Limitations

A few limitations of the FBDR should be considered when interpreting the information presented in this document. First, the FBDR's case definition is restricted to live births. The exclusion of other pregnancy outcomes, including stillbirths and spontaneous or elective terminations of pregnancy (TOPs), may result in underestimated prevalence rates, particularly for defects in which a large proportion of cases result in fetal mortality (e.g., anencephaly).

Second, the FBDR ascertains cases diagnosed up to the first year of life. Prevalence rates for defects diagnosed at birth or early in life (e.g., limb reduction defects) should be comparable to other state-based surveillance programs. However, we caution comparison of our rates to programs with longer ascertainment periods, particularly when assessing defects frequently identified after the first year of life (e.g., certain congenital heart defects). Third, the linkage algorithms employed by the FBDR require that the infant's birth certificate be able to be matched to administrative datasets, and this is often highly dependent upon the mother and/or infant having a social security number (SSN). This has the potential to result in disproportionate ascertainment of cases among subsets of the population that are more likely to have inaccurate or missing SSNs (e.g., immigrants, minorities).

Lastly, these administrative data sources are not created for the purposes of surveillance activities of a birth defects registry. For infants born with highly lethal defects (e.g., anencephaly, trisomy 18), the infant may die within minutes or hours after birth, not generating a hospital discharge record for the FBDR to access. Therefore, these infants may fall through the current FBDR's surveillance "net". The FBDR also presents challenges for case sensitivity and specificity. Currently, the passive surveillance system lacks the ability to verify and appropriately document birth defect diagnoses through medical record review and abstraction.

Relying solely upon reported ICD-9/10-CM codes without case confirmation may result in misclassified, non-specific, and false positive diagnoses. However, the FBDR is conducting enhanced surveillance of selected birth defects of major public health significance. Although enhanced surveillance is more labor and resource intensive than passive surveillance, its more thorough casefinding efforts can improve completeness, and its process of reviewing medical records to confirm suspected cases can dramatically increase the FBDR's diagnostic accuracy. In fact, the FBDR is currently in the process of utilizing data collected using enhanced surveillance activities to refine and improve the accuracy and completeness of the passive data currently reported.

Another important aspect of a birth defect surveillance system is the timeliness of data. Due to the registry's reliance on the acquisition, preparation, and linkage of source datasets, the final inventory of affected infants and dissemination of prevalence rates lags approximately two years behind present time. This has impacted implementation of effective primary, secondary, and tertiary recurrence prevention programs and timely referral to services.