U.S. Department of Health and Human Services
Centers for Disease Control and Prevention

*Print Date:* 5/17/22

**Title:** ED3N Newborn Screening Data Platform - Enhancing Disease-Detection in Newborns

**Project Id:** 0900f3eb81e2125c

**Accession #:** NCEH-DLS-6/7/21-4d4b6

**Project Contact:** Amy M Gaviglio

**Organization:** NCEH/ATSDR/DLS/NSB

**Status:** **Pending Regulatory Clearance**

**Intended Use:** **Project Determination**

**Estimated Start Date:** 09/01/2021

**Estimated Completion Date:** 12/31/2030

**CDC/ATSDR HRPO/IRB Protocol #:**

**OMB Control #:**

**Source System #:** 2020-0102

## Determinations

| Determination | Justification | Completed | Entered By & Role |
|---|---|---|---|
| HSC:<br>**Does NOT Require HRPO Review** | Not Research - Public Health Surveillance<br><br>*45 CFR 46.102(l)(2)* | 11/19/21 | Davis_Stephanie I. (sgd8) CIO HSC |
| PRA: | | | |

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|---|---|---|---|
| **PRA Applies** | | 11/19/21 | Davis_Stephanie I. (sgd8) CIO OMB / PRA |

# Description & Funding

## Description

**Priority:** Standard

**Date Needed:** 12/03/2021

**Determination Start Date:** 11/19/21

**Description:** Every year almost four million infants are born in the United States. Almost all newborns are screened by state and territorial newborn screening programs for diseases that can cause severe disability or death when untreated. The Newborn Screening and Molecular Biology Branch (NSMBB) at the CDC supports newborn screening (NBS) programs by creating quality assurance materials, developing new methods, and providing technical assistance and technology transfer for biochemical and molecular techniques aimed at identifying newborns at risk for congenital diseases. Both NSMBB and newborn screening programs are experiencing increased data analytic challenges associated with the continued expansion of the number of newborn diseases screened, the increased complexity of disease phenotype, and the difficulties in correlating disease markers with disease risk. To overcome analytic challenges and harmonize testing and interpretative processes, NSMBB and contractors are developing a Newborn Screening Data Platform. This Data Platform can serve as a secure, central, and national resource for the US and international newborn screening community, containing a data repository and data sharing resources for newborn screening data. The Data Platform will include three functional modules for biochemical, molecular, and clinical data and will support NBS Program best practices through 1) The evaluation of daily routine workflows at the individual patient level to enhance disease detection, 2) The exploration of aggregate data (e.g., pooled analyte, variant, and/or clinical data with potential identifiers removed) to test new workflows based on national normalized trends, and 3) The education of NBS program personnel on data analytic skills and best practices, and through a resource library of peer reviewed literature. Biochemical: Data for this module includes analyte values from tandem mass spectrometry and other biochemical tests, methods and instrumentation used, normalization techniques, and test cutoff values. Benefits of this data may include the harmonization of analytic values across NBS programs, assessment of demographic, analytic, and geographic influences on screening outcomes, comparison of test cutoff values, and the development of novel interpretative algorithms. Molecular: Data for this module includes variants identified through sequencing, the platform used, and the interpretation of the variants assigned by NBS programs. Benefits of this data may include improving variant interpretation, providing consistency around variant interpretation, correlation of biochemical and molecular data, and standardizing variant interpretation changes across programs. Clinical: Data for this module includes clinical information necessary for tracking health outcomes for infants screening positive for and/or afflicted by an NBS disorder. Benefits of this data may include the comparison of case definition algorithms and interpretation, the implementation and refining of public health surveillance case definitions, and creating an urgency index for NBS results. In addition, the data hub will collect demographic information that will crosscut between all three modules. Data for the demographic section include descriptive information about the infant and the mother that is necessary for appropriate interpretation of NBS results. An ATO and Privacy Impact Assessment have been completed and approved. This is a request to publish a 60-day Federal Register Notice.

**IMS/CIO/Epi-Aid/Lab-Aid/Chemical Exposure Submission:** No

**IMS Activation Name:** Not selected

**Primary Priority of the Project:** Not selected

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|---|---|
| **Secondary Priority(s) of the Project:** | Not selected |
| **Task Force Associated with the Response:** | Not selected |
| **CIO Emergency Response Name:** | Not selected |
| **Epi-Aid Name:** | Not selected |
| **Lab-Aid Name:** | Not selected |
| **Assessment of Chemical Exposure Name:** | Not selected |
| **Goals/Purpose** | The NSMBB ED3N Data Platform seeks to be a resource that NBS programs will use to improve and harmonize disease detection in newborn screening (NBS) programs. The state-based nature of NBS has led to a lack of uniformity in how NBS is administered and accomplished throughout the country, especially in the analytical and post-analytical phases of the NBS system. Additionally, the increasingly technological and interpretative complexities of NBS assays and diseases necessitates an innovative solution to compile data and share experiences across the country. National NBS harmonization and improved disease detection will be accomplished through a National Data Platform developed and operated by the CDC#s NSMBB, in conjunction with Booz Allen Hamilton and CI International. The Data Platform will serve as a secure, centralized resource for newborn screening partners to analyze and share biochemical and molecular data. It will support laboratory best practices through standardized workflows and robust, validated analytical tools. The Newborn Screening and Molecular Biology Branch (NSMBB) Enhancing Data-driven Disease Detection in Newborns (ED3N) Newborn Screening Data Platform (hereafter called ED3N) will help meet NSMBB's Mission of "Better detection of newborn disorders by implementing advanced technologies and capabilities that improve screening test performance, test result interpretation, quality assurance programs, training and technology transfer and by assisting state programs to expand screening to new conditions of high priority." The purpose of ED3N is to serve as a secure, central, and national resource for the state newborn screening community, containing a data repository and data sharing resources for newborn screening data. ED3N will collect infant and specimen level newborn screening (NBS) data to help overcome analytic challenges and harmonize testing and interpretive processes in order to reduce false positive and negative newborn screening results. |
| **Objective:** | The completion of the National Data Platform (ED3N) will allow for the NSMBB to meet several of the Branch#s priorities and address its congressional mandate. The expected impact of successfully completing this project is a site for sharing and analyzing molecular, biochemical, and clinical data that will enable newborn screening partners to accurately identify and appropriately act on newborns identified as being at-risk for life and health-threatening diseases. |
| **Does this project include interventions, services, or policy change work aimed at improving the health of groups who have been excluded or marginalized and /or decreasing disparities?:** | Yes |
| **Project does not incorporate elements of health equity science:** | Not Selected |
| **Measuring Disparities:** | Not Selected |
| **Studying Social Determinants of Health (SDOH):** | Not Selected |
| **Assessing Impact:** | Yes |
| **Methods to Improve Health Equity Research and Practice:** | Not Selected |
| **Other:** | Not Selected |

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|---|---|
| **Activities or Tasks:** | Secondary Data or Specimen Analysis ; Purchase, Use, or Transfer of Information, Data, Biospecimens or Materials ; Programmatic Work |
| **Target Populations to be Included/Represented:** | General US Population ; Children ; Neonates |
| **Tags/Keywords:** | DLS 2020-0102 ; NSMBB ED3N Data Warehouse |
| **CDC's Role:** | CDC employees or agents will obtain or use identifiable (including coded) private data or biological specimens ; CDC employees will participate as co-authors in presentation(s) or publication(s) ; CDC employees will provide substantial technical assistance or oversight ; CDC is provider of materials/services TO an institution ; CDC is recipient of private data/specimens FROM an institution |
| **Method Categories:** | Needs Assessment; Public Health Assessment; QA/QI; Surveillance Support; Technical Assistance |
| **Methods:** | ED3N will be developed in the AWS Gov Cloud using an iterative and phased release approach starting with the Molecular Module and ending with the Clinical Module. Throughout development, beta and pilot testing will be done with select states (less than 9 programs total) utilizing mock data and feedback obtained. Determination for this activity is also requested as part of this submission. Feedback will then be prioritized and additional development conducted prior to next pilot test. Development will utilize an Agile approach with Sprints. Once a Minimal Viable Product is developed, it will be moved to production. Access to the production site and consumption of actual patient data will require SAMS access and signed Data Sharing Agreements. Stakeholder engagement will be accomplished using a series of small discussion groups, update webinars, and a webpage. Small discussion groups will include less than 9 programs and/or will be convened using existing mechanisms through APHL. This activity, along with the webinars and webpage is under a separate STARS entry (0900f3eb81d44942). |
| **Collection of Info, Data or Biospecimen:** | ED3N will collect, store, or share the following types of information: * Newborn and parental demographic information (e.g., date of birth, time of birth, sex, birthweight, gestational age, etc.) * Specimen-specific information (e.g., date and time of collection, site of collection, etc.) * Newborn screening laboratory values and result interpretations (including all biochemical and molecular screening results) * Clinical evaluation, diagnostic, and treatment information (only for certain cases, when applicable) * Geographical information (e.g., zip code, state) User information will not be gathered directly by ED3N, as SAMS will be utilized to authenticate and allow user access. ED3N will consist of three functional modules: 1) Biochemical, 2) Molecular, and 3) Clinical Data sources for ED3N will include Newborn Screening Programs/Departments of Health as well as treating/managing clinicians. This is further delineated below in each module section. Data collected will not contain identifiers such as name, medical record number, or address. Dates (e.g., birth date and testing dates) will be collected. 1) Biochemical: Data for this module will flow from Newborn Screening Programs and include analyte values from tandem mass spectrometry and other biochemical tests, methods and instrumentation used, normalization techniques, and test cutoff values. 2) Molecular: Data for this module flow either from Newborn Screening Programs or from treating clinicians and will include variants identified through molecular analysis and the interpretation of the variants assigned by NBS programs or other diagnostic laboratories 3) Clinical: Data for this module will flow either from Newborn Screening Programs or from treating clinicians' electronic medical records and will include clinical information necessary for tracking final diagnoses and health outcomes for infants afflicted by an NBS condition. |
| **Expected Use of Findings/Results and their impact:** | The completion of the National Data Platform (ED3N) will allow for the NSMBB to meet several of the Branch#s priorities and address its congressional mandate. The expected impact of successfully completing this project is a site for sharing and analyzing molecular, biochemical, and clinical data that will enable newborn screening partners to accurately identify and appropriately act on disorders in newborns. An Assurance of Confidentiality is in process. |
| **Could Individuals potentially be identified based on Information Collected?** | Yes |
| **Will PII be captured (including coded data)?** | Yes |
| **Does CDC have access to the identifiers (including coded data)?:** | Yes |

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| **Is this project covered by an Assurance of Confidentiality?** | No |
| **Does this activity meet the criteria for a Certificate of Confidentiality (CoC)?** | Yes |
| **Is there a formal written agreement prohibiting the release of identifiers?** | Yes, *see supporting info* |

### Funding

| Funding Type | Funding Title | Funding # | Original Budget Yr | # Years Award | Budget Amount |
|---|---|---|---|---|---|
| CDC Funding Intramural | Project Funding and Partners | | | | $700000 |

## HSC Review

## Regulation and Policy

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|---|---|---|
| **Do you anticipate this project will be submitted to the IRB office** | No | |
| **Estimated number of study participants** | | |
| **Population - Children** | N/A | Protocol Page #: |
| **Population - Minors** | N/A | Protocol Page #: |
| **Population - Prisoners** | N/A | Protocol Page #: |
| **Population - Pregnant Women** | N/A | Protocol Page #: |
| **Population - Emancipated Minors** | N/A | Protocol Page #: |

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| **Suggested level of risk to subjects** | |
| **Do you anticipate this project will be exempt research or non-exempt research** | |

## Requested consent process waviers

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|---|---|
| **Informed consent for adults** | No Selection |
| **Children capable of providing assent** | No Selection |
| **Parental permission** | No Selection |
| **Alteration of authorization under HIPPA Privacy Rule** | No Selection |

## Requested Waivers of Documentation of Informed Consent

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|---|---|
| **Informed consent for adults** | No Selection |
| **Children capable of providing assent** | No Selection |
| **Parental permission** | No Selection |

## Consent process shown in an understandable language

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|---|---|
| **Reading level has been estimated** | No Selection |
| **Comprehension tool is provided** | No Selection |
| **Short form is provided** | No Selection |
| **Translation planned or performed** | No Selection |
| **Certified translation / translator** | No Selection |
| **Translation and back-translation to/from target language(s)** | No Selection |
| **Other method** | No Selection |

## Clinical Trial

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| **Involves human participants** | No Selection |
| **Assigned to an intervention** | No Selection |
| **Evaluate the effect of the intervention** | No Selection |
| **Evaluation of a health related biomedical or behavioral outcome** | No Selection |

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| **Registerable clinical trial** | No Selection |

### Other Considerations

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| **Exception is requested to PHS informing those bested about HIV serostatus** | No Selection |
| **Human genetic testing is planned now or in the future** | No Selection |
| **Involves long-term storage of identfiable biological specimens** | No Selection |
| **Involves a drug, biologic, or device** | No Selection |
| **Conducted under an Investigational New Drug exemption or Investigational Device Exemption** | No Selection |

## Institutions & Staff

### Institutions

| Name | FWA # | FWA Exp Date | IRB Title | IRB Exp Date | Funding # |
|---|---|---|---|---|---|
| Vermont Department of Health | | | | | |
| West Virginia Department of Health and Human Resources | | | | | |
| CI International, Center for Public Health Innovation | | | | | |
| Delaware Health and Human Services | | | | | |
| Georgia Department of Public Health | FWA00024279 | | | | |
| North Carolina Department of Health and Human Services | | | | | |
| Guam Department of Public Health & Social Services | | | | | |
| Kentucky Department of Health | | | | | |
| Alabama Department of Public Health | FWA00003283 | 12/16/24 | Alabama Dept Public Hlth-Panel A IRB #1 | 06/04/23 | |
| Alaska Division of Public Health | FWA00018306 | 11/12/24 | U Alaska Anchorage IRB #1 | 08/05/24 | |

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| Arizona Department of Health Services, Human Subjects Review Board | FWA00002311 | 01/03/25 | Arizona Department of Health Services, Human Subjects Review Board IRB #1 | 01/08/23 | |
| Arkansas Dept of Hlth | FWA00002961 | 04/12/22 | | | |
| Booz Allen Hamilton Inc. | FWA00014089 | 12/07/23 | Advarra, Inc. IRB #1 | 08/17/24 | |
| California Health & Human Services Agency | FWA00000681 | 04/16/24 | | | |
| Colorado Department of Public Health & Environment | FWA00003044 | 03/02/25 | | | |
| Connecticut Department of Public Health | FWA00026243 | 11/21/22 | | | |
| District of Columbia Dept of Hlth | FWA00003034 | 09/26/22 | | | |
| Hawaii State Department of Health | FWA00000118 | 05/05/26 | | | |
| Idaho Dept of Hlth & Welfare | FWA00010222 | 02/13/23 | | | |
| Illinois Department of Public Health | FWA00002005 | 07/01/24 | | | |
| Indiana State Department of Health | FWA00001959 | 03/18/26 | Indiana U Indianapolis IRB #1A - (IRB-01) | 03/02/23 | |
| Iowa Department of Public Health | FWA00027243 | 08/29/23 | U of Northern Iowa Graduate Coll IRB #1 | 11/12/22 | |
| Kansas Department of Health & Environment | FWA00000383 | 04/15/24 | Kansas Dept Hlth & Environment IRB #1 | 02/06/22 | |
| Louisiana Department of Health | FWA00026681 | 03/19/23 | Louisiana Department of Health IRB #2 | 03/29/24 | |
| Maryland Department of Health | FWA00002813 | 02/25/26 | Maryland Dept of Hlth & Mental Hygiene IRB #1 | 03/11/24 | |
| Massachusetts Department of Public Health | FWA00000786 | 03/04/25 | | | |
| Michigan Department of Health and Human Services | FWA00007331 | 09/12/24 | | | |
| Minnesota Department of Health | FWA00000072 | 03/24/22 | | | |
| Mississippi State Department of Health | FWA00021429 | 05/14/24 | Mississippi State Department of Health IRB #1 | 09/22/23 | |
| Missouri Dept of Hlth & Senior Services | FWA00001948 | 07/28/25 | Missouri Dept of Hlth & Senior Services IRB #1 | 03/10/24 | |
| Montana Dept Public Hlth & Human Services | FWA00005012 | 07/12/23 | | | |
| Nebraska Department of Health and Human Services | FWA00003679 | 10/15/24 | U of Nebraska Med Ctr IRB #1 | 09/21/24 | |

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| Nevada State Hlth Division | FWA00009908 | 05/24/26 | U Nevada - Reno IRB #1 Biomed | 02/12/24 | |
| New Hampshire Division of Public Health Services | FWA00004372 | 04/01/26 | | | |
| New Jersey Department of Health | FWA00029683 | 06/24/25 | Rowan University School of Osteopathic Medicine IRB #1 | 02/22/24 | |
| New Mexico Department of Health | FWA00030150 | 09/29/25 | New Mexico State U IRB #1 | 07/03/22 | |
| New York City Dept of Hlth & Mental Hygiene | FWA00009459 | 08/06/25 | New York City Dept of Hlth & Mental Hygiene IRB #1 | 08/06/23 | |
| North Dakota Dept Hlth | FWA00005027 | 02/17/22 | | | |
| Ohio Dept of Hlth | FWA00001963 | 01/29/25 | Ohio Dept of Hlth IRB #1 | 01/15/24 | |
| Oklahoma State Dept of Health | FWA00000183 | 08/23/26 | Oklahoma State Dept Hlth IRB #1 | 08/23/24 | |
| Oregon Health Authority - Public Hlth Division | FWA00000520 | 10/23/24 | DHS-Health Svces/Multnomah Co Public Hlth IRB #1 | 12/11/22 | |
| Pennsylvania Department of Health | FWA00028491 | 07/10/24 | Pennsylvania Dept Hlth IRB #1 | 06/11/22 | |
| Puerto Rico Dept Hlth | FWA00006333 | 02/17/22 | | | |
| Rhode Island Department of Health | FWA00006141 | 09/09/25 | RHODE ISLAND STE DEPT HLTH IRB #1 | 04/03/22 | |
| South Carolina Dept Hlth & Environmental Control | FWA00003803 | 08/30/22 | | | |
| South Dakota Dept. of Health | FWA00006900 | 01/14/26 | Centers for Disease Control & Prevention IRB #1 - A | 10/08/24 | |
| State of Florida, Dept of Hlth | FWA00004682 | 02/28/25 | | | |
| State of Maine Department of Health and Human Services | FWA00016239 | 03/11/25 | | | |
| Tennessee Department of Health | FWA00000379 | 02/07/22 | | | |
| Texas Dept of State Hlth Services | FWA00008616 | 02/28/25 | | | |
| Utah Department of Health | FWA00024843 | 02/02/26 | Utah Dept Hlth IRB #1 - Public Hlth | 06/03/24 | |
| Virginia Dept of Health | FWA00000274 | 03/29/24 | | | |
| Washington State Department of Health | FWA00000327 | 10/12/26 | | | |
| Wisconsin Dept of Health Services | FWA00002517 | 01/20/24 | | | |
| Wyoming Dept Hlth | FWA00005562 | 03/29/24 | Wyoming Dept Hlth IRB #1 | 08/05/24 | |

### Staff

| Staff Member | SIQT Exp. Date | CITI Biomedical Exp. Date | CITI Social & Behavioral Exp. Date | CITI Good Clinical Practice Exp. Date | Staff Role | Email | Phone | Organization |
|---|---|---|---|---|---|---|---|---|
| Amy Gaviglio | 08/10/2023 | 05/12/2025 | | | Project Officer | | - - | NEWBORN SCREENING BRANCH |
| Carla Cuthbert | 09/24/2023 | 08/21/2024 | | | Project Officer | | 770-488-7571 | DIVISION OF LABORATORY SCIENCES |
| Charles Pickens | 09/04/2023 | | | | Program Lead | ogh6@cdc.gov | 404-498-5586 | NEWBORN SCREENING TEAM 2 |
| Konstantions Petritis | 09/29/2023 | 03/07/2025 | | | Program Lead | nmo3@cdc.gov | 404-498-5248 | NEWBORN SCREENING BRANCH |
| Suzanne Cordovado | 09/28/2023 | 12/10/2021 | | | Program Lead | snc4@cdc.gov | 770-488-4048 | NEWBORN SCREENING BRANCH |

## Data

### DMP

**Proposed Data Collection Start Date:** 12/31/21

**Proposed Data Collection End Date:** 12/31/30

**Proposed Public Access Level:** Restricted

*Restricted Details:*

**Data Use Type:** Data Sharing Agreement

**Data Use Type URL:**

**Data Use Contact:**

Data is at an individual level and is secondary use from a mandated public health program where consent is not obtained. Access will be restricted to screening programs and clinicians needing to use the data for the purpose of patient care. NBS programs will be

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| **Public Access Justification:** | involved in determining who has access to their data. Data will be submitted to the NBS Data Hub by NBS programs, PCPs, and clinical specialists. Submitted data includes biochemical, molecular, and diagnostic/clinical information that could potentially identify an individual when combined with certain demographic data points. In addition, NBS programs consider NBS data sensitive in that it has the potential to compare performance between NBS programs. All data sharing at the record level is restricted to the DSA ratified between the CDC and each individual state or territorial NBS program De-identified, aggregate data will be made available to the public. |
| **How Access Will Be Provided for Data:** | Access will be provided through SAMS to individuals identified and authenticated by the data provider (e.g., NBS program) and CDC. ED3N will utilize defined user roles with specific data access rights and functionality and the ability for NBS programs to assign users to each role. In addition to user roles, each user may be assigned to data access groups (DAGs). Once assigned to a DAG the user can only interact with record-level data belonging to that DAG. System Administrators are the only users with the ability to assign and reassign users to jurisdictional DAGs. Both System Administrators and State Administrators can assign and reassign participants to other DAGs described below. The first level of DAG users may be assigned to is the jurisdiction for which the user represents. A user may be assigned to more than one DAG if that individual is a representative of more than one jurisdiction. The second level of DAG users may be assigned to is the program classification for which the user requires read and/or write access: 1) CDC (universal record access), 2) NBS System (state-level record access), 3) NBS Lab (state-level record access), 4) NBS Follow-Up (state-level record access), and 6) Clinicians (patient-level records). It is possible that a single user may be assigned to multiple DAGs from this level. The third level of DAG users may be assigned to is the data module(s) for which the user requires read and/or write access: 1) Biochemical, 2) Molecular, and 3) Clinical. A user may be assigned to more than one data module DAG. Users will not have access to record-level data from DAGs they are not assigned to but will have access to de-identified aggregate data within the ED3N user interface. System Administrator Role: The CDC will provide personnel assigned to the System Administrator role having universal access to all data records. These individuals can add and remove users at any level and assign them into the appropriate DAGs. System Administrators also function as national data quality monitors and are responsible for reviewing and verifying data submitted from each NBS program. Program Administrator Role: NBS programs will identify one or more individuals to serve in the Program Administrator user role (i.e., the individual(s) who manage all users for the jurisdiction). These individuals can assign and edit the rights granted to other jurisdiction-level users within their state or territory. Program Administrators have full read-write access to program-level records submitted by their NBS program. The State Administrators will be assigned to all three data module DAGs. NBS programs must notify the CDC of any change to the individuals identified as the State Administrators in writing. Any change will be effective no more than 30 days after the CDC receives the change request. Program Data Analyst Role: Users assigned to the State Data Analyst Role generally serve as data analysts for state or territorial NBS programs and require full read-write access rights to state-level records submitted by their state or territorial NBS Program. The State Data Analysts are assigned to all three data module DAGs. Program Data Entry Role: Users assigned to this role can add and edit records only for their assigned state or territorial DAG and data module DAG. Program Data Viewer Role: Users assigned to this role can read record level data for their assigned state or territorial DAG and data module DAG. |
| **Plans for Archival and Long Term Preservation:** | |

## Spatiality

Spatiality (Geographic Locations) yet to be added .....

## Dataset

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|---|---|---|---|---|---|---|---|---|---|

| Dataset Title | Dataset Description | Data Publisher /Owner | Public Access Level | Public Access Justification | External Access URL | Download URL | Type of Data Released | Collection Start Date | Collection End Date |
|---|---|---|---|---|---|---|---|---|---|
| Dataset yet to be added... | | | | | | | | | |