Birth Defects and NAS

Massachusetts Department of Public Health, Center for Birth Defects Research and Prevention

Boston, Massachusetts

Assignment Description

The mission of the Massachusetts Center for Birth Defects Research and Prevention (CBDRP) is to support surveillance, research, and dissemination of information aimed at preventing birth defects. The CBDRP houses two main large data sources: the Birth Defects Monitoring Program (BDMP) 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 BDMP was established in 1999 and is a statewide, population-based active surveillance program for birth defects among Massachusetts residents. The program uses multiple sources of ascertainment, including delivery and specialty care hospitals, laboratories and pathology departments, and reports of prenatally diagnosed birth defects. Vital records serve as an additional source of information, providing demographic information on cases, and acting as an additional source of case-finding. Potential birth defect cases, identified through these varied sources, are assigned to highly trained medical record abstractors who review maternal and infant medical records. They collect information on birth defect diagnoses, 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.

The CBDRP 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. 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.

In addition to data within the CBDRP, data outside the Center are available for analyses and include: birth certificates, fetal death certificates, infant and maternal death certificates, the Pregnancy to Early Life Longitudinal (PELL) data system, and data through such programs as the Massachusetts Home Visiting Initiative and Early Intervention. The CBDRP works closely with the Bureau of Infectious Diseases and Laboratory Sciences and the Fellow will have the opportunity to participate on collaborative projects related to pregnancy and infectious diseases. The Fellow will also have the opportunity to collaborate with staff and students from local universities including Harvard University, Boston University, Tufts University, the University of Massachusetts, and Brandeis University.

The Fellow will evaluate the BDMP surveillance system utilizing guidelines developed by the CDC. They will also be able to conduct epidemiological studies from a diverse array of data systems and the CBDRP 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 Birth Defects Monitoring Program External Advisory Committee, national conferences and meetings, local universities) and will be encouraged to publish their work in peer-reviewed journals.
**Day-to-Day Activities**

Day-to-day activities will include:

- Literature reviews
- Developing research plans
- Data cleaning and conducting analyses
- Preparing and presenting results of their analyses
- Preparing and leading meetings
- Submitting IRB applications as needed
- Meeting and providing updates to mentors and collaborators
- Writing, submitting, and reviewing manuscripts
- Presenting to internal and external collaborators
- Participating in meetings, trainings and webinars
- Joining one of the CBDRP’s quality improvement team projects
- Becoming familiar with the BDMP from reporting to ascertainment

**Potential Projects**

**Surveillance Activity**  
**Multisource Ascertainment in the Massachusetts Birth Defects Monitoring Program**

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 improves 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 Fellow will work with the BDMP staff to assess potential new sources of ascertainment (e.g., increasing the number of pathology departments and laboratories reporting, reports from ultrasound clinics, reports from out of the state hospitals). Once ascertainment sources have been identified, the Fellow will assess the potential yield of new cases, feasibility, and challenges in integrating the new ascertainment source into the BDMP. The Fellow may identify one of the ascertainment sources and lead the integration of that ascertainment source into the BDMP.
Surveillance Evaluation  
**Evaluation of the Massachusetts Birth Defects Monitoring Program and Maternal Prepregnancy Weight**

The Fellow will do an overall evaluation of the BDMP and then focus more specifically on prepregnancy weight. It has been suggested that the ability to detect birth defects prenatally through the use of ultrasounds may be limited in obese women. Given that maternal obesity has been associated with an increased risk of birth defects, the diminished ability to prenatally detect birth defects is among women who are at an increased risk of having a pregnancy with a birth defect. By examining the maternal prepregnancy weight as reported on the birth certificate compared to abstracted medical record data from BDMP, the Fellow will be able to assess the accuracy of this field. These data can then be utilized in the Fellow’s major project to adjust for potential measurement errors by conducting a bias analysis.

**Major Project**  
**Maternal Body Mass Index and Prenatal Detection of Birth Defects**

Prenatal ultrasound has evolved into an essential clinical tool for detection of birth defects. Studies have shown that the technical quality of the ultrasound examination is limited in women with obesity due to limited visualization, which leads to unsuccessful ultrasound examinations and increased need for further follow-ups. In the U.S., the National Center for Health Statistics data for 2011 to 2014 indicated that 34% of reproductive age-women have obesity which is defined as a body mass index (BMI) ≥30 kg/m^2, while the incidence of morbid obesity (BMI >40 kg/m^2) exceeded 7%. Performing the second trimester fetal anatomic survey at a later gestational age (20-22 weeks versus 18-20 weeks as usually performed) might improve visualization and increase the success rate of ultrasound examinations among women with obesity. However, screening at a later gestational age limits the time window for planning in the event that a birth defect is identified. The Fellow will utilize the BDMP data to assess if prenatal detection rates of birth defects vary by BMI and if the timing of diagnosis of birth defects differs among women with and without obesity in regards to gestational age and prenatal versus postnatal diagnosis. Additionally, the Fellow will evaluate if an increased number of prenatal ultrasounds or more advanced radiologic diagnostic methods such fetal MRI and fetal ECHO are needed for birth defect diagnosis among women with obesity. Given the increasing rates of obesity in the U.S., the information gathered from this study could have direct clinical implications and could suggest more imaging or advanced imaging may be necessary in this population.

**Second Major Project**  
**The Co-occurrence of Neonatal Abstinence Syndrome and Birth Defects**

Massachusetts and much of the nation is in the midst of an opioid epidemic and understanding the impacts of opioid use in pregnancy on infants is essential for public health planning and intervention. Opioid use in pregnancy may lead to neonatal abstinence syndrome (NAS) in infants; NAS is associated with gastrointestinal, respiratory, autonomic and central nervous system issues and leads to extended hospital stays. Opioid use in pregnancy has also been associated with an increased risk of specific birth defects. However, the co-occurrence between NAS and birth defects is not well understood. One study to date has suggested infants with NAS also have a higher prevalence of birth defects, with as much as a six-fold increase observed for certain defects. Utilizing data from birth certificates, early intervention,
and the BDMP, the Fellow will evaluate the overlap of NAS with specific birth defects to better understand the co-occurrence of these two conditions.

**Additional Project**

**Descriptive Assessment of the Co-occurrence of Birth Defects and Autism Spectrum Disorder**

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 maternal lifestyle and sociodemographic factors (e.g., alcohol consumption, cigarette smoking, maternal age). 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.

**Preparedness Role**

The Fellow will have the opportunity to work with the Division of Pregnancy, Infancy and Early Childhood, Division for Children and Youth with Special Health Needs, and the Bureau of Emergency Preparedness to 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 disaster. In addition, the Fellow will have the opportunity to participate in emergency preparedness exercises or responses (e.g., assisting with the Boston Marathon) or potentially work with the Bureau of Infectious Disease and Laboratory Sciences should there be an emerging issue related to pregnant women and infants.

**Additional Activities**

Health Disparity/Equity: the Bureau of Family Health and Nutrition (BFHN) is currently undertaking a Racial Equity Initiative with the following two goals:

1. Eliminate structural racism in all BFHN policies, programs, and practices to promote health equity and racial justice; and
2. Foster a healthy and equitable work environment in BFHN, where staff feel confident and supported to interact and communicate openly and respectfully.
The Fellow will have an opportunity to participate in this important endeavor and assist with data collection, analysis, and interpretation.

NBDPS/BD-STEPS: The Fellow will have the opportunity to see the day-to-day activities of an ongoing case-control study and participate in multi-Center conference calls that discuss questionnaire and methods, project management, and epidemiologic methods and analyses. Data from the studies are also available for research projects.

**Mentors**

**Primary**
Mahsa Yazdy BS, MPH, PhD
Director

**Secondary**
Susan Manning BA, MPH, MD
CDC Maternal and Child Health Epidemiology Assignee