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Project Requirements and Description

Requirements to submit AOI

Requirements to submit AOI (all answers must be "yes" to proceed)

A comprehensive review of previously published data has been completed	Yes
The specific aims are clear and focused	Yes
The investigator has appropriate experience and expertise to develop the concept proposal; if not, has identified a mentor or senior co-investigator.	Yes
The investigator agrees to develop an initial draft of the concept proposal within 6 weeks of approval of the AOI and to finalize the concept proposal within 6 months	Yes

Project Title Rare and Common Variation Associated with Primary Ovarian Insufficiency Risk in Survivors of Childhood Cancer

Planned research population (eligibility criteria)

For exome-based association analyses, we will use the combined cohort of SJLIFE and CCSS 5-year survivors with whole-exome sequencing (WES) data. Polygenic risk scores will be computed using matched SJLIFE whole-genome sequencing (WGS) data, CCSS WGS data (cancer diagnosed between 1987-1999), and CCSS imputed array data (cancer diagnosed between 1970-1986).

Proposed specific aims

Aim 1: Characterize the distribution of germline P/LP variants in putative POI-causative genes identified in the literature among long-term female survivors of childhood cancer by ovarian status and exposures to relevant treatment risk factors.

- The prevalence of P/LP variants in POI-causative genes among survivors will be evaluated against female SJLIFE community controls.

Aim 2: Evaluate whether carrying a P/LP variant in a putative POI-causative gene is associated with increased risk for developing POI or is modified by background POI risks conferred by common variants (e.g., PRS) or treatment exposures.

Aim 3: Conduct agnostic exome-wide association studies to identify novel loci associated with POI risk or that modify treatment-related POI risk among survivors.

Will the project require non-CCSS funding to complete? No

If yes, what would be the anticipated source(s) and timeline(s) for securing funding?

Does this project require contact of CCSS study subjects for:

Additional self-reported information	No
Biological samples	No
Medical record data	No

If yes to any of the above, please briefly describe.

What CCSS Working Group(s) would likely be involved? (Select all that apply)

Second Malignancy	
Chronic Disease	Secondary
Psychology/Neuropsychology	
Genetics	Primary
Cancer Control	
Epidemiology/Biostatistics	Secondary

Outcomes or Correlative Factors

Late Mortality	Correlative Factors
Second Malignancy	Correlative Factors

Health Behaviors

Tobacco	Correlative Factors
Alcohol	Correlative Factors

Physical Activity	Correlative Factors
Medical Screening	Correlative Factors
Other	Correlative Factors

If other, please specify

Psychosocial

Insurance	Correlative Factors
Marriage	Correlative Factors
Education	Correlative Factors
Employment	Correlative Factors
Other	Correlative Factors

If other, please specify

Medical Conditions

Hearing/Vision/Speech	
Hormonal Systems	Primary
Heart and Vascular	
Respiratory	
Digestive	
Surgical Procedures	Correlative Factors
Brain and Nervous System	
Other	

If other, please specify

Medications

Describe medications

Psychologic/Quality of Life

BSI-18	
SF-36	
CCSS-NCQ	
PTS	

PTG	
Other	

If other, please specify

Other

Pregnancy and Offspring	
Family History	
Chronic Conditions (CTCAE v3)	Primary
Health Status	

Demographic

Age	Correlative Factors
Race	Correlative Factors
Sex	Correlative Factors
Other	Correlative Factors

If other, please specify

Cancer Treatment

Chemotherapy	Correlative Factors
Radiation Therapy	Correlative Factors
Surgery	Correlative Factors

Anticipated Sources of Statistical Support

CCSS Statistical Center	No
Local Institutional Statistician	Yes

If local, please provide the name(s) and contact information of the statistician(s) to be involved.

Will this project utilize CCSS biologic samples?

Yes

If yes, which of the following?

Buccal cell DNA

If other, please explain

Other General Comments

Agree

I agree to share this information with St. Jude

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