Evaluation of Hospital Diagnostic Codes among Non-Liveborn Pregnancy Outcomes in New York State: 2017-2022

Poster Session Presenters: Adrian Michalski and Sarah Fisher
Co-Authors: Alicia Lee and Amanda St. Louis

Birth defects surveillance systems that exclude non-livebirths likely underestimate prevalence. Many birth defects surveillance systems use International Classification of Disease (ICD) codes to identify potential liveborn cases of birth defects, but ICD codes for diagnoses among non-liveborn cases are limited. We assessed the accuracy of ICD-10 fetal anomaly codes assigned to non-livebirths (stillbirths (fetal loss ≥20 weeks) and elective terminations).

We identified non-livebirths with a reported fetal anomaly in New York State during 2017-2022, based on ICD-10 codes reported to the New York Statewide Planning and Research Cooperative System (SPARCS). A nurse reviewed medical records to confirm the outcome and birth defect diagnoses. We identified 5,024 cases with ICD-10 codes indicating a non-livebirth and at least one fetal anomaly. We grouped fetal anomaly codes according to 8 body systems (central nervous system, craniofacial, cardiovascular, respiratory, gastrointestinal, genitourinary, musculoskeletal, chromosomal). For each category, we estimated the positive predictive value (PPV) and 95% confidence interval (CI), defined as the percent of confirmed cases among all cases with a given body system-specific code, and the false-negative rate (FNR), defined as the percent of confirmed cases that were not assigned the correct body system-specific ICD-10 code. We confirmed 4,945 birth defects across all 8 body systems, but the only body system-specific ICD-10 codes assigned were for central nervous system (n=524), chromosomal (n=1,867), and cardiovascular (n=4) defects. The PPV and FNR were 80.2% (95% CI: 76.7- 83.6) and 56.3%, respectively, for central nervous system defects; 83.2% (95% CI: 81.5- 84.9) and 25.5%, respectively, for chromosomal defects; and 50.0% (95% CI: 1.0-99.0) and 99.7%, respectively, for cardiovascular birth defects. PPV and NFR were 0% and 100%, respectively, for all other body systems. Codes that were assigned were generally confirmed by chart review, but most true cases were not assigned a body system-specific ICD-10 code. Currently in New York, ICD-10 codes alone are not useful for identifying specific birth defects among non-livebirths. Additional training for coding staff may improve the use and accuracy of hospital discharge codes for non-livebirths, so that birth defect surveillance systems with limited resources could rely on them for case-finding.

Title: An Evaluation of the Oklahoma Birth Defects Registry's Spina Bifida Coding Tool Poster Session

Poster Session Presenter: Lexi Richardson
Co-Authors: Flor Rodriguez, Kara Wilbur, and Lindsay Denson

The Oklahoma Birth Defects Registry (OBDR) utilizes a birth defect coding system adapted from the British Pediatric Association (BPA). This adapted BPA guide contains 21 spina bifida codes. The descriptions for these codes contain outdated terms and vague verbiage, which has historically resulted in coding errors and inconsistent interpretations among registered nurse abstractors (RNAB). To decrease coding errors and improve analysis, the OBDR created a spina bifida coding tool adapted from the Birth Defects Study to Evaluate Pregnancy exposureS (BD-STEPS) Spina Bifida Codes guide. This modified 270-code, multi-level search tool was created in Excel and contains an "if/then" logic formula to assist in efficiently selecting the code that best describes the following spina bifida characteristics: 1) type of lesion, 2) lesion level, 3) Arnold Chiari presence, and 4) open/closed lesion status. The OBDR evaluated the new coding tool by comparing the coding accuracy of spina bifida cases pre- and post-implementation (birth years 2013-2017 and 2018-2022, respectively). To mitigate bias, the evaluation was blinded to include only the pre/post status, BPA code, and verbatim defect description. An RNAB who routinely performs clinical case review conducted the first round of evaluation and categorized cases as "Accurate," "Not Accurate," or "For Review." Cases categorized as For Review, all of which were pre-implementation, were then jointly reviewed by the OBDR Program Manager and the RNAB for consensus. A final unblinded evaluation was conducted utilizing all case information to best categorize the For Review cases as Accurate or Not Accurate. After the final review, 75% (82/109) of the pre-implementation cases and 84% (77/92) of the post-implementation cases were coded accurately.

Although a chi-square test indicated this was not a statistically significant difference ($p<0.14$), the new tool provides superior defect description and resulted in an 11.25% increase in the proportion of cases coded accurately. Additionally, defect descriptions for a majority of the post-implementation cases included relevant documentation of characteristics in the order in which they appear in the coding tool. This suggests the tool also serves as an aid to prompt RNABs to document pertinent details, which will likely result in reduced quality assurance re-abstraction requests.

Effects of modified algorithm for pulse oximetry screening on missed critical congenital heart defects in Utah infants: a retrospective analysis

Poster Session Presenters: Stephanie Pocius and Aubree Boyce
Co-Authors: Stephanie Robinson

The American Academy of Pediatrics (AAP) recommended a new algorithm for critical congenital heart defect (CCHD) screening starting January 2025. The new algorithm requires a passing oxygen saturation threshold of >= 95% in both pre-ductal and post-ductal measurements and recommends one retest instead of two for those infants who did not pass the first screen. Missed CCHD diagnosis can lead to increased risk of morbidity and mortality in infants. The goal of this analysis is to determine if factors associated with missed CCHD diagnosis can be identified and potentially mitigated. The new algorithm was applied to pulse oximetry screening data from Utah infants born between 2018 and 2022. A total of 82 infants born with a CCHD in Utah were missed by pulse oximetry screening when following the original algorithm. Eleven infants that were originally missed were captured when applying the updated AAP guidelines, representing a 13% decrease in the number of missed CCHD cases.

Descriptive statistics and chi-square analysis showed a significant majority of the additional CCHD cases that were captured using the updated AAP guidelines were screened in urban facilities with Level II+ NICUs and higher birth volumes (82%). Certain diagnoses, such as coarctation of the aorta, were also more frequently caught using the updated AAP guidelines. The results from this analysis highlight the benefit of following the updated AAP guidelines, as additional CCHD cases can be identified and treated in a timely manner to prevent severe morbidity and mortality. However, additional investigation will be needed to determine why there was not a significant decrease in missed CCHD cases in rural settings with Level I NICUs and lower birth volume.

Birth Defects and Newborn Screening Longitudinal Follow-up: A look back on a fruitful partnership for Critical Congenital Heart Disease

Poster Session Presenter: Heather Pint
Co-Authors: Sara Lammert, Barbara K Frohnert, and Sook Ja Cho

The Children and Youth with Special Health Needs (CYSHN) program at the Minnesota Department of Health incorporated a Critical Congenital Heart Disease (CCHD) Follow-up Specialist Public Health Nurse when CCHD screening was added to the Minnesota (MN) newborn screening panel. This position is shared between the Newborn Screening Longitudinal Follow-Up (LFU) unit and the Birth Defects (BD) unit both located in the CYSHN program. The poster describes the benefits of ongoing collaboration related to surveillance and follow-up spanning identification through transition to early adulthood between BD and LFU.

The relationship provides a bridge between LFU and BD. A swim lane diagram was developed to map the process flow of information between units. Both units have public health evaluation measures that require finding babies with CCHDs both for surveillance and connecting families to resources. By streamlining overlapping processes both areas improve surveillance and family outreach outcomes.

The CCHD Follow-up Specialist:

- Provides expertise using clinical and public health best practice recommendations and training.
- Provides timely reports to birth defects surveillance of newborns with CCHDs identified through newborn screening (i.e. those not identified prenatally).
- Builds strong relationships with pediatric cardiologists performing clinical review of surveillance cases.
- Develops cardiac specific resources for families with a child with a CCHD.
- Leads workgroups of cardiac specialist staff, community organizations and parent support groups, and families with CCHDs.

The cardiac-focused collaboration between LFU and BD surveillance allows for improved and streamlined processes and timely referral to local public health agencies for connecting families to community resources. Through this collaboration, the teams participate in projects such as the CHSTRONG KIDS survey to improve understanding of families' needs for healthcare use, education, social experiences, and quality of life. Published studies and surveillance standards in turn provide data to improve the systems that serve the entire cardiac population. The teams are also exploring new opportunities, such as electronic case reporting (eCR) for case finding efficiency and expanding the Early Childhood Longitudinal Data System (ECLDS) to include children with CCHDs to answer questions about development and learning outcomes.

Folic Acid Distribution in Oklahoma

Poster Session Presenter: Flor Rodriguez
Co-Authors: Laura Shodall, Mary Green, and Lindsay Denson

The Oklahoma Birth Defects Registry (OBDR) relies on local county health departments (CHDs) to provide neural tube defect (NTD) prevention education and distribute folic acid to women across the state. However, some women of childbearing age still lack access to, or awareness of, the benefits of folic acid use before and during early pregnancy for NTD prevention. The OBDR sought to increase awareness of NTD prevention, promote CHD resources, and expand folic acid distribution in Oklahoma by developing a direct folic acid distribution program. In 2023, the OBDR collaborated with multiple departments within the Oklahoma State Department of Health (OSDH) including Nursing Service, Office of General Counsel, Pharmacy, and Office of Minority Health to develop protocols for a direct folic acid distribution program. In addition to obtaining a standing order from the OSDH Chief Medical Officer, updating OSDH Nursing Service public health nursing protocols, obtaining consent language from general counsel, implementing a process to obtain multivitamins from the OSDH pharmacy, and incorporating approved Spanish translations, the OBDR created a Microsoft Power App for documentation of pertinent demographics and consent. From March 7, 2023-November 20, 2024, OBDR staff directly distributed 466 bottles of 100-day supply multivitamins with folic acid at health fairs, at community events, and to university students across the state.

The majority of the bottles were distributed to women 25-34 years of age (41%) and to women who lived in the Central region of the state (46%). From March 7, 2023-November 20, 2024, CHDs distributed 25,560 bottles of 100-day supply multivitamins with folic acid. Unlike the OBDR direct distribution program, the majority of these bottles were distributed to women less than 25 years of age (46%) and to women who lived in the Northeast region of the state (25%). Chi-square tests for homogeneity indicated both age and region of residence differed when comparing the CHD and OBDR multivitamin distribution programs ($p<0.0001$).

These comparisons suggest the OBDR achieved the goal of providing NTD prevention education and distributing folic acid to a different subset of the population than those served at CHDs, which also provided an opportunity for the OBDR to promote CHD resources.

Gastroschisis-Related Mortality in Oklahoma

Poster Session Presenter: Lindsay Denson
Co-Authors: Amanda E. Janitz, Stephanie L. Pierce, Jennifer D. Peck, Daniel Zhao, and Deirdra R. Terrell

It is unknown if gastroschisis-related mortality in Oklahoma has decreased over time, similar to national trends, or if gastroschisis-related mortality differs between the two largest cities in Oklahoma. The goal of this study was to expand the almost non-existent literature regarding gastroschisis-related mortality in Oklahoma. Gastroschisis cases were identified by the Oklahoma Birth Defects Registry and linked with both birth and death certificate data. Gastroschisis-related mortality was defined as the total number of liveborn infants with gastroschisis who died within the first year of life divided by the total number of liveborn infants with gastroschisis born in the same birth year, 1994-2019.

Join point regression was used to assess changes in gastroschisis-related mortality trends. Modified Poisson regression was used to estimate risk ratios to determine if gastroschisis-related mortality differed by delivery location or by maternal and infant characteristics. In Oklahoma from 1994-2019, the one-year gastroschisis-related mortality was 3.9% (95% confidence interval [CI] 2.6-5.7) and the annual percent change in gastroschisis- related mortality was -8.65% (95% CI -11.51, -5.70). When compared to infants with gastroschisis born in Oklahoma City, infants born with gastroschisis in Tulsa were not more likely to die within one year of birth (risk ratio 0.53 [95% CI 0.20-1.40]). After adjusting for birth year and birth weight, infants born to minority women were 2.81 (95% CI 1.31-6.06) times more likely to die within one year of birth compared to those born to non-Hispanic White women. These results suggest an increase in prenatal detection, allowing for better delivery planning, and improvement in postnatal treatment in Oklahoma over time.

The finding of no difference in gastroschisis-related mortality between deliveries in the two largest cities in Oklahoma may give families the confidence to deliver at specialty care-appropriate hospitals closer to home, reducing the financial and emotional burdens on families impacted by gastroschisis. Future studies are needed to further investigate the racial disparities related to mortality among infants born with gastroschisis in Oklahoma.

Assessment of Burden of Additional CDC Reportable Fields on Case Abstraction Process in Minnesota

Poster Session Presenter: Sook Ja Cho
Co-Authors: Michele Hort, Jasmin Wenigha, and Barbara Frohnert

Sixteen new fields were added to the MN surveillance in May and September 2022 to meet the reporting requirement for the birth defects surveillance grant (Notice of Funding Opportunity CDC-RFA-DD21-2101) received from the Centers for Disease Control and Prevention (CDC). This resulted in more time needed to abstract new cases and created a large backlog of cases already completed that needed maternal chart reviews to backfill the new fields.

Timeliness was assessed to determine how the additional CDC reportable fields impacted our case abstraction process.

Any case born in 2018 through 2024 were classified into two groups based on the eligibility for CDC reporting.

Monthly means in days were calculated using three date fields to measure the routine case abstraction process, and extra time needed to collect the additional CDC fields: 1) Date case entered into the surveillance system, 2) date regular abstraction complete, and 3) complete date for additional CDC fields. The number of cases eligible for CDC reporting with incomplete data fields was measured over time to assess how extra workload tapered off as staff caught up the case backlog.

About 25% (3,325/13,516) of cases in the analysis were eligible for CDC reporting.

Over 1,600 cases with incomplete fields were identified in 12/2022 shortly after new fields were added. This decreased to about 90 cases by 12/2023, and became steady in 2024, indicating over a year to catch up the case backlog.

For CDC reportable cases, monthly means in days from date case entered in the surveillance to case abstraction complete stayed stable (range: 13-111 days), similar to non-CDC reportable cases.
For cases completed before the new fields were added, it took hundreds of days (range: 150-1,580 days) until additional CDC fields were completed, while this extra time decreased substantially for cases initiated after the new fields were added.

Although thousands of previously completed cases had incomplete fields shortly after the new fields were added, staff caught up the backlog in just over a year. The extra time is still needed to complete the additional CDC fields, which may affect other timeliness measures, such as overall case completion.

Orofacial Cleft Case Review – A Pilot Study in Louisiana

Poster Session Presenter: Julie Johnston
Co-Author: Tri Tran

Understanding risk factors and causes of birth defects plays an important role in primary prevention. Risk factors and causes of birth defects may be identified through case review, a critical analysis of patient's medical history. Currently, Louisiana does not have a standardized case review model for birth defects. A pilot study using orofacial cleft defects was conducted to develop the case review model. The purpose of the study was to (1) identify the causes of orofacial clefts through case review, and (2) evaluate effectiveness of the case review model in identifying causes.

The case review used multiple data sources including hospital discharge reports, vital records (birth, death and fetal death), medical records and Medicaid data. Medical records included prenatal, delivery, postnatal records including genetic and autopsy reports. Maternal exposures included medication, substance misuse, environmental, occupation, fevers from illness and infections, pregnancy-related conditions, family history of clefts, pre-conception period factors, earliest HbA1C and prenatal screening, tests or procedures. A case review tool with compiled data facilitated the review. Review panelists included pediatricians, epidemiologists, Maternal Fetal Medicine and public health stakeholders. Staff conducted reviews in October 2022. Cases were limited to children without chromosomal defects and with mothers diagnosed with chronic diabetes, a known risk factor. Among 2017-2019 births, 291 children (1.6 per 1,000 live births) had orofacial clefts. Of those, nine cases had the following characteristics: two cleft palate without cleft lip, four cleft lip without cleft palate, three cleft lip with cleft pate, all live births and singletons, four black, two white, two Hispanic, five male, three preterm birth and two deceased.

Resulting review did not identify one specific cause for all cases due to lack of information from available data sources, and lack of a clear pathway connecting the birth defects with causes. Study lessons learned indicated an unrealistic initial goal of identifying causes within the practice of birth defects case review. Revised goals should target timeliness of diagnosis, treatment and referral to services. These variables exist in medical records and do not require new sources of data.

Exploring the Utility of Electronic Case Reporting Data in Birth Defects Surveillance: Lessons Learned from Three Pilot Projects

Poster Session Presenter: Rebecca Howell
Co-Authors: Rachel Allred and Charles Shumate

Background: The introduction of birth defects into the electronic initial case reporting (eICR) framework is a recent innovation with implications for birth defects registries. While potentially useful for case identification to improve timeliness, the utility of eICR in birth defects surveillance is currently unknown. The purpose of this study was to identify initial challenges in using eICR data through three pilot assessments: 1) for repeat eICRs, determining if the eICR with the latest timestamp was the most comprehensive [i.e., included all data from eICRs triggered at earlier timestamps], 2) for one chosen case with three birth defect triggering codes, determining if all birth defects were included across all eICRs, and 3) describing average eICR file size by page count.

Methods: (1) Ten pairs of repeat eICRs (i.e., two eICRs with the same admission date but differing in timestamp by at least one second) were compared using document comparison software (Adobe and Microsoft Word). (2) Eight eICRs for a selected case with three unique birth defect triggering codes (limb reduction, gastroschisis, cleft lip with cleft palate) were manually reviewed to determine if all three birth defects were present in each eICR. (3) A random sample of 15 unique eICRs was used to calculate average eICR page count by encounter type and facility.

Results: For assessment (1), data cumulativeness varied in an eICR section-specific manner, where some sections (e.g., Results) were cumulative, while others were not (e.g., Medications Administered and Miscellaneous Notes). For assessment (2), among the eight eICRs, none included all three defects. For assessment (3), for the same facility, average eICR page number was >1000 for inpatient encounters and <20 for outpatient and emergency encounters. For the same encounter type, average page number varied by facility.

Conclusions: These pilot assessments highlight the potential of eICR data to support birth defects surveillance while identifying key challenges such as data completeness, file size variability, and discrepancies between facilities in data structure and richness. Findings suggest opportunities to enhance surveillance processes through integration of eICR data, but data organization improvements, standards, and definitions are needed.

Surveillance of Fetal Deaths Affected by Birth Defects in New Jersey

Poster Session Presenter: Jing Shi
Co-Authors: Elena Napravnik, Shaila Montero Castro, and Lisa Stout

Background: Accurately assessing the prevalence of major structural birth defects requires comprehensive surveillance of both live births and fetal deaths. Birth defects are significant contributors to fetal mortality, and failure to include fetal deaths in birth defects surveillance can result in underreporting of the true burden of these conditions. Including fetal deaths will improve prevalence rates estimates, especially for defects in which a large proportion of cases result in fetal deaths.

Methods: New Jersey has the statutory authority to obtain reports of fetal deaths affected by birth defects for fetuses greater than or equal to 15 weeks' gestation. We began ascertainment of fetal deaths in November 2021 by developing a separate module in the Birth Defects and Autism Reporting System – our electronic database. In this study we examined the percentages of contribution of fetal deaths for different birth defect conditions and other characteristics for fetal deaths affected by birth defects.

Results: We have 5% of birth defects come from fetal deaths and 95% come from live births. The percentage of fetal deaths among all registrations is highest for Trisomy 18 (35%), Trisomy 13 (31%), Anencephaly (30%), followed by Hypoplastic left heart syndrome (15%), Omphalocele (13%), Turner syndrome (13%) and Transposition of the great arteries (10%). For all fetal death registrations with birth defects 57% are affected by chromosomal disorders, followed by cardiovascular conditions (13%). 19% have birth defects affecting multiple body systems; 43% are male, 48% are female and 9% are with unknown gender; 6% are multiple births and 94% are single births. Most fetal death registrations do not have the birth weight information, however for those with the information, 68% are with birth weight under 1000 grams.

Discussion: The results underscore the importance of including both live births and fetal deaths in birth defect surveillance to obtain an accurate estimation of the prevalence and impact of congenital anomalies. Limitations to case ascertainment include, the de-identification of fetal death cases in the birth defects registry, which makes it difficult to further validate the data.

Barriers to Accessing Specialized Genetic Services Faced by Families of Children with Birth Defects: Perspectives from Pediatric Genetics Providers

Poster Session Presenter: Dayana Betancourt
Co-Authors: Caitlyn Yantz, Henal Gandhi, Charles Shumate, and A.J. Agopian

Background: Clinical geneticists and genetic counselors can help families of children with birth defects understand their child's condition and guide their medical treatment; however, our recent survey of 400 families of young children with select birth defects in Texas found that 65% reported not accessing a clinical geneticist, genetic counselor, or visiting a genetics clinic. To contextualize these results, understand barriers to accessing services, and inform future initiatives, we conducted interviews with clinical geneticists and genetic counselors in Texas.

Methods: Structured interviews were conducted, guided by questions organized into four domains: 1) processes by which clinical referral systems should ideally operate, 2) barriers to accessing genetic services faced by families, 3) guidance for the development of an intervention, and 4) contextualizing our survey results. Transcripts for each interview were reviewed independently by two reviewers to identify and summarize recurring themes within each domain.

Results: Seven interviews have been completed. Based on thematic analysis, barriers to accessing genetic services faced by families were reported to be multi-factorial, including insurance limitations, long waitlists, lack of referrals from primary care providers (PCPs), and limited awareness about the benefits of genetic testing and counseling among families.

Geographic challenges were reported to particularly affect access for families residing in rural areas or along the Texas-Mexico border, compounded by transportation difficulties and a national shortage of geneticists. Suggested future interventions include culturally tailored multi-lingual outreach campaigns (social media campaigns, information materials) about the benefits of genetic services for children with birth defects and targeted outreach for PCPs to increase referrals.

Conclusion: Our initial findings suggest that barriers to accessing clinical geneticists and genetic counselors are complex and our findings may inform development of a future outreach campaign focused on increasing awareness about the benefits of genetic services among families of children with birth defects and other healthcare providers.

Evaluation of Data Sources for Critical Congenital Heart Defect Case Ascertainment in North Carolina

Poster Session Presenter: Helen Parker Atkinson
Co-Authors: Kristin Bergman and Nina Forestieri

Critical congenital heart defects (CCHDs) have significant morbidity and mortality.
Surveillance of these defects is important for research and public health practice. The recent implementation of universal pulse oximetry (POX) testing may provide an additional useful resource for improving the completeness of surveillance data. This analysis evaluates the utility of multiple data sources, including POX records, for identification of potential CCHD cases. Using the cohort of CCHD cases confirmed by the North Carolina Birth Defects Monitoring Program (NCBDMP) born 2018-2021 (N=1035), sensitivity values were calculated for the birth, infant death, and fetal death certificates; hospital discharge records; and POX records. Sensitivity was defined as the percentage of confirmed CCHD cases identifiable from a given data source. The following were criteria for a case being considered identifiable: for infant and fetal death certificates and discharge records, designation of any cause of death or diagnosis code indicating a heart defect; for the birth certificate, selection of the CCHD checkbox; and for the POX record, a result of Fail, Not Screened with an abnormal echo, infant deceased, or other reason with supplemental information indicating a heart defect. Sensitivity values were also stratified by demographic and clinical characteristics of cases.

The sensitivity of the data sources for case identification varied widely: birth certificate (6.7%; 95% confidence interval [CI]: 5.1-8.3); fetal death certificate (29.8%; 95% CI: 18.0-41.7); POX record (59.5%; 95% CI: 56.2-62.8); infant death certificate (73.0%; 95% CI: 66.7- 79.3); and discharge record (88%; 95% CI: 85.9-90.1). Sensitivity values for some data sources were significantly higher for cases delivered in larger hospitals or transferred after delivery (discharge record), admitted to the neonatal intensive care unit (birth certificate, POX record, and discharge record), or prenatally diagnosed (POX record and discharge record). The hospital discharge record was the most sensitive overall. The POX record was only moderately sensitive, in part due to missing records, limiting its added value for case finding, though improvements in reporting may increase its utility. To help optimize staff time and resources, surveillance programs should consider the utility of supplemental data sources for CCHD case ascertainment.

Comparing Congenital Heart Disease Prevalence in States with Varying Health Coverage, Access, and Affordability for Women of Reproductive Age

Poster Session Presenter: Adam Zbib
Co-Author: Abeer Hamdy

When compared to other congenital anomalies, Congenital Heart Disease (CHD) was highly associated with mortality before the age of fourteen. The etiology of CHD is believed to be multifactorial with notable maternal pregnancy risk factors being obesity, smoking, alcohol use, diabetes, rubella, teratogenic medication use, and organic solvent exposure. However, healthcare access and coverage for women of reproductive age is known to be widely variable between states. The purpose of this study was to examine the relationship between CHD prevalence and state-dependent healthcare coverage, access, and affordability for women of reproductive age.