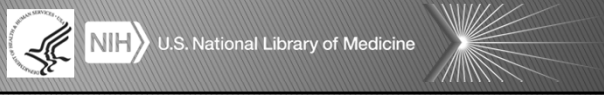
Diagnostic Coding and Case Definition for Critical Congenital Heart Disease (CCHD) Detected by Newborn Screening

APHL Newborn Screening and Genetic Testing Symposium, Atlanta GA, May 8, 2013

Alan Zuckerman[1], Swapna Abhyankar[1], Tiffany Colarusso[2], Richard Olney[2], Kristin Burns[3], Marci Sontag[4]

[1]National Library of Medicine, NIH, Bethesda, MD, USA, [2]Centers for Disease Control and Prevention, Atlanta, GA, USA, [3]National Heart Lung and Blood Institute, NIH, Bethesda, MD, USA, [4]Colorado School of Public Health/NewSTEPs, Denver, CO, USA



Disclosures

- *The findings and conclusions in this report are those of the authors and do not necessarily represent the official position of the Centers for Disease Control and Prevention, the National Institutes of Health, or the Health Resources and Services Administration*

Problem

- Critical congenital heart disease (CCHD) is the latest addition to the Recommended Uniform Screening Panel (RUSP).
- Unlike other newborn screening (NBS) conditions, it is not a single condition, but a group of disorders based on cardiac physiology that requires intervention soon after birth.
- However, a diagnosis at birth can evolve based on changing physiology, subsequent evaluations, or therapeutic interventions.
- Birth defects surveillance programs, pediatricians or nursery staff, and cardiac specialists often use diverse terms and codes for similar CCHD conditions to meet their specific needs and available data.

Adding CCHD Accompanied by Targeted Recommendations for Federal Agencies

- National Heart, Lung, and Blood Institute (NHLBI) to conduct research, including screening technologies and health outcomes for infants diagnosed with CCHD based on screening
- Centers for Disease Control and Prevention (CDC) to evaluate surveillance data and ability to monitor effectiveness of CCHD screening, conduct cost-effectiveness analyses of screening, and leverage opportunities for data use from electronic health records (EHRs).
- National Library of Medicine (NLM) to develop the standard terminology for screening and diagnosis to help facilitate health information exchange.
- Health Resources and Services Administration (HRSA) to guide the development of screening standards, protocols, and infrastructure to guide state public health programs.

Special Features of CCHD Screening

- The point-of-service (POS) test used for newborn CCHD screening is pulse oximetry, which non-invasively estimates oxygen saturation as a reflection of hypoxemia.
- Hypoxemia in a newborn can have several causes, one of which is the presence of a CCHD.
- Thus, it is important to distinguish between a positive screen for hypoxemia versus for CCHD, and to define the meaning of "false positive" accordingly.
- Newborn hearing screening also uses a POS test and is considered a single NBS condition with many different etiologies, some of which are heritable.

Importance of Uniform Diagnostic Coding

- Uniform diagnostic coding is important for all NBS conditions to facilitate public health activities, long-term follow-up, and research.
- For CCHD, uniform coding will enable:
  - Understanding of the phenotypic mix of those cases detected and undetected by NBS
  - Facilitate monitoring long-term outcomes
  - Enable comparisons between regions that have population-based screening and those that do not
- Uniform case definitions are important for accurate reporting, quality assurance and enrollment in studies.

Methods

- We mapped the seven conditions considered to be the primary targets for newborn CCHD screening by the Secretary's Advisory Committee for Heritable Disorders in Newborns and Children's Evidence Review Committee:
  - Hypoplastic left heart syndrome (HLHS)
  - Pulmonary atresia (PVA)
  - Truncus arteriosus (TA)
  - Total anomalous pulmonary venous connection (TAPVC)
  - d-Transposition of the great arteries (TGA)
  - Tetralogy of Fallot (TOF)
  - Tricuspid atresia (TVA)
- These are conditions that almost always produce hypoxemia and that are almost always cases of CCHD.

Other Targets of CCHD Newborn Screening Were Also Explored

- These are conditions that include both critical and non-critical variants.
- They are less likely to produce hypoxemia and may not be detected by pulse oximetry newborn screening.
- Examples of conditions tracked by some states include:
  - Coarctation of the aorta (COA)
  - Double outlet right ventricle (DORV)
  - Ebstein's anomaly (EA)
  - Interrupted aortic arch (IAA)
  - Severe pulmonary valve stenosis (PVAS)
  - Severe tricuspid valve stenosis (TVAS)
  - Single ventricle (SV)
- Non-cardiac conditions, such as Respiratory Distress Syndrome are also secondary targets of pulse oximetry screening.

The NLM Approach to Mapping Codes for CCHD

- The NLM used the same approach for CCHD coding that was used for other NBS conditions, and provided the codes and code mappings on its NBS coding website: http://newbornscreeningcodes.nlm.nih.gov
- Common code systems used by stakeholders were identified.
- The Unified Medical Language System (UMLS) Metathesaurus identified potential codes and mapping between code systems, and variants and modifications were explored.
- Domain-specific and genomic databases outside the UMLS, such as Online Mendelian Inheritance in Man® (OMIM), International Pediatric and Congenital Cardiac Code (IPCCC), and Society of Thoracic Surgeons (STS), were included as needed.
- Domain experts were consulted for distinguishing uncertain mappings.

Stakeholders for CCHD Diagnostic Coding

- Patients
  - Knowing and understanding the specific diagnosis and related information
- Newborn nursery and neonatal intensive care unit (NICU) clinicians
  - Conduct CCHD screening and record data in their paper and electronic clinical records
- Hospitals
  - Act as a site of screening, bill for services and report birth defects
- Cardiologists
  - Evaluate infants who screen positive and make the initial diagnosis
- Cardiac surgeons
  - Provide treatment and maintain a national congenital heart surgery database
- Pediatric and other primary care providers
  - Provide a medical home and primary care services

Stakeholders for CCHD Diagnostic Coding (cont.)

- State newborn screening programs
  - Track all NBS, including point-of-care testing
- State birth defects surveillance programs
  - Track all congenital heart disease including CCHD, whether detected by NBS or other means
- CDC
  - Fund and conduct birth defects surveillance activities
- Researchers
  - Seek to improve screening, diagnostic, and treatment strategies
- HRSA and NewSTEPs (Newborn Screening Technical Assistance and Evaluation Program)
  - Provide technical assistance to states and maintain national statistics on all NBS programs

Administrative Coding Systems for CCHD

- ICD-9-CM (International Classification of Diseases, 9th Revision, Clinical Modification)
  - Used by hospitals for billing and public health reporting
- CDC/BPA (Centers for Disease Control and Prevention/British Paediatric Association (BPA))
  - Modification of ICD-9-CM
  - Uses six-digit codes for birth defects surveillance
- ICD-10-CM (International Classification of Diseases, 10th Revision, Clinical Modification)
  - Alphanumeric
  - Federally mandated for adoption in EHRs in 2015

Clinical Coding Systems for CCHD Newborn Screening

- SNOMED CT (Systematized Nomenclature of Medicine Clinical Terms)
  - Maintained by the International Health Terminology Standards Development Organization
  - Required for EHR Problem Lists under Meaningful Use incentive programs starting in 2014
- OMIM (Online Mendelian Inheritance in Man®)
  - Maintained by Johns Hopkins University
  - Part of National Center for Biotechnology Information Genomic Databases
  - Catalogs of specific genes and chromosome loci
- UMLS (Unified Medical Language System®)
  - Maintained by the National Library of Medicine
  - Uses a Concept Unique Identifier (CUI) to link coding systems and terms

Specialized Cardiac Coding Systems

- IPCCC (International Pediatric and Congenital Cardiac Code)
  - Very detailed, multi-axial coding uses multiple codes to capture full detail of the lesion
  - Often requires data available only at the time of surgery or cardiac catheterization
  - Strongly encouraged by NIH/NHLBI for cardiac research
- STS (Society of Thoracic Surgeons)
  - A four-digit subset and mapping of IPCCC for the STS Congenital Heart Surgery Database
  - May become a good resource for long term follow-up of CCHD and tracking patients who are treated outside of their State of birth or residence

Hypoplastic Left Heart Syndrome (HLHS)

Code System	Code	Description
CDC/BPA	746.700	Hypoplastic left heart syndrome (HLHS)
ICD-9-CM	746.7	Hypoplastic left heart syndrome
ICD-10-CM	Q23.4	Hypoplastic left heart syndrome
OMIM	241550	Hypoplastic left heart syndrome
OMIM	214350	Hypoplastic left heart syndrome 1; HLHS1
OMIM	614435	Hypoplastic left heart syndrome 2; HLHS2
SNOMED CT	62067003	Hypoplastic left heart syndrome
STS IPCCC	730	Hypoplastic left heart syndrome (HLHS)
UMLS CUI	C0152101	Hypoplastic left heart syndrome

Pulmonary [Valve] Atresia (PVA)

Code System	Code	Description
CDC/BPA	746.000	Atresia, hypoplasia of pulmonary valve
ICD-9-CM	746.01	Atresia of pulmonary valve, congenital
ICD-10-CM	Q22.0	Pulmonary valve atresia
OMIM	265150	Pulmonary atresia with intact ventricular septum
SNOMED CT	204342004	Congenital atresia of pulmonary valve
STS IPCCC	330	Pulmonary atresia, intact ventricular septum
UMLS CUI	C0242855	Congenital atresia of pulmonary valve

Truncus Arteriosus (TA)

Code System	Code	Description
CDC/BPA	745.000	Persistent truncus arteriosus
ICD-9-CM	745.0	Common truncus
ICD-10-CM	Q20.0	Common arterial trunk
OMIM	217095	Conotruncal heart malformation; CTHM; Truncus arteriosus included
SNOMED CT	61959006	Common truncus arteriosus (disorder)
STS IPCCC	160	Truncus arteriosus
UMLS CUI	C0041207	Truncus arteriosus, persistent

Total Anomalous Pulmonary Venous Connection [Return] (TAPVC/TAPVR)

Code System	Code	Description
CDC/BPA	747.420	Total anomalous pulmonary venous return (TAPVR)
ICD-9-CM	747.41	Total anomalous pulmonary venous connection
ICD-10-CM	Q26.2	Total anomalous pulmonary venous connection
OMIM	106700	Total anomalous pulmonary venous return 1; TAPVR1
SNOMED CT	204456001	Subdiaphragmatic total anomalous pulmonary venous return
SNOMED CT	204457005	Supradiaphragmatic total anomalous pulmonary venous return
STS IPCCC	200	Total anomalous pulmonary venous connection, Type 1 (supracardiac)
STS IPCCC	210	Total anomalous pulmonary venous connection, Type 2 (cardiac)
STS IPCCC	220	Total anomalous pulmonary venous connection, Type 3 (infracardiac)
STS IPCCC	230	Total anomalous pulmonary venous connection, Type 4 (mixed)
UMLS CUI	C0265917	Supradiaphragmatic total anomalous pulmonary venous return
UMLS CUI	C0265921	Subdiaphragmatic total anomalous pulmonary venous return

d-Transposition of the Great Arteries (d-TGA)

Code System	Code	Description
CDC/BPA	745.100	Transposition of great vessels, complete, no ventricular septal defect (VSD)
CDC/BPA	745.110	Transposition of great vessels, incomplete, with VSD
CDC/BPA	745.140	Double outlet right ventricle w/ transposed great vessels
CDC/BPA	745.190	Unspecified transposition of great vessels
ICD-9-CM	745.10	Complete transposition of great vessels
ICD-10-CM	Q20.3	Discordant ventriculoarterial connection
OMIM	608808	Transposition of great arteries 1, Dextro-loop 1; DTGA1
OMIM	613853	Transposition of great arteries 2, Dextro-loop 2; DTGA2
OMIM	613854	Transposition of great arteries 3, Dextro-loop 3; DTGA3
SNOMED CT	26146002	Complete transposition of great vessels (disorder)
SNOMED CT	204297006	Total great vessel transposition
STS IPCCC	800	TGA-IVS (intact ventricular septum)
STS IPCCC	890	TGA-IVS-LVOTO(left ventricular outflow tract obstruction)
STS IPCCC	900	TGA-VSD
STS IPCCC	910	TGA-VSD-LVOTO (left ventricular outflow tract obstruction)
UMLS CUI	C0040761	Transposition of Great Vessels

Tetralogy of Fallot (TOF)

Code System	Code	Description
CDC/BPA	745.200	Fallot's tetralogy
CDC/BPA	745.210	Fallot's pentalogy (tetralogy + atrial septal defect (ASD))
CDC/BPA	747.310	Pulmonary atresia with VSD (severe TOF)
ICD-9-CM	745.2	Tetralogy of Fallot (incl. pentalogy)
ICD-10-CM	Q21.3	Tetralogy of Fallot
OMIM	187500	Tetralogy of Fallot; TOF
SNOMED CT	86299006	Tetraology of Fallot (disorder)
STS IPCCC	290	TOF
STS IPCCC	340	Pulmonary atresia (including TOF, PA)
UMLS CUI	C0039685	Tetralogy of Fallot

Tricuspid [Valve] Atresia (TVA)

Code System	Code	Description
CDC/BPA	746.100	Tricuspid atresia
ICD-9-CM	746.1	Tricuspid atresia and stenosis, congenital
ICD-10-CM	Q22.4	Congenital tricuspid stenosis (incl atresia)
OMIM	605067	Tricuspid atresia
SNOMED CT	204354004	Congenital tricuspid atresia and stenosis
STS IPCCC	390	Tricuspid stenosis
UMLS CUI	C0158616	Congenital tricuspid atresia and stenosis

Summary Results of Code Mapping

- While we could map each condition to every coding system, the mapping between systems was imperfect due to variations in term definitions.
- Coding system differences which hindered mapping included:
  - Having multiple codes in one system to specify anatomic variants of a single condition
  - Specifying different genomic associations for some conditions
  - Inability to distinguish critical and non-critical disease
    - For example, some codes are broad and do not allow differentiation between critical and non-critical disease, such as ICD-9-CM code 746.1 for both tricuspid atresia and stenosis.

Special CCHD Coding Challenges

- Often clinical data beyond the initial screening diagnosis would be needed to map to a specific code, such as subdiaphragmatic total anomalous pulmonary venous return or dextro-transposition of the great arteries. Stakeholders select codes to meet their specific needs and available data.
- Modifications to hospital-based ICD-9-CM codes, used by CDC and some state birth defect surveillance programs, improve the specificity of CCHD coding and ability to do population-based surveillance and research.
- Some states may choose to track more than the seven primary target conditions, so it is also important to aggregate all CCHD conditions.

Conclusions

- CCHD is a spectrum of conditions beyond the seven primary targets of newborn screening.
- Case definitions and descriptions will be important to evaluate the types of CCHD detected by screening and create a foundation for public health reporting, long-term follow-up, and closing evidence gaps.
- While different coding systems serve the purposes for which they were created, a crosswalk between the codes, highlighting similarities and differences, could help facilitate meaningful data exchange between stakeholders.

Conclusions (cont.)

- Code mapping is feasible and will facilitate data exchange among stakeholders.
- Migration to ICD-10-CM will not create significant barriers as the ICD-9-CM codes appear well-aligned to ICD-10-CM for these conditions.
- Detailed specialized cardiac coding from the STS, the CDC, and from birth defects surveillance programs will help capture details available after NBS and assist long term follow-up and research.
- The UMLS will help link future updates and additions to coding and new terms or synonyms.

For More Information

- http://newbornscreeningcodes.nlm.nih.gov
- Key References:
  - Mai C. et al "Selected Birth Defects Data from Population-based Birth Defects Surveillance Programs in the United States, 2005-2009: Featuring Critical Congenital Heart Defects Targeted for Pulse Oximetry Screening" *Birth Defects Research (Part A) 2012; 94:970-983.*
  - Houyel et al, "Population-based evaluation of a suggested anatomic and clinical classification of congenital heart defects based on the International Paediatric and Congenital Cardiac Code" *Orphanet Journal of Rare Diseases 2011, 6:64*
- Contact:
  - Alan Zuckerman: Alan.Zuckerman@nih.gov
  - Swapna Abhyankar: Swapna.Abhyankar@nih.gov