

**ORPHAN DISEASE CENTER  
BLOOM SYNDROME  
2022 GRANT PROGRAM**

The ODC Grant Program will provide two-year grants to support research related to Bloom Syndrome. Up to four grants may be awarded.

## **Background**

Bloom syndrome is an ultra-rare multi system disorder that substantially increases the risk of developing cancer at an early age. While only about ~300 cases have been reported to date, Bloom syndrome is part of a group of rare DNA damage response and repair disorders. The gene *BLM*, located at 15q26.1, encodes for the BLM protein, a RecQ DNA Helicase family protein. A recurrent mutation, known as *BLMAsh*, is responsible for over 90% cases of Bloom syndrome among Ashkenazi Jews.

*BLM* pathogenic variants that cause Bloom syndrome results in DNA repair defects which results in a chromosome breaks and rearrangements. The abnormal DNA repair is responsible for the increased risk for cancer. Greater knowledge of its mechanisms will be beneficial not only for patients of Bloom syndrome but also could be translated to the clinic for cancer therapy

We are seeking grant applications to increase the rate of discovery of the mechanisms of Bloom and, importantly, lead to treatments and/or cures for Bloom Syndrome. While the RFA is broad in scope, priority will be given to grants that cover the following areas:

- 1) **Improve knowledge of tumor biology**, including, but not limited to tumor models, tumor sequencing to determine a tumor signature for Bloom Syndrome, molecular surveillance for tumors, as well as a centralized registry of validated tissue (coordination of sampling, storing and distribution of samples). **Two-year grant for \$150,000 (total cost).**
- 2) **Novel therapeutic approaches for Bloom Syndrome**, including, but not limited to, techniques in genome editing, RNA-based mechanisms, biologics, novel cell-based therapeutics, and development of novel therapeutic compounds, including through small molecule repurposing or screening against validated phenotypes in human cellular systems. **Two-year grant for \$100,000 (total cost).**

## Eligibility

**Non-profits.** All individuals holding a faculty-level appointment at an academic institution or a senior position at a non-profit institution or foundation are eligible to respond to this RFA.

**Commercial entities.** We will accept applications from commercial entities as long as a principal investigator of appropriate experience is identified.

Intellectual property created as a result of this funding will be assigned to the home institution of inventors which presumably would be employees of the company. We expect grants provided to commercial entities be matched dollar for dollar. Note that the budget should cover the entire scope of work and should include the ODC award and the company's match. We will need assurance in the grant contract that the company will diligently develop the project.

## Letter of Interest Instructions

All applicants must first submit a Letter of Interest (LOI) to be reviewed for consideration of a full application submission. Please visit our [website](#) to submit your LOI, which can also be found [here](#).

Format for the 1-page LOI:

- Project Title
- PI and Co-PIs, and associated institutions or organizations
- Overall goal of the project
- Why application is responsive to the RFA: please note that the applicant will not be invited to submit a full application if relevance to the RFA is not clearly described in the LOI.
- Brief background
- Specific Aims provided as a brief list
- Requested resources in terms of 2 years of funding. The total award amount is \$150,000 or \$100,000 (including direct and indirect costs). It is strongly preferred that applicant institutions waive indirect costs allowing the total amount to go towards research, but indirect costs up to a maximum of 10% could be accepted in exceptional cases.

**LOI Due Date:** LOI document is to be uploaded no later than 5pm (EST) on **Friday, April 15, 2022**, via [ONLINE FORM](#).

Applicants will be notified via email with a decision regarding their LOI, which, if successful, will invite the applicant to submit a full application.

**Full Application Instructions and Review Procedure**  
**NOTE: Full Application is by Invitation only after review of LOI**

Full Proposal Due Date: **Friday, May 27, 2022**, no later than 5pm (EST). Full application documents are to be uploaded on our [WEBSITE](#)

**FORMAT for documents:**

*Font and Page Margins:* Use Arial typeface, a black font color, and a font size of 11 points. A symbol font may be used to insert Greek letters or special characters. Use 0.5 inch margins (top, bottom, left, and right) for all pages, including continuation pages. Print must be clear and legible; all text should be single-spaced.

*Header:* There should be a header at the top right on all pages of the PDF indicating the full name of the PI (e.g., **PI: Smith, John D.**).

For your convenience, a continuation page template is included at the end of the application document.

*File names:* ALL files to be uploaded should start with the LAST NAME of the PI followed by the brief name of the document. Examples: SMITH CV, SMITH Cover Page, SMITH Budget. **If files are not labeled properly, you will be asked to resubmit the PDFs before your application can be considered.**

**CONTENT to be uploaded:**

**Cover Page/Checklist/Institutional Signature Page [PDF].** An application template will be provided.

**Biosketch/CV, with key personnel supporting the project (5 pages max). [PDF]**

The PI must include accurate and complete information regarding all other sources of grant support (current and pending), including title, abstract, annual and total amount of grant, inclusive funding period, and percent effort. PI should add a section listing key personnel working on the project.

**Detailed Budget and Justification. [combined into one PDF]**

Complete Excel budget sheet (to be provided). Describe justifications in a Word document including all subcontracts to co-investigators.

Proposed funding period: July 1, 2022 – June 30, 2024

Awarded amounts will not exceed Award Total

### Allowable direct costs

- Salary for PI
- Salary/stipend and related benefits for graduate student/postdoctoral fellow/technical support
- Travel (up to \$1500)
- Laboratory supplies and other research expenses

### Unallowable costs

- Consultant costs
- Tuition
- Professional membership dues
- Equipment >\$5,000
- General office supplies, institutional administrative charges (e.g., telephone, other electronic communication, IT network, etc.)
- Pre-award charges
- Any other expenses not directly related to the project
- Indirect cost

### **Research Plan** (5 pages max) and **Bibliography** (1 page max) **[combined into one PDF]**

Include the following sections: Specific Aims, Background and Significance, Preliminary Studies/Data, Research Design and Methods. Research plan should address the following questions: 1) Do you require access to reagents, animal models, patient blood samples, IRB/ethical board approvals, and/or equipment necessary to complete work? If so, please describe your plan to gain access within the time-frame of this grant period. 2) Have you identified qualified personnel to complete this project within the grant period? If not, please provide your plan to do so. Text citations should use a numbered format. Include all author names in the reference list.

### **Appendix [combined into one PDF]**

Limited to 5 pages of supplemental information pertaining to proposal or preliminary data only; a maximum of 3 relevant reprints are also acceptable. Include IRB and/or IACUC approval letters if relevant.

### **Project Disclosures and No Cost Extensions (NCE):**

- NCEs will be granted at the discretion of the ODC
- Awardees will be limited to 1 NCE request for their award
- Maximum NCE time awarded will be 6 months
- NCEs will be granted after a formal request through [this form](#) found on the ODC website prior to the NCE deadline with adequate justification

- If granted a NCE, you are still required to submit an interim scientific report 6 months into the duration of the original award period, regardless of your new project end date
  
- In your letter of interest, you will be required to certify that you have identified qualified personnel to complete this project within the grant period **PRIOR** to the start date of the award. If you have not, you will be required to provide your plan to engage said personnel. Only under extenuating circumstances will personnel issues be considered for NCE requests
  
- In your letter of interest, you will also be required to state whether or not you require access to reagents, cell lines, animal models, IRB/ethical board approvals, and/or equipment necessary to complete your work. If so, you will be required to describe your plan to gain access within the time-frame of this grant period

**Fund Disbursement:**

Funds will be issued through a cost reimbursement mechanism executed by purchase order from the University of Pennsylvania. Details of invoicing schedules and reporting requirements will be made available upon award. For additional information, please contact Deborah Requesens at [dreques@upenn.edu](mailto:dreques@upenn.edu).