

# Neonatal Research Network Terminology Harmonization

A Formative Research Initiative of the  
National Children's Study

National Children's Study  
Metadata Repository Workshop

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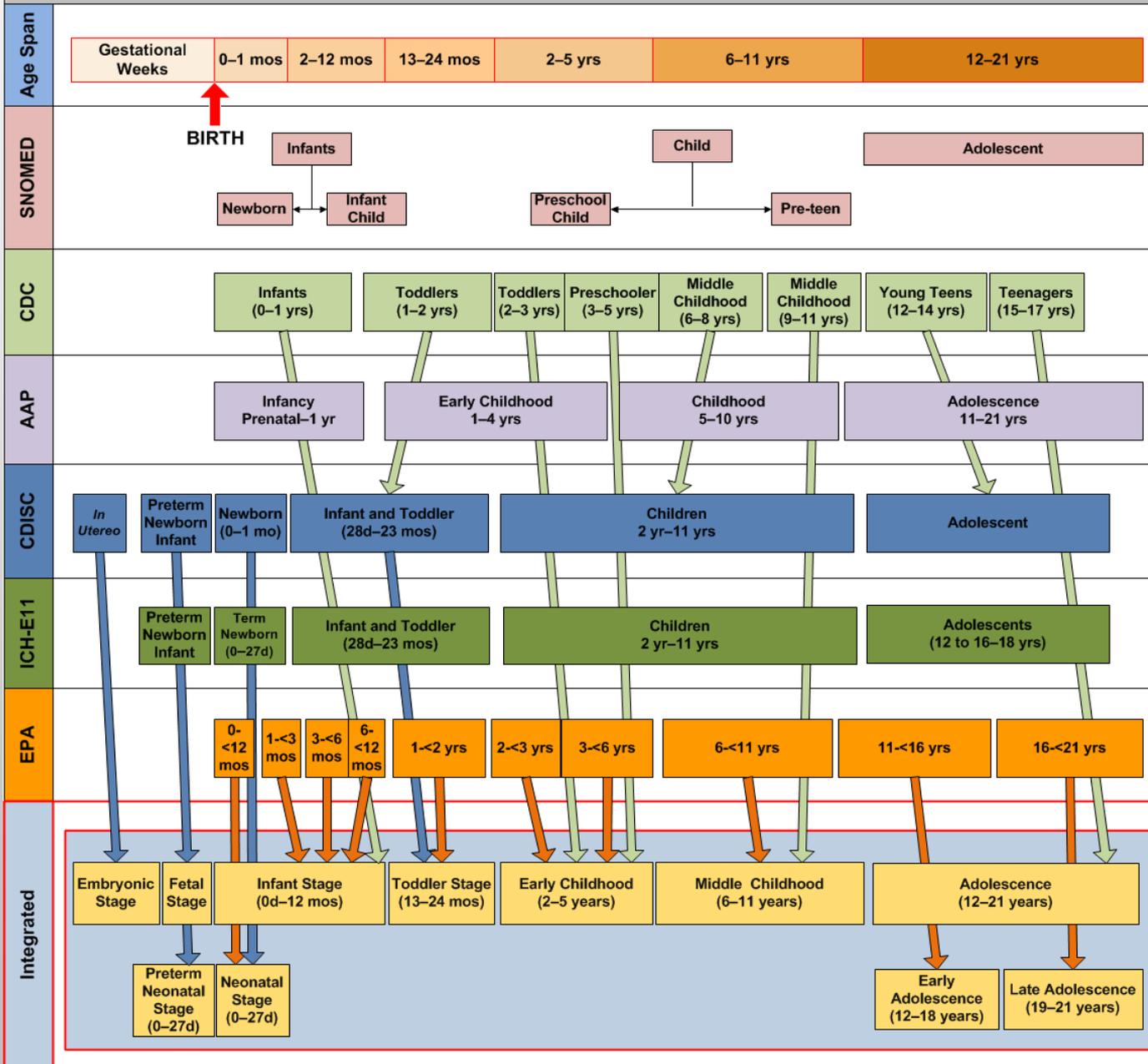
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# NCS Formative Research: Terminology

- focused, time-limited activities to address specific technical or methodological questions
- “*capacity building to support a scalable infrastructure*”
- Study Logistics Analysis and Improvements
  - Terminology
    - Data sharing and integrated analyses rely on ***harmonizing*** (or standardizing) critical **concepts**, specific terms and data fields
    - Child health research needs a consistent terminology to bridge concepts among multiple disciplines and across developmental stages

**Integrated Child Life Stages for NICHD Pediatric Terminology mapped to existing classifications used by medical standards/ organizations**



- SNOMED:** Systematized Nomenclature of Medicine
- CDC:** Centers for Disease Control and Prevention
- AAP:** American Academy of Pediatrics
- CDISC:** Clinical Data Interchange Standards Consortium
- ICH-E11:** International Conference on Harmonization
- EPA:** Environmental Protection Agency

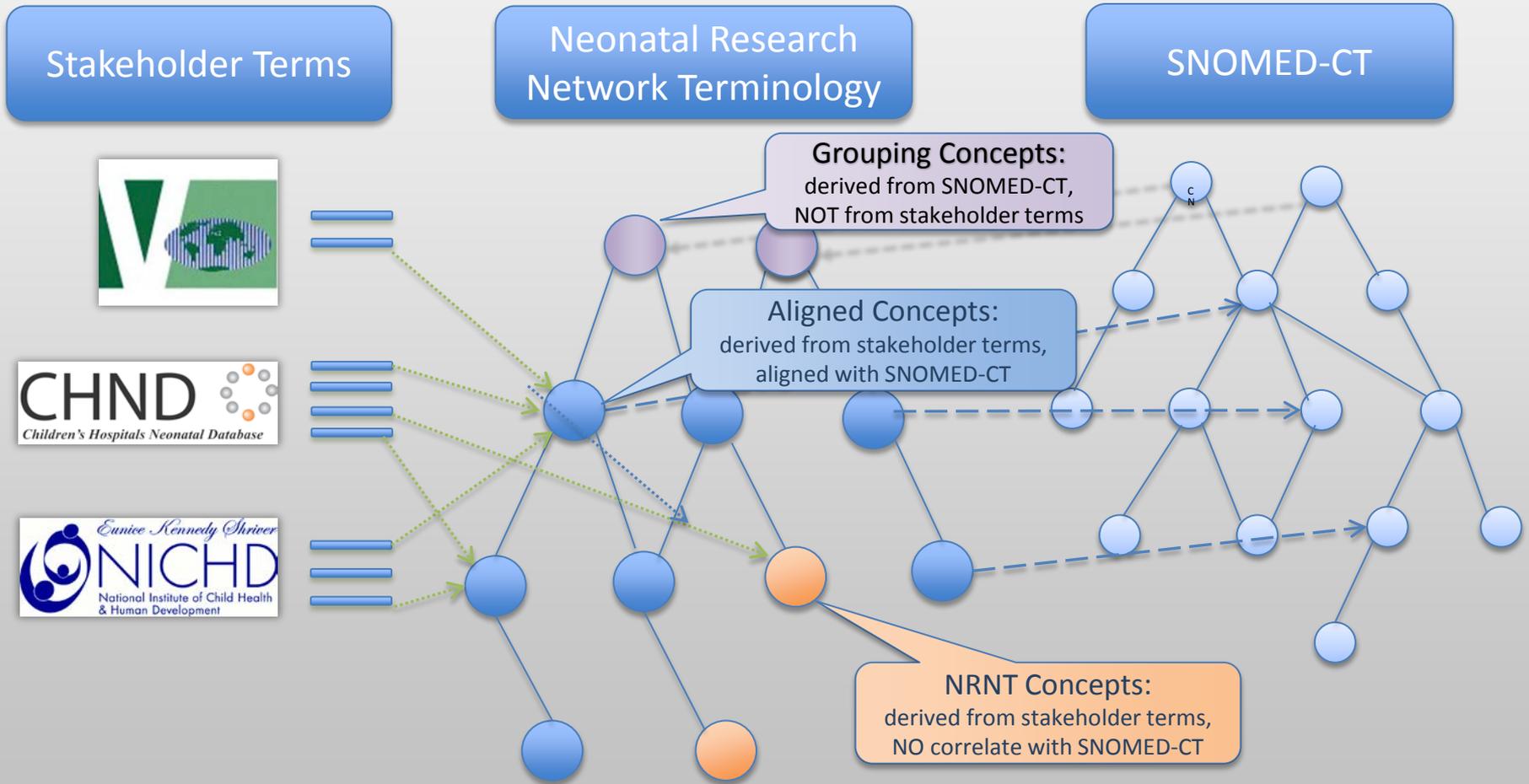
# The Neonatal Research Networks

- Vermont-Oxford Network (VON)
  - co-morbidities of very low birth weight (VLBW) infants
- Children's Hospitals Neonatal Database (CHND)
  - therapies & outcomes of premature and term infants referred to tertiary centers for surgical/subspecialty care
- National Institute of Child Health and Development (NICHD)
  - data fields from prospective clinical trials (RCTs)

# NRNT Harmonization Project Team

- Subject Matter Experts
  - Stakeholders:
    - Children's Hospital Neonatal Database (CHND)
    - Vermont Oxford Network (VON)
    - National Institute of Child Health & Human Development (NICHD)  
Neonatal Research Network
  - Additional clinicians with informatics & terminology expertise
- Project oversight: Apelon, Inc. & Booz Allen Hamilton
- Aim: Harmonize concepts among these networks
  - Develop common mapping of terms and concepts
    - map to reference terminologies (i.e., *SNOMED-CT*)
    - integrated hierarchy of concepts among the networks for use in the NCS and future research activities

# Development of a Harmonized Terminology



# NRN Terminology Mapping

## Disorder of Blood Vessel - Proposed Concepts

	Forum Thread	Approval	Reference Number 	Proposed NRNT Concept	Proposed NRNT Parent(s)	SNOMED CT Mapping	Corresponding Network Term(s)
	Disorder of Blood Vessel	Yes		<b>Please edit this row and click "Yes" if you wish to approve all proposed concepts in this table.</b>			
	Disorder of Blood Vessel		100	Great vessel abnormalities, other	Congenital vascular disorder		Other Great Vessel Abnormalities [CHND]
	Disorder of Blood Vessel		101	Transposition of great vessels	Congenital vascular disorder	Discordant ventriculoarterial connection (disorder)	Transposition of the Great Vessels [VON]; Transposition of the Great Vessels, Transposition unspecified [CHND]; Transposition of the Great Vessels [NICHD NRN]
	Disorder of Blood Vessel		102	Dextro- transposition of the great vessels	Transposition of great vessels	D - transposition of the great vessels (disorder)	
	Disorder of Blood Vessel		103	Dextro-transposition of the great vessels with intact ventricular septum	Dextro-transposition of the great vessels		Transposition of the Great Vessels, D-TGA with intact Ventricular Septum [CHND]
	Disorder of Blood Vessel		104	Dextro-transposition of the great vessels with ventricular septal defect	Dextro-transposition of the great vessels		Transposition of the Great Vessels, D-TGA with VSD [CHND]
	Disorder of Blood Vessel		105	Levo - transposition of the great vessels	Transposition of great vessels	L - transposition of the great vessels (disorder)	Transposition of the Great Vessels, L-TGA [CHND]
	Disorder of Blood Vessel		106	Transposition of the great vessels, other	Transposition of great vessels		Transposition of the Great Vessels, Transposition, unspecified [CHND]
	Disorder of Blood Vessel		111	Partial anomalous pulmonary venous return	Congenital anomaly of pulmonary veins	Partial anomalous pulmonary venous connection	Partial Anomalous Pulmonary Venous Return (PAPVR) [CHND]

# NRNT Hierarchy

- Congenital anomaly
  - Congenital anomaly of cardiovascular system
    - Congenital heart disease
      - Cardiac septal defects
        - Atrial septal defect
        - Ventricular septal defect
        - Single ventricle
      - Complete atrioventricular canal defect
        - Complete atrioventricular canal defect (balanced)
        - Complete atrioventricular canal defect (unbalanced)
      - Complex congenital heart disease with left ventricle only
      - Complex congenital heart disease with right ventricle only
      - Congenital abnormality of atria and atrial septum
        - Cor triatriatum
      - Congenital abnormality of ventricles and ventricular septum
        - Left ventricular outflow tract abnormality, other specified
        - Left ventricular outflow tract obstruction
        - Tetralogy of Fallot
          - Tetralogy of fallot - complete av canal defect
          - Tetralogy of fallot - type unknown
          - Tetralogy of fallot with pulmonary atresia
          - Tetralogy of fallot with pulmonary stenosis
    - Congenital anomaly of heart valve
      - Aortic valve atresia
        - Hypoplastic left heart syndrome with aortic atresia
      - Congenital atresia of the pulmonary valve
      - Congenital atresia of tricuspid valve
        - Congenital atresia of tricuspid valve with pulmonary atresia
      - Congenital insufficiency of aortic valve
      - Congenital insufficiency of pulmonary valve
      - Ebstein's anomaly
      - Mitral atresia
        - Hypoplastic left heart syndrome with mitral atresia
      - Pulmonary atresia
        - Congenital atresia of tricuspid valve with pulmonary atresia
  - Dextrocardia

# NRNT by Organ-system Domains

Body System	NRNT Concepts	Stakeholder Terms	SNOMED CT Mappings	NRNT-Specific Concepts
CNS	249	162	197	50
Cardiovascular	209	141	155	50
Endocrinology	80	40	76	4
Genetics	163	97	153	9
Gastrointestinal	158	119	135	23
Genitourinary	89	56	79	10
Hematology	125	66	117	8
Metabolic	112	67	110	2
Musculoskeletal	70	44	66	4
Oncology	33	13	31	2
Ophthalmology	83	41	58	25
Respiratory	114	94	84	29
Skin	105	38	97	8

\* The same concept will be counted in multiple rows if it is included in multiple body systems

# Lessons Learned

- Addressing Gap between NRNT Terms and SNOMED-CT
  - Approximately 20% of concepts not represented
  - Request addition of concepts to SNOMED-CT
  - Consider adding additional reference terminologies for mapping
- Assess Intra-network Alignment of Data Definitions
  - Critical for meta-analyses, studies spanning multiple networks
  - Optimal while stakeholder subject matter experts are closely reviewing terms (& definitions)

# Intra-network Concept Alignment: Capturing Precision of Data Definitions

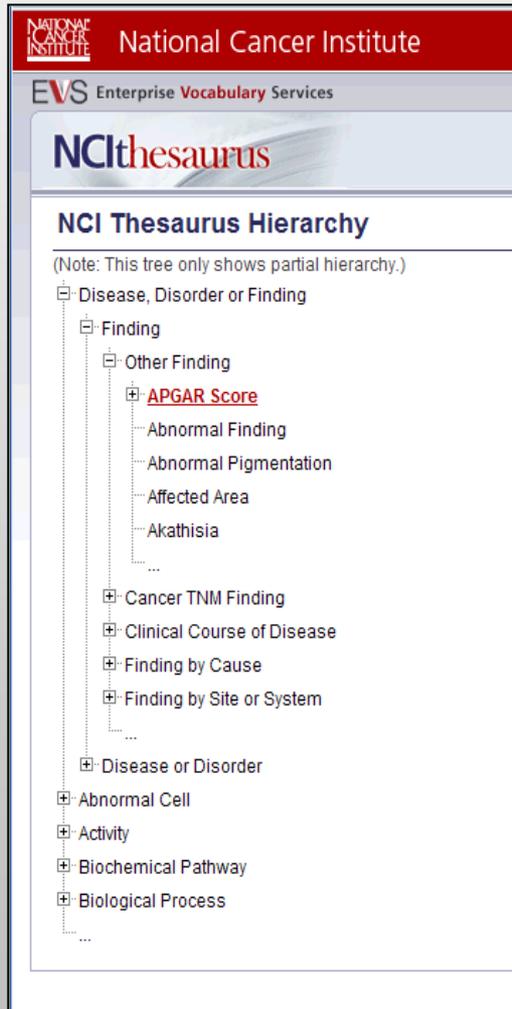
- Necrotizing Enterocolitis*

Data Definition	VON	CHND	NICHD	SNOMED-CT Representation
VON	----	Exact	Approximate	Yes
CHND	Exact	----	Approximate	Yes
NICHD	Approximate	Approximate	-----	Yes

- Retinopathy of Prematurity*

Data Definition	VON	CHND	NICHD	SNOMED-CT Representation
VON	----	Exact	Exact	Yes
CHND	Exact	----	Exact	Yes
NICHD	Exact	Exact	-----	Yes

# Promoting Access to NRNT



The screenshot shows the NCI Thesaurus Hierarchy interface. At the top, there is a red header with the National Cancer Institute logo and the text "National Cancer Institute". Below this is a grey header with "EVS Enterprise Vocabulary Services" and the "NCIthesaurus" logo. The main content area is titled "NCI Thesaurus Hierarchy" and includes a note: "(Note: This tree only shows partial hierarchy.)". The hierarchy is a tree structure starting with "Disease, Disorder or Finding", which is expanded to show "Finding". Under "Finding", there are several sub-categories, including "Other Finding", which is further expanded to show "APGAR Score" (highlighted in red), "Abnormal Finding", "Abnormal Pigmentation", "Affected Area", and "Akathisia". Other categories under "Finding" include "Cancer TNM Finding", "Clinical Course of Disease", "Finding by Cause", and "Finding by Site or System". Below "Finding" are "Disease or Disorder", "Abnormal Cell", "Activity", "Biochemical Pathway", and "Biological Process".

- NRNT to be incorporated into NICHD Pediatric Terminology in the NCI Thesaurus
  - tagged subset of NRNT terms
  - coordination with NCI EVS team
  - instance of the Pediatric Terminology is also available on BioPortal for dissemination to a broader community.