Appendix 4A: Common Health Information Classification and Coding Systems in Birth Defects Surveillance

Table 1. Classification and Coding Systems for Diagnoses

System	Description	Strengths	Limitations
ICD	The **I**nternational **C**lassification of **D**iseases and Related Health Problems (ICD) is a global standard for coding and classifying diseases, causes of death, and health conditions. It was developed and continues to be monitored by the ***World Health Organization (WHO)***. New versions are released as health care systems change, and diagnoses expand.	Considered a global standard for codes related to clinical care, research, and (in some countries including the US) medical billing and resource allocation. ICD is used in over 100 countries and 43 languages. The standardized ICD system is compatible with Health Level 7 (HL7) messaging, interoperability, and electronic case reporting (eCR). ICD codes can be utilized as trigger codes. ICD codes facilitate international comparability in collecting, processing, classifying, and presenting disease-related data. Diagnoses are organized clinically (i.e., grouped by disease, body system, etiology, or other clinical similarities). The code set is comparable across different health sectors, for uses including disease index, clinical data set, discharge data set, etc.	There is a time lag between WHO acceptance of the ICD, and acceptance for use in the US. Since the codes are organized clinically, the entire alpha-numeric code system changes with each new version. In the US, ICD is used for mortality coding, and ICD-CM is used as an administrative data set for insurance, billing, and reimbursement. Federal disease coding rules affect the assignment of the disease code at a data source. ICD and ICD-CM lack specificity, with ICD-CM being marginally more detailed. There is problematic grouping of multiple birth defects into one code. The ICD-CM code set is updated at the start of each federal fiscal year. Data sources that use the ICD-CM system activate new codes and/or new exclusion criteria, and deactivate the "voided" codes. This impacts data collection (i.e., trigger codes and record selection criteria) and affects the diagnosis codes selected for statistical analysis. Birth defects surveillance programs need to be aware of ICD-CM updates that affect surveillance functions.
ICD-CM	**ICD-C**linical **M**odification (ICD-CM) is a version developed by National Center for Health Statistics (NCHS), authorized by the WHO, for use in the United States (US) to classify disease diagnoses in the inpatient and outpatient setting.		

Table 1. Classification and Coding Systems for Diagnoses, *continued*

System	Description	Strengths	Limitations
CDC/BPA	In 1979, the **B**ritish **P**ediatric **A**ssociation (BPA) developed a classification of diseases by modifying ICD-9-CM (British). In 1983, staff in the US **C**enters for **D**isease **C**ontrol and Prevention (CDC) birth defects branch modified the BPA coding system and developed a classification system specific to birth defects coding, termed the CDC/BPA system. The six-digit code is a classification system that allows coding of more detailed descriptions of birth defects and related conditions. The CDC maintains and updates the CDC/BPA codes, which can be accessed for public use through the CDC website.	A relatively small set of specialized diagnostic codes, specific to birth defects. Used within birth defects surveillance programs, primarily by those who assign diagnosis codes to birth defects. Some birth defects surveillance programs further modify the CDC/BPA codes to their own programmatic objectives. Arranged into general categories (body systems, medical conditions), with the first 4 digits identical to the ICD-9-CM code, enabling birth defects surveillance programs to use ICD-9 codes from the hospital data set as a starting point. The codes are hierarchical, so that the 5th and 6th digit can reflect more detail (e.g., laterality, severity, or status as a probable versus confirmed diagnosis).	The structure of this coding system is tied to the now-outdated ICD-9 codes. It does not significantly change with implementation of later ICD versions (ICD-10, ICD-11). The CDC/BPA code is not standardized at the national or international level. This means the codes are not used by data sources, nor is the code set compatible with HL7 messaging/interoperability or eCR. The CDC/BPA codes (and other modified versions) are not recognized as trigger codes.

Table 1. Classification and Coding Systems for Diagnoses, *continued*

System	Description	Strengths	Limitations
BD-STEPS **NBDPS**	The **B**irth **D**efects **St**udy to **E**valuate **P**regnancy Exposure**s**, and the **N**ational **B**irth **D**efects **P**revention **S**tudy are major national research efforts conducted by the Centers for Birth Defects Research and Prevention (funded by the CDC), focused on 17 categories of structural defects. The BD-STEPS/NBDPS codes correspond to the 17 structural defects categories used as the focus of the research efforts. They were based on the CDC/BPA codes, with further modifications to increase their specificity for research purposes.	A relatively small set of diagnostic codes focused on birth defects of high interest to public health research. Used for two major birth defects studies in the US. The BD-STEPS coding system is designed to be specific, detailed, and tailored to research objectives. It may be useful to inform some routine surveillance functions (e.g., inclusion and exclusion criteria for a birth defect, abstracting, and clinical reviews).	The BD-STEPS codes are not standardized at the national or international level. This means the codes are not used by data sources, nor is the code set compatible with HL7 messaging/interoperability or eCR. The BD-STEPS codes are not recognized as trigger codes. The system only encompasses codes for the 17 categories of interest to the research effort: • Anomalous pulmonary venous return • Anophthalmia/microphthalmia • Anotia/microtia • Cleft lip with/without cleft palate • Cleft palate • Coarctation of the aorta • Diaphragmatic hernia • Esophageal atresia • Gastroschisis • Hypoplastic left heart syndrome • Pulmonary atresia • Spina bifida • Tetralogy of Fallot • Transposition of the great arteries • Transverse limb deficiency • Tricuspid atresia • Truncus arteriosus

Table 2. Classification and Coding Systems for Terminology

System	Description	Strengths	Limitations
SNOMED CT	**S**ystematized **No**menclature of **Med**icine **C**linical **T**erms (SNOMED CT) is a multilingual dictionary of standardized clinical terms, encompassing terminology describing symptoms, signs, diagnoses, procedures, clinical findings, anatomical structures, pharmaceuticals, specimens, and medical devices. Developed, maintained, and distributed by the International Health Terminology Standards Development Organization. Each term in the SNOMED database is defined by 3 criteria: • **Concept:** the unique clinical meaning. • **Description:** the full name, and synonyms. • **Relationship:** defining how concepts are related to each other.	Considered the most comprehensive glossary of standardized clinical terminology in the world, with over 350,000 concepts each identified by a unique number/code. Searchable, sortable, categorized, and based on term concepts, language descriptors, etc. The standardized SNOMED CT number codes are compatible with HL7 messaging, interoperability, and eCR. SNOMED CT codes can be utilized as trigger codes. SNOMED can be cross-mapped with other international standards (e.g., ICD-10) to facilitate interoperability. Since 2013, this health information classification and coding system has been a mandatory standard for electronic exchange of health information in the US.	SNOMED CT is a licensed product. The National Library of Medicine holds the US license and distributes SNOMED CT at no cost to US users. In 2025, the implementation of SNOMED CT in US clinical settings remains relatively new. There is a large and involved IT component and the learning curve to understand, use, and implement SNOMED CT continues to evolve for its use as a tool in clinical settings, public health efforts, and data modernization initiatives.

Table 3. Classification and Coding Systems for Clinical or Surgical Procedures

System	Description	Strengths	Limitations
CPT	**C**urrent **P**rocedural **T**erminology (CPT) codes are developed and maintained by the American Medical Association to describe medical services and procedures performed by physicians and other health care professionals. CPT codes fall into 4 categories. In general, only Category I (procedures or services, with code range 00100-99499) are relevant to birth defects surveillance work.	CPT codes provide a uniform language (in the form of distinct 5-digit numeric or alphanumeric codes) to report medical, surgical, radiology, laboratory, anesthesiology, genomic sequencing, and clinical evaluation & management services received by a patient. The code set applies to services and procedures performed in any health care setting (e.g., inpatient, outpatient, etc.).	Potential for misinterpretation of codes due to complex terminology, nuanced procedures, and lack of uniformity in how different providers define services. CPT codes are also used for insurance reimbursement, which may impact which code(s) a health care provider decides to assign.
ICD-PCS	The ICD-10 **P**rocedure **C**oding **S**ystems (ICD-10-PCS) was developed for use in the US by the Centers for Medicare and Medicaid, in collaboration with the NCHS to accompany the WHO ICD-10 diagnosis classification. ICD-10-PCS is the medical classification system used in the US to catalogue inpatient procedures within the hospital setting. The definitive guide for this procedure classification system is the ICD-10-PCS Reference Manual, produced by the Centers for Medicare & Medicaid Services.	The alphanumeric code is 7 characters, with each character having 34 choices to describe the details of the medical procedure or action. ICD-10-PCS is a common data set for the electronic health record, and comprises part of the data elements for the discharge data set. The code set is comparable across different US based health sectors, for uses including disease index, clinical data set, discharge data set, etc. The ICD-10-PCS code may be used as a data quality tool by understanding how the encoded procedure relates to diagnosis or treatment of a birth defect of interest.	Although standardized nationally, the ICD-10-PCS code is not used outside of the US. The code set only applies to inpatient procedures within the hospital setting and thus excludes events in outpatient/ambulatory or clinical settings. There is a steep learning curve in learning and interpreting the information in the 7-digit procedure codes.

Table 4. Classification and Coding Systems for Tests and Results

System	Description	Strengths	Limitations
ISCN	**I**nternational **S**ystem for **C**ytogenomic **N**omenclature (ISCN) is a standardized system for describing any genomic rearrangement. The classification system is comprised of specific symbols, terms, and abbreviations.	ISCN provides a common language for interpreting and reporting results consistently across different cytogenetic laboratories. The system applies to cytogenetic techniques (e.g., karyotype, DNA sequencing, FISH, microarray, region-specific assays, etc.) *and* to the findings obtained (e.g., chromosome banding, structural aberrations, numerical changes). Used in clinical and research settings. Cytogenetics labs may provide the complete ISCN nomenclature, to describe the specimen results, as well as the more familiar karyotype, FISH, microarray description and interpretation.	Only encompasses cytogenetic tests and results, not the broader range of results from other study modalities. (However, some cytogenetics labs provide several types of specimen analysis and will record the ISCN code per pertinent analysis.)
LOINC	**L**ogical **O**bservation **I**dentifiers **N**ames and **C**odes (LOINC) is a database for clinical results (e.g., lab tests, imaging tests, and measurement-related clinical observations or patient outcomes), which provides a unified terminology for results obtained at different sites. LOINC terminology has 2 main parts: • **Laboratory LOINC:** lab tests and micro-biology values • **Clinical LOINC:** non-lab values, such as obstetric ultrasound, ECG results, etc.	Considered more interoperable than other health information coding systems for lab values. LOINC is compatible with HL7, and is also used by other standards (e.g., ASTM, E1238, IHE, CEN/TC251) to electronically transfer results. Database has over 73,000 terms to describe lab results and observations.	Each term in the database represents a single test, observation, or measurement. They are arranged with a computable hierarchy, but there is no exposed semantic format so it can be difficult for a terminology server to interpret the hierarchy. The evolution and development of lab tests, in coordination with accompanying technologies, is constantly changing. This requires manual resources to process incoming case reports (including those produced for an eCR), and to manage tracking and mapping.