Representation of Disorders of the Newborn Infant by SNOMED CT®

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SNOMED Clinical Terminology

Comprehensive, concept based, clinical terminology

Semantic model based on description logic

Unique identification and description of clinically relevant concepts
SNOMED structure . . .

Polyhierarchial structure, multiple parent-child relationships

Relationships across different subtype hierarchies

Relationships define meaning of a concept relative to other SNOMED concepts
SNOMED contents . . .

Contains more than 370,000 active concepts

More than 1,000,000 active descriptions

Nearly 1,500,000 relationships

SNOMED expressions are references to SNOMED concepts
SNOMED clinical terms . . .

Clinical terms may be represented as

- a single, pre-coordinated SNOMED expression
- a post-coordinated SNOMED compositional expression of two or more SNOMED concepts

SNOMED CT is the most sophisticated reference terminology currently available for the representation of health care
Representation of clinical terms

Evolution from SNOP into SNOMED, SNOMED RT, and SNOMED CT over a period of forty years

Content largely determined by voluntary contributions by many diverse clinical groups

Clinically relevant terms of specialised clinical domains may not be represented in SNOMED CT
Evolution of SNOMED . . .

<table>
<thead>
<tr>
<th>Year</th>
<th>Terminology</th>
<th>Comments</th>
</tr>
</thead>
<tbody>
<tr>
<td>1965</td>
<td>SNOP</td>
<td>Focus on morphology [10,990 concepts]</td>
</tr>
<tr>
<td>1974</td>
<td>SNOMED</td>
<td>Focus on morphology, etiology and location</td>
</tr>
<tr>
<td>1979</td>
<td>SNOMED II</td>
<td>[30,547 concepts]</td>
</tr>
<tr>
<td>1993</td>
<td>SNOMED Version 3.0</td>
<td>Focus on disorders, morphology, living organisms and social context [96,042 concepts]</td>
</tr>
<tr>
<td>1997</td>
<td></td>
<td>LOINC codes integrated into SNOMED</td>
</tr>
<tr>
<td>1998</td>
<td>SNOMED Version 3.5</td>
<td></td>
</tr>
<tr>
<td>2000</td>
<td>SNOMED RT</td>
<td>Logic based approach for definition of SNOMED concept, axes became hierarchies, polyhierarchical approach to classification [128,030 concepts]</td>
</tr>
<tr>
<td>2002</td>
<td>SNOMED CT</td>
<td>Merger of SNOMED RT and NHS Clinical Terms, version 3 [325,857 concepts]</td>
</tr>
<tr>
<td>2007</td>
<td></td>
<td>Transfer of SNOMED CT to the newly formed International Health Terminology Standards Development Organization</td>
</tr>
</tbody>
</table>
Representation of disorders of the newborn infant . . .

Systematic examination of SNOMED CT to assess the representation of disorders of the newborn infant by the terminology

- SNOMED CT [July 2006 release]

- CliniClue terminology Browser [version 2006.2.8, October 7, 2006]
Terms for neonatal disorders

434 terms for neonatal disorders

Extracted from the Clinical Information Management System (CIMS), Neonatal Intensive Care Unit, Hospital for Sick Children, Toronto

Structured data entry since 2003 for clinical, research and administrative purposes
Representative CIMS terms for neonatal disorders...

<table>
<thead>
<tr>
<th>Frequently used CIMS terms</th>
</tr>
</thead>
<tbody>
<tr>
<td>Transient tachypnoea of the newborn, meconium aspiration syndrome, respiratory distress syndrome, bronchopulmonary dysplasia</td>
</tr>
<tr>
<td>Ventricular septal defect, patent ductus arteriosus, transposition of the great arteries, pulmonary stenosis, coarctation of aorta</td>
</tr>
<tr>
<td>Gastroesophageal reflux, necrotising enterocolitis duodenal atresia, tracheo-esophageal atresia/fistula</td>
</tr>
<tr>
<td>Urinary tract infection, hypospadias, renal failure</td>
</tr>
<tr>
<td>Anaemia, haemolytic disease of the newborn</td>
</tr>
<tr>
<td>Hypoglycaemia, hypocalcaemia, hypothyroidism</td>
</tr>
<tr>
<td>Hypoxic-ischaemic encephalopathy, intraventricular haemorrhage, periventricular leucomalacia, retinopathy of prematurity, seizures</td>
</tr>
<tr>
<td>Congenital dislocation of the hip, talipes equinovarus</td>
</tr>
<tr>
<td>Group B streptococcal infection, RSV infection</td>
</tr>
<tr>
<td>Intrauterine growth retardation, cephalohaematoma, Erb's palsy, non-immune fetal hydrops</td>
</tr>
</tbody>
</table>
Representative CIMS terms for neonatal disorders . . .

<table>
<thead>
<tr>
<th>Less frequently used CIMS terms</th>
</tr>
</thead>
<tbody>
<tr>
<td>Pulmonary interstitial emphysema, subglottic stenosis, congenital diaphragmatic hernia, pulmonary hypoplasia, pulmonary agenesis</td>
</tr>
<tr>
<td>Atrioventricular canal defect, hypoplastic left heart syndrome, interrupted aortic arch, total anomalous pulmonary venous drainage, congenital heart block</td>
</tr>
<tr>
<td>Meconium ileus, gastroschisis, malrotation, Hirschsprung's disease, anorectal malformation</td>
</tr>
<tr>
<td>Renal agensis, epispadias, extrophy of the bladder</td>
</tr>
<tr>
<td>Congenital thrombocytopenia</td>
</tr>
<tr>
<td>Congenital adrenal hyperplasia, phenylketonuria</td>
</tr>
<tr>
<td>Anencephaly, encephalocele, holoprosencephaly, cerebellar haemorrhage, transient myasthenia gravis, transverse venous sinus thrombosis</td>
</tr>
<tr>
<td>Submucous cleft palate, septic arthritis</td>
</tr>
<tr>
<td>Congenital rubella syndrome</td>
</tr>
<tr>
<td>Neonatal drug withdrawal, subarachnoid haemorrhage, skull fracture due to birth injury</td>
</tr>
</tbody>
</table>
## Criteria for representation

<table>
<thead>
<tr>
<th>Representation of ClMS terms within SNOMED</th>
<th>Criterion for degree of representation</th>
</tr>
</thead>
<tbody>
<tr>
<td>Complete representation</td>
<td>Matching SNOMED concept</td>
</tr>
<tr>
<td>• simple match</td>
<td>• single, pre-coordinated SNOMED expression</td>
</tr>
<tr>
<td>• compositional match</td>
<td>• post-coordinated SNOMED compositional expression</td>
</tr>
<tr>
<td>Partial representation</td>
<td>Related SNOMED concept</td>
</tr>
<tr>
<td></td>
<td>• related concept too broad</td>
</tr>
<tr>
<td></td>
<td>• related concept too narrow</td>
</tr>
<tr>
<td></td>
<td>• related concept different</td>
</tr>
<tr>
<td>Absent representation</td>
<td>No representation within SNOMED</td>
</tr>
</tbody>
</table>
Search strategy . . .

No restrictions applied to the search

Initial search for an exact lexical match

Subsequent search for a semantic match

At least three separate searches for terms not represented in SNOMED CT
Data collection . . .

<table>
<thead>
<tr>
<th>Fully specified name</th>
</tr>
</thead>
<tbody>
<tr>
<td>Concept Id</td>
</tr>
<tr>
<td>Description Id</td>
</tr>
<tr>
<td>Concept Status [current or limited]</td>
</tr>
<tr>
<td>Status of the definition for the Concept [fully defined or primitive]</td>
</tr>
<tr>
<td>Hierarchy tag</td>
</tr>
<tr>
<td>Number of synonyms</td>
</tr>
</tbody>
</table>
# Representation of CIMS terms

<table>
<thead>
<tr>
<th>Category of representation</th>
<th>CIMS terms (N)</th>
<th>CIMS terms (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Complete representation</td>
<td>394</td>
<td>90.8%</td>
</tr>
<tr>
<td>Partial representation</td>
<td>28</td>
<td>6.4%</td>
</tr>
<tr>
<td>Absent representation</td>
<td>12</td>
<td>2.8%</td>
</tr>
</tbody>
</table>
Categorisation of CIMS terms . . .

CIMS terms categorised as congenital or acquired

Representation of 100% terms categorised as congenital

Representation of 96.4% terms categorised as acquired
Unique neonatal terms . . .

CIMS term/SNOMED concept includes the word neonatal, or use of the CIMS term is restricted to the neonatal period.

Representation of 97.8% unique neonatal terms

Representation of 98% general terms
Properties of SNOMED concepts for represented terms . . .

<table>
<thead>
<tr>
<th>Properties of SNOMED concepts</th>
<th>CIMS terms (N)</th>
<th>CIMS terms (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>current Concept Status</td>
<td>341</td>
<td>86.5%</td>
</tr>
<tr>
<td>Pre-coordinated SNOMED concept</td>
<td>379</td>
<td>96.2%</td>
</tr>
<tr>
<td>Post-coordinated SNOMED concept</td>
<td>15</td>
<td>3.8%</td>
</tr>
<tr>
<td>Fully defined concept</td>
<td>483</td>
<td>40.1%</td>
</tr>
<tr>
<td>Synonyms present</td>
<td>179</td>
<td>45.4%</td>
</tr>
</tbody>
</table>
CIMS terms not represented

<table>
<thead>
<tr>
<th>CIMS term</th>
<th>Category</th>
</tr>
</thead>
<tbody>
<tr>
<td>Birth injury to peripheral nervous system, unspecified</td>
<td>Unique neonatal</td>
</tr>
<tr>
<td>Craniofacial</td>
<td>General</td>
</tr>
<tr>
<td>Hemolytic diseases of fetus and newborn, unspecified</td>
<td>Unique neonatal</td>
</tr>
<tr>
<td>Non-hemolytic jaundice</td>
<td>General</td>
</tr>
<tr>
<td>Other neonatal stroke, unspecified</td>
<td>Unique neonatal</td>
</tr>
<tr>
<td>Perinatal depression</td>
<td>Unique neonatal</td>
</tr>
<tr>
<td>Suboptimal vascular access</td>
<td>General</td>
</tr>
<tr>
<td>Transient disorders of calcium and magnesium metabolism</td>
<td>General</td>
</tr>
<tr>
<td>Unique perinatal diagnosis</td>
<td>Unique neonatal</td>
</tr>
<tr>
<td>Unspecified disturbance of potassium balance of newborn</td>
<td>Unique neonatal</td>
</tr>
<tr>
<td>Unspecified disturbance of sodium balance of newborn</td>
<td>Unique neonatal</td>
</tr>
<tr>
<td>Unspecified intrauterine growth restriction</td>
<td>Unique neonatal</td>
</tr>
</tbody>
</table>
CIMS terms for diagnosis . . .

More than 97% of the CIMS terms for diagnosis are represented within SNOMED CT

Half of the CIMS terms that are not represented should not exist within a contemporary clinical terminology

Nearly 90% of the completely represented CIMS terms for diagnosis have a current Concept Status
Complete representation . . . . .

More than 95% of the CIMS terms for diagnosis represented by a single, pre-coordinated SNOMED expression

Only 40% of the terms for diagnosis are represented by a fully defined SNOMED concept

Nearly 50% of the terms for diagnosis have synonyms
An observation . . .

Respiratory Distress Syndrome

Concept Status: Current

Descriptions
- F respiratory distress syndrome in the newborn (disorder)
- P respiratory distress syndrome in the newborn
- S cardiorespiratory distress syndrome of newborn
- S congenital alveolar dysplasia
- S HMD – hyaline membrane disease
- S hyaline membrane disease
- S IRDS – idiopathic respiratory distress syndrome
- S IRDS of newborn
- S idiopathic respiratory distress syndrome
- S idiopathic respiratory distress syndrome of newborn
- S neonatal respiratory distress syndrome
- S pulmonary hypoperfusion syndrome of newborn
- S RDS - respiratory distress syndrome of newborn
- S RDS of newborn
- S respiratory distress syndrome of newborn
- S wet lung disease of newborn
Erroneous synonyms...

Respiratory Distress Syndrome

Concept Status: Current

Descriptions

- respiratory distress syndrome in the newborn (disorder)
- respiratory distress syndrome in the newborn
- cardiopulmonary distress syndrome of newborn
- congenital alveolar dysplasia
- HMD – hyaline membrane disease
- hyaline membrane disease
- IRDS — idiopathic respiratory distress syndrome
- IRDS of newborn
- idiopathic respiratory distress syndrome
- idiopathic respiratory distress syndrome of newborn
- neonatal respiratory distress syndrome
- pulmonary h ypoperfusion syndrome of newborn
- RDS - respiratory distress syndrome of newborn
- RDS of newborn
- respiratory distress syndrome of newborn
- wet lung disease of newborn
A simple update . . .

Respiratory Distress Syndrome

Concept Status: Current

Descriptions

F respiratory distress syndrome in the newborn (disorder)
P respiratory distress syndrome in the newborn
S HMD – hyaline membrane disease
S hyaline membrane disease
S neonatal respiratory distress syndrome
S RDS - respiratory distress syndrome of newborn
S RDS of newborn
S respiratory distress syndrome of newborn
Another observation. . .

Respiratory Distress Syndrome

Concept Status: Current

Descriptions

Definition: Primitive

is_a respiratory condition of fetus OR newborn
is_a perinatal respiratory distress
is_a neonatal respiratory system disorder
  is_a neonatal disorder
    is_a perinatal disorder
      is_a disorder of fetus OR newborn
        is_a disease
          is_a disorder of respiratory system
An ontological error . . .

Respiratory Distress Syndrome

Concept Status: Current

Descriptions

Definition: Primitive

is_a respiratory condition of fetus OR newborn
is_a perinatal respiratory distress
is_a neonatal respiratory system disorder
is_a neonatal disorder
is_a perinatal disorder
is_a disorder of fetus OR newborn
is_a disease
is_a disorder of respiratory system
Perinatal and neonatal . . .
An easy correction . . .

Respiratory Distress Syndrome

Concept Status: Current

Descriptions

Definition: Primitive

is_a respiratory condition of fetus OR newborn

is_a perinatal respiratory distress

is_a perinatal respiratory system disorder

is_a perinatal disorder

is_a disorder of fetus OR newborn

is_a disease

is_a disorder of respiratory system
Another easy correction . . .

Respiratory Distress Syndrome

Concept Status: Current

Descriptions

Definition: Primitive

- is_a respiratory condition of fetus OR newborn
- is_a perinatal respiratory distress
- is_a neonatal respiratory system disorder
  - is_a neonatal disorder
    - is_a disorder of fetus OR newborn
      - is_a disease
    - is_a disorder of respiratory system
The root anomaly . . .

Neonatal disorder
Concept Status: Current

Descriptions
F neonatal disorder (disorder)
P neonatal disorder
S neonatal disease

Definition: Fully defined by . . .
  is_a
    is_a perinatal disorder
      is_a disorder of fetus OR newborn
        is_a disease
          is_a clinical finding
            is_a SNOMED CT concept

occurs in:
  neonatal
    early neonatal period
    late neonatal period
Neonatal may not be perinatal

Birth

-7d

Perinatal

7d

Neonatal

28d
Perinatal may not be neonatal

Birth

-7d

7d

28d

Perinatal

Neonatal
The definitive correction . . .

Respiratory Distress Syndrome

Concept Status: Current

Descriptions

Definition: Primitive

is_a_respiratory_distress_of_fetus_OR_newborn

is_a_perinatal_respiratory_system_disorder

is_a_perinatal_disorder

is_a_disorder_of_fetus_OR_newborn

is_a_disease

is_a Disorder_of_respiratory_system

is_a_neonatal_respiratory_system_disorder

is_a_neonatal_disorder

is_a_disorder_of_fetus_OR_newborn

is_a_disease

is_a Disorder_of_respiratory_system
Conclusion

SNOMED CT provides structured representation based on description logic for the majority of this set of terms that are used for disorders of the newborn infant.

An increase in the number of fully defined SNOMED concepts is necessary to enhance subsumption, auto-classification and other computational processes.

An increase in the number of SNOMED concepts with one or more synonyms should enhance the interface functionality of SNOMED CT.
Future directions . . .

Future activities should focus on increasing the number of fully defined SNOMED concepts that represent disorders of the newborn infant, and the number of synonyms for those concepts.

Ontological errors need identification and correction.
Contact information . . .

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