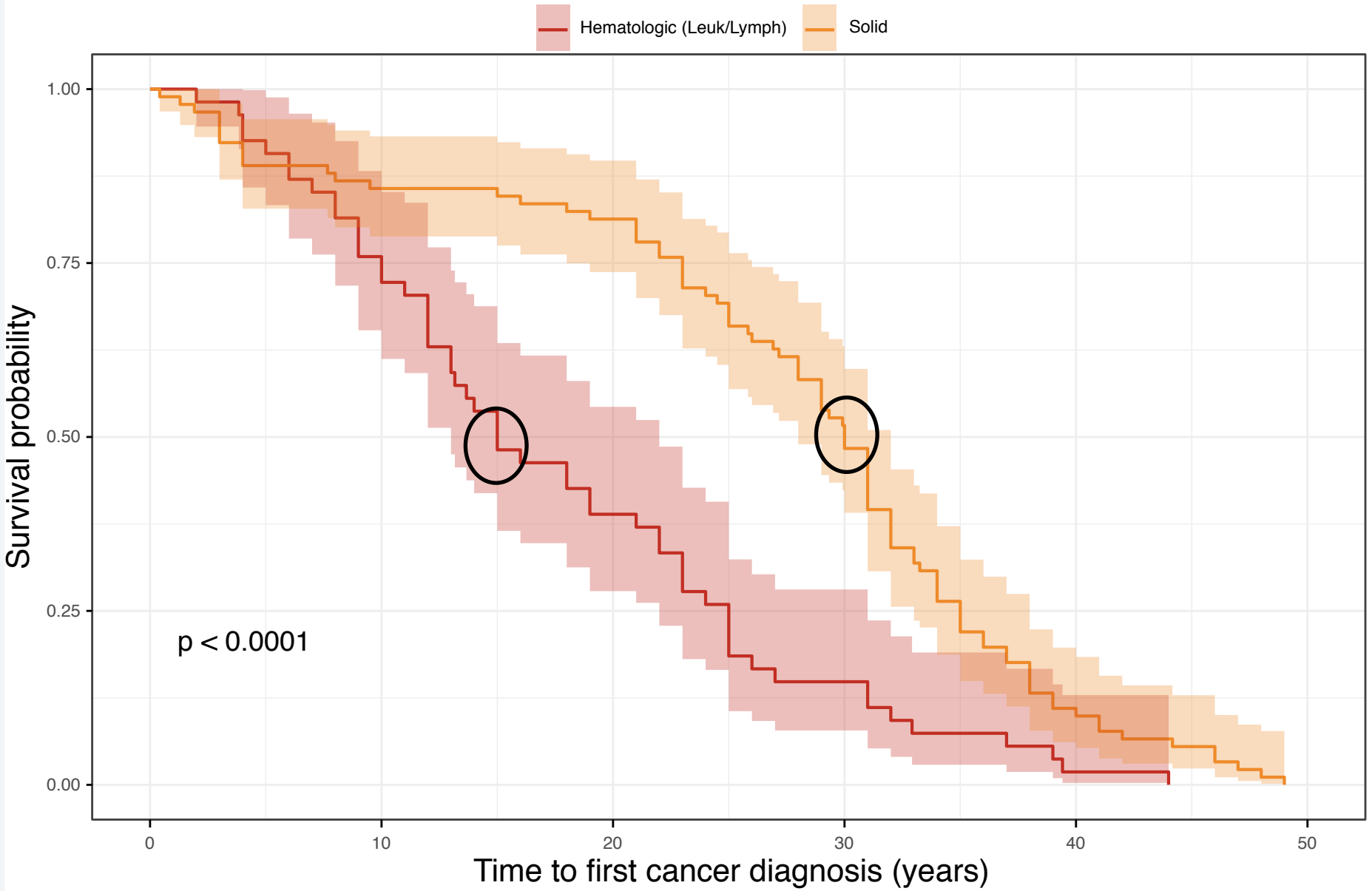
Only those with a cancer diagnosis on or after the year 2000

First Cancer Analysis

Time to First Cancer Diagnosis

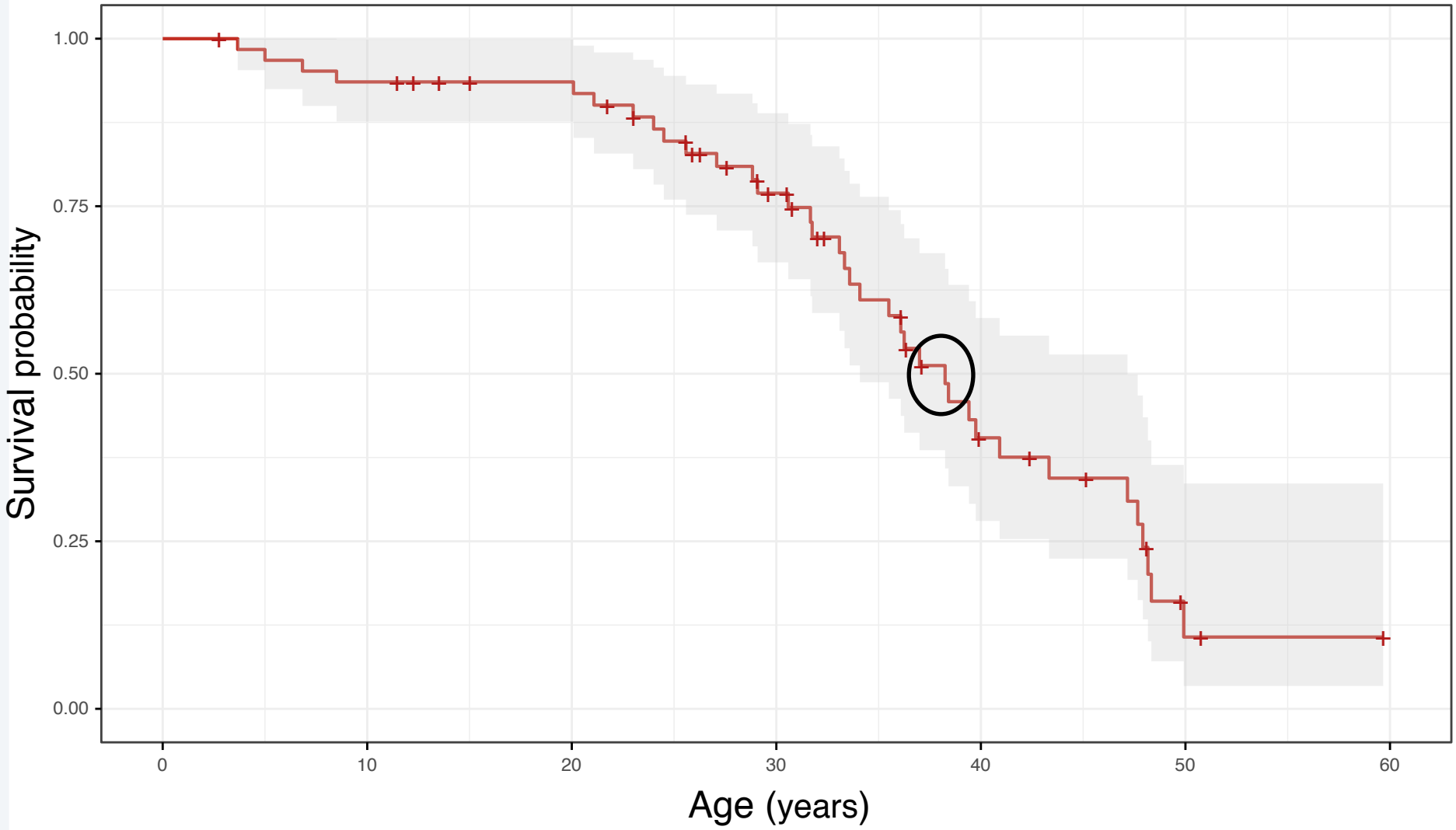


Time to First Cancer Diagnosis

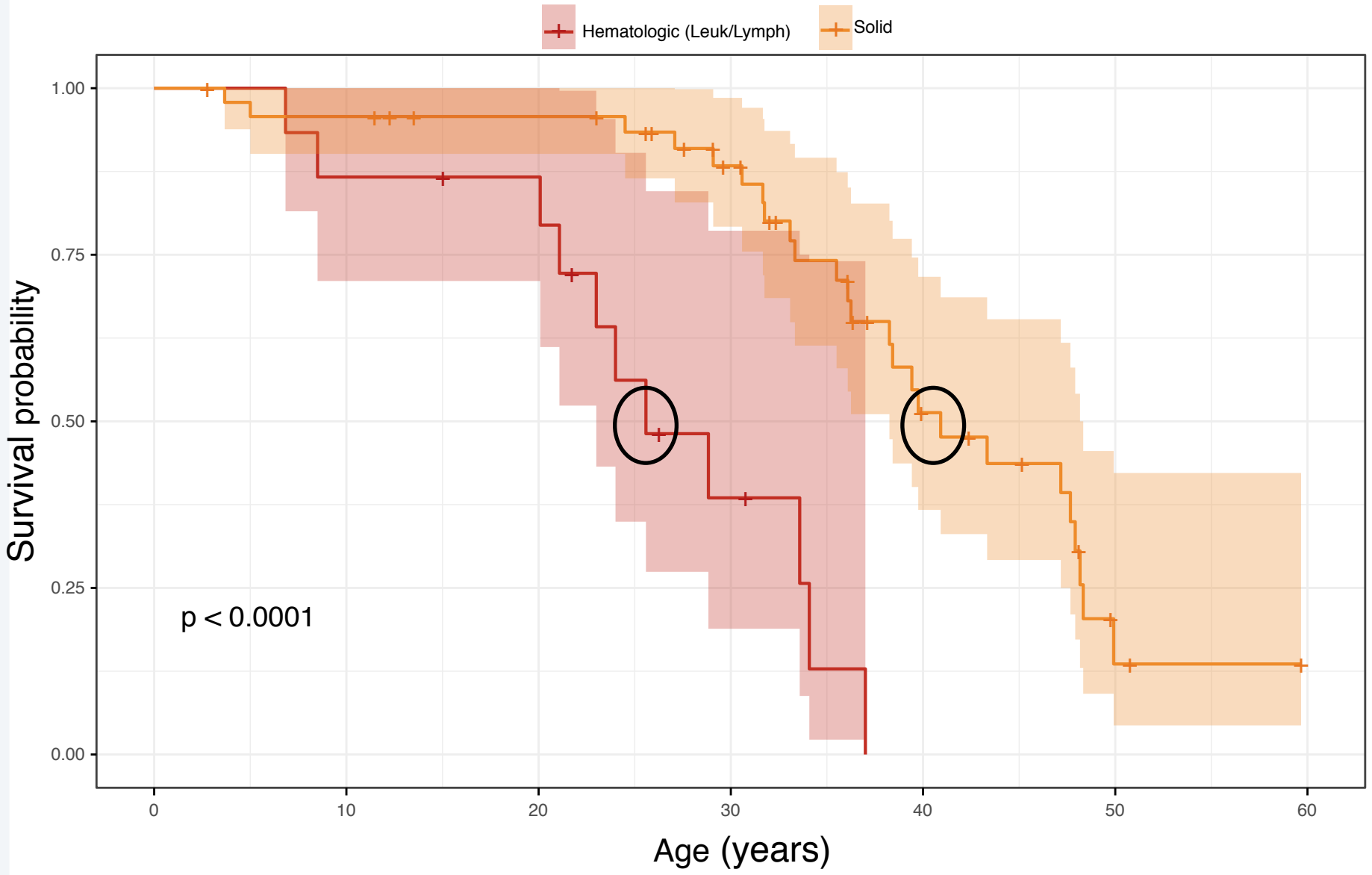


Survival Analysis

Cancer-related survival



Cancer-specific survival



Discussion

Younger age of diagnosis

Increased frequency

Multiple cancers

No differences based on genotype

Younger age of Diagnosis

National Cancer Institute:

- Leukemia: 67
  - ALL: 17
  - AML: 68
- Lymphoma (NHL, HL): 67, 39
- Colorectal: 67
- Breast: 62

Bloom syndrome:

- Leukemia: 15
  - ALL: 13.7
  - AML: 20
- Lymphoma: 15
- Solid tumors: 30
- Colorectal: 36
- Breast: 26

Cancer frequency: how does it compare to the general population?

SEER Cancer Statistics Review annual age adjusted average rate from 2013-2017

- Leukemia: 14.1 per 100,000 people
- Lymphoma (NHL): 19.6 per 100,000 people
- Colorectal cancer: 38.2 per 100,000 people

Bloom Syndrome cumulative rate by age 40

- Hematologic cancer: 23 per 100 people
- Solid tumors: 29.7 per 100 people

Multiple cancer types

147 registry participants with cancer:

- 49 (33%) with 2nd cancer
- 6 with 3rd cancer
- 1 with 4th cancer
- 6 with 5th cancer
- 3 with 6th cancer

Average 6 years between 1st and 2nd cancers

Differences based on genotype

Genotype groups:

- Ashkenazi founder variant blm^{Ash}
- Nonsense variants
- Missense variants

No significant difference between blm^{ash} and non-blm^{ash} age of first cancer diagnosis or survival

Conclusions

Personalized treatment regimens

Standardized tumor surveillance

Medical team guidance/teaching

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