Request for Proposal

NBSTRN Longitudinal Pediatric Data Resource and Genomics Warehouse (LPDR)

Issued February 6, 2019
Brief Statement Indicating Intent to Apply Requested by February 14, 2019
Responses Due by March 16, 2019
Award Date Estimated April 1, 2019

American College of Medical Genetics and Genomics
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General Information

Background

Each year the 4 million babies born in the US undergo laboratory and hospital-based screening for 60 genetic conditions. The laboratory portion of screening is conducted in state-based public health departments, and birthing hospitals screen newborns for hearing and congenital heart disorders. Screening is just the beginning and newborn screening (NBS) is considered a "system" involving prenatal education, neonatal screening, diagnosis, clinical referral, and treatment. The majority of NBS conditions are rare, and while NBS prevents morbidity and mortality, most of the conditions require lifelong clinical care and management. NBS began over 50 years ago and treatments have evolved from dietary interventions to medically complex and invasive procedures like stem cell transplant, gene therapy, intrathecal enzyme replacement using lumbar puncture, and genotype-directed therapies. Throughout the years, a better understanding of each of the 60 conditions has been possible because screening on a population basis enables unbiased ascertainment and the ability to longitudinally collect health information on each diagnosed newborn. These longitudinal natural history studies have profoundly improved knowledge of the diseases' etiology, pathophysiology, phenotypic heterogeneity, and comorbidities.

Federal Efforts

The *Eunice Kennedy Shriver* National Institute of Child Health and Human Development (NICHD) has played a role in NBS since the beginning but their leadership and role in NBS research were formalized in the NBS Saves Lives Act of 2008 and reaffirmed in the NBS Saves Lives Reauthorization Act of 2014. This legislation established and authorized the Hunter Kelly NBS Research Program within NICHD to carry out, coordinate, and expand research in NBS. The legislation also established and funded related efforts at the Centers for Disease Control and Prevention (CDC) and the Health Resources and Service Administration (HRSA), including continued support for the HHS Secretary Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC). ACHDNC included representation for genomics and industry in 2005, and ACHDNC is the body that reviews evidence and makes recommendations for which conditions should be part of nationwide screening. The current process is for any candidate condition, including genomics, to be submitted to ACHDNC for a formal evidence review. It is important to note that the NBS is state based, so although HHS recommends a uniform panel, each state determines which conditions to include in screening.

ACMG

Founded in 1991, the American College of Medical Genetics and Genomics (ACMG) is the only nationally recognized medical society dedicated to improving health through the clinical practice of medical genetics and genomics. ACMG provides education, resources and a voice for more than 2,200 biochemical, clinical, cytogenetic, medical and molecular geneticists, genetic

counselors and other healthcare professionals, nearly 80% of whom are board certified in the medical genetics specialties. The College's mission is to develop and sustain genetic initiatives in clinical and laboratory practice, education and advocacy. To obtain more information about ACMG please visit https://www.acmg.net.

NBSTRN

Through two consecutive five-year contracts, the ACMG has led the development and implementation of a key component of the Hunter Kelly NBS Research Program, the NBS Translational Research Network (NBSTRN). To learn more about NBSTRN, please visit https://www.nbstrn.org. The NBSTRN began as an effort to engage a variety of stakeholders across the NBS system and has matured into a dynamic and committed network comprised of researchers, public health professionals and clinicians with expertise in:

- The discovery and validation of novel technologies to screen and diagnose disease;
- The clinical care of newborns including execution of clinical trials and application of cutting-edge treatments and management strategies;
- Pilots of new technologies and treatments;
- The collection, analysis and dissemination of longitudinal health and genomic data; and
- The ethical, legal and social implications of NBS research.

In September 2018 ACMG was awarded a one-year contract as a base plus four option years to continue the NBSTRN. The NBSTRN contract outlines a series of resources, information technology tools and expert workgroups to be maintained and enhanced by ACMG.

RFP for NBSTRN Longitudinal Pediatric Data Resource and Genomics Warehouse (LPDR)

This RFP is intended to solicit proposals to subcontract portions of the enhancement and maintenance of the NBSTRN Longitudinal Pediatric Data Resource and Genomics Warehouse (LPDR https://nbstrn.org/research-tools/longitudinal-pediatric-data-resource). The RFP will focus on enabling the upload, analysis, transfer, and display of genomic and associated phenotypic data.

Contact for RFP

Responses to the RFP should be submitted to both Dr. Michael Watson (mwatson@acmg.net) and Dr. Amy Brower (abrower@acmg.net).

Timeline for RFP

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Suggested Project Phase	Timeline
Development- Phase I	Complete first phases of a genomics data repository with analytical and visualization capabilities and training draft manual by September 25, 2019
*Development- Phase II	A final product with training manual to be completed by May 31, 2020.
*Additional Enhancements	June 2020 - September 2020
*Maintenance	October 2020- September 2021
*Maintenance	October 2021- September 2022
*Maintenance	October 2022- September 2023

**Note: Development-Phase II and following phases will be awarded based on option years.*

Preparing and submitting a response

Contractor Information Required to be Included with Response

The proposal must contain the full legal name and signature of the authorized representative of the contractor. Telephone, fax, email and business address of the point of contact person needs to be included.

Response Submittal

A very brief statement indicating intent to apply is requested by 5:00 pm EST on February 14, 2019. Complete responses must be submitted **by 5:00 pm EST**, **on March 16, 2019**. Responses can be sent via email to Michael Watson mwatson@acmg.net and Amy Brower abrower@acmg.net. It is the responsibility of the applicant to ensure that the response is received by this deadline.

Responses should be limited to 20 pages of text. References, charts and graphics are not included as part of the page limit.

Submitters will receive a confirmation of receipt of their response by ACMG. ACMG may terminate or modify the RFP process at any time during the response period. All changes to the

RFP will be posted to the ACMG Website. No changes will be made to the RFP within one week of due date. Responses that are not received by the stated deadline shall be determined to be non-responsive and, at ACMG's discretion, may not be considered in the review of respondents.

Submission Timeline

Applications are due by 5:00 pm EST on March 16, 2019 and will be evaluated for a two-week period following submission. ACMG may request meetings or a teleconference with the respondents prior to announcement of final contract recipients.

General information, assumptions and references

This section on general issues, assumptions and preferences, while not requirements, provide an overview of the non-functional requirements and preferences that are important issues for consideration by respondents since they describe key aspects of the NBSTRN LPDR that would be most appropriate.

- Develop the technical requirements of a genomics data module within the LPDR in collaboration with the NBSTRN team.
- Both the interactive website and the web interface to the LPDR must continue to meet modern web standards and be browser independent; must not require any plug-ins so it will work on every platform; must be HTML5 and 508 compliant; and must operate within a FISMA Moderate IT environment.
- Complete first phases of a genomics data repository with analytical and visualization capabilities by September 25, 2019. A final product with functionality to upload data to 3rd party databases (e.g. dbGaP, DASH) to be completed by May 31, 2020.
- The respondent can subcontract out any portion of the website or repository creation with prior approval from ACMG and NICHD, but is accountable for all project deadlines.
- Ability for the NBSTRN team, including their web developer, to provide incremental adjustments and feedback.
- Participation in the NBSTRN Steering Committee by regularly attending committee calls.

General requirements of proposal

Instructions: Respondents must provide a point-by-point response to each of the General Requirements specified in this RFP. For each requirement, respondents should provide documentation as requested. The documentation demonstrates they understand the requirement and agree to the requirement. Responses to general requirements must be in sequence and numbered as they appear in the RFP.

Executive summary

Provide a narrative summary of the proposal being submitted. The summary should identify the services that are being offered in the proposal including their design and development.

Organization/management capabilities and financial stability

Respondents should describe their company's organizational structure and provide a brief history of their business.

References

Respondents should include a list of clients, organizations or institutions that can be used as references. At least one reference should be from an organizational client where a similar project was developed. Selected references may be contacted to determine the quality of work performed, competency of personnel assigned to the project, etc. The results of the reference checks will be provided to the evaluation and may be used in scoring the written proposal.

These references should be capable of verifying information supplied by the respondent in their proposal. The data required for each reference should include:

Company Name
Address
Contact person name, email, phone number and address
Description of the type of work performed and the results

Examples of successful implementation

Respondents should include a description of their relevant experience developing and implementing web-based resource systems. Please provide information about specific platforms used, dates implemented, and customer satisfaction. Testimonials are encouraged.

Staff qualifications

The respondent must provide a project team organizational chart, followed by resumes of key personnel that will be assigned to the system development and implementation. Each key personnel must be an employee of the respondent or be identified as a subcontractor. Include roles, responsibilities and estimated time allocated for each key personnel.

Budget

A budget should be presented for all phases of this project including development and maintenance. The respondent must provide a cost application to accomplish the scope of work. The budget must encompass all design, production, and software acquisitions necessary for the development and maintenance of the website and data repository. Please include any costs associated with additional maintenance, support, upgrades and long-term annual maintenance. This budget is not binding but will provide information to ACMG for project development.

Technical requirements

The respondent must discuss the scope of the project with regards to enhancing and maintaining the LPDR in a FISMA Moderate IT environment.

1. *Genomics Data Repository -* The applicant for the enhanced LPDR must describe how his/her organization will meet following specifications.

    a. Design and implement a scalable, 508-compliant, FISMA Moderate compliant online environment for NBSTRN users to store, analyze, visualize and share genomic and phenotypic data. Note that this includes password protected account access as well as a method of two-factor authentication.

    b. Outline the requirements and specifications of the hosting environment.

    c. Storage of case level data that includes information about samples, variants, phenotypes, -omics data, NBS screening results, NBS diagnosis results, and other data relevant to the NBSTRN user base.

    d. A framework to import clinical, phenotypic and genomic data that includes data cleaning, personal health information monitoring, consent compliance, and summary reports.

    e. A web-based platform that allows NBSTRN users to query, analyze, share and report accumulated phenotypic and genotypic data using a robust permissioning scheme which allows for case-level and project-level data access.

    f. A cohort builder with an easy to use visual user interface for querying and sharing datasets across NBSTRN users and projects at cohort, subject and sample levels. The

cohort builder should work in conjunction with the query and analysis tool and not require knowledge of any database querying languages or underlying structure.

g. A visualization framework that enables NBSTRN users to visualize aggregate data, drill down to case level data, navigate across genomic regions, and retrieve NBSTRN annotations for individual genomic elements.

h. Identifiable and/or de-identified data upload capability to 3rd party platforms including NIH dbGaP and Data Commons, ClinVar/ClinGen, NICHD Data and Specimen Hub (DASH) and others as determined by the NBSTRN team.

i. Enable uploading of both phenotypic and genomic data, and transfer of data to dbGaP as appropriate, including data from the Newborn Sequencing in Genomic Medicine and Public Health (NSIGHT) effort.

j. Incorporate FISMA Moderate IT criteria in design and execution, including providing materials for ACMG FISMA Moderate submissions as needed.

k. Conduct weekly and/or biweekly meetings with research teams using the LPDR and implement a support desk for user support, training modules for each clinical domain and application

l. Develop and disseminate a written and online guide to NBS researchers for best practices for genomic data.

m. Capture and report website usage data using an analytics platform such as Google Analytics as well as tool usage data including the number of users, sessions, projects, etc. to the NBSTRN team on a monthly, quarterly and annual basis.

n. Participate in and develop communications and marketing materials as appropriate.

o. Ensure compatibility with other NIH and NICHD websites (e.g. https://dash.nichd.nih.gov/; https://cde.nlm.nih.gov/; https://www.ncbi.nlm.nih.gov/clinvar/)

p. Ensure compatibility with the REDCap projects already included in LPDR. Note that the REDCap data dictionaries are available upon request.

q. Maintenance of an online documented code repository.

r. Delivery to ACMG, upon the expiration date of the contract, all source code and object code developed, modified, and/or enhanced under this contract.

s. Provision of all documentation to properly compile the source code and related data on ACMG's internal infrastructure with hardware requirements.

2. *Security Requirements*

    a. The respondent must include requirements for security and discuss processes that will be developed to ensure compliance with HHS Enterprise Performance Life Cycle (EPLC) requirements.

    b. Comply with the Privacy Act Requirements.

    c. The respondent must include requirements for security and compliance and discuss processes that will be developed to ensure data integrity.

    d. Hosting location and storage/backup details should be discussed. Hosting location should take into account FISMA and FIPS 140-2 requirements as well as proposed linkages between tools and websites.

3. *Other Specifications*

    a. Respondent must include a training plan, to include administrator/configuration training as well as user training, and should address training at initial implementation and any training necessary as a result of new development or ACMG staffing changes.

    b. Respondent must discuss their plan for making a test environment available for NBSTRN staff testing and validation.

Evaluation Criteria and Subcontracting Reports

Applications will be evaluated based on budget, demonstration of technical understanding of the application, and creativity in the response. ACMG will evaluate each application based on the description of the General, Technical and Budget requirements listed in the RFP. The evaluation team will review each applicant and provide recommendations to the NBSTRN Director.

Subcontracting Reports - The subcontractor should provide the following deliverables to the NBSTRN team using the format specified below:

a. Cover page

    i. All reports shall include a cover page prepared in accordance with the following format:

1. Contract Number and Project Title
2. Period of performance being reported
3. Contractor's name and address, telephone and fax numbers
4. Author(s)
5. Date of submission
6. Delivery Address

b. Monthly reports

This report shall include: An analysis of web traffic and tool use, budget update, a very brief overview of the work completed for the reporting period and/or justification for intended work not completed

c. Quarterly progress report

This report shall include: A statement of intended work for the reporting period, a brief overview of the work completed for the reporting period and/or justification for intended work not completed, and a brief overview of any problems (technical or financial) that occurred during the current reporting period and proposed solutions.

d. Annual Progress Report

This report includes a summation of the results of the entire contract work for the period covered. An Annual Progress Report will not be required for the period when the Final Report is due. A Quarterly Progress Report shall not be submitted when an Annual Progress Report is due. This report should include:

- Summary of work completed
- Maintenance and proposed upgrades
- Brief overview of any problems (technical or financial)
- Summary of access and use analytics
- Proposed future efforts
- Summary of changes in staffing

e. Final Report

This report includes a summation of work performed and the results obtained for the entire contract period of performance. This report shall be in sufficient detail to describe comprehensively the results achieved. An annual report will not be required for the period when the Final Report is due.

Disclaimer

This RFP is neither an agreement nor an offer to enter into an agreement with any respondent. Once evaluation is complete, ACMG may choose to enter into a definitive contract with the selected RFP applicant.

Each respondent shall bear its own costs associated with or relating to the preparation and submission of its application. All such costs and expenses will remain with such respondent and ACMG will not be liable for these or for any other costs or other expenses incurred by a respondent in preparation or submission of its application, regardless of the conduct or outcome of the response period or the selection process.

Note

Subcontractors may respond to more than one RFP issued as part of the NBSTRN. If responding to more than one RFP, please explain the combined approach and any cost reduction.