ICARE
Interagency Collaborative to Advance Research in Epilepsy
NIH Campus, Bethesda, MD
April 14, 2016

Ontologies: Applications to Epilepsy

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Lister Hill National Center for Biomedical Communications
Bethesda, Maryland - USA
Outline

- Why do we need ontologies?
- Epilepsy in standard clinical ontologies
  - ICD-10
  - SNOMED CT
  - LOINC
  - RxNorm
- Beyond standard ontologies
Why do we need ontologies?
London Bills of Mortality

A general Bill for this present year, ending the 19 of December 1665, according to the report made to the King's most Excellent Majesty, by the Company of Parish Clerks of London, &c.

<table>
<thead>
<tr>
<th>Disease and Casualties this year</th>
<th>Male</th>
<th>Female</th>
</tr>
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<tbody>
<tr>
<td>A</td>
<td>612</td>
<td>30</td>
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<td>L</td>
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<tr>
<td>M</td>
<td>669</td>
<td>65</td>
</tr>
<tr>
<td>N</td>
<td>668</td>
<td>65</td>
</tr>
</tbody>
</table>

Locality: Mailed

Number of the Plague: 65

In the Bills in the 132 Parishes and at the Pest-House this year: 9,520

Increased of the Plague in the 132 Parishes and at the Pest-House this year: 9,520
To support epidemiology

- **John Graunt (1620-1674)**
  - Analyzes the vital statistics of the citizens of London

- **William Farr (1807-1883)**
  - Medical statistician
  - Improves Cullen’s classification
  - Contributes to creating ICD

- **Jacques Berthillon (1851-1922)**
  - Chief of the statistical services (Paris)
  - Classification of causes of death (161 rubrics)
“The advantages of a uniform statistical nomenclature, however imperfect, are so obvious, that it is surprising no attention has been paid to its enforcement in Bills of Mortality. Each disease has, in many instances, been denoted by three or four terms, and each term has been applied to as many different diseases: vague, inconvenient names have been employed, or complications have been registered instead of primary diseases. The nomenclature is of as much importance in this department of inquiry as weights and measures in the physical sciences, and should be settled without delay.”

– William Farr

First annual report.
From “bad air” to “bad water” (John Snow)
Epilepsy in standard clinical ontologies
Revision (ICD-10)-WHO Version for 2016

Chapter VI
Diseases of the nervous system
(G00-G99)

Episodic and paroxysmal disorders
(G40-G47)

G40  Epilepsy

Excl.: Landau-Kleffner syndrome (F60.3)
seizure (convulsive) NOS (R56.8)
status epilepticus (G41.-)
Todd paralysis (G83.8)

G40.0  Localization-related (focal)(partial) idiopathic epilepsy and epileptic syndromes with seizures of localized onset
Benign childhood epilepsy with centrotemporal EEG spikes
Childhood epilepsy with occipital EEG paroxysms

G40.1  Localization-related (focal)(partial) symptomatic epilepsy and epileptic syndromes with simple partial seizures
Attacks without alteration of consciousness
Simple partial seizures developing into secondarily generalized seizures

G40.2  Localization-related (focal)(partial) symptomatic epilepsy and epileptic syndromes with complex partial seizures
Attacks with alteration of consciousness, often with automatisms
Complex partial seizures developing into secondarily generalized seizures

G40.3  Generalized idiopathic epilepsy and epileptic syndromes
Benign:
- myoclonic epilepsy in infancy
- neonatal convulsions (familial)
Childhood absence epilepsy [petit mal]
Epilepsy with grand mal seizures on awakening
Juvenile:
- absence epilepsy
- myoclonic epilepsy [petit mal]
Nonspecific epileptic seizures:
- atonic
- clonic
- myoclonic
- tonic
- tonic-clonic

G40.4  Localization-related (focal)(partial) symptomatic epilepsy and epileptic syndromes
Benign:
- myoclonic epilepsy in infancy
- neonatal convulsions (familial)
Childhood absence epilepsy [petit mal]
Epilepsy with grand mal seizures on awakening
Juvenile:
- absence epilepsy
- myoclonic epilepsy [petit mal]
Nonspecific epileptic seizures:
- atonic
- clonic
- myoclonic
- tonic
- tonic-clonic
Internal Classification of Diseases

G40.4 Other generalized epilepsy and epileptic syndromes
Epilepsy with:
- myoclonic absences
- myoclonic-astatic seizures
Infantile spasms
Lennox-Gastaut syndrome
Sokol syndrome
Symptomatic early myoclonic encephalopathy
West syndrome

G40.5 Special epileptic syndromes
Epilepsia partialis continua [Kozhevnikov]
Epileptic seizures related to:
- alcohol
- drugs
- hormonal changes
- sleep deprivation
- stress

Use additional external cause code (Chapter XX), if desired, to identify drug, if drug-induced.

G40.6 Grand mal seizures, unspecified (with or without petit mal)
G40.7 Petit mal, unspecified, without grand mal seizures
G40.8 Other epilepsy
Epilepsies and epileptic syndromes undetermined as to whether they are focal or generalized

G40.9 Epilepsy, unspecified
Epileptic:
- convulsions NOS
- fits NOS
- seizures NOS

G41 Status epilepticus
G41.0 Grand mal status epilepticus
Tonic-clonic status epilepticus
Excl.: epilepsy partialis continua [Kozhevnikov] (G40.5)
G41.1 Petit mal status epilepticus
Epileptic absence status
G41.2 Complex partial status epilepticus
G41.8 Other status epilepticus
G41.9 Status epilepticus, unspecified
<table>
<thead>
<tr>
<th>ICD-10</th>
<th>ICD-10-CM</th>
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</thead>
<tbody>
<tr>
<td>G40 Epilepsy (10 codes)</td>
<td>G40 Epilepsy and recurrent seizures (77 codes)</td>
</tr>
<tr>
<td>G41 Status epilepticus (5)</td>
<td>no G41</td>
</tr>
</tbody>
</table>

**G40.4 Other generalized epilepsy and epileptic syndromes**

- Epilepsy with:
  - myoclonic absences
  - myoclonic-astatic seizures
- Infantile spasms
- Lennox-Gastaut syndrome
- Salaam attacks
- Symptomatic early West syndrome

<table>
<thead>
<tr>
<th>Code</th>
<th>Description</th>
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<tr>
<td>G40.811</td>
<td>Lennox-Gastaut syndrome, not intractable, with status epilepticus</td>
</tr>
<tr>
<td>G40.812</td>
<td>Lennox-Gastaut syndrome, not intractable, without status epilepticus</td>
</tr>
<tr>
<td>G40.813</td>
<td>Lennox-Gastaut syndrome, intractable, with status epilepticus</td>
</tr>
<tr>
<td>G40.814</td>
<td>Lennox-Gastaut syndrome, intractable, without status epilepticus</td>
</tr>
</tbody>
</table>

**Specific code(s) for**

- Rare forms
- Tractability
SNOMED CT

211 descendants

SNOMED CT

Parents
- Seizure disorder (disorder)

Finding site → Cerebral structure
Has definitional manifestation → Seizure

Epilepsy (disorder)
SCTID: 84757009
84757009 | Epilepsy (disorder)
- Epilepsy
- Epileptic fits
- Epileptic seizures
- Epileptic attack
- Epileptic disorder
- Epileptic convulsions
- Epileptic
- EP - Epilepsy
- Epilepsy (disorder)

A disorder characterized by recurrent seizures

Children (23)
- Atonic epilepsy (disorder)
- Atypical absence epilepsy (disorder)
- Benign neonatal convulsions (disorder)
- Contre-cerebral epilepsy (disorder)
- Cursory seizure (disorder)
- Drug-induced epilepsy (disorder)
- Epilepsy in mother complicating childbirth (disorder)
- Epilepsy in mother complicating pregnancy (disorder)
- Epilepsy undetermined whether focal or generalized (disorder)
- Epilepsy, not refractory (disorder)
- Generalized epilepsy (disorder)
- Kohlschutter's syndrome (disorder)
- Localization-related epilepsy (disorder)
- Post-cerebrovascular accident epilepsy (disorder)
- Post-traumatic epilepsy (disorder)
- Psychosensory epilepsy (disorder)
- Reflex epilepsy (disorder)
- Refractory epilepsy (disorder)
- Somatosensory epilepsy (disorder)
- Status epilepticus (disorder)
- Tonic-clonic epilepsy (disorder)
- Visceral epilepsy (disorder)
- Visual epilepsy (disorder)
SNOMED CT

Parents

- Seizure related finding (finding)

Children (31)

- Abdominal seizure (finding)
- Afebrile seizure (finding)
- Akinetic seizure without atonia (finding)
- Alcohol-related fit (finding)
- Anoxic seizure (finding)
- Brief atomic seizure (finding)
- Central convulsion (finding)
- Childhood seizure (finding)
- Drug withdrawal seizure (finding)
- Dysnemic seizure (disorder)
- Eclamptic seizure (finding)
- Epileptic cry (finding)
- More...

Finding site → Structure of nervous system

59 descendants
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<th>Component</th>
<th>Property</th>
<th>Timing</th>
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<th>Method</th>
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<tr>
<td>45424-3</td>
<td>Epilepsy [Minimum Data Set]</td>
<td>Epilepsy</td>
<td>Find</td>
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<td>39060-7</td>
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<td>EPM2A gene mutation analysis</td>
<td>Pnd</td>
<td>Pt</td>
<td>Bid/Tiss</td>
<td>Nom</td>
<td>Moigen</td>
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<td>65263-7</td>
<td>Have you ever had, or has anyone ever told you that you had, a seizure disorder or epilepsy [PhenX]</td>
<td>Have you ever had, or has anyone ever told you that you had, a seizure disorder or epilepsy</td>
<td>Find</td>
<td>Pt</td>
<td>*Patient</td>
<td>Ord</td>
<td>PhenX</td>
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<td>62763-8</td>
<td>PhenX - epilepsy screener protocol</td>
<td>PhenX - epilepsy screener protocol</td>
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<td>*Patient</td>
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<td>39494-4</td>
<td>VA Compensation and Pension (C and P) examination epilepsy/narcolepsy</td>
<td>VA C&amp;P exam.epilepsy &amp; or narcolepsy note</td>
<td>Find</td>
<td>Pt</td>
<td>(Setting)</td>
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<td>ADNFE gene mutation analysis</td>
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<td>34490-3</td>
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<td>MT-TK gene mutation analysis</td>
<td>Pnd</td>
<td>Pt</td>
<td>Bid/Tiss</td>
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<td>41062-9</td>
<td>MT-TK gene mutations found [Identifier] in Blood or Tissue by Molecular genetics method Nominal</td>
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<td>Bid/Tiss</td>
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<td>Moigen</td>
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<td>21713-3</td>
<td>MT-TK gene m.8344A&gt;G [Presence] in Blood or Tissue by Molecular genetics method</td>
<td>MT-TK gene m.8344A&gt;G</td>
<td>Pr</td>
<td>Pt</td>
<td>Bid/Tiss</td>
<td>Ord</td>
<td>Moigen</td>
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<td>49831-5</td>
<td>ATN1 gene allele 1.CAG repeats [Entitic number] in Blood or Tissue by Molecular genetics method</td>
<td>ATN1 gene allele 1.CAG repeats</td>
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<td>49032-3</td>
<td>ATN1 gene allele 2.CAG repeats [Entitic number] in Blood or Tissue by Molecular genetics method</td>
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<td>On</td>
<td>Moigen</td>
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<td>21758-2</td>
<td>ATN1 gene CAG repeats [Presence] in Blood or Tissue by Molecular genetics method</td>
<td>ATN1 gene CAG repeats</td>
<td>Threshold</td>
<td>Pt</td>
<td>Bid/Tiss</td>
<td>Ord</td>
<td>Moigen</td>
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<td>MT-TK gene m.8296A&gt;G</td>
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<td>55427-9</td>
<td>Seizure disorder tracking panel</td>
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<td>86386-4</td>
<td>Has the child had a seizure or a brain problem [PhenX]</td>
<td>Has the child had a seizure or a brain problem</td>
<td>Find</td>
<td>Pt</td>
<td>Patient</td>
<td>Ord</td>
<td>PhenX</td>
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<td>86387-6</td>
<td>Have you ever had, or has anyone ever told you that you had, a seizure, convolution, fit or spell under any circumstances [PhenX]</td>
<td>Have you ever had, or has anyone ever told you that you had, a seizure, convolution, fit or spell under any circumstances</td>
<td>Find</td>
<td>Pt</td>
<td>Patient</td>
<td>Ord</td>
<td>PhenX</td>
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<td>70339-7</td>
<td>I am afraid of having a seizure - convolution - in the past 7 days [FACIT]</td>
<td>I am afraid of having a seizure - convolution - in the past 7D</td>
<td>Find</td>
<td>7D</td>
<td>Patient</td>
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<td>86385-9</td>
<td>Did anyone ever tell you that you had a seizure or convolution caused by a high fever when you were a child [PhenX]</td>
<td>Did anyone ever tell you that you had a seizure or convolution caused by a high fever when you were a child</td>
<td>Find</td>
<td>Pt</td>
<td>Patient</td>
<td>Ord</td>
<td>PhenX</td>
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<tr>
<td>86386-7</td>
<td>Have you ever had, or has anyone ever told you that you had, a seizure disorder or epilepsy [PhenX]</td>
<td>Have you ever had, or has anyone ever told you that you had, a seizure disorder or epilepsy</td>
<td>Find</td>
<td>Pt</td>
<td>Patient</td>
<td>Ord</td>
<td>PhenX</td>
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<tr>
<td>86387-8</td>
<td>Have you had a seizure, brain, or other nervous system problem [PhenX]</td>
<td>Have you had a seizure, brain, or other nervous system problem</td>
<td>Find</td>
<td>Pt</td>
<td>Patient</td>
<td>Ord</td>
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<td>32472-3</td>
<td>History of Seizure activity disorders</td>
<td>History of symptoms &amp; diseases</td>
<td>Find</td>
<td>Pt</td>
<td>Procedure</td>
<td>Cerebral cortex</td>
<td>Qn</td>
<td>EEG</td>
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<td>50847-1</td>
<td>Seizure count Cerebral cortex Electroencephalogram (EEG)</td>
<td>Seizure count</td>
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<td>Patient</td>
<td>Ord</td>
<td>MDS</td>
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<td>45852-4</td>
<td>Seizure disorder [Minimum Data Set]</td>
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<td>Ord</td>
<td>MDS</td>
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<td>74153-3</td>
<td>Seizure Disorder action plan</td>
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<td>Patient</td>
<td>Ord</td>
<td>MDS</td>
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<td>54814-9</td>
<td>Seizure disorder in last 7 days [MDG3]</td>
<td>Seizure disorder in last 7D</td>
<td>Find</td>
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<td>Ord</td>
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<td>Seizures per month</td>
<td>Seizures per month</td>
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<td>Ord</td>
<td>MDG3</td>
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<tr>
<td>83822-7</td>
<td>When you stopped, cut down, or went without drinking, did you ever have fits, seizures, or convulsions, where you lost consciousness, fell to the floor, and had difficulty remembering what happened [PhenX]</td>
<td>When you stopped, cut down, or went without drinking, did you ever have fits, seizures, or convulsions, where you lost consciousness, fell to the floor, and had difficulty remembering what happened</td>
<td>Find</td>
<td>Pt</td>
<td>Patient</td>
<td>Ord</td>
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</table>
65365-9  Did anyone ever tell you that you had a seizure or convulsion caused by a high fever when you were a child [PhenX]

<table>
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<tr>
<th>NAME</th>
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<tbody>
<tr>
<td>Fully-Specified Name: Did anyone ever tell you that you had a seizure or convulsion caused by a high fever when you were a child</td>
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<th>BASIC ATTRIBUTES</th>
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<tr>
<td>Class/Type: PHENX/Clinical</td>
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<tr>
<td>Created On: 2011/04/29</td>
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<tr>
<td>Last Updated in Version: 2.44</td>
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<td>Status: Trial – caution, may change</td>
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<td>Answer</td>
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<td>1</td>
<td>Yes</td>
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<tr>
<td>2</td>
<td>No</td>
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<tr>
<td>3</td>
<td>Possible</td>
</tr>
<tr>
<td>4</td>
<td>Don't know</td>
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<th>SURVEY QUESTION</th>
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<tr>
<td>Text: Did anyone ever tell you that you had a seizure or convulsion caused by a high fever when you were a child?</td>
</tr>
<tr>
<td>Source: PhenX.130401010000</td>
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Generated from LOINC version 2.54.
# 55427-9  Seizure disorder tracking panel

## Panel Hierarchy

<table>
<thead>
<tr>
<th>LOINC#</th>
<th>LOINC Name</th>
<th>R/O/C</th>
<th>Cardinality</th>
<th>Ex. UCUM Units</th>
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<td>Seizure disorder tracking panel</td>
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<td></td>
</tr>
<tr>
<td>55414-7</td>
<td>Seizures per month</td>
<td>O</td>
<td></td>
<td>/mo</td>
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<tr>
<td>3432-2</td>
<td>Carbamazepine [Mass/volume] in Serum or Plasma</td>
<td>C</td>
<td></td>
<td>ug/mL</td>
</tr>
<tr>
<td>3487-6</td>
<td>Clobazam [Mass/volume] in Serum or Plasma</td>
<td>C</td>
<td></td>
<td>ug/mL</td>
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<td>Clonazepam [Mass/volume] in Serum or Plasma</td>
<td>C</td>
<td></td>
<td>ug/mL</td>
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<td>3616-0</td>
<td>Ethosuximide [Mass/volume] in Serum or Plasma</td>
<td>C</td>
<td></td>
<td>ug/mL</td>
</tr>
<tr>
<td>6899-9</td>
<td>Felbamate [Mass/volume] in Serum or Plasma</td>
<td>C</td>
<td></td>
<td>ug/mL</td>
</tr>
<tr>
<td>9738-6</td>
<td>Gabapentin [Mass/volume] in Serum or Plasma</td>
<td>C</td>
<td></td>
<td>ug/mL</td>
</tr>
<tr>
<td>6948-4</td>
<td>Lamotrigine [Mass/volume] in Serum or Plasma</td>
<td>C</td>
<td></td>
<td>ug/mL</td>
</tr>
<tr>
<td>30471-7</td>
<td>Levetiracetam [Mass/volume] in Serum or Plasma</td>
<td>C</td>
<td></td>
<td>ug/mL</td>
</tr>
<tr>
<td>35331-8</td>
<td>Oxcarbazepine [Mass/volume] in Serum or Plasma</td>
<td>C</td>
<td></td>
<td>ug/mL</td>
</tr>
<tr>
<td>3948-7</td>
<td>Phenobarbital [Mass/volume] in Serum or Plasma</td>
<td>C</td>
<td></td>
<td>ug/mL</td>
</tr>
<tr>
<td>2968-5</td>
<td>Phenytoin [Mass/volume] in Serum or Plasma</td>
<td>C</td>
<td></td>
<td>ug/mL</td>
</tr>
<tr>
<td>47414-8</td>
<td>Pregabalin [Mass/volume] in Serum or Plasma</td>
<td>C</td>
<td></td>
<td>ug/mL</td>
</tr>
<tr>
<td>3978-4</td>
<td>Primidone [Mass/volume] in Serum or Plasma</td>
<td>C</td>
<td></td>
<td>ug/mL</td>
</tr>
<tr>
<td>21565-7</td>
<td>Tiagabine [Mass/volume] in Serum or Plasma</td>
<td>C</td>
<td></td>
<td>ug/mL</td>
</tr>
<tr>
<td>1713-9</td>
<td>Topiramate [Mass/volume] in Serum or Plasma</td>
<td>C</td>
<td></td>
<td>ug/mL</td>
</tr>
<tr>
<td>4086-5</td>
<td>Valproate [Mass/volume] in Serum or Plasma</td>
<td>C</td>
<td></td>
<td>ug/mL</td>
</tr>
<tr>
<td>30042-6</td>
<td>Vigabatrin [Mass/volume] in Serum or Plasma</td>
<td>C</td>
<td></td>
<td>ug/mL</td>
</tr>
<tr>
<td>29620-2</td>
<td>Zonisamide [Mass/volume] in Serum or Plasma</td>
<td>C</td>
<td></td>
<td>ug/mL</td>
</tr>
</tbody>
</table>
## RxNorm / RxClass

### RxNorm Name | RXCUI  | Status  
--- | --- | ---  
Carbamazepine | 2002 | Both  
nesicarbazepine | 1482502 | Both  
Ethosuximide | 4135 | Both  
Ethotoin | 4136 | Both  
ezogabine | 1112990 | Both  
felbamate | 24812 | Both  
gabapentin | 25480 | Both  
lacosamide | 623400 | Both  
lamotrigine | 28439 | Both  
Levetiracetam | 114477 | Both  
Mephenytoin | 6757 | Both  
Mephobarbital | 6758 | Both  
methsuximide | 47858 | Both  
oxcarbazepine | 32624 | Both  
perampanel | 1356552 | Both  
Phenytoin | 8183 | Both  
Primidone | 8691 | Both  
rufinamide | 69036 | Both  
tiagabine | 31914 | Both  
topiramate | 38404 | Both  
Trimethadione | 10827 | Both  
Valproate | 40254 | Both  
Vigabatrin | 14851 | Both  
zonisamide | 39996 | Both  
clobazam | 21241 | Class1 only  
Brivaracetam | 1739745 | Class2 only  
Clonazepam | 2598 | Class2 only  
fosphenytoin | 72236 | Class2 only  
Phenobarbital | 8134 | Class2 only  
pregabalin | 187832 | Class2 only  
sulthiame | 10240 | Class2 only  

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Class 1 Only | Both | Class 2 Only

- **Class 1**: **ANTICONVULSANTS (N0000029145)** in NDFRT (has_VAClass)
- **Class 2**: **ANTIEPILEPTICS (N03A) in ATC**
- # Class 1 Only: 1
- # Class 2 Only: 6
- # Both: 24
- Equivalence Score: 0.76
- Inclusion Score: -0.69
Beyond standard ontologies
Epilepsy ontologies

- Specialized ontologies
  - EpSO, ESSO
  - Finer-grained
  - Specific features (e.g., ILAE classification)
  - Useful for specific research studies/protocols

- But... unlikely to be used in mainstream EHR systems
  - Disconnect between healthcare and research
  - Limits secondary use of EHR data for epilepsy research

- Also consider extending standard ontologies (e.g., SNOMED CT) for better coverage of epilepsy
Medical Ontology Research

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