ESSO

Epilepsy Syndrome and Seizure Ontology

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Goal

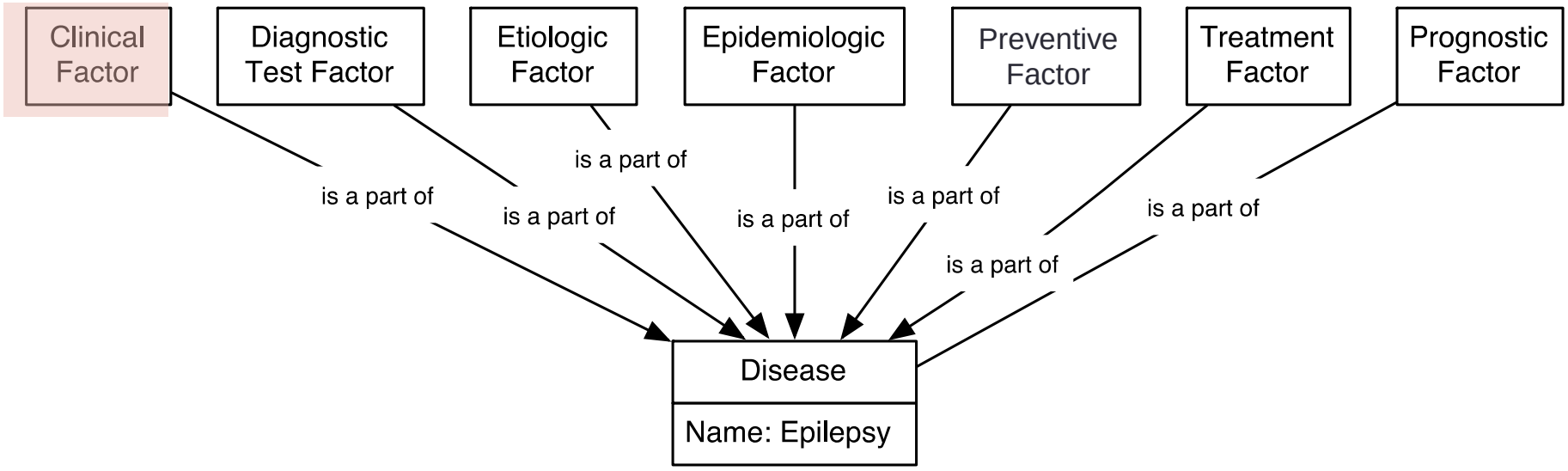
Create an **epilepsy ontology**
that *appeals to* and is *useful to*
practicing physicians and researchers:

A framework to organize knowledge
about epilepsy that is
machine readable and understandable by
domain experts and other users.

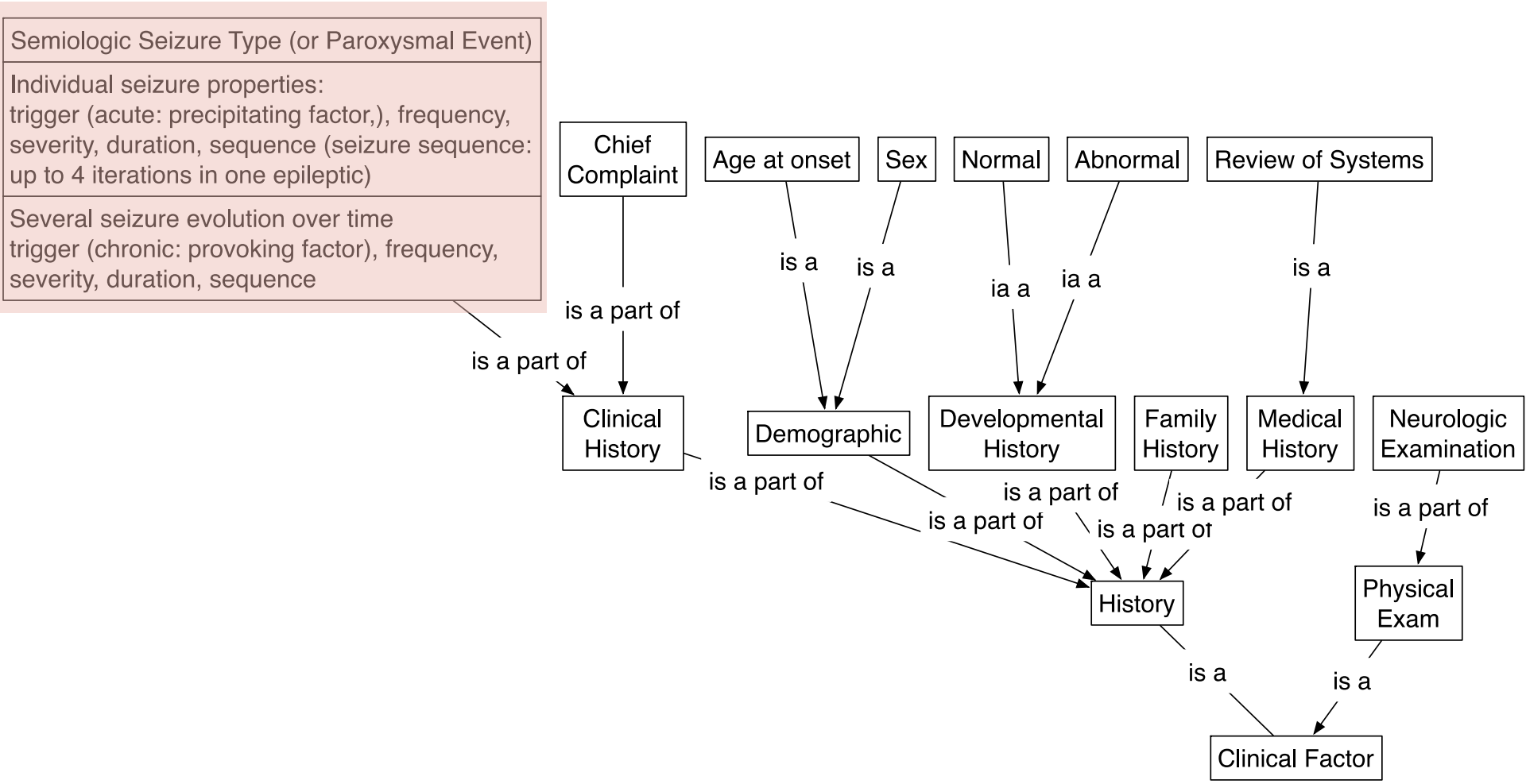
Design principles & approach

- Design Principles
  - What does the domain expert think is important?
  - How would he/she define the different concepts and relationships?
  - What would be the observations that a domain expert make/need?
  - How would he/she want to query the information? (multi-axial)
- Ontology Creation Steps
  1. Define ontology domain and scope
  2. Review existing ontologies
  3. Select upper ontology (based on design principles)
  4. Create classes, properties, relationships
  5. Validate (via expert review / comment)
  6. Revise classes, properties, relationships
  7. Repeat 5 and 6, as needed…
  8. Evaluate (via case studies or other applications)

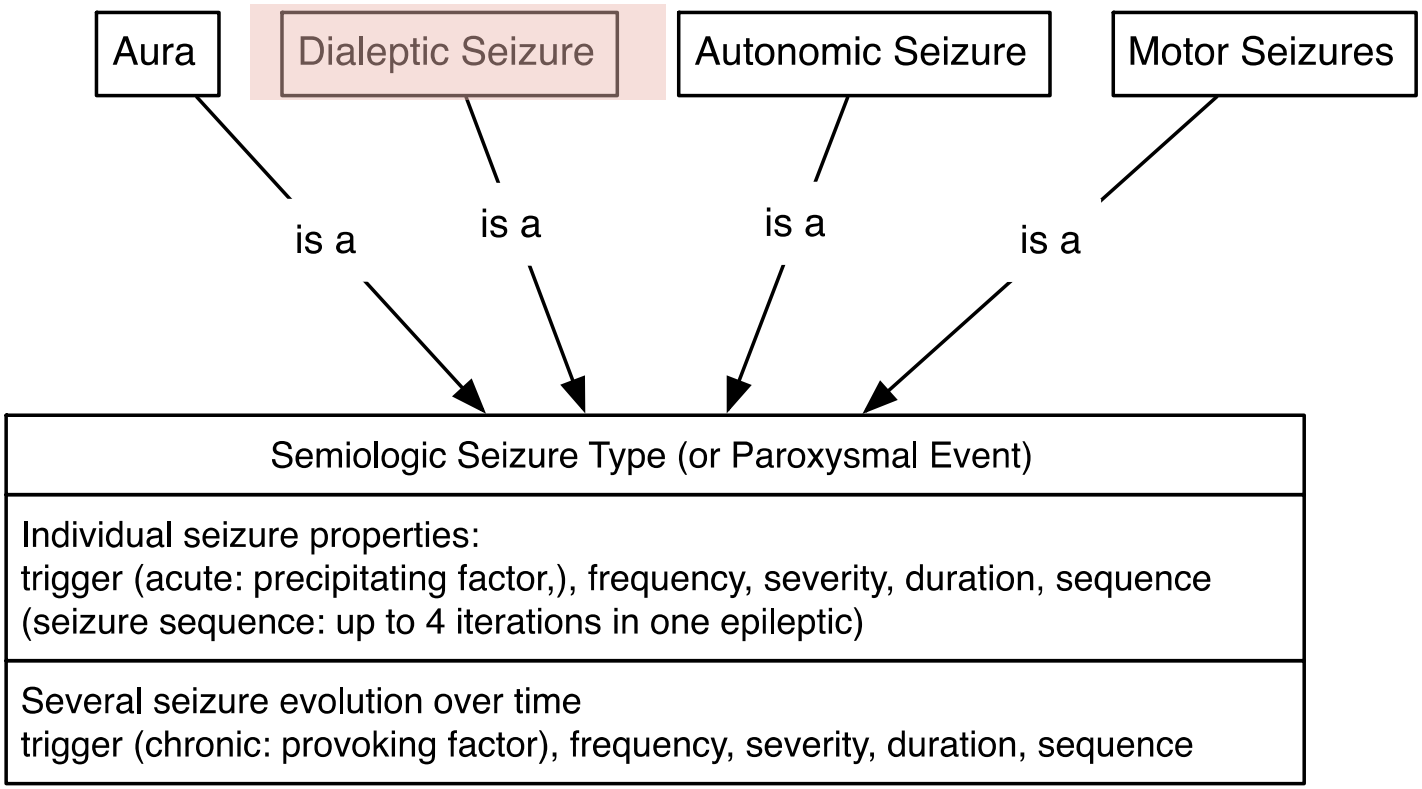
Upper ontology – Epilepsy as Disease

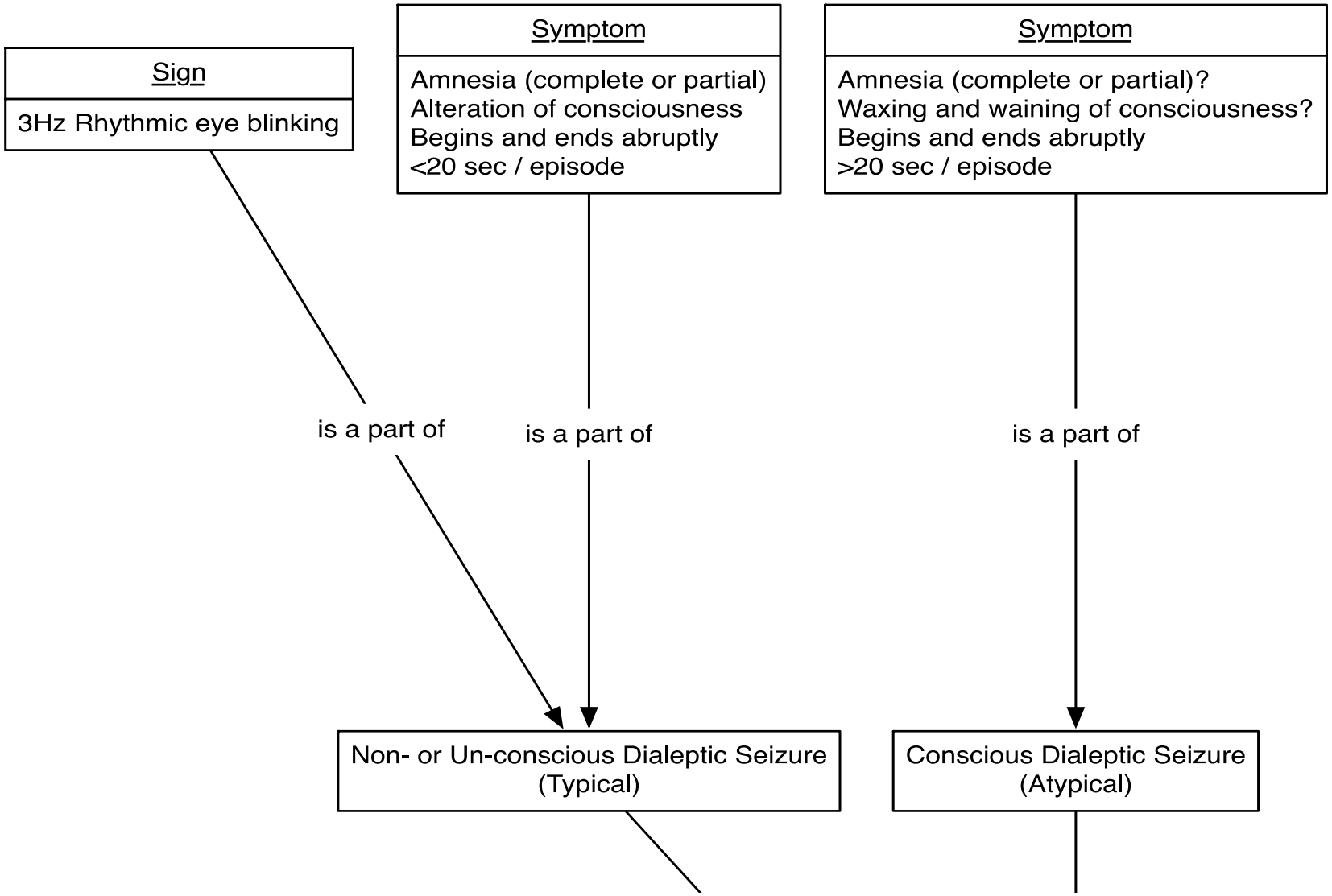


Classes, properties and relationships

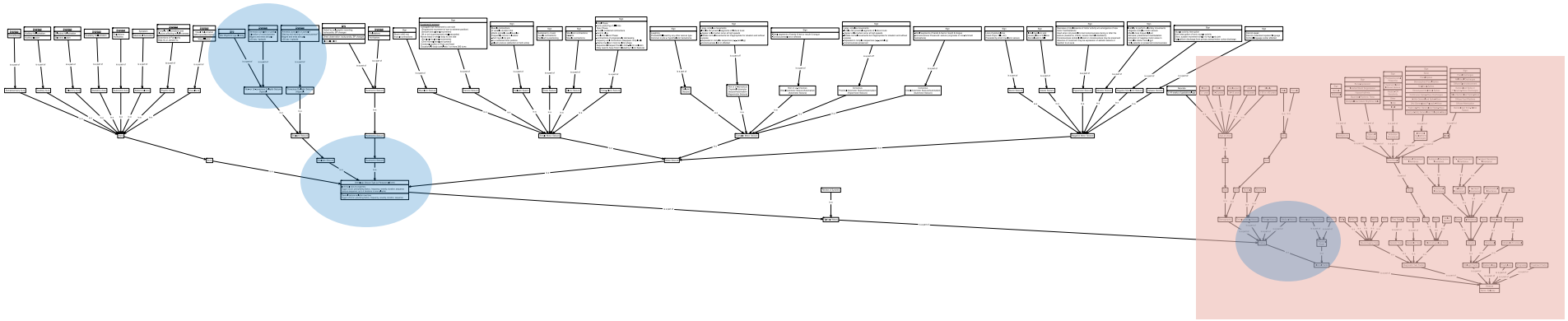


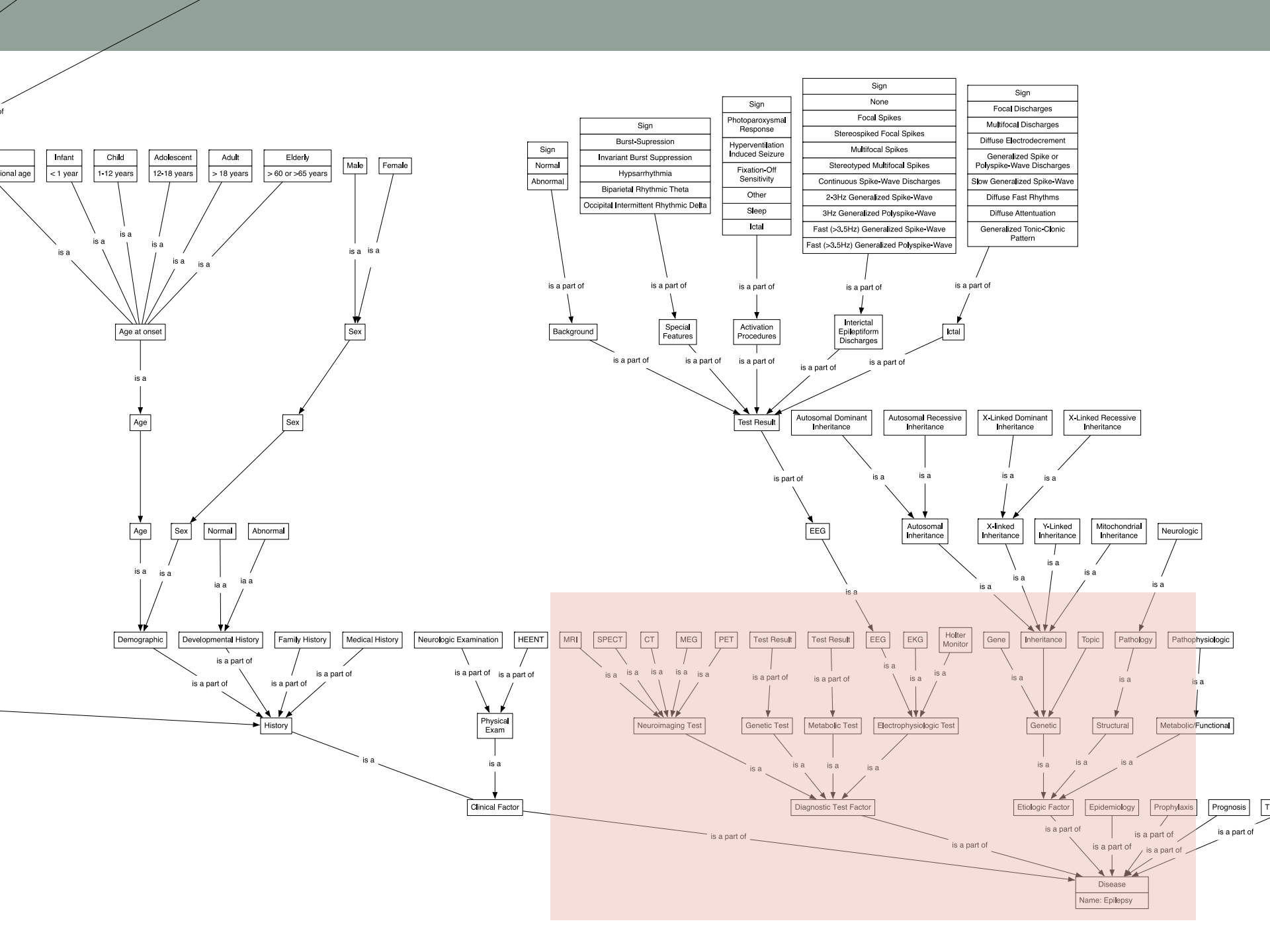
Layered details – finer granularity

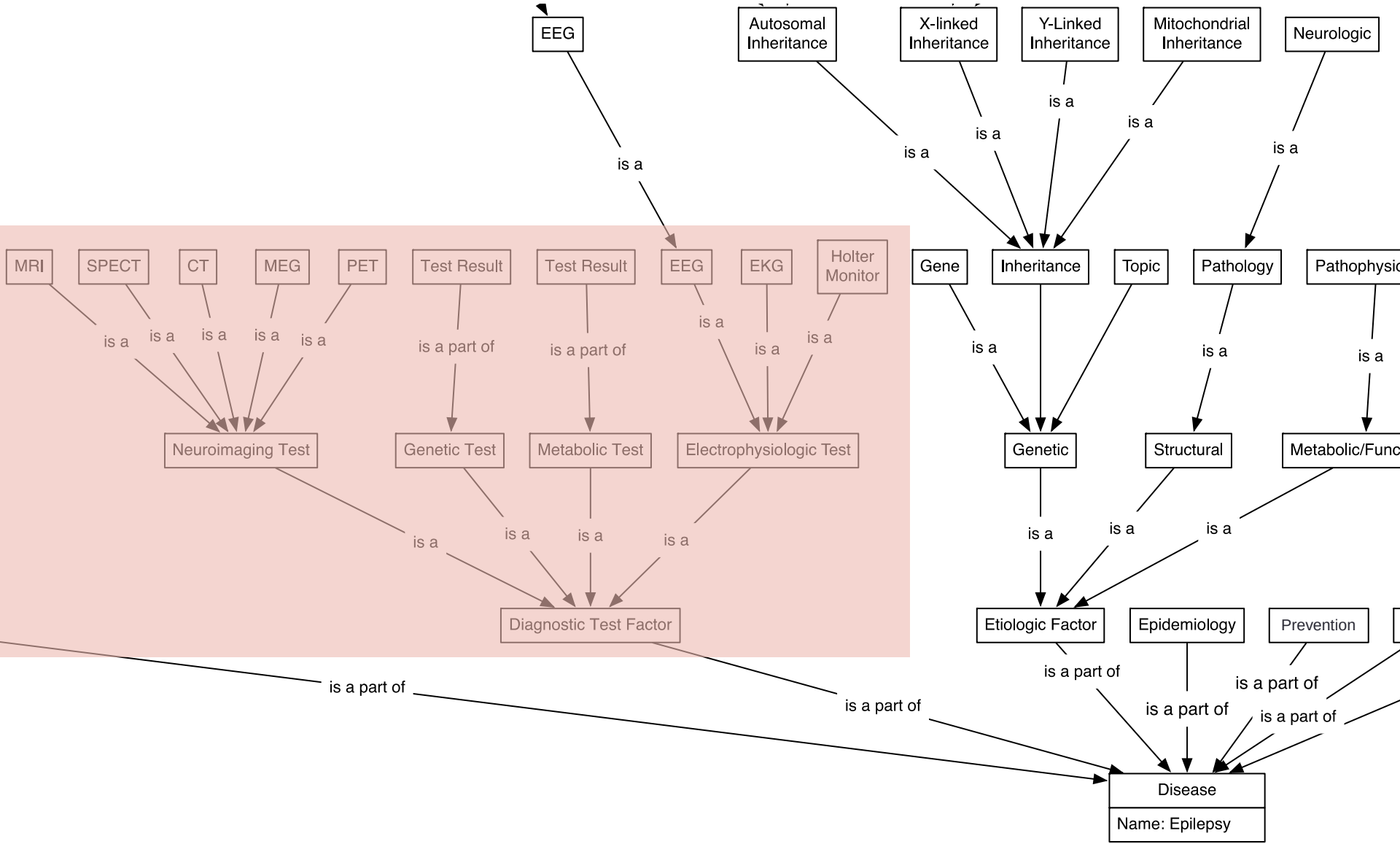



Sign
3Hz Rhythmic eye blinking
Symptom
Amnesia (complete or partial)
Alteration of consciousness
Begins and ends abruptly
<20 sec / episode
Symptom
Amnesia (complete or partial)?
Waxing and waining of consciousness?
Begins and ends abruptly
>20 sec / episode
is a part of
is a part of
is a part of
Non- or Un-conscious Dialeptic Seizure
(Typical)
Conscious Dialeptic Seizure
(Atypical)

WARNING: Do not attempt to read




Infant
< 1 year
Child
1-12 years
Adolescent
12-18 years
Adult
> 18 years
Elderly
> 60 or >65 years
Male
Female
is a
Age at onset
Sex
Age
Sex
Normal
Abnormal
Demographic
Developmental History
Family History
Medical History
Neurologic Examination
HEENT
is a part of
History
Physical Exam
Clinical Factor
Sign
Normal
Abnormal
Sign
Burst-Supression
Invariant Burst Suppression
Hypsarrhythmia
Biparietal Rhythmic Theta
Occipital Intermittent Rhythmic Delta
Sign
Photoparoxysmal Response
Hyperventilation Induced Seizure
Fixation-Off Sensitivity
Other
Sleep
Ictal
Sign
None
Focal Spikes
Stereospiked Focal Spikes
Multifocal Spikes
Stereotyped Multifocal Spikes
Continuous Spike-Wave Discharges
2-3Hz Generalized Spike-Wave
3Hz Generalized Polyspike-Wave
Fast (>3.5Hz) Generalized Spike-Wave
Fast (>3.5Hz) Generalized Polyspike-Wave
Sign
Focal Discharges
Multifocal Discharges
Diffuse Electrodecrement
Generalized Spike or Polyspike-Wave Discharges
Slow Generalized Spike-Wave
Diffuse Fast Rhythms
Diffuse Attentuation
Generalized Tonic-Clonic Pattern
Background
Special Features
Activation Procedures
Interictal Epileptiform Discharges
Ictal
Test Result
Autosomal Dominant Inheritance
Autosomal Recessive Inheritance
X-Linked Dominant Inheritance
X-Linked Recessive Inheritance
EEG
Autosomal Inheritance
X-Inked Inheritance
Y-Linked Inheritance
Mitochondrial Inheritance
Neurologic
MRI
SPECT
CT
MEG
PET
Test Result
Test Result
EEG
EKG
Holter Monitor
Gene
Inheritance
Topic
Pathology
Pathophysiologic
Neuroimaging Test
Genetic Test
Metabolic Test
Electrophysiologic Test
Genetic
Structural
Metabolic/Functional
Diagnostic Test Factor
Etiologic Factor
Epidemiology
Prophylaxis
Prognosis
Disease
Name: Epilepsy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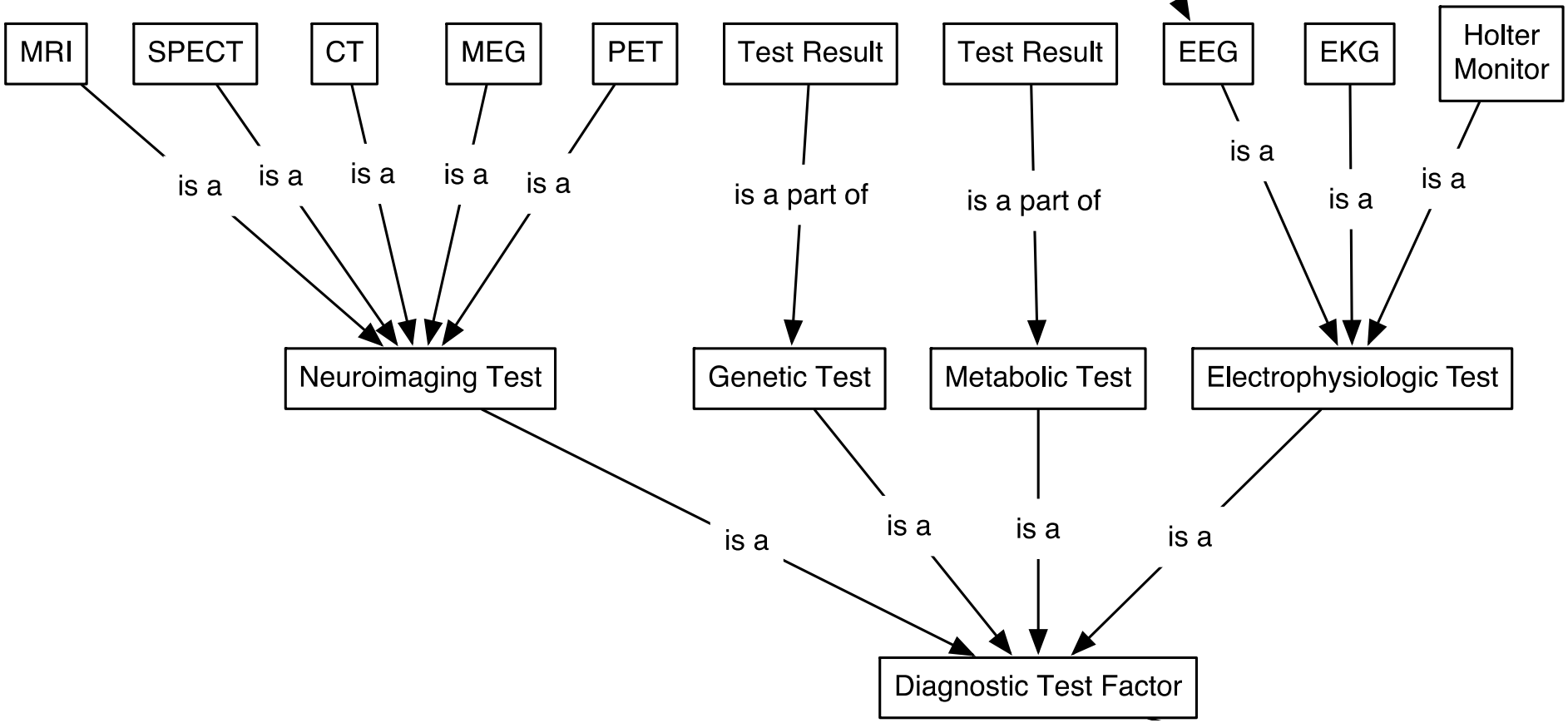
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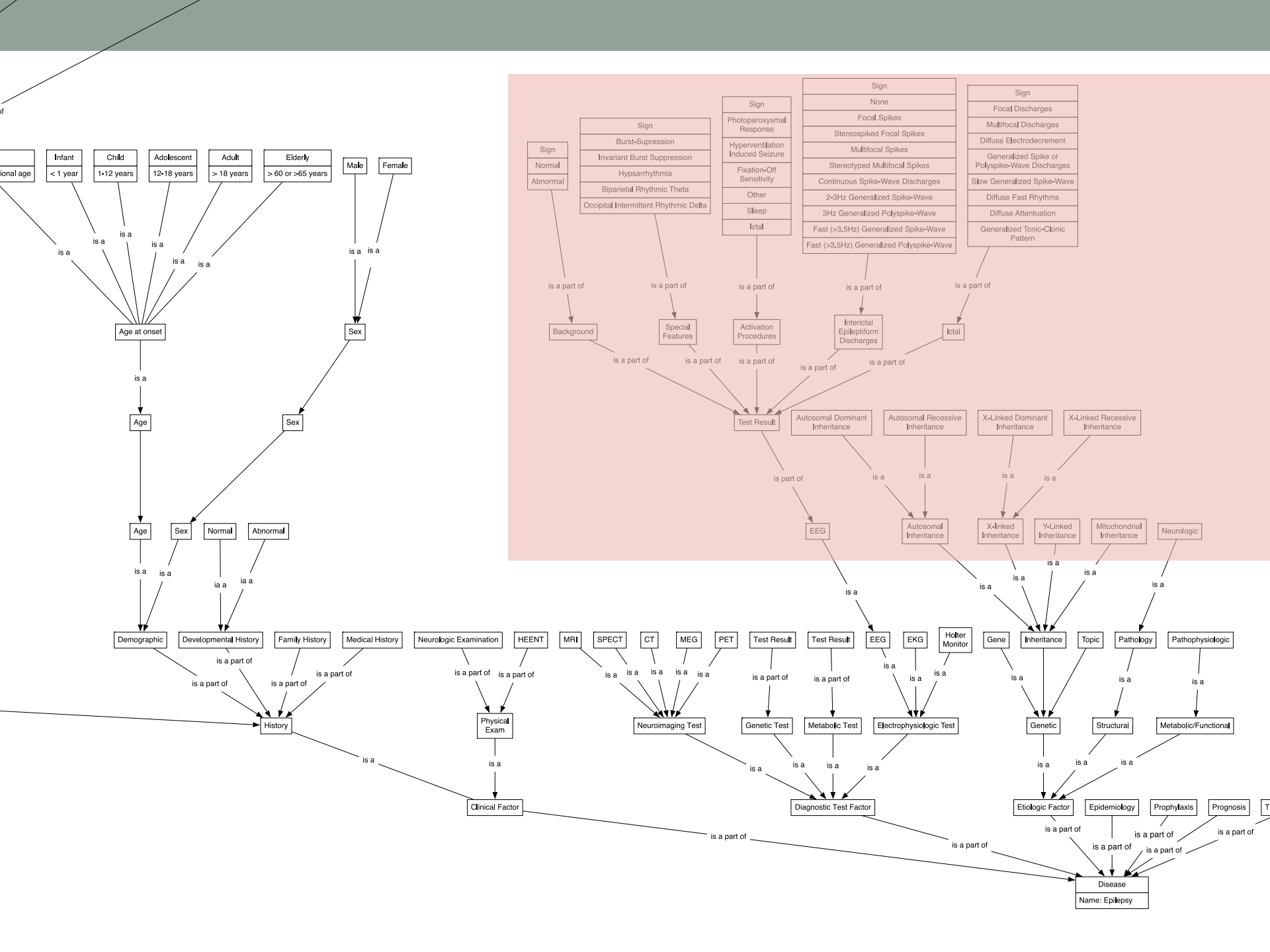
is a
Diagnostic Test Factor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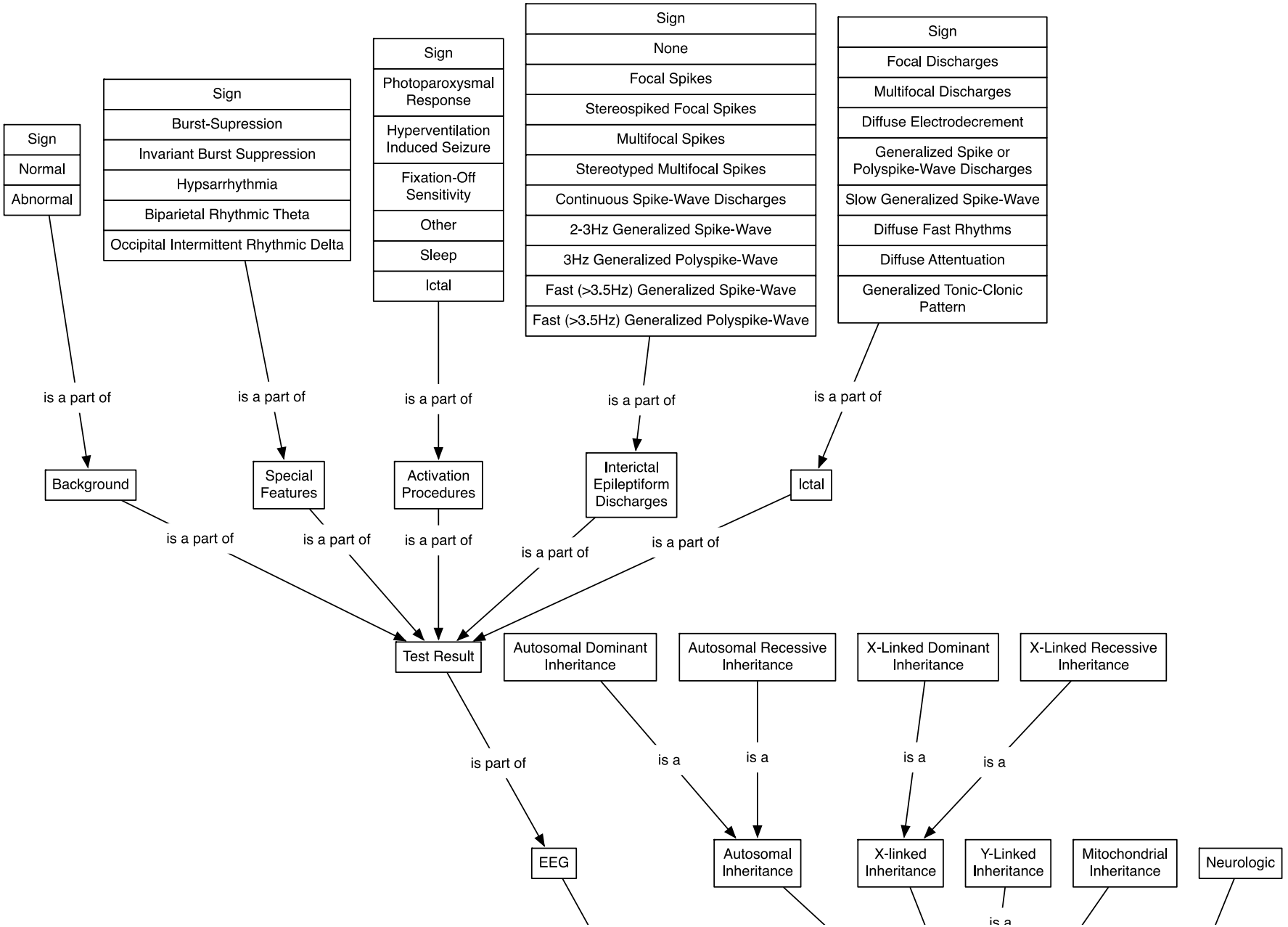
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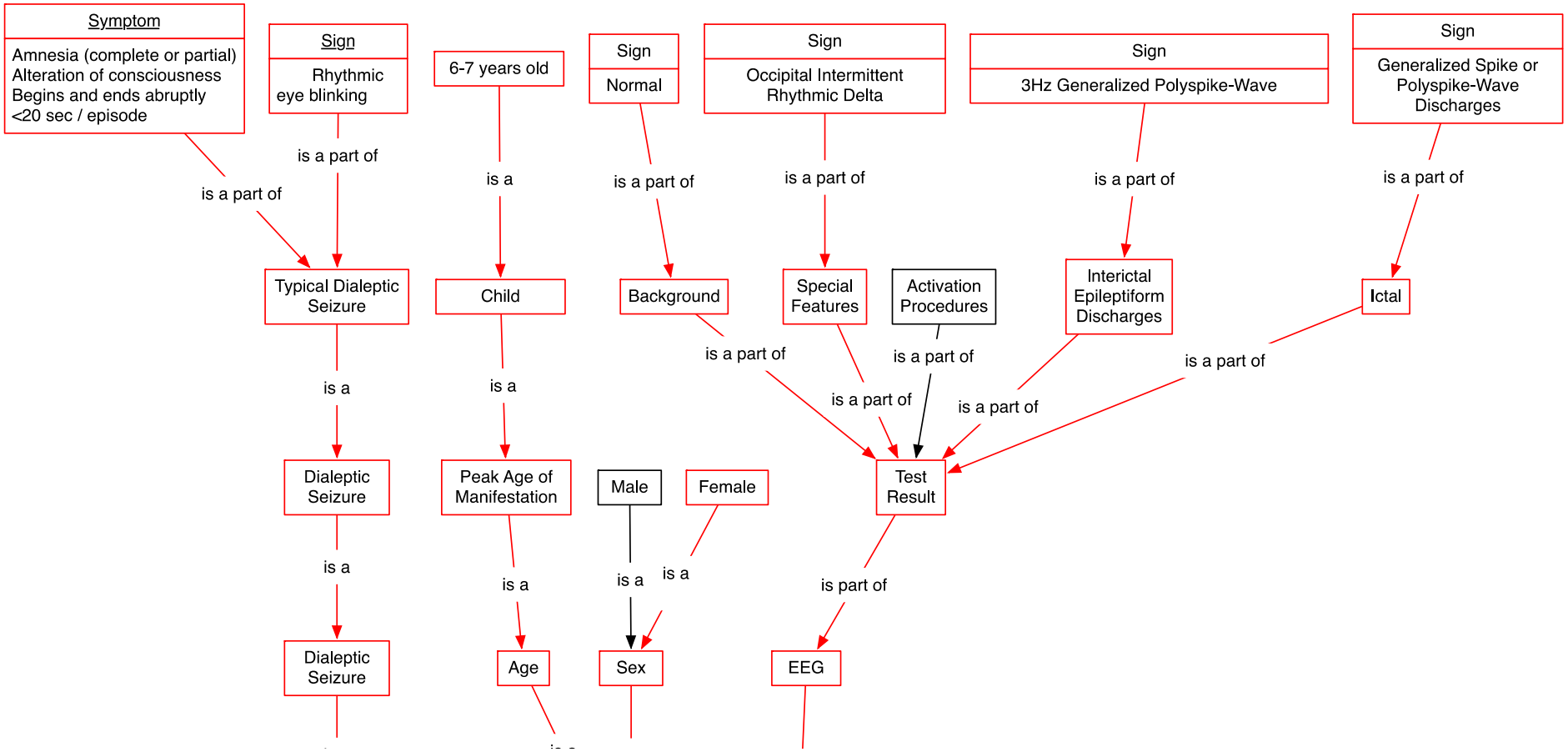
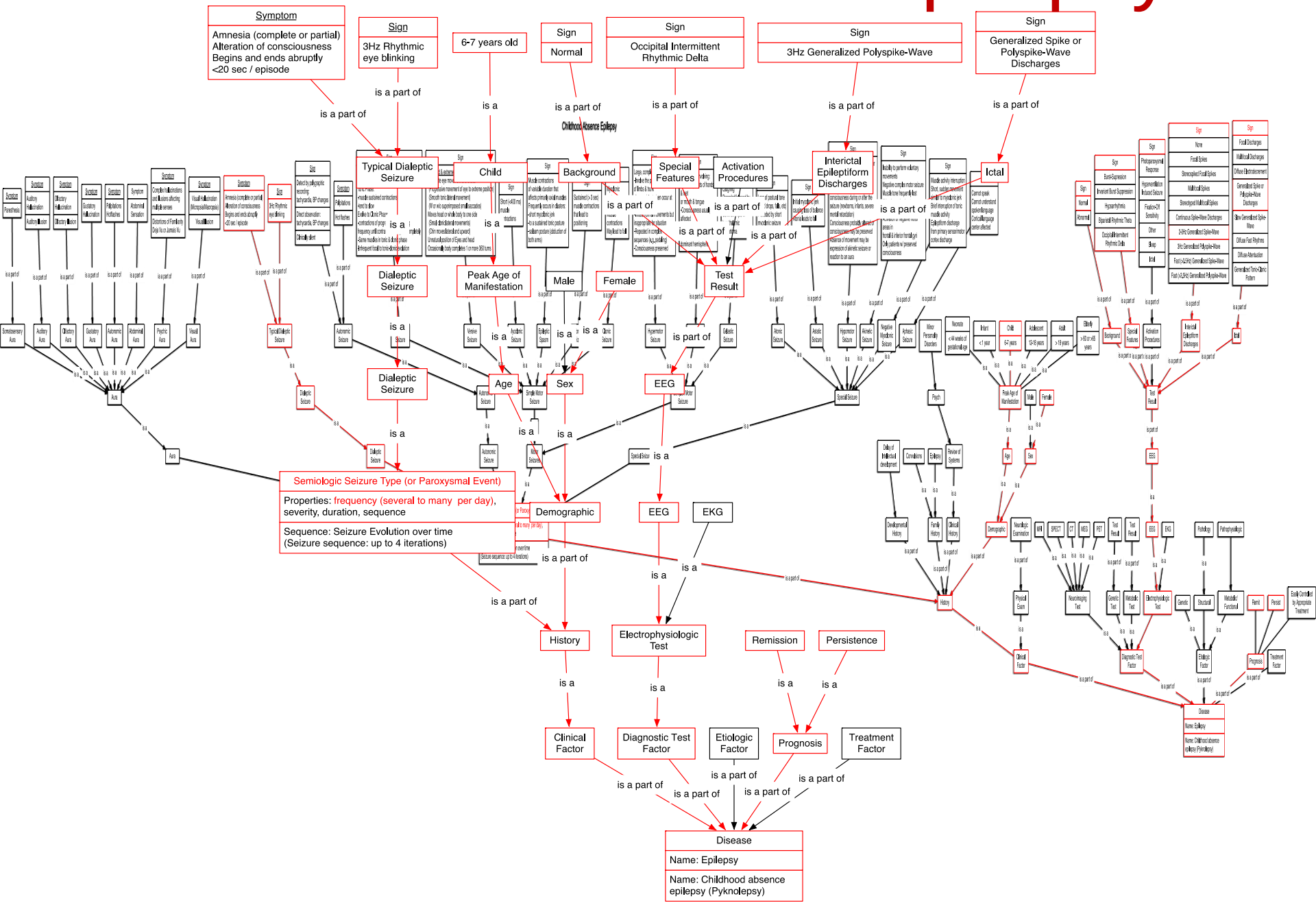
is part of
is a
is a
is a
is a
EEG
Autosomal Inheritance
X-linked Inheritance
Y-Linked Inheritance
Mitochondrial Inheritance
Neurologic
is a

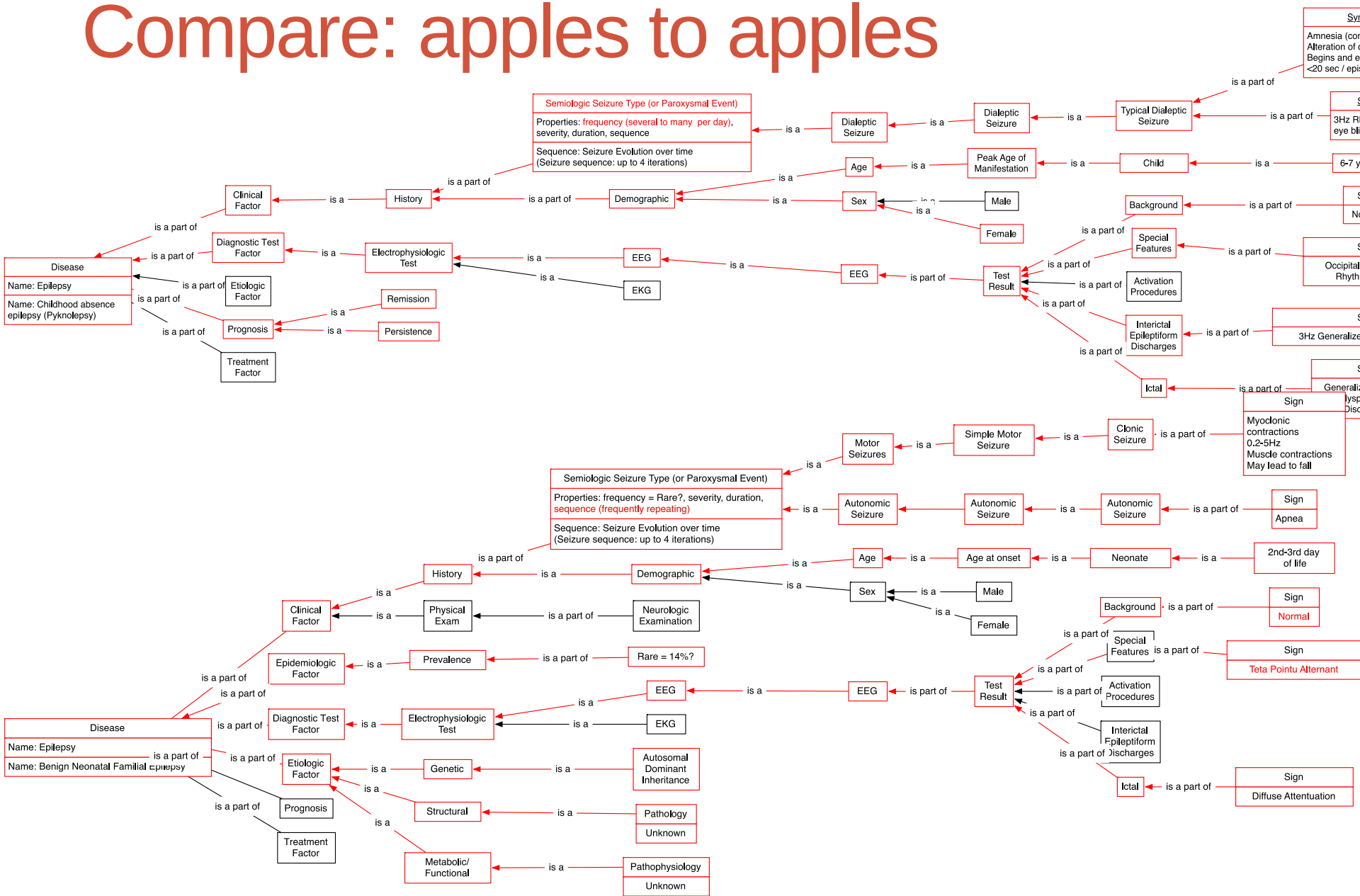
Annotating the ontology – clinical data



Childhood Absence Epilepsy



Compare: apples to apples



Current progress

- **Still in development**
- General framework in place
- Discussion is open for review by experts
- Concepts include references to sources (other past/current classifications, descriptions)
- Extensive validation ongoing
- Openly available in Bioportal (http://bioportal.bioontology.org/ontologies/ESSO)


https://www.google.com/
Epilepsy Syndrome Seizur
Epilepsy Syndrome Seizur
bioportal.bioontology.org/ontologies/ESSO/?p=classes&conceptid=http%3A%2F%2Fwww.semanticweb.org%2Frjyy
Apps
UnoTelly SmartDNS a
Accesos Online
https://www.faceboo
Testing DB
Raynen - QA
Proj Status
DSTRB
Arena
Backlog
BioPortal
Browse
Search
Mappings
Recommender
Annotator
Resource Index
Epilepsy Syndrome Seizure Ontology
Summary
Classes
Properties
Notes
Mappings
Widgets
Jump To:
Clinical_Factor
History
Clinical_History
Paroxysmal_Event
Epileptic_Event
Epileptic_Seizure_Event
Aura
Autonomic_Seizure
Dialeptic_Seizure
Dyscognitive_Seizure
Motor_Seizure
Reflex_Seizure
Status_Epilepticus_Event
Non-Epileptic_Event
Seizure_Factor
Semiology
Action_Myoclonus
Affective_Symptom
Autonomic_Sign
Blank_Stare
Details
Visualization
Notes (0)
Class Mappings (0)
Preferred Name
Clinical_Factor
ID
http://www.semanticweb.org/rjyy/ont
prefLabel
Clinical_Factor
sources
Bancaud 1981 Clinic ILAE 1989 Clinica
of disease perceptible to the examinin
by the patient (symptom). Manifestatio
deviation from clinical normality that e
observable. Observability includes obs
special instruments. Preclinical Manife
prior to its becoming detectable in a cl
Manifestation of a Disease: A manifest
or physical examination. Phenotype =d
organism determined by the interactio
Clinical Phenotype =def. – A clinically
abnormal phenotype that is characteri
being observed. Indeed, as technology
disease phenotype will expand. The fu
phenotypes realized at each stage in th

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Text Classification towards Detecting Misdiagnosis of an Epilepsy Syndrome in a Pediatric Population

Ryan Sullivan, MS,[1] Robert Yao,[1] Randa Jarrar, MD,[2] Jeffrey Buchhalter, MD, PhD,[3] and Graciela Gonzalez, PhD[1]

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Abstract

Go to:

When attempting to identify a specific epilepsy syndrome, physicians are often unable to make or agree upon a diagnosis. This is further complicated by the fact that the current classification and diagnosis of epilepsy requires specialized training and the use of resources not typically available to the average clinician, such as training to recognize specific seizure types and electroencephalography (EEG)[1–4]. Even when training and resources are available, expert epileptologists often find it challenging to identify seizure types and to distinguish between specific epilepsy syndromes[5]. Information relevant to the diagnosis is present in narrative form in the medical record across several visits for an individual patient. Our ultimate goal is to create a system that will assist physicians in the diagnosis of epilepsy. This paper explores, as a baseline, text classification methods that attempt to correlate the narrative text features to the diagnosis of West syndrome (Infantile Spasms), using data from Phoenix Children's Hospital (PCH). We tested these methods against a

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  - Robert Greenes, MD, PhD – Arizona State University
  - Matthew Scotch, PhD – Arizona State University
  - Joseph Sirven, MD – Mayo Clinic Arizona