ID: 88809463
Birth Defects and NAS - Host Site Description
Massachusetts Department of Public Health

Assignment Location: Boston, US-MA
Massachusetts Department of Public Health
Bureau of Family Health and Nutrition/Division for Family Health Data and Analytics

Primary Mentor: Mahsa Yazdy, BS, PhD, MPH
Director
Massachusetts Department of Public Health

Secondary Mentor: Eirini Nestoridi, BS, MD
Associate Director
Massachusetts Department of Public Health

Work Environment
Hybrid

Assignment Description

The Fellow will join the Division for Family Health Data and Analytics (DFHDA) team, within the Bureau of Family Health and Nutrition (BFHN) at the Massachusetts Department of Public Health (MDPH). The Division's mission is to gather, analyze, and interpret maternal and child health data to inform policy and programs and prepare tomorrow's researchers to carry the Division's work forward. The Division houses multiple surveillance and research efforts: the Birth Defects Monitoring Program (BDMP), Neonatal Abstinence Syndrome (NAS) Surveillance System, the Maternal Mortality and Morbidity Review (MMMR) Team, and data from two national case-control studies on birth defects (the National Birth Defects Prevention Study [NBDPS] and the Birth Defects Study To Evaluate Pregnancy exposureS [BD-STEPS]), the Pregnancy Risk Assessment Monitoring System (PRAMS) and the Pregnancy to Early Life Longitudinal (PELL) data system.

The BDMP was established in 1999 and is a statewide, active, population-based active surveillance program for birth defects among Massachusetts residents. Highly trained medical record abstractors who review maternal and infant medical records, collect information on diagnostic tests, autopsy reports, and lab results to confirm diagnoses. Relevant demographic, clinical, family history and birth characteristics are also recorded. The BDMP is a rich data source that can be utilized to monitor the prevalence of birth defects across the state and to better understand the causes of birth defects. Launched in April 2020, the NAS surveillance system is also a state-wide, active, population-based system that collects data on infant signs and treatment, medication or substance use during pregnancy, and social factors such as housing stability for all Massachusetts residents to provide timely and accurate data on NAS in MA in order to better understand the impact of NAS.

The Division participated in the NBDPS and currently participates in the BD-STEPS; both of these are case-control studies aimed at understanding the causes of birth defects. BD-STEPS was recently expanded to also focus on understanding risk factors associated with stillbirths without birth defects. Together NBDPS and BD-STEPS are the largest population-based studies on birth defects ever undertaken in the U.S. and include data from over 43,000 telephone interviews. These data are available for etiologic research examining risk factors for birth defects, as well as risk for stillbirths.

The Division began working closely with the Bureau of Infectious Diseases and Laboratory Sciences (BIDLS) at the time of the Zika Virus epidemic and has continued that collaboration through the CDC's Surveillance for Emerging Threats to Mothers and Babies Network (SET-NET). As part of SET-NET, the Division led the COVID-19 Pregnancy Surveillance and assessed the impact of COVID-19 on pregnant people and their infants while currently collaborates on hepatitis C virus

pregnancy surveillance. The Fellow will have the opportunity to participate in the SET-NET efforts, as well as on collaborative projects related to pregnancy and infectious diseases.

The Division is also home to PRAMS, a CDC-Massachusetts Department of Public Health (MDPH) collaboration regarding experiences, attitudes, and beliefs shortly before, during, and after pregnancy. MDPH has been conducting PRAMS since 2007 and surveys approximately 2,500 new mothers each year on topics including preconception health, prenatal care, maternal mental health, oral health, infant sleep practices, breastfeeding, and social support. Data are weighted to represent Massachusetts births and findings are shared regularly with our clinical and community partners including Massachusetts Perinatal-Neonatal Quality Improvement Network (PNQIN) and March of Dimes, through our PRAMS Advisory Committee and posted at mass.gov.

The Pregnancy to Early Life Longitudinal (PELL) database is also housed in DFHDA. PELL comprises the linkage of the maternal hospital delivery administrative record, the infant's birth certificate, and the infant's birth hospitalization administrative record. To this, both maternal and infant subsequent hospital-based healthcare administrative records are linked (hospitalizations, emergency department visits, and observational stays) longitudinally. These records date from births in 1998 and can be linked to PRAMS to provide a robust dataset of maternal and infant experiences. PELL is also linked to records from the BDMP, and records are also linked to the state cancer registry, Early Intervention, and the state Universal Newborn Hearing Screening data systems as needed. The Fellow will have access to these datasets as needed to support projects.

In addition to data within the Division, data outside the Division are available for analyses and include data through such programs as the Massachusetts Home Visiting Initiative and Early Intervention. The Fellow will also have the opportunity to collaborate with staff and students from the MDPH, local universities including Harvard University, Boston University, Tufts University, and the University of Massachusetts.

The Fellow will be able to conduct epidemiological studies from a diverse array of data systems and the Division will work with the Fellow to tailor their experience based on their interest and expertise to ensure a fulfilling experience. In addition, the Fellow will have the opportunity to present their work internally as well as externally (e.g., the Advisory Committees, national conferences and meetings, local universities) and will be encouraged to publish their work in peer-reviewed journals.

DFHDA also conducts needs assessments and provides crucial data and decision support for HRSA's Title V MCH Block Grant and State Systems Development Initiative annual application and report. The Division is committed to performance management and quality improvement and works closely with the Office of Population Health to implement the Public Health Data Warehouse (PHD). PHD is a longitudinal dataset that is a partnership with other state agencies including Medicaid, Department of Mental Health, Department of Corrections, Department of Transitional Assistance, MA Cancer Registry and the Prescription Monitoring Program, and various programmatic data systems including data from the Women, Infants, and Children (WIC) Nutrition program, and the Early Intervention program. The Division is home to 10 epidemiologists, 9 abstractors, and 5 support staff. While most staff hold an MPH, there are also 5 doctoral-level epidemiologists and 1 MD.

Day-to-day activities will include:

- Literature reviews
- Developing research and analytic plans
- Data cleaning and conducting analyses including data linkages
- Preparing and presenting results of their analyses
- Preparing and leading meetings
- Writing and submitting IRB applications, as needed

- Meeting and providing updates to mentors and collaborators
- Working with programs and initiatives to use the Division's data to inform their work
- Writing, submitting, and reviewing reports and manuscripts
- Presenting to internal and external collaborators
- Participating in meetings, trainings and webinars
- Joining one or more of the Division's quality improvement team projects
- Assisting with mentoring student interns

Describe Statistical and Data Analysis Support, Such as Databases, Software, and Surveillance Systems Available to the Fellow

The Division for Family Health Data and Analytics (DFHDA) recently migrated the majority of our datasets to SAS Studio, allowing for web-based analyses and data sharing. Support using SAS is available both within DFHDA and through a MDPH-wide SAS user group. Additionally, once a month the Division has research meetings where epidemiologists solicit feedback on their projects and are able to discuss any data issues or programming challenges they have encountered.

The Birth Defects Monitoring Program (BDMP) data are currently stored in an Access database and the Fellow will have the opportunity to learn to access, query, and retrieve data from the system. The system is being upgraded to an Apex database and the Fellow will have the opportunity to participate in trainings on how to utilize this state-of-the-art data system; once the transition is complete, the Fellow will be able to actively use and query and retrieve data from the new system.

The Division currently uses REDCap databases for the NAS Surveillance System; therefore, the Fellow will gain familiarity with querying REDCap, pulling data from the system, as well as modifying the data collections tools, as needed.

Projects

Surveillance Activity Title: Expanding Multisource Ascertainment in the Massachusetts Birth Defects Monitoring Program

Surveillance Activity Description:
The Massachusetts Birth Defects Monitoring Program (BDMP) is an active statewide surveillance system for birth defects. Potential cases of birth defects are ascertained from multiple sources including: birthing and non-birthing hospitals, clinical geneticists and genetic counselors, commercial laboratories, selected outpatient records, emergency departments, pathology departments, and vital records. Medical records for all reported cases are reviewed and if a case meets the inclusion criteria, the data are abstracted by highly-trained abstractors who pursue various sources to confirm the diagnosis of a birth defect and collect relevant demographic, clinical, family history and birth characteristics. The BDMP strives for complete ascertainment of infants and fetuses diagnosed with a birth defect, which is why multisource reporting and ascertainment are essential. As testing and screening improve over time, birth defects are being diagnosed earlier in pregnancy; therefore, the potential for missing cases due to early losses is always a concern.

The Division has already identified a potential new source of reporting through external laboratories that conduct genetic tests; the Division plans to pilot the incorporation of two external laboratories into BDMP's reporting sources. Specifically, for 15 years, the BDMP has received reports from one LabCorp laboratory that provide essential confirmatory evidence for chromosomal abnormalities identified through genetic testing during pregnancy, on products of conception, and in infants. Labcorp operates multiple specialized laboratories across the U.S., each offering different types of genetic tests and screening. To improve completeness of reporting, we have begun discussions with Labcorp to add two additional data sources: (1) the laboratory that performs only non-invasive prenatal screening (NIPS) and (2) the

laboratory that conducts pregnancy related genetic testing not currently captured in our existing reports. We are particularly interested in adding NIPS because it has been widely used since 2012 and is now universally offered to pregnant people, making it a valuable addition to BDMP's surveillance. Although NIPS is a screening test, its positive predictive value is very high for Trisomy 21, 13, and 18, which allows BDMP to count early pregnancy losses with NIPS-only results, without any additional clinical or diagnostic evidence, as true cases for these conditions.

Incorporating these additional genetic test and screening results is also important because one of BDMP's current ascertainment sources, hospital reports, provides us early pregnancy losses miscarriage-related ICD-10 CM codes not specific to a chromosomal abnormality. Access to these new laboratory data sources would allow us to identify which potential cases should be prioritized for medical record abstraction, thereby increasing timeliness and overall productivity. The Fellow would support the integration of these new reporting sources as well as assess the feasibility and challenges in integrating the new ascertainment source into the BDMP, and the potential yield of new cases. The Fellow would also evaluate if these new data sources allow us to better estimate the true prevalence of chromosomal abnormalities and streamline medical record abstraction to increase timeliness and productivity.

Surveillance Activity Objectives:
Project Objectives:

- The objective of this project is to broaden BDMP's ascertainment sources by incorporating additional external laboratory data sources to ensure that chromosomal abnormalities diagnosed early in pregnancy, including those resulting in early pregnancy losses, are not missed and can be more accurately identified, prioritized for review, and used to better estimate their true prevalence.

Expected Deliverables:

- Integrate two new ascertainment sources into the BDMP.
- Assess if case ascertainment has improved and if cases were identified that would otherwise have been missed.
- Evaluate if the new sources reduce the workload of medical record abstraction.

Surveillance Activity Impact:
Accurate estimates of the prevalence of birth defects are essential for assessing the impact of new medications, new vaccines such as COVID-19 vaccine, or infections such as SARS-CoV-2 on fetal development, identifying potential community clusters, and planning for public health services. However, prevalence estimates are only as accurate as the completeness and quality of the underlying data. When cases, particularly those diagnosed early in pregnancy or resulting in early pregnancy loss, are missed, prevalence estimates underestimate the true burden and may obscure important trends or emerging conditions. By expanding BDMP's ascertainment sources to include additional external laboratory data, especially NIPS, this project aims to ensure to increase the completeness of case ascertainment. Doing so will improve our ability to more closely estimate the true prevalence of chromosomal abnormalities and enhance confidence in the data used for surveillance, research, and public health decision-making.

Surveillance System Evaluation Title: Evaluation of the Massachusetts Hepatitis C Virus (HCV) Pregnancy Surveillance System

Surveillance System Evaluation Description:
The Division for Family Health and Data Analytics (DFHDA) has been collaborating with the Bureau of Infectious Diseases and Laboratory Sciences' (BIDLS) on the hepatitis C virus (HCV) pregnancy surveillance as part of the Surveillance for Emerging Threats to Mothers and Babies (SET-NET). For this surveillance effort, detailed information on the pregnant person-infant dyad is collected through two years of life. This work is based on data linkages (i.e., vital records and laboratory reporting), as well as medical record abstraction; together these sources provide essential insight into maternal infection, infant follow-up, and early childhood outcomes.

ID: 88809463
Birth Defects and NAS - Host Site Description
Massachusetts Department of Public Health

To strengthen the quality and efficiency of HCV pregnancy surveillance, the Fellow will focus on evaluating the current HCV pregnancy surveillance through SET-NET and identify the most reliable data sources for key maternal and infant variables.

Surveillance System Objectives:
Project Objectives:

- The objective of this project is to streamline data collection to better support analysis and dissemination of data on the monitoring of HCV in pregnancy.

Expected Deliverables:

- Assess the completeness of selected variables on the HCV pregnancy surveillance system.
- Assess the validity and utility of each data source, including laboratory reporting data, birth certificate data, and medical record abstractions, for specific maternal and infant variables.
- Recommend the preferred data source for each variable of interest, helping to optimize workflows and improve the accuracy and completeness of HCV pregnancy surveillance.

Surveillance System Impact:
Rates of hepatitis C virus (HCV) infection among reproductive-aged adults in the U.S. have increased in recent years, reflecting the rise in injection-drug use associated with the opioid crisis. Although national guidelines now recommend universal HCV screening during each pregnancy, data remain limited and inconsistent regarding the broader impact of maternal HCV infection on pregnancy and infant outcomes beyond perinatal transmission. This underscores the urgent public health need for reliable, timely surveillance systems capable of producing accurate, complete, and actionable data.

Strengthening and streamlining data collection, analysis, and dissemination is essential. High-quality surveillance data enable public health systems to monitor trends, allocate resources effectively, evaluate prevention strategies, and ensure that pregnant patients and their infants receive timely, evidence-based care.

Major Project Title: Outcomes among families with an infant diagnosed with neonatal abstinence syndrome (NAS)

Major Project Description:
Studies have found that children with NAS have a higher rate of death, in particular from external causes. Additionally, the 10-year mortality risk among birthing parents who had a child with NAS has been found to be 10-12 times higher than control populations. Massachusetts NAS surveillance data shows that 27 infants diagnosed with NAS delivered between 4/1/2020-12/31/2024 died within approximately 6 months after birth. The aim of this project is to assess mortality among families affected with NAS. The family will be the unit of measure and mortality will be evaluated among the birthing parent, the non-birthing parent, and the infant. We will also explore the ability to link to siblings and assess their outcomes as well. In addition, the fellow will compare the characteristics of families affected by NAS who experienced a death to the overall population of families affected by NAS in Massachusetts and assess service utilization (e.g., Early Intervention) for these children. Given that Massachusetts has a Maternal Mortality and Morbidity Review (MMMR) and Child Fatality Review Committees as well as a Fetal Infant Mortality Review Committee, that is currently under development, we will also explore whether we can obtain more detailed cause of death and recommendations that come from reviewing any of the deaths identified in this population. This project will also provide the fellow with the opportunity to closely collaborate with the current CSTE fellow whose work focuses on the Fetal and Infant Mortality Review Committee.

ID: 88809463
Birth Defects and NAS - Host Site Description
Massachusetts Department of Public Health

Major Project Objectives:
Project Objectives:

- The objective is to assess mortality among infants with NAS and their family members, including birthing parents, non-birthing parents, and siblings, by linking vital records and by comparing characteristics and service utilization between families who experienced a death and those who did not, to identify risk factors, disparities, and opportunities for prevention.

Expected Deliverables:

- Conduct a comprehensive literature review on mortality and long-term outcomes among families affected by NAS or perinatal substance exposure.
- Perform linkage between birth certificates of infants with NAS and death records for infants, birthing parents, non-birthing parents, and, if feasible, siblings.
- Calculate mortality rates and compare them to appropriate control or statewide populations.
- Summarize findings from relevant review committees for deaths occurring within this cohort.
- Analyze cause of death patterns and identify any common risk factors.
- Assess service utilization patterns (e.g., Early Intervention enrollment) and compare families with and without a death.
- Prepare a written report and develop presentations for internal and external partners, including committees involved in maternal and child health surveillance.

Major Project Impact:
This project fills a critical gap by examining mortality across the entire family unit affected by NAS, rather than only focusing on outcomes of infants diagnosed with NAS. Understanding mortality patterns among infants diagnosed with NAS, birthing parents, non-birthing parents, and siblings will help identify disparities and unmet needs. These findings can guide prevention strategies, inform service and resource allocation, and strengthen public health recommendations aimed at improving long-term outcomes and reducing preventable deaths among families affected by NAS in Massachusetts.

Additional Project #1 Title: Immunization Coverage among Children with Birth Defects
Project #1 Type: Major Project

Project #1 Description:
Immunizations are regarded as one of the most effective public health interventions for preventing illness in young children and have been associated with substantial reductions in morbidity and mortality. Vaccinations remain a cost-effective and safe means of decreasing the risk of vaccine-preventable infectious diseases and interrupting their transmission and outbreaks in the community. In Massachusetts, MDPH maintains a longstanding commitment to promoting evidence-based immunization practices and currently recommends that clinicians use the American Academy of Pediatrics (AAP) Recommended Child and Adolescent Immunization Schedule as the primary reference for routine immunization of infants, children, and adolescents. The AAP schedule is developed by pediatric experts, updated regularly, and reflects the most current scientific evidence and clinical best practices.

In the current environment, where misinformation, inconsistent guidance, and non-evidence-based recommendations can undermine public confidence, clear, science-based immunization schedules are essential for protecting children and communities. Monitoring immunization coverage is equally critical, as delays or gaps in vaccination can lead to preventable infections, hospitalizations, long-term complications, and death. Despite the fact that chronic underlying conditions, including birth defects, may make children more susceptible to infections and complications, little is known about vaccine coverage among children with birth defects. Limited studies have reported lower vaccination rates among children with Down syndrome or congenital heart defects compared with children without birth defects, underscoring the need for timely and accurate monitoring.

ID: 88809463
Birth Defects and NAS - Host Site Description
Massachusetts Department of Public Health

This project will explore immunization coverage among children with birth defects in Massachusetts to help ensure that all children receive the protection they need and to support evidence-based public health decision-making.

Project #1 Objectives and Expected Deliverables:
The Massachusetts Immunization Information System (MIIS) is a complete, accurate, secure, real-time immunization record for residents of Massachusetts of all ages. The Fellow will facilitate linkage of immunizations records of all children with MA birth certificates and BDMP records for a period of 10 years between 2014 and 2023 and then compare the vaccination coverage between children with selected birth defects and children without a birth defect. They would examine whether the rates of vaccination of children with birth defects differ by vaccine type, by birth defect severity (i.e., isolated versus complex versus multiple birth defects), or by sociodemographic characteristics (e.g., maternal age, race/ethnicity, country of birth, education, insurance type, and delivery hospital level). The Fellow would have the opportunity to explore changes in vaccination coverage before, during, and after the COVID-19 pandemic to identify the impact of COVID-19 pandemic, due to the disruption of pediatric health services, in immunization coverage among children in MA, especially among children with birth defects. Depending on the timeline of the project, the Fellow may also be able to explore any recent changes in vaccine coverage that coincide with the recent changes in federal immunization recommendations, particularly given the confusion these changes might have introduced for clinicians and families. This work will support ongoing efforts to ensure that all children receive timely, evidence-based protection against vaccine-preventable diseases.

Project #1 Impact:
Data on vaccination coverage among children with birth defects are valuable for informing communication messaging to health care providers and emphasizing the importance of routine vaccination among these children. In the current environment, these data are essential for monitoring emerging trends that may be influenced by recent shifts in federal recommendations and the confusion they have created for some clinicians and families. This project will allow us to assess if there are any disparities in vaccine uptake among different groups, the results of which could be utilized by vaccination campaigns to develop targeted interventions to increase vaccine uptake and protect children with birth defects and their communities from vaccine-preventable infectious diseases.

Additional Project #2 Title: Descriptive Assessment of the Co-occurrence of Birth Defects and Autism Spectrum Disorder
Project #2 Type: Other

Project #2 Description:
Autism spectrum disorder (ASD) is a biologically based neurodevelopmental disorder characterized by persistent deficits in social communication and social interaction and restricted, repetitive patterns of behavior, interests, and activities. The causal mechanisms underlying ASD are to a large extent not known. However, previous studies suggest that both genetic predisposition and environmental risk factors might play a role. Consideration of co-occurring birth defects with ASD may provide clues to gestational periods or developmental stages in pregnancy that may be important in its etiology. A number of studies have examined the association and while most have found a positive association between birth defects and ASD, the specific classes of birth defects exhibiting overlap with ASD have been inconsistent. In addition, birth defects and ASD have been reported to be associated with multiple risk factors including pregnancy exposures and sociodemographic factors (e.g., alcohol consumption, cigarette smoking, maternal and paternal age). This project will consider whether birth defects identified before one year of age, by class and specific defect where possible, occur more frequently in children later diagnosed with an autism spectrum disorder (ASD) than in children not diagnosed with ASD.

ID: 88809463
Birth Defects and NAS - Host Site Description
Massachusetts Department of Public Health

Project #2 Objectives and Expected Deliverables:
Project Objectives:

- To assess the co-occurrence of birth defects and ASD, the Fellow will link BDMP data with Early Intervention (EI) data.

Expected Deliverables:

- The EI data will be used to identify children with ASD or other distinct entities based on the World Health Organization ICD-10 such as atypical autism, Asperger's syndrome, other childhood disintegrative disorder, and other pervasive developmental disorder. The Fellow will assess if specific birth defects occur more frequently in children diagnosed with ASD than in children not diagnosed with ASD when considering parental characteristics and other risk factors as covariates. In addition, they will assess temporal trends in the co-occurrence of birth defects and ASD, as well as evaluate whether adjusted prevalence ratios of ASD among children with birth defects versus without birth defects varies by other developmental disabilities including intellectual disabilities.

Project #2 Impact:
The prevalence of ASD has risen to about to 1 in 31 children. It affects an estimated 1.2 million American children and is increasing at a rate of approximately 10-20% every two years. Despite being the most common developmental disability in the U.S., little is known about the causal mechanisms underlying this complex disorder and there is currently no cure. Understanding the co-occurrence of birth defects and ASD may shed some light on the potential etiologic relationship and advance our understanding of ASD.

Additionally, the results of this project could improve early diagnosis of ASD and earlier initiation of appropriate developmental services; advise increased surveillance of targeted populations (e.g., children with specific birth defects) by primary care practitioners and EI personnel; enhance the definition of phenotypes that might require special or different testing and interventions and might require additional instruction to EI staff; advance the classification of severity of ASD (a combination of autism and certain birth defect conditions may indicate a level of severity that one diagnosis alone would not imply); and inform EI and other public health programs, policies and practices and ultimately improve health and developmental outcomes for children with ASD.

Please Describe the Fellow's Anticipated Role in Preparedness and Response Efforts – Include Activities and Time Allocation (Required Competency of Fellowship)

Emergency Preparedness and Response: Currently Massachusetts does not have the capacity to address the needs of women of reproductive age (WRA), especially pregnant and postpartum women, infants, and children with special health needs and their families, during public health emergencies and disasters. A public health emergency can include an infectious disease outbreak, natural disaster, human-caused disaster, or other event or incident that requires a jurisdictional response to protect the public's health or to recover from mass injury, loss of life, or widespread property damage. The Fellow will have the opportunity to work with the Title V Program, the Division of Pregnancy, Infancy and Early Childhood, the Division for Children and Youth with Special Health Needs, and the Bureau of Emergency Preparedness on a strategic plan with a focus on special population groups such as pregnant women and children with special health needs that may require additional assistance beyond what the general population needs in a public health emergency or disaster. In addition, the Fellow will have the opportunity to participate in emergency preparedness exercises or responses (e.g., assisting with the Boston Marathon, participating in MA Responds). Time allocation can range from several part-time days to a full week of training.

ID: 88809463
Birth Defects and NAS - Host Site Description
Massachusetts Department of Public Health

Please Describe the Fellow's Anticipated Role in Cluster and Outbreak Investigations – Include Activities and Time Allocation (Required Competency of Fellowship)

The Fellow will have the opportunity to collaborate with colleagues in the Bureau of Infectious Disease and Laboratory Sciences (BIDLS) to lead an outbreak investigation. Past Fellows have conducted investigations of food-borne illness, other gastrointestinal illness of unknown etiology, an investigation of birth defects due to contaminated groundwater, and an outbreak of SARS-CoV-2 infections among vaccinated people in Barnstable County, MA. The time allocation for the Cluster and Outbreak Investigation is approximately one to two weeks. Activities will be specific to the nature of the investigation but may include meeting with key BIDLS staff, other Bureau staff, conducting record reviews, interviewing patients, creating a database of the cluster/outbreak, data analysis, and presentation of findings.