

Birth Defects Surveillance Methods in the U.S. 2022-2024: A Landscape Analysis Describing Variation in Surveillance Methods Across U.S. Programs

Victoria Markwalter¹, Nicole Lindsey¹, Jason Salemi², Nina Forestieri³, Dayana Betancourt⁴, and Laura Pabst¹ on behalf of the National Birth Defects Prevention Network

¹ National Center on Birth Defects and Developmental Disabilities, Centers for Disease Control and Prevention, Atlanta, Georgia

² Department of Epidemiology, College of Public Health, University of South Florida, Tampa, Florida

³ Birth Defects Monitoring Program, North Carolina Department of Health and Human Services, Raleigh, North Carolina

⁴ Birth Defects Epidemiology and Surveillance Branch, Texas Department of State Health Services, Austin, Texas

August 2026

Introduction

Birth defects surveillance data are used for producing national prevalence estimates, monitoring for changes in prevalence, evaluating prevention strategies, and planning and providing services for affected families. Birth defects are rare conditions and combining data across programs is important to gain a thorough understanding of the true impact and landscape of birth defects nationally. Variation in surveillance methods may influence ascertainment sensitivity, case classification, and the comparability of prevalence estimates across jurisdictions. Understanding methodological variation is particularly important for pooled multi-state analyses, temporal trend monitoring, and rapid assessment of emerging teratogenic or environmental threats.

The National Birth Defects Prevention Network (NBDPN), in collaboration with CDC, conducted a survey of birth defects surveillance programs in 2025 to collect information on surveillance methods across jurisdictions. The survey, referred to as the Performance Summary, was distributed to U.S. birth defects programs in conjunction with NBDPN's biennial call for data. In this document, the 2025 survey results are summarized to describe the landscape of birth defects surveillance methods throughout the country. Findings will allow the birth defects surveillance community to understand where programs are similar and different, identify opportunities to modify current surveillance methods to promote consistency, contextualize data, provide technical assistance, resource development, and education.

Additionally, the landscape analysis may inform revisions to the NBDPN Guidelines for Conducting Birth Defects Surveillance. The Performance Summary survey tool may also be revised as those updated guidelines are published and implemented for use in future data collections.

Methods

Survey questions centered around case definition criteria and surveillance methodology utilized by programs from 2023-2024. The Performance Summary also included questions surrounding data quality and utility, though those areas are outside the scope of this analysis. Each surveillance method survey question included pre-defined responses, and space was provided for programs to leave additional information to expound on responses. Survey responses and comments were reviewed in the data cleaning process and programs were contacted, when necessary, to clarify responses and confirm the accuracy of proposed response reclassifications. Standardized decision rules were applied during data cleaning to improve consistency of response categorization across jurisdictions. For each survey question, the proportion of programs using the same case definition criteria or surveillance methodology was calculated. Proportions and one-way frequencies were calculated in RStudio and replication was conducted in SAS. Analyses were descriptive and exploratory in nature. Maps of responses by jurisdiction are presented to visualize commonalities and variations in methods across jurisdictions.

Survey Responses

Birth defects surveillance programs are established in 48 U.S. jurisdictions encompassing 45 states/territories, and three regional systems (California, the Metropolitan Atlanta Congenital Defects Program [MACDP] and the Department of Defense [DoD]). California collects birth defects data in 10 counties, MACDP is administered by the Centers for Disease Control and Prevention (CDC) and conducts

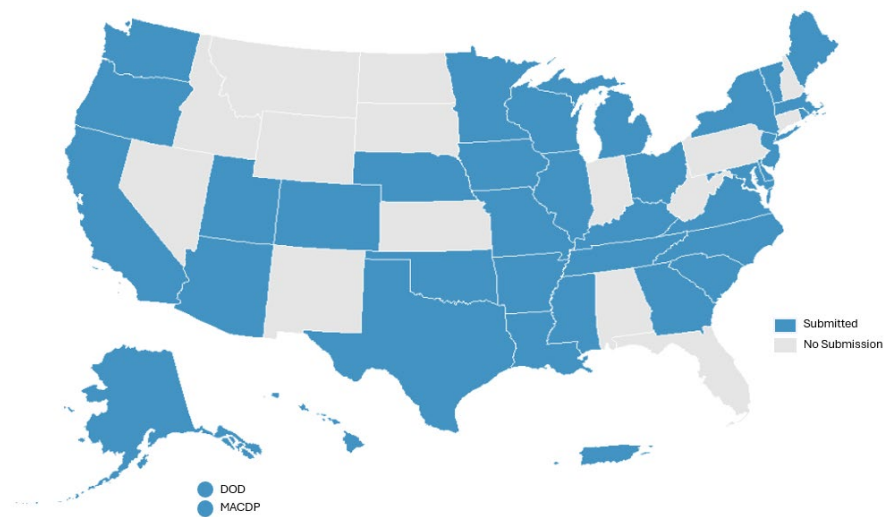


Figure 1 Map of Programs that Completed the Performance Summary

surveillance in three counties in the metropolitan Atlanta area, and the DoD conducts surveillance among births to US military beneficiaries domestically and abroad. Among these 48 jurisdictions, 39 (81%) responded to this survey.

Case Definitions - Programmatic Inclusion Criteria

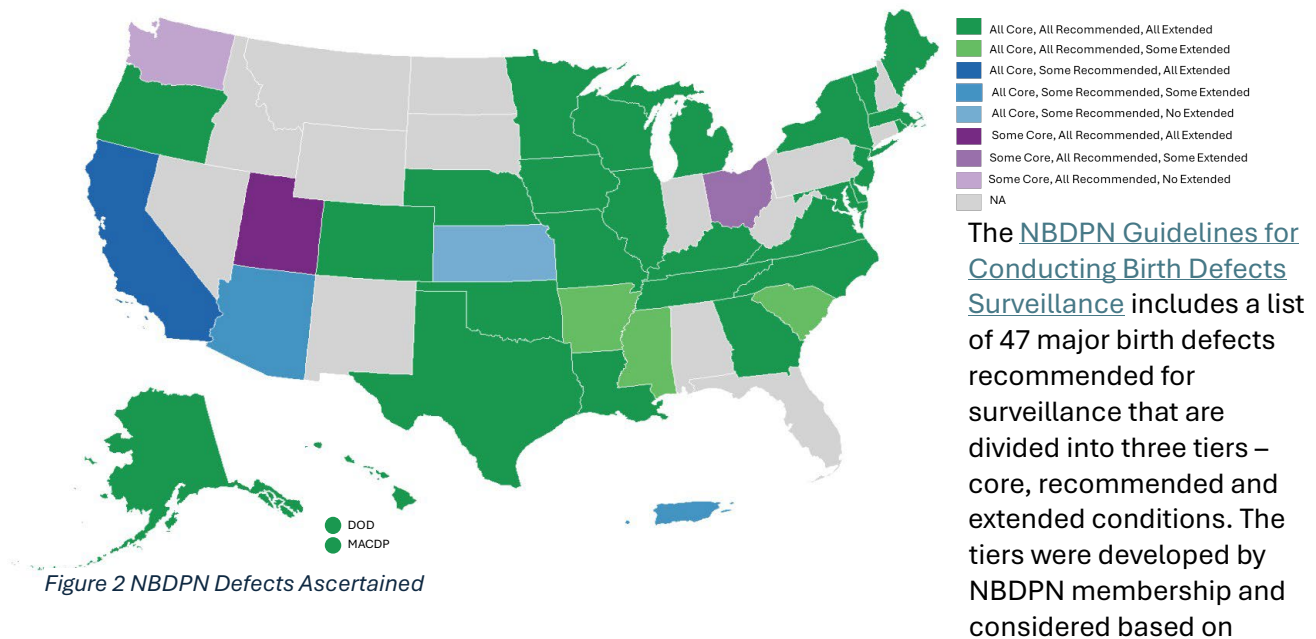
The Performance Summary survey asked about defects for which each program conducts surveillance. It also included four multi-part questions about programmatic inclusion criteria, which are operational surveillance rules that generally apply to all birth defects under surveillance and determine which diagnosed cases are eligible for inclusion in a surveillance program.

Programmatic inclusion criteria include:

1. Defects under surveillance,
2. Age of diagnoses of cases included in the surveillance system,
3. Residency criteria, and
4. Pregnancy outcomes.

These criteria are distinct from defect-specific case definition criteria, which were not included in this survey. The Performance Summary used the term “Case Definitions” to represent what will be described as programmatic inclusion criteria questions for the remainder of this landscape analysis.

Defects Under Surveillance



defect severity, defects readily apparent at the time of delivery, and public health priorities. Of the 39 respondents to the Performance Summary, 36 programs (92%) included all core defects within routine surveillance activities, while the remaining 3 programs included at least some of the core defects (Table 1). Eighty-two percent of programs (n=32) included all recommended defects, with the remaining 18% (n=7) including at least some of the recommended defects. Eighty percent of programs (n= 31) included all extended conditions. Nearly three quarters of NBDPN programs conducted surveillance for all core, recommended, and extended defects (Table 1).

Two-thirds (n=26) of all programs collect defects outside of those on the NBDPN defects lists. Some programs noted that “more than 200 additional conditions” were routinely tracked and multiple programs noted that all “Q Codes” (birth defects ICD-10-CM codes indicative of a birth defect) were part of their programmatic inclusion. In some states, many of the conditions collected outside of the NBDPN lists were not structural birth defects, but rather other conditions related to infant health and development including neonatal abstinence syndrome (NAS), congenital hearing loss, and congenital syphilis.

Age of Diagnosis

While most birth defects may be apparent at the time of delivery, some defects may not be detected until a child is older. Differences in age-at-diagnosis criteria may meaningfully affect ascertainment of conditions commonly diagnosed after infancy, including milder congenital heart defects, and certain genetic syndromes. Programs ascertain defects up to different ages throughout the network (range <1-6 yrs). Thirty-nine percent (n=15) of programs reported limiting cases to children diagnosed through one year of age (Table 1). Twenty-six percent included through two years. The remainder of programs n=12 (31%) included cases diagnosed through ages 3-6 (Table 1).

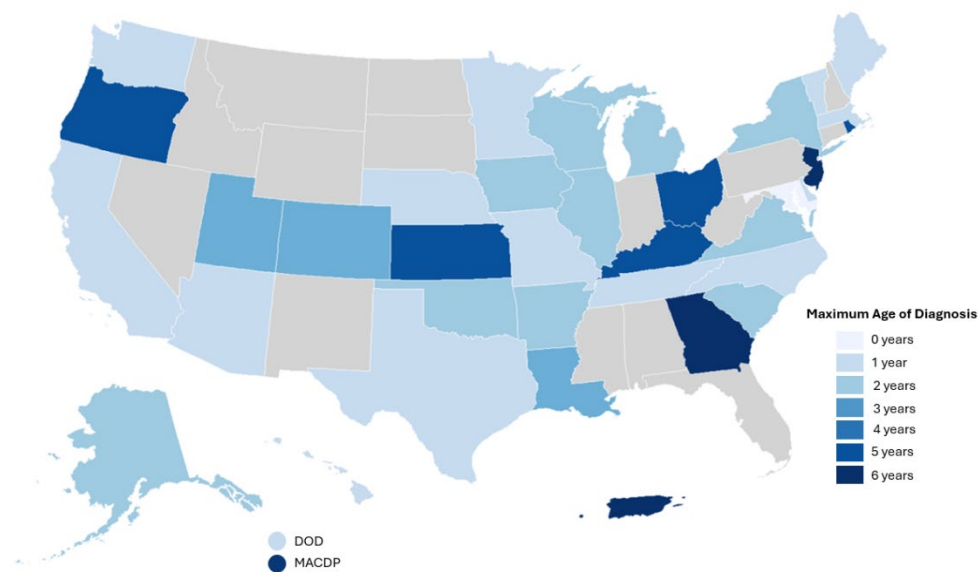


Figure 3 Maximum Age at Diagnosis

Pregnancy Outcomes

Some birth defects are more likely to result in fetal deaths or other non-live pregnancy outcomes; therefore, inclusion or exclusion of non-live outcomes may impact prevalence estimates for these defects.

All programs collected information on live births, 82% conducted surveillance for fetal deaths, and fewer than half (39%, n=15) collected all pregnancy outcomes (Table 1). Notably, all programs that collected other pregnancy outcomes also collected information on fetal deaths. Seven programs (18%) collected live births only. Programs used different criteria to define fetal deaths and other pregnancy outcomes, for example some included cases at any gestational age or birthweight while others limited to cases >20 weeks gestation, >500 grams, >350 grams, or other program-specific criteria.

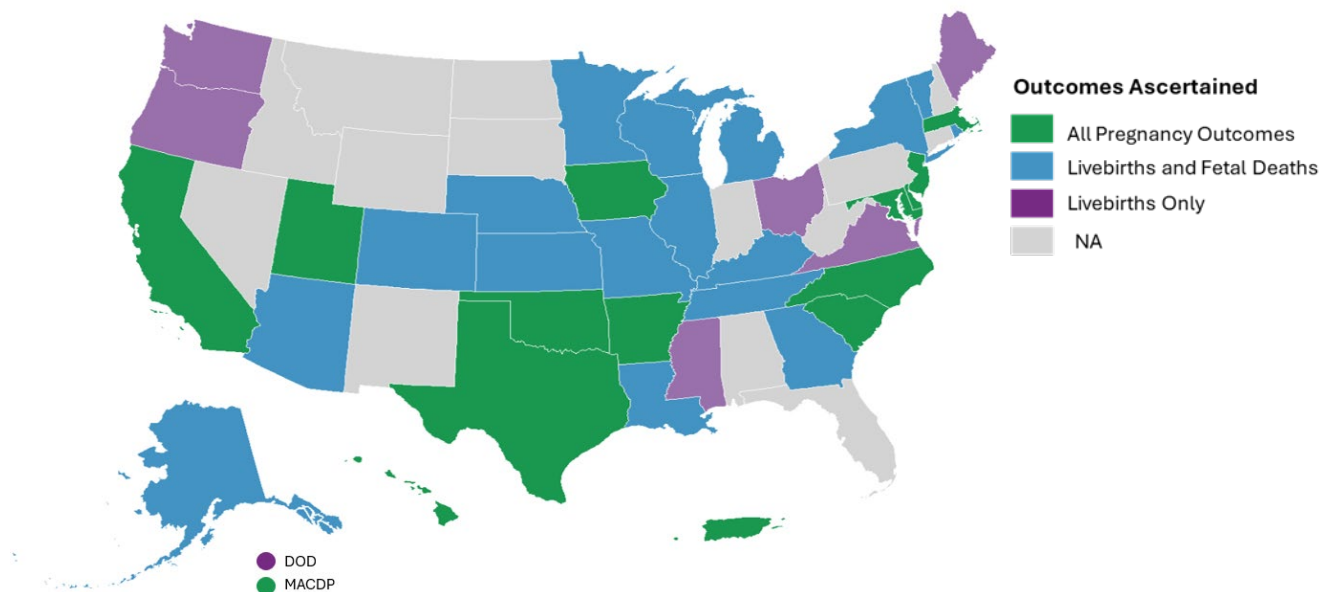


Figure 4 Pregnancy Outcomes

Residency Criteria

All programs collected data on deliveries that occur within their jurisdiction to mothers who were residents of the jurisdiction at the time of delivery. Many programs also collected data on their states' residents who delivered out of state (64%, n=25), while few collect information on in-state deliveries to out-of-state residents (26%, n=10) (Table 1). Data sharing agreements between jurisdictions, especially those with large healthcare systems located near state borders, can help ensure that birth defects case information is not lost. However, this approach can present difficulties with ensuring that cases are duplicated, highlighting the importance of interstate coordination and standardized deduplication procedures.

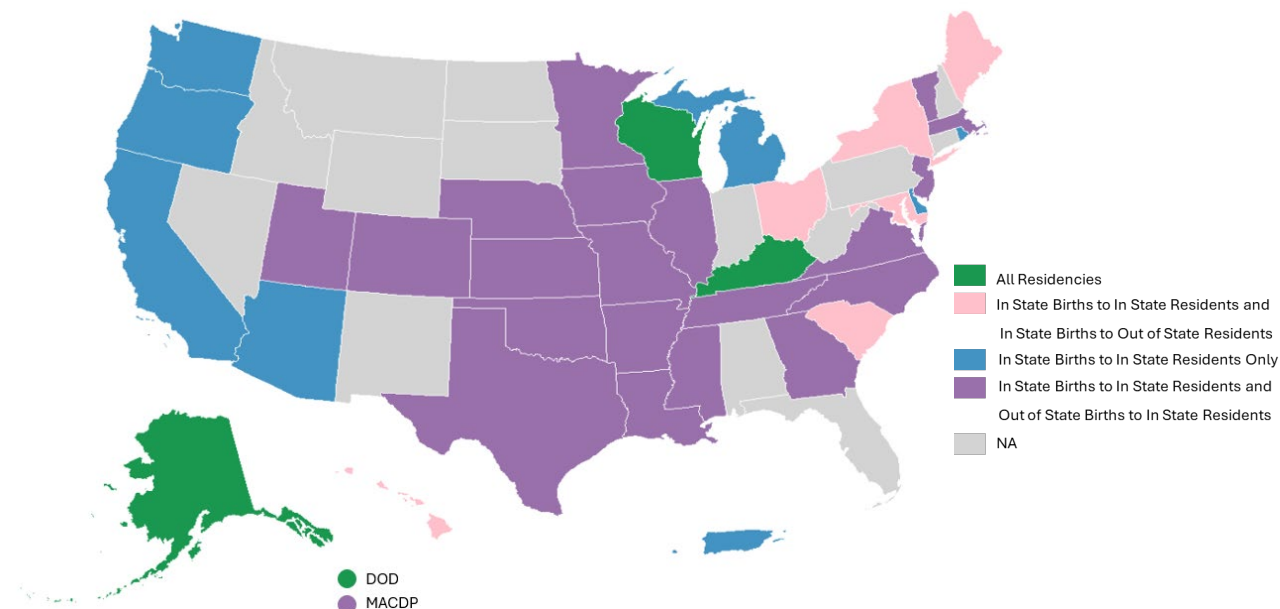


Figure 5 Residency Criteria

Program Inclusion Criteria: Summary Findings

Four programmatic inclusion criteria (birth defects under surveillance excluding other conditions, age of diagnosis, pregnancy outcomes collected, and residency criteria) impact which birth defects cases are included in surveillance systems. Taken together, survey respondents had 29 distinct combinations of these inclusion criteria across the 39 programs (Figure 8).

Any one of these programmatic inclusion criteria can impact data collection and prevalence estimates. That impact may be amplified when differences occur across all four criteria. We acknowledge that some criteria may have substantially greater effects on ascertainment sensitivity and prevalence estimation than others. Additional research may inform which criteria have the greatest impact on prevalence to allow programs to balance data quality with resource availability. See Appendix A for a map that displays all 29 combinations of the programmatic inclusion criteria and Appendix B for programmatic inclusion criteria by reporting surveillance program.

Surveillance Methods

This section of the survey asked about operational surveillance methodologies including criteria used to identify potential cases, methods to receive case notifications, data sources for case information, medical record abstraction, and clinical review.

Case Identification Criteria

Programs use multiple different criteria to identify potential cases for inclusion in their surveillance systems. All programs utilize at least one criterion that explicitly indicates that a birth defect is present for case identification purposes. The most common criteria used to identify potential birth defects cases was the presence of an ICD-10-CM code indicating a birth defect, which was used by 95% (n=37) of reporting jurisdictions (Table 2). Check boxes indicating birth defects on vital records were also commonly used with 56% (n=22) of programs using fetal death certificates, and 54% (n=21) of programs using birth and/or death certificates (Table 2).

In addition to criteria indicating the presence of a birth defect, 64% (n=25) of programs also used case identification criteria that did not specifically indicate presence of a birth defect. The most common criterion in this category was ICD-10-CM codes (Table 2). Examples of codes provided by programs included H codes (hearing loss), A50 (congenital syphilis), D82.1 (DiGeorge syndrome), P021 (placental separation and hemorrhage). Almost half of responding programs reported reviewing all fetal deaths, even if no birth defect was indicated, as a source of potential cases. A small number of programs review cases of infants with low APGAR scores, NICU admissions, and low birth weights (regardless of birth defects indicated) as potential cases (Table 2).

Among the 39 programs that responded, there were 31 variations in the criteria where a birth defect was indicated. Among the criteria where there was no birth defect indicated; there were 25 variations. The mean number of case identification criteria per program was 5.3 (range 1-12).

Case Identification Sources

A case identification source is a facility/organization that provides information on birth defects cases to birth defects surveillance programs. Nearly all programs (97%) identify cases through delivery and pediatric/tertiary hospitals (Table 2). Many programs (77%, n=30) also identify cases through vital records. While the majority of programs included both delivery and pediatric hospitals and vital records, there was wide variation in additional sources programs used, including maternal/fetal medicine providers, midwifery facilities, third party payers, other state registries, and other pediatric facilities (Table 2), with 28 variations of sources used to identify cases. Additional case identification sources may improve ascertainment completeness for certain defects or populations but may also increase operational complexity and inter-program variability.

Notification of Potential Case

Many programs (56%, n=22) reported using multiple methods to receive case reports from reporting partners, such as hospitals and clinics. The majority of programs (77%, n=30) receive files containing multiple case reports such as disease indices (Table 2). Disease indices (DIs) are lists of patients discharged from the healthcare facility/system that include children with diagnosis codes that meet reporting criteria.

About half of programs (54%, n=21) receive individual case reports (Table 2). Very few programs reported receiving case reports using HL7 messaging, including electronic case reporting (eCR) and

non-eCR interoperability mechanisms. As interoperability infrastructure expands nationally, HL7-based electronic case reporting may provide opportunities to improve surveillance timeliness, standardization, and reporting efficiency.

Medical Record Abstraction

Medical record abstraction may improve completeness and accuracy of data elements, including specific information to verify or correct assigned diagnoses. Forty-four percent (n=17) of programs abstract all cases and about one-third of programs (n=13) abstract some but not all cases, nearly one quarter (n=9) of programs do not abstract any cases (Table 2).

Among programs indicating that they only abstract some cases, there was variability in how programs determined which cases would undergo abstraction, including review only for special studies or criteria-based selection, for example reviewing all congenital heart defects.

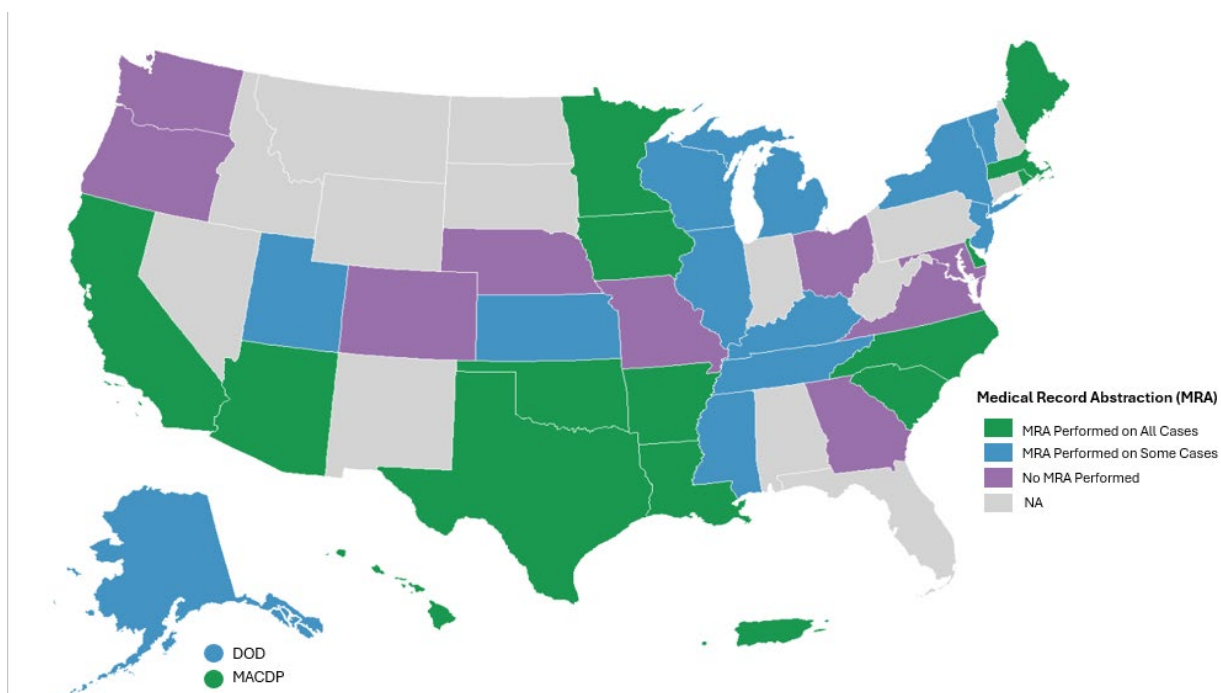
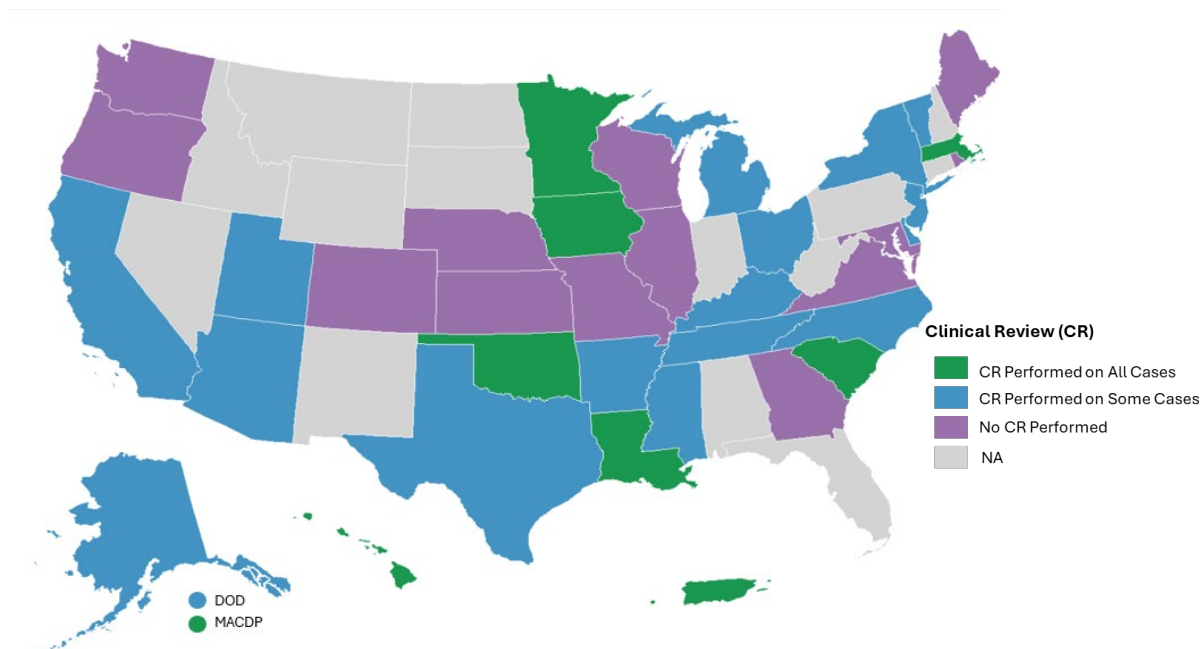


Figure 6 Medical Record Abstraction

Clinical Review

Clinical review is an additional quality assurance step used by some programs to ensure diagnosis codes are medically accurate and reflect all diagnosed birth defects for each case. Twenty-three percent (n=9) of programs clinically reviewed all cases, while one-third (n=12) of programs did not clinically review any cases (Table 2).



Nine programs conduct both medical record abstractions and clinical review for all cases, while 8 programs do not do either for any of their cases.

Key Findings

This survey found substantial variability in programmatic inclusion criteria and surveillance methods across U.S. birth defects surveillance programs, which can introduce challenges to aggregating data to produce multi-site prevalence estimates, monitoring temporal trends, and identifying geographic variation across jurisdictions. While some of these factors may be pre-determined by program statutes and unable to be changed, others may reflect modifiable operational practices. This survey did not evaluate whether differing programmatic inclusion criteria or surveillance methods were associated with differences in data quality or resource utilization. Additional research is needed to identify which practices most meaningfully influence prevalence estimation. For example, further research is needed to understand when clinical review improves the accuracy of certain defect diagnoses.

Additionally, some surveillance methodology recommendations may be universal while others are appropriately program-specific, such as the in-state/out-of-state residency criteria. The impact of these recommendations is therefore likely program dependent.

In evaluating and updating birth defects methodology and best practices, we recognize the need to balance impact on surveillance quality and comparability with programmatic resources. For example, identifying additional cases that minimally affect prevalence estimates but require many

additional hours of staff time may not represent the most efficient use of limited surveillance resources.

This survey offers a solid foundation for monitoring progress across NBDPN member programs and may provide opportunities to better align surveillance practices moving forward, thereby strengthening comparability, data interpretation, and the overall evidence base.

Strengths

These data represent one of the first systematic efforts to characterize programmatic inclusion criteria and surveillance methods across U.S. birth defects surveillance programs using a standardized survey instrument. Participation included 39 of 48 eligible jurisdictions, representing the majority of state, territorial, and metropolitan-area surveillance programs in the United States. The survey collected information on multiple dimensions of surveillance practices, including case inclusion criteria, residency requirements, pregnancy outcomes, case identification methods, medical record abstraction, and clinical review, providing a comprehensive snapshot of current surveillance approaches nationwide.

Limitations

This analysis is subject to at least three limitations. First, pre-defined response options within the survey questionnaire may not have fully reflected the nuanced surveillance practices by responding programs. Second, the survey was completed by 39/48 (81%) regional, state, and territorial birth defects surveillance programs throughout the U.S. Nonparticipating programs may differ systematically from responding programs with respect to surveillance infrastructure, methodology, or available resources. Third, survey responses were self-reported and were not independently verified through review of program protocols, surveillance manuals, or operational procedures. As a result, responses may not fully reflect actual surveillance practices in every jurisdiction. Lastly, although this survey was piloted with a small group of U.S. programs, this was the first distribution of this survey nationwide. It is possible that programs may have interpreted and responded to questions differently than what was intended. For example, the wording of the question “What is the maximum age of diagnosis for cases of birth defects your program includes in routine surveillance?” appears to have created confusion, as some programs interpreted “2” as indicating surveillance through the child’s second birthday while others interpreted it as including the entire period during which the child was two years old but before their third birthday. This ambiguity may have affected comparability of reported age-at-diagnosis inclusion criteria across responding programs.

Conclusions and Recommendations

Currently, there are no universally implemented national standards governing programmatic inclusion criteria and surveillance methods across U.S. birth defects surveillance programs. Thus, each program develops its own surveillance methodology and case definitions, largely influenced by their state's statutes and programmatic resources, healthcare infrastructure, and jurisdiction-specific priorities. The diversity of methods employed makes aggregating data across programs and accurately interpreting prevalence estimates, temporal trends, and geographic variation in the U.S. challenging. Harmonization of surveillance methods across the country is needed to improve our ability to aggregate data, monitor trends, and more readily assess emerging threats to maternal and infant health. The findings of this survey improve understanding of the current landscape of birth defects surveillance methods and will assist in the development of updated birth defects surveillance guidelines and national standards.

There is some literature *<insert hyperlink to literature review to be posted on the NBDPN website>* to inform best practices in the anticipated updates to the NBDPN surveillance guidelines, but little to no data to inform other areas. Additionally, the literature represents only a few programs. Further study would benefit the birth defects surveillance community and may improve approaches to conducting surveillance across the country.

Approaches to surveillance vary widely across the country. Historically, programs have often been categorized as either *active* or *passive* systems. However, thanks to advancements in electronic health records, interoperability infrastructure, and electronic case reporting, the delineation between *active* and *passive* systems has blurred and many programs now describe themselves as operating under *hybrid* models. These findings suggest that birth defects surveillance systems may be better conceptualized along a continuum of ascertainment intensity, verification practices, abstraction methods, and data integration strategies rather than as strictly “active” or “passive” systems. While hybrid and evolving surveillance models may improve data completeness and operational efficiency for individual programs, they may also introduce additional complexity when attempting to harmonize data across jurisdictions.

Importantly, not all methodological differences identified in this analysis are likely to have equivalent impact on ascertainment sensitivity, case classification, prevalence estimation, or data comparability. Future work is needed to identify which surveillance practices most meaningfully influence surveillance quality and comparability, as well as which methodological components may be most feasible and impactful to harmonize across programs. Continued collaboration across programs will be critical to strengthening the national evidence base and improving the comparability, interpretability, and utility of U.S. birth defects surveillance data.

Table 1. Programmatic Inclusion Criteria Among U.S. Birth Defects Surveillance Programs (n=39)

Collected Defects	All	Some	None
Core	36 (92%)	3 (8%)	0 (0%)
Recommended	32 (82%)	7 (18%)	0 (0%)
Extended	31 (80%)	6 (15%)	2 (5%)
Non-NBDPN Defects	---	26 (67%)	13 (33%)
Age at Diagnosis			
<1 year*	1	3%	
1 year	15	39%	
2 years	10	26%	
3 years	3	8%	
5 years	5	13%	
6 years	4	10%	
Not reported	1	3%	
Collected Pregnancy Outcomes			
Live births	39	100%	
Fetal deaths	32	82%	
Other outcomes	15	39%	
Residency Criteria			
In-state births to in-state residents	39	100%	
Out-of-state births to in-state residents	25	64%	
In-state births to out-of-state residents	10	26%	

* Program reported including infants identified “in the newborn period”

Table 2. Surveillance Methods Among U.S. Birth Defects Surveillance Programs (n=39)

Reporting Criteria for Case Identification		
Birth defects indicated		
ICD codes	37	95%
Fetal death certificates	22	56%
Birth certificates	21	54%
Death certificates	21	54%
Prenatal records	17	44%
Medical chart	11	28%
Other	6	15%
No specific birth defect indication		
ICD codes	23	59%
Fetal death certificates	17	44%
Neonatal death	9	23%
Other pregnancy outcomes	8	21%
Medical chart	5	13%
Other	4	10%
Procedure codes	4	10%
Apgar score	2	5%
NICU admission	2	5%
Low birth weight	1	3%
Case Identification Sources		
Delivery hospitals	38	97%
Pediatric/Tertiary Hospitals	37	95%
Vital Records	30	77%
Other Pediatric Facilities	18	46%
State Registries	16	41%
Maternal/Fetal Medicine Providers	12	31%
Midwifery Facilities	11	28%
Third Party Payers	7	18%
Other	10	26%
Case Notification Mechanisms		
Multiple Case Reports, e.g. DIs	30	77%
Individual Case Reports	21	54%
Other non-eCR HL7 Reporting	5	13%
Electronic Case Reports	3	8%
Other	8	21%

Medical Record Abstraction

All	17	44%
Some	13	33%
Criteria-based selection	7	18%
Special studies	3	8%
Random sample	1	3%
Other	7	18%
None	9	23%

Clinical Review

All	9	23%
Some	17	44%
Criteria-based selection	5	13%
Special studies	5	13%
Random sample	5	13%
Other	7	18%
None	12	31%

Surveillance System Type

Standalone system	26	67%
Part of larger agency information system	11	28%
Other	4	10%

Appendix A

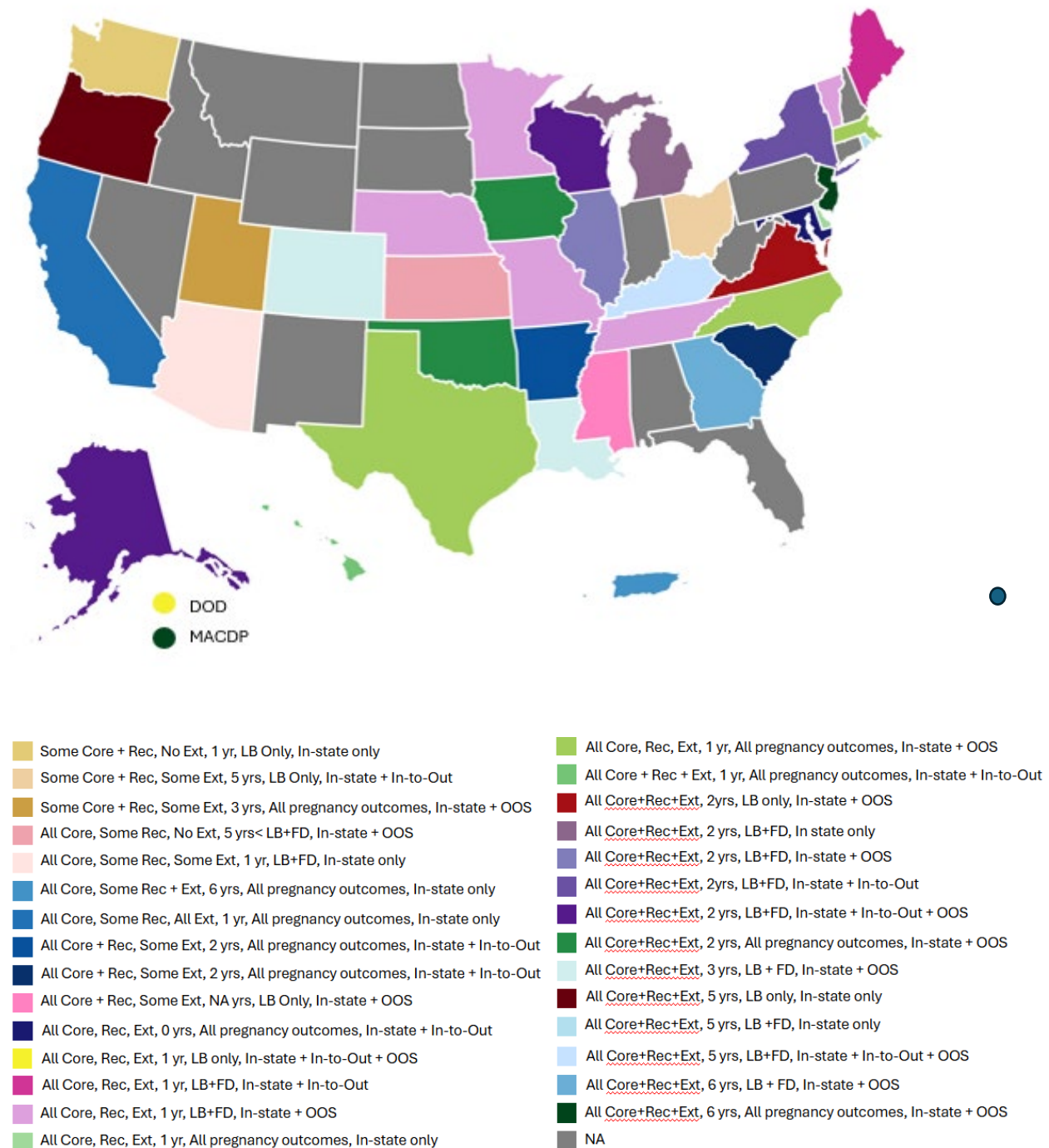


Figure 8 Variety of Programmatic Inclusion Criteria throughout NBDPN

Appendix B

Program	Core	Recommended	Extended	Maximum age of diagnosis	Outcome	In-state births to in-state residents	In-state births to out-of-state residents	Out-of-state births to in-state residents
Alaska	All	All	All	2	LB, FD	Yes	Yes	Yes
Arizona	All	Some	Some	1	LB, FD	Yes	No	No
Arkansas	All	All	Some	2	All	Yes	No	Yes
California	All	Some	All	1	All	Yes	No	No
Centers for Disease Control and Prevention	All	All	All	6	All	Yes	No	Yes
Colorado	All	All	All	3	LB, FD	Yes	No	Yes
Delaware	All	All	All	1	All	Yes	No	No
Department of Defense	All	All	All	1	LB	Yes	Yes	Yes
Georgia	All	All	All	6	LB, FD	Yes	No	Yes
Hawaii	All	All	All	1	All	Yes	Yes	No
Illinois	All	All	All	2	LB, FD	Yes	No	Yes
Iowa	All	All	All	2	All	Yes	No	Yes
Kansas	All	Some	None	5	LB, FD	Yes	No	Yes
Kentucky	All	All	All	5	LB, FD	Yes	Yes	Yes
Louisiana	All	All	All	3	LB, FD	Yes	No	Yes
Maine	All	All	All	1	LB	Yes	Yes	No
Maryland	All	All	All	<1*	All	Yes	Yes	No
Massachusetts	All	All	All	1	All	Yes	No	Yes
Michigan	All	All	All	2	LB, FD	Yes	No	No
Minnesota	All	All	All	1	LB, FD	Yes	No	Yes
Mississippi	All	All	Some	**	LB	Yes	No	Yes
Missouri	All	All	All	1	LB, FD	Yes	No	Yes

Program	Core	Recommended	Extended	Maximum age of diagnosis	Outcome	In-state births to in-state residents	In-state births to out-of-state residents	Out-of-state births to in-state residents
Nebraska	All	All	All	1	LB, FD	Yes	No	Yes
New Jersey	All	All	All	6	All	Yes	No	Yes
New York	All	All	All	2	LB, FD	Yes	Yes	No
North Carolina	All	All	All	1	All	Yes	No	Yes
Ohio	Some	Some	Some	5	LB	Yes	Yes	No
Oklahoma	All	All	All	2	All	Yes	No	Yes
Oregon	All	All	All	5	LB	Yes	No	No
Puerto Rico	All	Some	Some	6	All	Yes	No	No
Rhode Island	All	All	All	5	LB, FD	Yes	No	No
South Carolina	All	All	Some	2	All	Yes	Yes	No
Tennessee	All	All	All	1	LB, FD	Yes	No	Yes
Texas	All	All	All	1	All	Yes	No	Yes
Utah	Some	Some	All	3	All	Yes	No	Yes
Vermont	All	All	All	1	LB, FD	Yes	No	Yes
Virginia	All	All	All	2	LB	Yes	No	Yes
Washington	Some	Some	None	1	LB	Yes	No	No
Wisconsin	All	All	All	2	LB, FD	Yes	Yes	Yes

* Program reported including infants identified “in the newborn period”

**Not reported