Glossary of Terms: NBDPN Guidelines

age-at-diagnosis: The age of the child when a birth defect was diagnosed, or at which the signs or symptoms were first recognized as potentially representing the birth defect.

agenesis: Failure of an organ or body part to form.

anomaly: Deviation or departure from what is normal or typical.

aplasia: Absence of a tissue or organ due to lack of cell proliferation.

atresia: Abnormal complete closure or absence of a body opening or passage.

association: A nonrandom pattern of anomalies that occur together more frequently than expected by chance alone, but for which no underlying common etiology has been demonstrated. Use of the term association does not indicate that a specific diagnosis has been made. *For example: the **VACTERL** association (**V**ertebral, **A**nal, **C**ardiac, **T**racheo-**E**sophageal, **R**enal, and **L**imb anomalies).*

authorizing language: Legislative language giving authority for birth defects surveillance program operations. Language varies by state and program, and may include defining the purpose of the surveillance activities, specifying the kinds of data or information to be collected, and designating who is responsible for the program activities.

birth defects: For the purposes of the NBDPN Guidelines, birth defects are major congenital anomalies, chromosomal abnormalities, and genetic syndromes. The terms congenital abnormality, congenital anomaly, and congenital malformation are often used as synonyms for birth defect. By definition, the condition is present at birth, even if signs and symptoms take weeks, months, or years to fully manifest for diagnosis or treatment.

birth defect case definition criteria: A set of uniform criteria specific to each birth defect diagnosis used to define the condition for public health surveillance. Birth defect case definitions include the diagnosis, further delineated by the type of surveillance performed and the level of certainty that the diagnosis is correct. Surveillance case definitions enable public health to classify and count cases consistently across reporting jurisdictions. (Of note: surveillance case definitions are not intended for use by healthcare providers for making a clinical diagnosis or determining how to meet an individual patient's health needs.)

Birth Defects Study to Evaluate Pregnancy Exposures (BD-STEPS): A national, multi-site, population-based, case-control study investigating the risk factors for and causes of birth defects and stillbirths in the United States. This study builds upon the National Birth Defects Prevention Study (NBDPS).

birth defects surveillance program: A program that facilitates the essential activities of data collection, data analysis and evaluation, information dissemination, and data utilization for a specific jurisdiction. The data collected by each birth defects surveillance program can be aggregated with that of other programs to enable standardized, national surveillance of birth defects. Of note: some programs may refer to their work as a surveillance registry or system, rather than a surveillance program.

case: In birth defects surveillance, a case refers to an individual with characteristics that fit into defined parameters, including the birth defect diagnosis itself (see birth defect case definition criteria), along with the programmatic case inclusion criteria.

case ascertainment: Identifying and gathering information on potential cases of a disease within a population. NBDPN supports a systematic approach to case ascertainment that involves a four-part process: case identification, case investigation, case verification, and case inclusion review, as defined below in this glossary.

case classification: Evaluating a potential case using standardized criteria to assign the case an official status (i.e., determining whether the case represents an included diagnosis for any of the birth defects in the surveillance program's scope, and whether the level of certainty about that diagnosis is *Accepted* versus *Confirmed*).

case identification: The process of identifying potential cases that may possibly meet a birth defects surveillance program's programmatic case inclusion criteria and birth defect case definition criteria, in order to bring those cases to the attention of the surveillance program for further review.

case inclusion review: Reviewing the information related to a potential birth defect case to ensure that it meets programmatic case inclusion criteria and birth defect case definition criteria, and thus warrants inclusion as a case in the birth defects surveillance database. The case inclusion review process occurs throughout the entire arc of case identification, case investigation, and case verification.

case investigation: The process of gathering abstracting and consolidating data on a potential birth defect case, with a key focus on collecting every data element needed for coding, case classification, and case verification.

case verification: The process of ensuring completeness and accuracy of the data collected during the case identification and investigation process.

catchment area: A defined geographic area from which cases for surveillance are collected.

Centers for Disease Control and Prevention (CDC): CDC is the nation's leading science-based, data-driven, service organization that protects the public's health.

Centers for Disease Control and Prevention/British Pediatric Association (CDC/BPA) codes: A set of six-digit diagnostic codes, based on ICD-9-CM and the BPA Classification of Diseases extensions, then further modified by CDC, offering detailed information and specificity about congenital birth defects (e.g., laterality, severity, or subtype) for birth defects surveillance programs in the United States.

comparability: The use of standardized sources to enable data to be pooled at a national level, in order to accurately compare data across jurisdictions.

computer assisted coding (CAC): A form of natural language processing developed to scan and process clinical documentation for key words, terms, phrases, or codes, then use the compiled information to suggest the diagnosis code(s). CAC is used at many hospitals, clinics, and other health care provider settings.

congenital: Arising during intrauterine development and therefore present at birth (although not necessarily clinically evident at birth).

congenital anomaly: Structural or functional abnormality arising during intrauterine development regardless of the underlying cause. See also: *birth defect.*

Current Procedural Terminology (CPT): A standardized coding system developed by the American Medical Association for coding medical procedures (e.g., surgeries, prenatal tests, etc.), used in billing and data exchange. The CPT code set includes many procedures, imaging tests, and laboratory tests related to birth defect diagnosis and treatment.

data collection: The systematic process of collecting, analyzing, and interpreting data elements from data sources to address specific questions. Birth defects surveillance programs ultimately use the collected data to improve public health.

data element: A unique unit of information that can be collected from a data source (e.g., date of birth), abstracted from medical records (e.g., age-at-diagnosis), derived based upon other information collected (e.g., month prenatal care began), or created/assigned by the birth defects surveillance program for operational purposes (e.g., unique case ID). Each data element has a defined definition, format, and acceptable range of values. See also: *data variable.*

data mining: Use of data science methods (e.g., epidemiology, statistics, computer technologies) to analyze large data sets, identify patterns, and perform advanced analytics.

data source: An entity that can provide information related to a potential birth defect case, including demographics or medical records, which can be used by a birth defects surveillance program to complete the case ascertainment process.

data variable: A unique unit of information that can be collected from a data source (e.g., date of birth), abstracted from records (e.g., age-at-diagnosis), derived based upon other information collected (e.g., mother's age at delivery), or created/assigned by the birth defects surveillance program for operational purposes (e.g., unique case ID). Each data variable has a defined definition, format, and acceptable range of values. See also: *data element.*

deformation: A major anomaly that results from molding of an anatomical structure by mechanical forces (e.g., oligohydramnios; or intrauterine crowding in twin, triplet, or higher order pregnancies). Deformation occurs during intrauterine development after the structure's initial formation, usually over a prolonged time. *Examples of deformations include the compression (Potter's) facies in bilateral renal agenesis, and some instances of clubfoot.*

diagnosis code: A unique set of letters and/or numbers that indicate a certain disease, disorder, symptom, injury, or other reasons for patient encounters in the health care setting.

diagnosis/code-based surveillance: A method of birth defects surveillance in which cases are reviewed and classified based only on coded or written diagnosis data reported by one or more data sources, without further review of additional information in the medical record.

disruption: A major anomaly that results from alteration of a structure during intrauterine development after its initial formation. The resulting structure may have an altered shape and configuration, abnormal division or fusion of its component parts, or loss of parts that were previously present. *An example of a disruption-type defect is intestinal atresia.*

dysgenesis: Anomalous or disorganized formation of an organ.

dysplasia: Disorganized cell structure or arrangement within a tissue or organ.

early fetal death: See listing under *fetal death.*

electronic case reporting (eCR): An automated exchange of case report information between electronic health records and public health agencies.

electronic health record (EHR): The systematized collection of patient information in a digital format, including medical history of tests, diagnoses, and treatments, along with patient demographics and billing information. See also: *electronic medical record (EMR).*

electronic initial case report (eICR): An initial case report delivered through an electronic case reporting (eCR) system when patient data matches certain preset criteria (see: *trigger code*). The eICR is intended to convey sufficient information for case identification.

electronic medical record (EMR): The systematized collection of patient information in a digital format, including medical history of tests, diagnoses, and treatments, along with patient demographics and billing information. See also: *electronic health record (EHR).*

embryonic period: The first 8 weeks after fertilization, during which most organs are formed.

epidemiology: The study of the distribution and determinants of health-related states or events in specified populations, and the application of this study toward the control of health problems.

extremely low birth weight (ELBW): Birth weight < 1,000 grams, regardless of gestational age.

Fast Healthcare Interoperability Resource (FHIR): A healthcare data standard developed as part of Health Level 7 (HL7) to securely exchange information (e.g., SNOMED terms, ICD codes) across electronic systems, to enhance interoperability and streamline data sharing.

fetal death: A delivery in which death occurred prior to the complete expulsion or extraction of a product of human conception from its mother, irrespective of pregnancy duration.

early fetal death (miscarriage): Spontaneous loss of a fetus at < 20 completed weeks gestation. The terms miscarriage, spontaneous abortion, and early pregnancy loss are used interchangeably, and there is no consensus on terminology.

late fetal death: A fetal death delivered at ≥ 28 completed weeks of gestation.

stillbirth: Spontaneous delivery of a fetus at ≥ 20 weeks completed gestation (or at > 350 grams if gestational age is unknown) that does not exhibit signs of life. Transient cardiac contractions and fleeting respiratory efforts or gasps are not necessarily considered signs of life by all programs.

fetal period: The period from the 9th week after fertilization through delivery.

geocode: The process of entering an address and returning an actual or calculated latitude/longitude coordinate. The precision of a geocode will depend on the accuracy of the address to which the latitude and longitude are assigned.

gestational age: A measure of the duration of a pregnancy. Gestational age may be derived in different ways (with variable accuracy) based on measurement of the fetus by prenatal ultrasound, the date of the last menstrual period, or the date of *in vitro* fertilization if applicable. Gestational age can indicate a point in time during gestation (e.g., "amniocentesis was performed at 17 weeks of gestation"), or ultimately to indicate the age of the fetus at the time of pregnancy outcome (e.g., "delivered at 37 weeks gestation.")

Guidelines (or Guidelines for Conducting Birth Defects Surveillance): The written manual produced and maintained by NBDPN which provides practical guidance for planning, implementing, and enacting surveillance processes and operations to ensure timely, accurate, and complete data, particularly for the purpose of prevalence estimates and monitoring.

health information classification system: A structured framework (e.g., ICD, or CDC/BPA) for categorizing disease diagnoses, clinical and surgical procedures, test results, pharmaceuticals, devices, and other health-related terminology. Health information classification systems use consistent rules and codes, enabling standardized surveillance and analysis.

Health Level 7 (HL7): A set of global technical standards for the transfer of clinical and administrative health data between electronic applications with the aim of improving patient outcomes and health system performance.

hyperplasia: Overgrowth of a tissue or organ due to excess proliferation of otherwise normal cells.

hypoplasia: Undergrowth of a tissue or organ due to insufficient proliferation of otherwise normal cells.

infant death: Death of a live-born infant starting at 29 days after birth, and ending when the infant reaches 1 year (12 completed months) of age.

International Classification of Disease and Related Health Problems (ICD): A medical classification system developed and maintained by the World Health Organization, used internationally for coding and classifying diseases, causes of death, and health conditions.

International Classification of Disease-Clinical Modification (ICD-CM): A medical classification system developed by the National Center for Health Statistics (adapted from the global ICD system, and authorized by the World Health Organization). ICD-CM is used only in the United States to classify disease diagnoses in the inpatient and outpatient setting.

International Classification of Disease-Procedure Coding Systems (ICD-PCS): A medical classification system specifically designed for classifying inpatient procedures performed in hospitals in the United States.

International System for Cytogenomic Nomenclature (ISCN): A standard system of human genomic nomenclature for classifying results of cytogenetic techniques ranging from karyotyping to microarray, genomic mapping, fluorescence *in situ* hybridization (FISH), various region-specific assays, and DNA sequencing.

interoperability: The ability of different information systems, devices, and applications/systems to access, exchange, integrate and cooperatively use data in a coordinated manner, within and across a birth defects surveillance program's catchment area, to provide timely and seamless portability of information.

isolated anomaly: A single major congenital anomaly, or a sequence of major anomalies (see: *sequence*), that occurs without any other major anomalies present. Most (~75% in the aggregate) major congenital anomalies are isolated. Isolated major anomalies can be associated with one or more minor anomalies.

late fetal death: See listing under *fetal death.*

legislation: A law enacted by an elected body.

live birth: Spontaneous delivery of an infant that exhibits signs of life including heartbeat, spontaneous breathing, and movement of voluntary muscles.

Logical Observation Identifiers Names and Codes (LOINC): A standard system with identifiers, names, and codes for clinical results of lab tests, imaging tests, and measurement-related clinical observations or patient outcomes.

low birth weight (LBW): Birth weight less than 2,500 grams, but greater or equal to 1,500 grams regardless of gestational age.

major anomaly: A congenital abnormality that requires medical or surgical treatment, has a serious adverse effect on health and development, and/or has significant cosmetic impact. Individual major anomalies occur in less than 1% of the population. Together, they are seen in approximately 3% percent of all births. *Examples include cleft lip and tracheo-esophageal fistula, among many others.*

malformation: A major anomaly that arises during the initial formation of a structure (i.e., during organogenesis). For most organs, this occurs during the first 8 weeks after fertilization. The resulting structure may be abnormally formed, incompletely formed, or may fail to form altogether. The term 'congenital malformation' is used more broadly by some to indicate any major anomaly regardless of the underlying cause. *Examples of malformations include spina bifida and hypoplastic left heart.*

medical record abstraction: A process in which staff trained to identify birth defects search through data sources (virtually or in person) to gather information about the mother and infant/fetus, and then format the information in a standardized manner for use by the birth defects surveillance program.

medical record abstraction-based surveillance: A method of birth defects surveillance in which trained staff perform further review of additional information in the medical record (e.g., clinical notes, laboratory testing, and/or imaging data), beyond the coded diagnosis data reported by the data sources. The process of case investigation, verification, and classification is based on that medical record information.

minor anomaly: A congenital structural change that poses no significant health problem and tends to have limited social or cosmetic consequences for the affected individual. Intervention or surgery might be performed if desired (e.g., tying off an extra digit skin tag, or utilizing molds for unusually shaped ears). Individual minor anomalies generally occur in less than 4% of the population. The presence of multiple minor anomalies in the same child may provide clues to the timing of a prenatal insult and may prompt closer examination to assess for an undiagnosed major anomaly, syndrome, or functional deficit.

miscarriage: See listing under *fetal death.*

National Birth Defects Prevention Network (NBDPN): A group of individuals involved in birth defects surveillance, research, and prevention. NBDPN was created to establish and maintain a national network of state and population-based programs. Data from networked programs helps assess the impact of birth defects upon children, families, and health care; identifies factors that can be used to develop primary prevention strategies; and assists families and their providers in prevention of secondary disabilities.

National Birth Defects Prevention Study (NBDPS): One of the largest multi-state case-control studies to identify the causes of birth defects in the United States, conducted as a collaborative effort between the CDC and Centers for Birth Defects Research and Prevention (CBDRP). A dedicated set of diagnosis codes were developed for the purpose of the study, and data from the NBDPS findings are available for further research.

National Center for Health Statistics (NCHS): The principal health statistics agency in the United States, and part of the Centers for Disease Control and Prevention. NCHS develops and maintains the ICD-CM coding system for the United States.

natural language processing (NLP): A type of machine language scanning that uses specially developed software to 'read' information and extract key pieces of data. NLP software can be customized for public health applications, including extracting and analyzing text from medical records or health data repositories, and proposing diagnosis codes.

NBDPN Core Conditions: A list of specific birth defects that are designated as high priority by NBDPN for national birth defects surveillance. NBDPN recommends that all birth defects surveillance programs include all of the Core Conditions in their scope of data collection.

NBDPN Enhanced Conditions: A list of specific birth defects which NBDPN has designated as the next priority for national birth defects surveillance, when birth defects surveillance programs are already meeting quality standards for surveillance of NBDPN Core Conditions, and when program policies and resources permit.

NBDPN Recommendation: Specific guidance that NBDPN strongly suggests incorporating into every birth defects surveillance program to ensure timely, accurate, and complete data.

NBDPN Standard: The activities (i.e., outputs and performance indicators) expected of a birth defects surveillance program. Each NBDPN Standard is presented as a tiered rubric for birth defects surveillance program activities, displayed in table format, with up to three tiers:

Core tier: The minimum criteria needed for surveillance activities to ensure a birth defects surveillance program meets an acceptable level of performance.

Enhanced tier: The minimum criteria (Core tier) *plus* some additional activities a birth defects surveillance program might engage in based on available resources and local policies.

Extended tier: The archetype of what an ideal birth defects surveillance program's activities would encompass.

neonatal death: Death of a live-born infant within the first 28 days after birth.

Early neonatal death refers to death during the first 7 days after birth.

Late neonatal death refers to death at any point from 8 to 28 days after birth.

neonatal (newborn) period: The first 28 days following delivery of a live-born infant.

nomenclature: A systematic structure of naming things within a particular field to ensure clarity and consistency.

non-live birth: A delivery in which embryonic or fetal death occurred prior to the complete expulsion or extraction of a product of human conception from its mother, irrespective of pregnancy duration. See also: *fetal death.*

normal variant: A structural change that occurs in approximately 4% or more of the population and is not considered abnormal. *Examples of normal variants include webbing of the second and third toes only, or a single umbilical artery.*

perinatal: The time closely surrounding delivery (before, during, and after). The exact timing of the perinatal period may vary, starting at 20 to 28 completed weeks of gestation, and ending 7 to 28 days after delivery, depending on the context in which the term is used.

population-based surveillance: Surveillance that identifies a population under study (usually defined by geopolitical boundaries), which establishes the denominator from which cases are obtained. A data source that is population-based covers the entire population within a defined area. *Examples of population-based data sources include vital records (birth, death, and fetal death), statewide newborn genetic screening programs, and statewide newborn hearing screening programs.*

positive predictive value (PPV): A statistical analysis that calculates the proportions of true positive diagnosis among individuals who test positive for a disease. In other words, given that a certain number of individuals tested positive for a disease, PPV is the proportion of those individuals who actually have the disease.

postnatal: After delivery.

pregnancy outcome: Outcome of a pregnancy, which can include either live birth, fetal death (i.e., stillbirth at $\geq$ 20 weeks of gestation, or miscarriage at < 20 weeks of gestation), or termination. Depending on the available information for a specific birth defects surveillance case, the pregnancy outcome may also be listed as an unspecified non-live birth, or unknown.

prenatal: The time from conception through delivery.

pre-term infant: An infant born before 37 completed weeks of gestation.

post-term infant: An infant born at or after 42 completed weeks of gestation.

procedure code: A standardized alphanumeric code used to describe specific medical, surgical, or diagnostic procedures performed by healthcare providers.

programmatic case inclusion criteria: Criteria that ensure a birth defects surveillance program only collects data on cases within its scope and jurisdiction. These include geographic catchment area, residency status of the mother, pregnancy outcome, and age-at-diagnosis.

public health authority: An agency or authority of the United States government, or a state, territory, political subdivision of a state or territory, or tribe that is responsible for public health matters as part of its official mandate. May also refer to a person or entity acting under a grant of authority from, or under a contract with, a public health agency.

Public Health Informatics Institute (PHII): An institute that guides public health organizations to effectively access, use, and share information to advance health outcomes worldwide.

public health surveillance: The ongoing systematic collection, analysis, and interpretation of health data essential to the planning, implementation, and evaluation of public health practice, closely integrated with the timely dissemination of data to those who need to know.

public health surveillance system: An organization with the functional capacity for data collection, analysis, interpretation, and dissemination linked to public health programs for prevention, treatment, and support.

Reportable Conditions Knowledge Management System (RCKMS): A real-time portal to enhance disease surveillance by providing comprehensive information on public health reporting criteria. RCKMS includes a decision support tool and curated content that are part of the national electronic case reporting (eCR) infrastructure.

record linkage: The process of connecting information from multiple sources for the same individual (e.g., linking or merging a birth record with a hospital record).

regulations: Rules created by agencies to shape how legislation is implemented.

residence: The place of residence of an individual with one or more birth defects. This serves as a component of the programmatic case inclusion criteria, and usually refers to the mother's place of residence at the time of the pregnancy outcome of the case.

sequence: A pattern of anomalies that results from a single primary anomaly or mechanical factor, in cascade fashion. The presence of the initial anomaly or factor leads to one or more secondary anomalies, which may then lead to one or more tertiary anomalies, etc. *Examples include Robin sequence (micrognathia, posterior displacement of the tongue, cleft soft or hard palate, and airway obstruction); or the oligohydramnios or Potter sequence (pulmonary hypoplasia, flattened facies, abnormal positioning of the limbs).*

stenosis: Abnormal narrowing of a body opening or passage.

syndrome: A pattern of multiple anomalies with a specific underlying etiology in common (usually genetic), which together form a specific diagnosis for which the natural history and recurrence risk are usually known. *Example: Down Syndrome (Trisomy 21); Turner Syndrome.*

Systematized Nomenclature of Medicine Clinical Terms (SNOMED CT): An international system of medical terms (including clinical findings, symptoms, diagnoses, procedures, body structures, specimens, pharmaceuticals, devices, etc.) for communicating clinical data across specialties and sites of care to create a standard global healthcare vocabulary.

term infant: An infant born at ≥ 37 completed weeks of gestation, but before 42 completed weeks of gestation.

trigger code: Specific codes used to initiate the reporting of health conditions. In birth defects surveillance, each trigger code can be a diagnosis code, procedure code, or other key words that indicate the potential presence of a birth defect. When a trigger code is entered in the electronic record at the data source, the presence of that code causes automatic transmission of a standardized set of case data (e.g., an electronic initial case report) to the birth defects surveillance program to notify them of the potential case. Surveillance programs can also have trigger codes for internal programmatic usage (e.g., to identify medical records for abstraction or clinical review, or to route specific cases for inclusion in surveillance data).

use case flow diagram: A technical tool that depicts the processes and procedures for data flow, key decision points/interactions involving the system staff, and expected information, data outputs, or data extracts. Use case flow diagrams are frequently used for information technology (IT) to determine system requirements, and to map out sequences of user interactions involved in a given task or event.

Value Set Authority Center (VSAC): A repository and authoring tool for public value sets created by external programs. The VSAC is provided by the National Library of Medicine (NLM), in collaboration with the Office of the National Coordinator for Health Information Technology (ONC) and Centers for Medicare & Medicaid Services (CMS).

very low birth weight (VLBW): Birth weight < 1,500 grams, but ≥ 1000 grams, regardless of gestational age.

vital records: Legal documents that attest to the occurrence of a life event and contain a set of standardized data elements pertinent to the life event. These include certificates for birth, death, and fetal death issued by authorized United States government agencies.

World Health Organization (WHO): An international organization of member states and health professionals with the goal of coordinating the world's response to health emergencies, promoting well-being, preventing disease and expanding access to health care by connecting nations, people, and partners to scientific evidence.