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Expanding the Massachusetts Birth Defects Monitoring Program to include additional pregnancy outcomes: Programmatic efforts and impacts on case ascertainment, 2012–2020

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Abstract

Background: Birth defects affect 1 in 33 infants in the United States and are a leading cause of infant mortality. Birth defects surveillance is crucial for informing public health action. The Massachusetts Birth Defects Monitoring Program (MBDMP) began collecting other pregnancy losses (OPLs) in 2011, including miscarriages (<20 weeks gestation) or elective terminations (any gestational age), in addition to live births and stillbirths (≥20 weeks gestation). We describe programmatic changes for adding OPLs and their impact on prevalence estimates.

Methods: Using population-based, statewide, data from the MBDMP (2012–2020), we assessed prevalence per 10,000 live births and 95% confidence intervals (CIs) with and without OPLs overall and for specific birth defects by time period, maternal age, and race/ethnicity.

Results: Including OPLs required amending a state statute and promulgating regulations, new data sources, and additional data processing, cleaning, and verification. Overall prevalence with OPLs increased from 257.4 (95% CI: 253.5–261.4) to 333.9 (95% CI: 329.4–338.4) per 10,000; increases were observed in all time periods, age, and race/ethnicity groups. After including OPLs, the prevalence increased for neural tube defects [3.2 (2.7–3.6) to 8.3 (7.6–9.0)], and trisomies 13 [0.5 (0.3–0.7) to 4.1 (3.6–4.6)], 18 [1.5 (1.2–1.9) to 8.2 (7.5–8.9)], and 21 [12.3 (11.4–

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CONFLICT OF INTEREST STATEMENT

The authors declare no conflicts of interest.

SUPPORTING INFORMATION

Additional supporting information can be found online in the Supporting Information section at the end of this article.

13.2) to 28.9 (27.6–30.2)]. Cardiovascular defects increased slightly, while prevalence of eye/ear, respiratory, and gastrointestinal defects remained similar.

Conclusions: Adding OPLs required substantial programmatic efforts and resulted in more complete case ascertainment, particularly for certain birth defects. More complete case ascertainment will allow for improved research, screening, and resource allocation.

Keywords

birth defects; chromosomal defects; neural tube defects; other pregnancy losses; prenatal reporting; surveillance

1 | INTRODUCTION

Ongoing, systematic birth defects surveillance is crucial for informing public health action. Surveillance data can help monitor trends, raise public awareness, target resources, and inform screening and prevention efforts and etiologic research. Including birth defects across all identifiable pregnancy outcomes improves ascertainment and can therefore increase prevalence estimates, especially for birth defect types where there is a substantial proportion of non-live births (National Birth Defects Prevention Network, 2004; Yeung et al., 2016). In the United States, population-based birth defects surveillance programs vary by pregnancy outcomes included (National Birth Defects Prevention Network, 2022; Yeung et al., 2016). Few studies have described programmatic changes needed to expand birth defects surveillance programs to include additional pregnancy outcomes or directly measured impacts on prevalence estimates.

Since 1999, Massachusetts has conducted active, state-wide, population-based surveillance on major structural birth defects and chromosomal abnormalities (Massachusetts Department of Public Health, 2023). An update to the Massachusetts birth defects surveillance statute in 2002, followed by regulations promulgated by the Department of Public Health in 2009, allowed for inclusion of pregnancy outcomes beyond live births and stillbirths (fetal death ≥20 weeks gestation or birth weight ≥ 350 g) (Massachusetts Department of Public Health, 2019). In 2011, ascertainment began for birth defects among other pregnancy losses (OPLs), including elective terminations at any gestational age (GA) and miscarriages <20 weeks GA (Massachusetts Department of Public Health, 2016). We describe efforts to incorporate birth defects diagnosed in OPLs and evaluate impacts on prevalence.

2 | METHODS

We used data from the Massachusetts Birth Defects Monitoring Program (MBDMP), described previously (Liberman et al., 2014). Briefly, case reports are submitted regularly from multiple sources, including delivery and specialty care hospitals, and prenatal reporters (e.g., healthcare professionals, including genetic counselors, maternal-fetal medicine specialists). Potential cases are reviewed by highly trained abstractors, undergo clinical review by a clinical geneticist trained in pediatric cardiology to confirm diagnoses, and are linked to vital records whenever possible (Table 1). Reports of potential cases are

submitted to the MBDMP in International Classification of Diseases, 10th edition, Clinical Modification (ICD-10-CM). However, when cases are confirmed through programmatic and clinical review they are coded in ICD-9-CM/modified British Pediatric Association (ICD-9-CM/BPA), a more specified coding system for birth defects (Table S1).

Programmatic materials documenting changes (e.g., surveillance reports, advisory committee presentations, and program reference materials) were reviewed to assess steps taken to improve prenatal reporting and include birth defects diagnosed in OPLs in the MBDMP system.

Live birth, stillbirth, and OPL cases with one or more birth defects ascertained by the MBDMP with deliveries between 2012 and 2020 were evaluated. Case counts and prevalence per 10,000 live births with 95% confidence intervals (CIs) were estimated with and without including OPLs overall, by maternal age and race/ethnicity, for specific types of birth defects, and by time period. CIs were estimated using the Poisson distribution. Statistical analyses were conducted using SAS 9.4 (SAS Institute, Inc., Cary, NC, USA).

3 | RESULTS

3.1 | Process evaluation

The MBDMP leveraged partnerships with collaborators and birth defects researchers to develop regulations allowing data collection on birth defects diagnosed in OPLs. Details on data collection and processing are described in Table 1. Comparing standard program procedures to those specific to OPLs showed that many processes are similar. Optimizing prenatal data reporting was expected to improve ascertainment across all pregnancy outcomes through karyotype and other diagnostic confirmation but was thought to be especially important for birth defects diagnosed in OPLs, as prenatal reports are the primary and sometimes the only data source. Our document review showed that MBDMP worked with prenatal reporters to establish consistent and comprehensive prenatal reporting. To address initial challenges in data submission processes, a formal protocol and structured data submission template were developed to improve consistency across facilities and enhance data quality. These procedures help ensure that prenatal reporters submit required data elements (e.g., type of testing, results), but do not eliminate needs for additional data processing, since some data elements include unstructured text. Regular meetings were held with healthcare professionals at prenatal reporting facilities to identify, design, and implement new quality improvement measures.

Medical records abstraction for birth defects diagnosed in OPLs also required adaptations. Prenatally diagnosed birth defects in OPLs lack vital records in Massachusetts, so data elements typically extracted from or verified with these data sources are not available. The abstraction tool is intentionally flexible to allow for free text responses for some data elements and with auto-filling of “not applicable” into fields not relevant (e.g., birth weight, head circumference). All MBDMP staff are highly trained and have additional protocols for abstracting birth defects diagnosed in OPLs, including extra efforts to identify all prenatal testing and autopsy reports to ensure data quality and completeness.

3.2 | Case counts and prevalence

Including birth defects diagnosed in OPLs resulted in ascertainment of 4848 additional birth defects cases between 2012 and 2020, a 29.7% increase. OPL inclusion resulted in additional case ascertainment in all time periods and across all maternal demographics examined (Figure 1a; Table S2), although some subgroups saw larger increases (e.g., maternal age ≥ 35 years: 51.6%).

The overall prevalence after including OPLs increased from 257.4 (95% CI: 253.5–261.4) to 333.9 (95% CI: 329.4–338.4) per 10,000 live births, and impact differed by defect type (Figure 1b; Table S3). Prevalence of ear, eye, gastrointestinal, genitourinary, orofacial, and respiratory defects remained similar. Conversely, prevalence of cardiovascular, central nervous system, chromosomal, and musculoskeletal birth defects increased. Specifically, prevalence was more than two times higher for trisomies 13 [0.5 (0.3–0.7) to 4.1 (3.6–4.6)], 18 [1.5 (1.2–1.9) to 8.2 (7.5–8.9)], and 21 [12.3 (11.4–13.2) to 28.9 (27.6–30.2)] and for neural tube defects (NTDs) [3.2 (2.7–3.6) to 8.3 (7.6–9.0)]. The overall prevalence without OPLs also increased from 203.4 (197.5–209.5) in 2012–2014 to 293.8 (286.5–301.2) in 2018–2020, and chromosomal defects increased from 30.8 (28.5–33.2) to 42.3 (39.6–45.2).

4 | DISCUSSION

Expanding the MBDMP to include birth defects diagnosed in OPLs required statute and regulation changes and modifications to system processes including additional data collection, abstraction, and verification procedures. Efforts resulted in improved case ascertainment overall, and particularly for NTDs and trisomies. Adding OPLs also increased overall ascertainment among live births and stillbirths, and especially for chromosomal defects and defects.

While collecting prenatal data is critical to ensuring complete ascertainment of birth defects diagnosed in OPLs, there are challenges. Clinical information from prenatal reporters is often unstructured free-text, making processing and verification less automatable and more time-intensive. Continuous quality improvement efforts and process updates, informed by regular meetings with abstractors and programmers, have allowed prenatal reports with more submission variability (e.g., quarterly) and free-text variables to be incorporated into automated deduplication and case matching procedures, reducing redundancy. These and other data cleaning/verification procedures were developed and implemented to ensure more complete and accurate identification of birth defects in OPLs.

Including OPLs differentially impacted certain birth defects, particularly NTDs and trisomies. This improved case ascertainment was expected, as these severe conditions often end in early miscarriage or termination that would not be captured without including OPLs. Comparing MBDMP data prior to including OPLs demonstrated that their addition improved case ascertainment in live births and stillbirths as well (Massachusetts Department of Public Health, 2019). We observed substantial increases in overall prevalence and particularly in chromosomal defects between the earliest and latest time periods, likely from continued improvements in prenatal reporting.

Our findings are consistent with other studies among US birth defects surveillance programs including elective terminations (Peller et al., 2004; Velie & Shaw, 1996) or elective terminations and miscarriages <20 weeks (Allen et al., 1996; Cragan & Gilboa, 2009; Ethen & Canfield, 2002; Forrester et al., 1998), where higher prevalence of birth defects was observed when these outcomes are included, particularly for severe birth defects (e.g., NTDs, chromosomal abnormalities). Collectively, these reports suggest that including birth defects diagnosed in these other outcomes can lead to improved ascertainment (National Birth Defects Prevention Network, 2022; Yeung et al., 2016).

Expanding eligible pregnancy outcomes allowed the MBDMP to increase their data's utility. Since the addition of OPLs, prevalence estimates for trisomies 13, 18, and 21, anencephaly, and spina bifida in Massachusetts more closely resemble national prevalence estimates (Massachusetts Department of Public Health, 2023). Improved case ascertainment allows better monitoring of new threats to pregnant people, such as the Zika outbreak (Cragan et al., 2017), should lead to more accurate assessment of health care resource needs and will enhance etiologic research. While including birth defects among terminations and early miscarriages provides more complete case ascertainment, it may not be feasible for some programs. Legal authority, programmatic capacity, and limited resources can pose barriers to including additional pregnancy outcomes.

This study used data from the MBDMP, a statewide, high-quality, population-based, active surveillance system with clinical reviews of all cases. However, some limitations exist. Deliveries out of state may not be captured, especially for OPLs. Demographic information in OPLs may be less accurate and often cannot be verified; due to small numbers we could not evaluate demographics among specific birth defect types. Data availability on very early miscarriages varies by reporting site; thus, birth defects prevalence in OPLs may be underestimated. Reporting prenatal diagnoses in text rather than codes could result in misclassification of some defects. Even with improved case ascertainment, gaps may still exist, particularly for birth defects occurring at very early GAs and without any diagnostic testing.

5 | CONCLUSIONS

Including OPLs required substantial programmatic efforts and led to more complete case ascertainment. Nearly 30% more birth defects cases were ascertained after including OPLs, with particularly noticeable increases among NTDs and trisomies. Including prenatal data sources and pregnancy outcomes can improve the utility of surveillance data and aid in targeting research, screening, and prevention efforts.

Supplementary Material

Refer to Web version on PubMed Central for supplementary material.

DATA AVAILABILITY STATEMENT

Research data are not shared.

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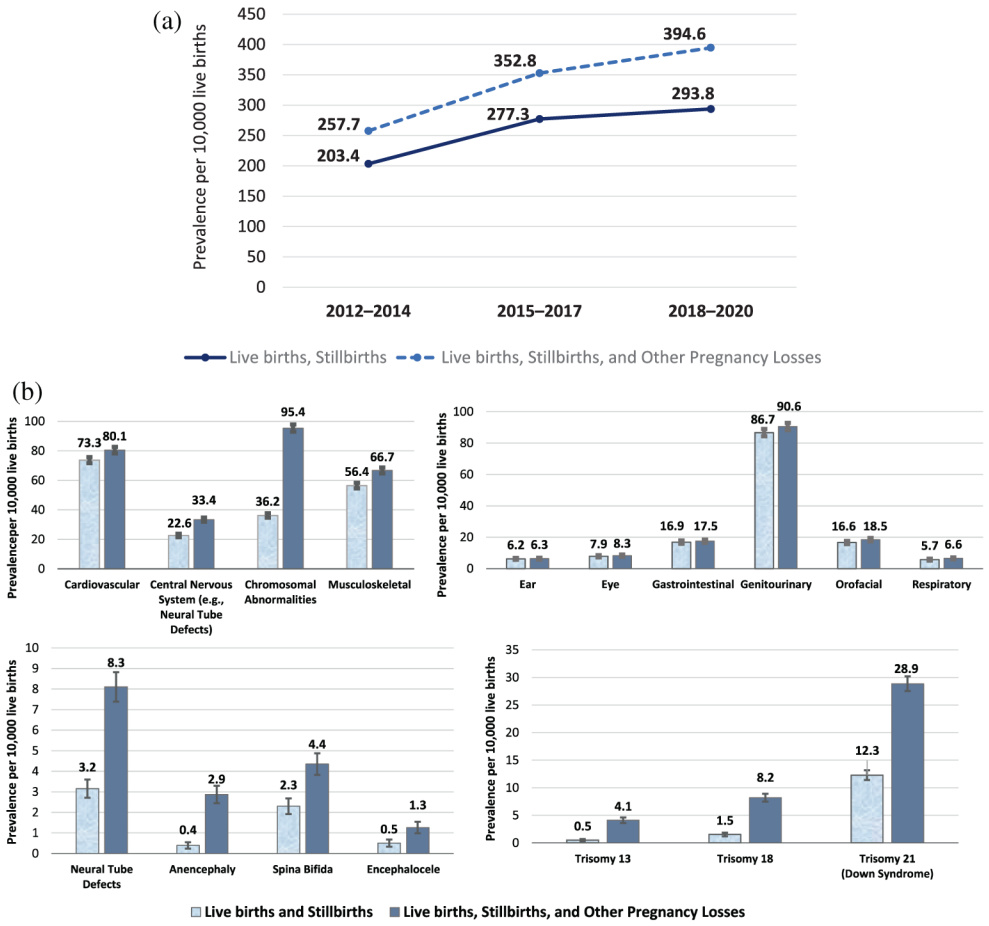

FIGURE 1.

Changes in birth defects prevalence (a) over time and (b) by birth defect types after adding other pregnancy losses to the Massachusetts Birth Defects Monitoring Program.

TABLE 1

System attributes and data flow for the Massachusetts Birth Defects Monitoring Program, and considerations for birth defects diagnosed in other pregnancy losses.

	General system attributes	**Considerations for birth defects diagnosed in other pregnancy losses**[a]
Data sources	Monthly reports are submitted from all birthing hospitals in the state, tertiary referral hospitals, a children's hospital, a non-birthing hospital specializing in eye and ear conditions, emergency departments, pathology departments, commercial laboratories, and prenatal testing sites. Prenatal reports include results of prenatal testing independent of pregnancy outcome. Prenatal reports are regularly provided from all tertiary delivery hospitals, one Level 2 hospital, one standalone ultrasound group, several commercial laboratories and pathology departments, and some genetic counseling sites.	Prenatal reports are the primary data source, but ICD-10-CM codes submitted through regular monthly reporting by any reporting facility can also contribute.
Data linkages	Surveillance cases are linked to vital records (e.g., birth certificates, fetal death reports, and infant death certificates) when possible, to obtain demographic and clinical information.	Birth defects diagnosed in OPLs cannot be linked to vital records as vital records in Massachusetts only includes live births or fetal deaths ≥20 weeks or ≥350 g. Most demographic and clinical information comes from medical records.
Data format for submission	Standardized .txt data files with ICD-10-CM codes and other information are electronically reported by facilities directly to the program.	Prenatal reports are electronically submitted in a standardized Microsoft Excel format and include text summaries of results of tests/procedures performed.
Data elements	Automated reports from facilities: • ICD-10-CM codes Prenatal reports: • Patient data (e.g., name, date of birth, address, medical record number, and genetics counselor's name, if applicable) • Prenatal test information (imaging reports) • Non-imaging procedures and results (e.g., amniocentesis, chorionic villus sampling, other) • Non-imaging lab tests (e.g., chromosome, microarray, molecular, others) • Other pregnancy information as available (e.g., estimated delivery hospital and date, plurality) Vital records: • Demographic and clinical variables obtained from vital records include maternal age, region of maternal residence, race/ethnicity, birth weight, and gestational age. Surveillance information is collected through medical records and entered into electronic confidential reporting and abstraction form (eCRAF): • Infant characteristics (e.g., sex, diagnostic procedures and testing, detailed imaging, autopsy results) • Diagnoses based on verbatim descriptions are coded using modified ICD-9-CM/BPA coding system	Automated reports with codes not available, and variables differ in formats and uses. • Prenatal reports include free text. • Vital records information usually not available • Demographic, pregnancy, and delivery information often more limited • Birth weight, head circumference, and length are auto filled to N/A Because postnatal imaging and other confirmatory tests are not available or not performed, detailed imaging and autopsy results, if done, are incredibly important and we make every effort to obtain these. Additional precautions are implemented when reviewing multiple gestations.

	General system attributes • Maternal characteristics (e.g., medications, procedures, medical conditions before and during pregnancy) • Family history of birth defects	**Considerations for birth defects diagnosed in other pregnancy losses**[a]
Challenges	Some residents of MA are known to both receive prenatal care and give birth at facilities outside of MA, which we cannot always capture. However, some MA residents who deliver out of state, receive medical care in MA, which would allow them to enter the MBDMP program through a different MA-based facility. With electronic access to health records for >92% of the facilities reporting to the MBDMP, the MBDMP abstractors are often able to abstract data from out of state records that have been linked to in-state medical records.	There is a greater likelihood of incomplete ascertainment of OPLs, especially those occurring at very early gestational age without any diagnostic testing. Quality and completeness of prenatal reporting varies by facility. Some specific data items are more challenging to collect and submit by prenatal reporters because prenatal diagnoses do not have established codes. Out of state deliveries to MA residents are less likely to be captured in this group.

Abbreviations: ICD-9-CM/BPA, International Classification of Diseases, 9th edition, Clinical Modification/modified British Pediatric Association; ICD-10-CM, International Classification of Diseases, 10th edition, Clinical Modification; MA, Massachusetts; MBDMP, Massachusetts Birth Defects Monitoring program, OPL, other pregnancy losses.

[a]Other pregnancy losses (OPLs) include miscarriage [<20 weeks gestational age (GA)] or elective terminations (any GA).

Source: Adapted with permission from Massachusetts Department of Public Health (2016, 2019, 2023).