



Freiburg

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Ontologias Biológicas para Endemias

XX CONGRESSO BRASILEIRO DE PARASITOLOGIA
Avanços e Desafios
Recife 1 / 11 / 07




Recife

O termo “Ontologias”

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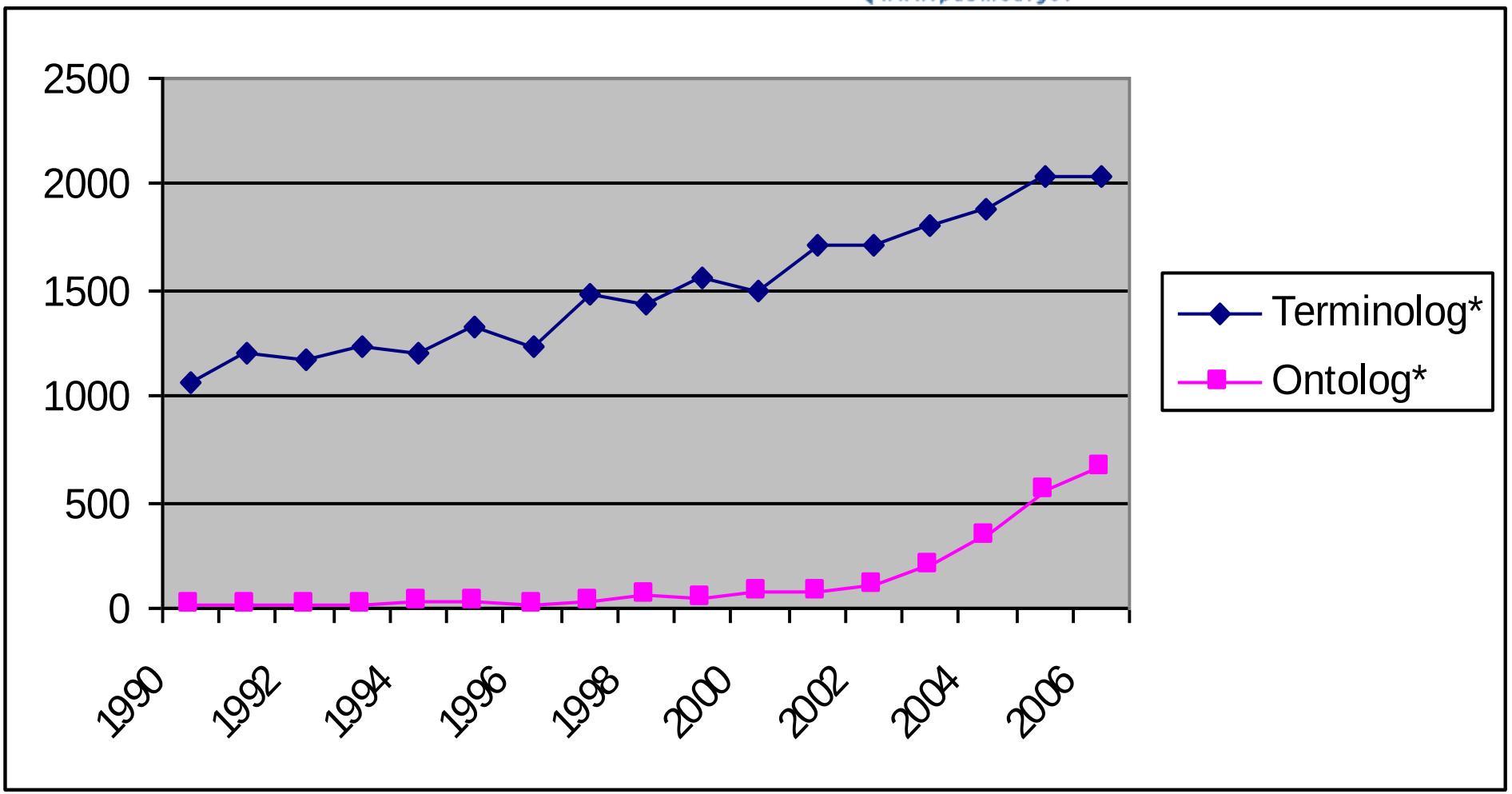
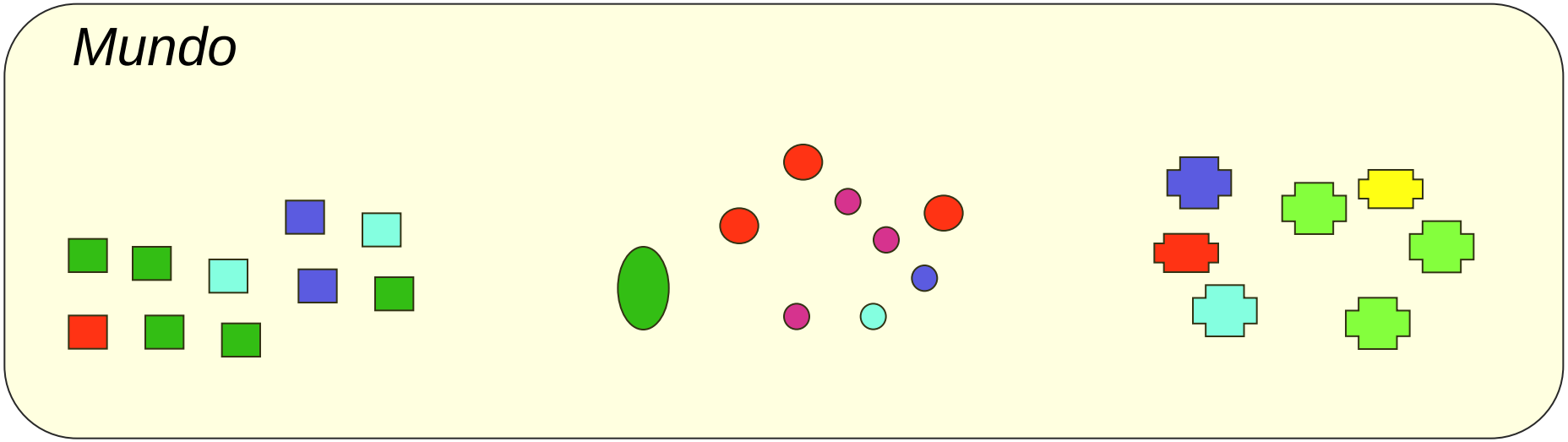


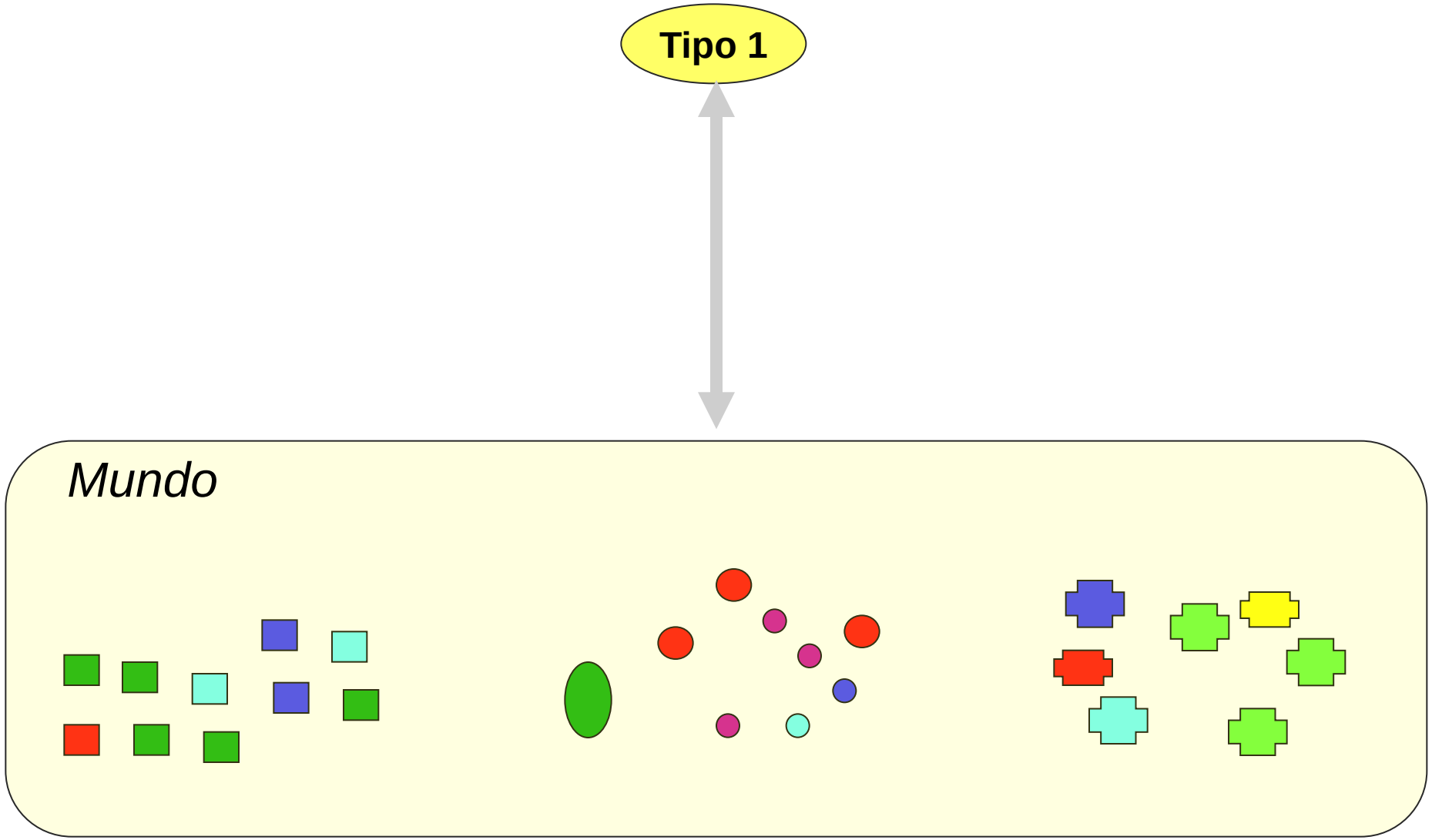
Ontologia - Definições

- Ontologia é o estudo do que existe
- Ontologias formais são teorias as quais buscam dar precisas formulações matemáticas das propriedades e relações de certas entidades. (Stanford Encyclopedia of Philosophy)
- Ontologia: sistema de organizar, descrever, e classificar as entidades (objetos, processos, etc.) de um domínio

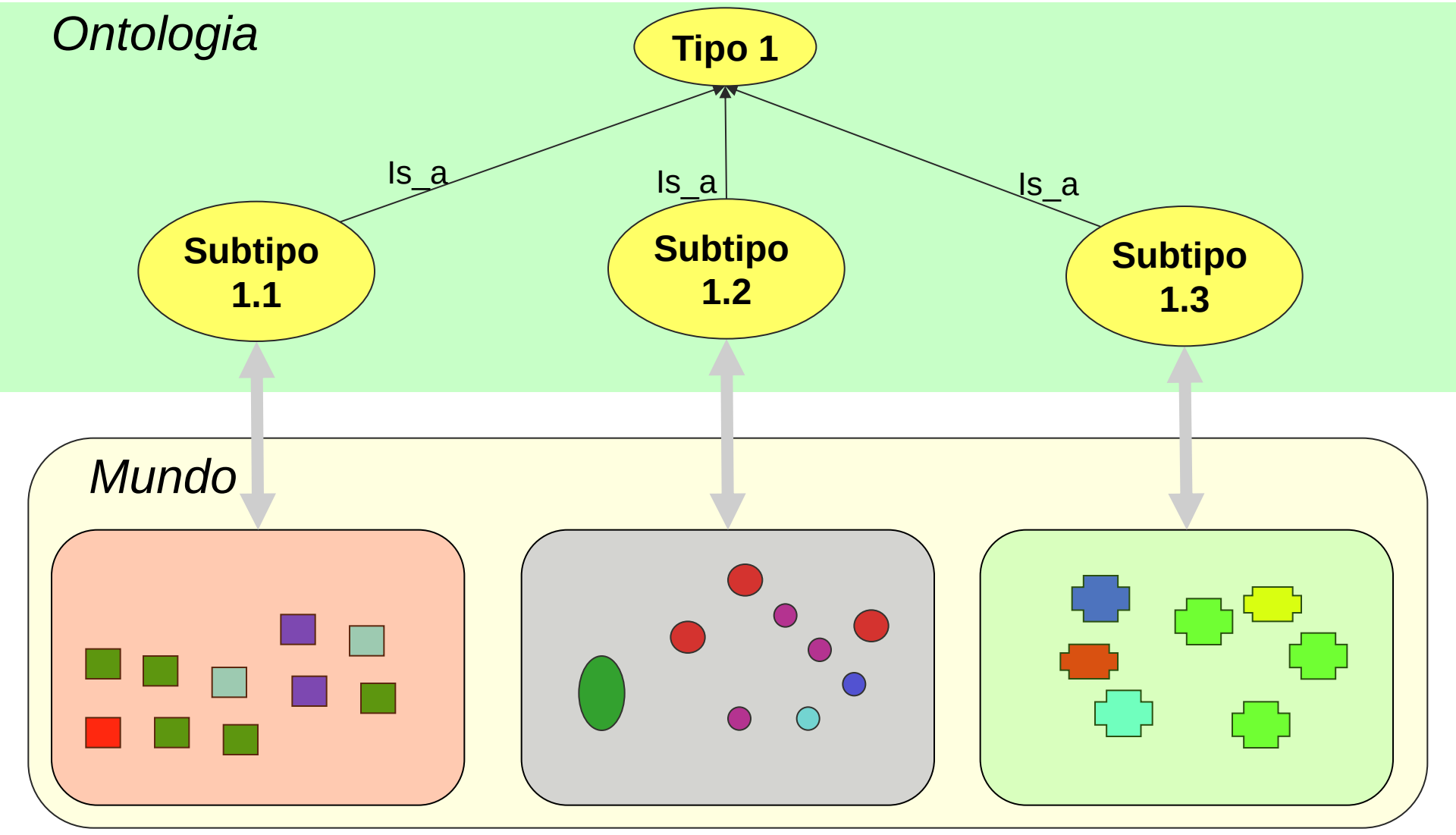
Hierarquias, Tipos, Classes, Indivíduos



Hierarquias, Tipos, Classes, Indivíduos

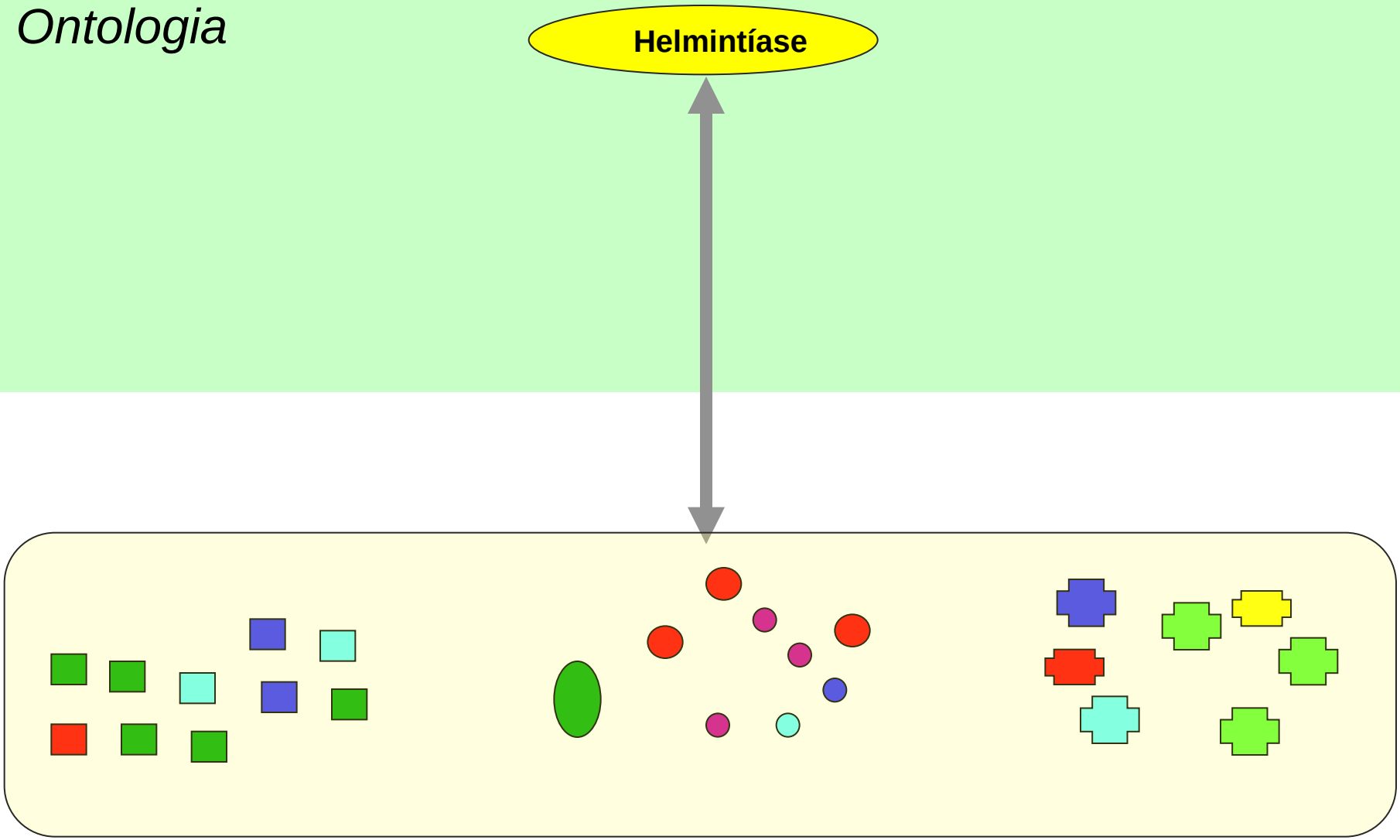


Hierarquias, Tipos, Classes, Indivíduos

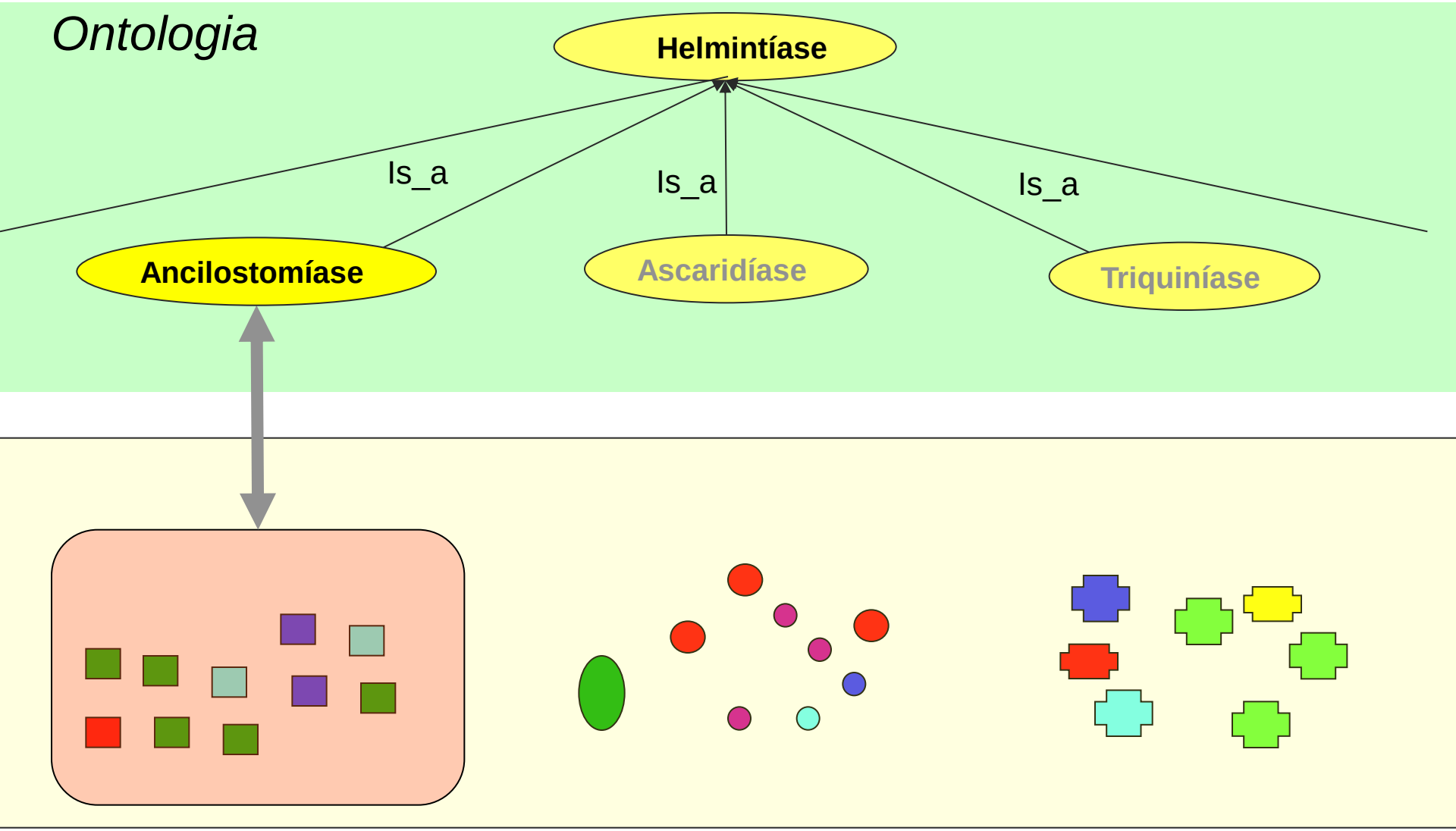


Hierarquias, Tipos, Classes, Indivíduos

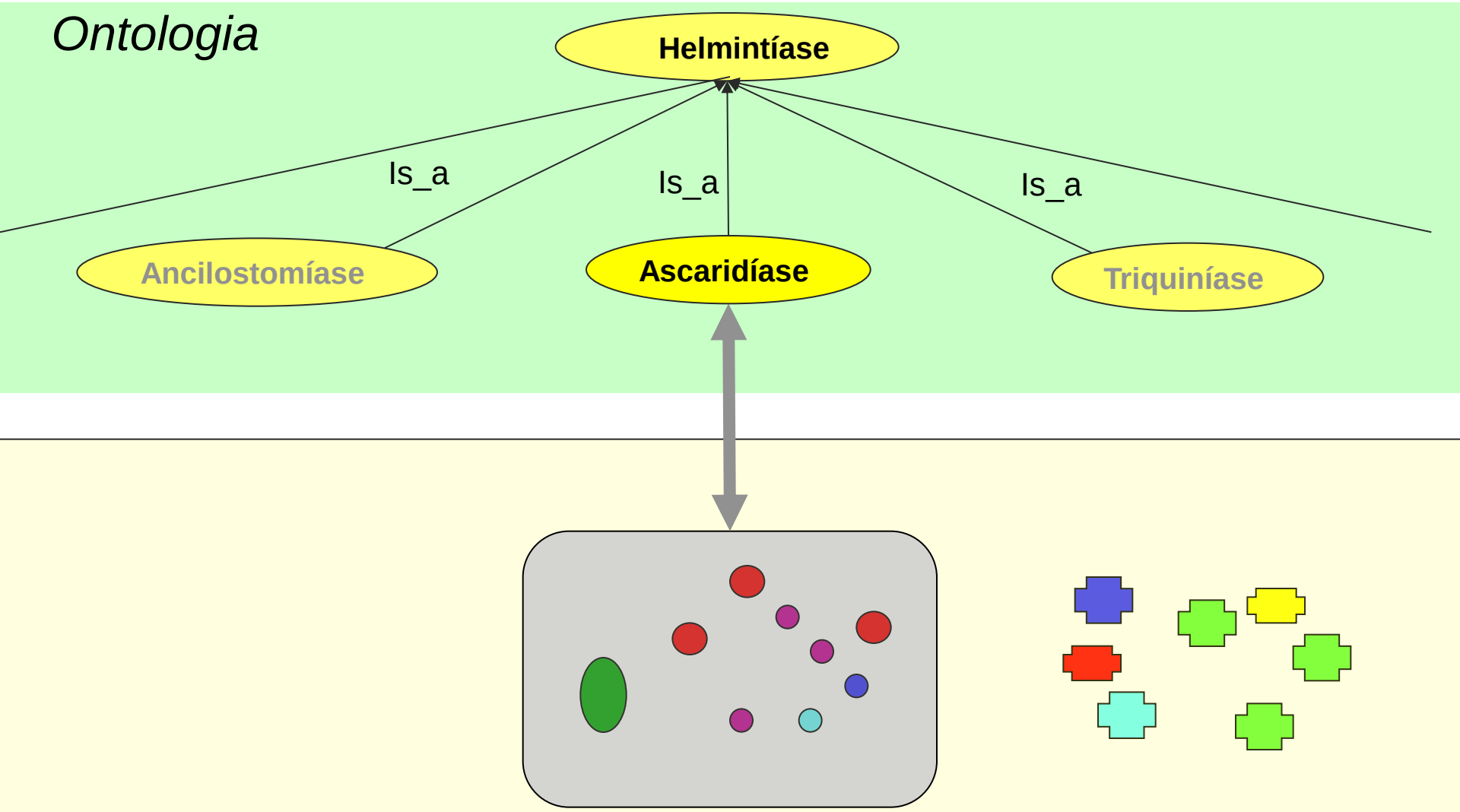
Ontologia



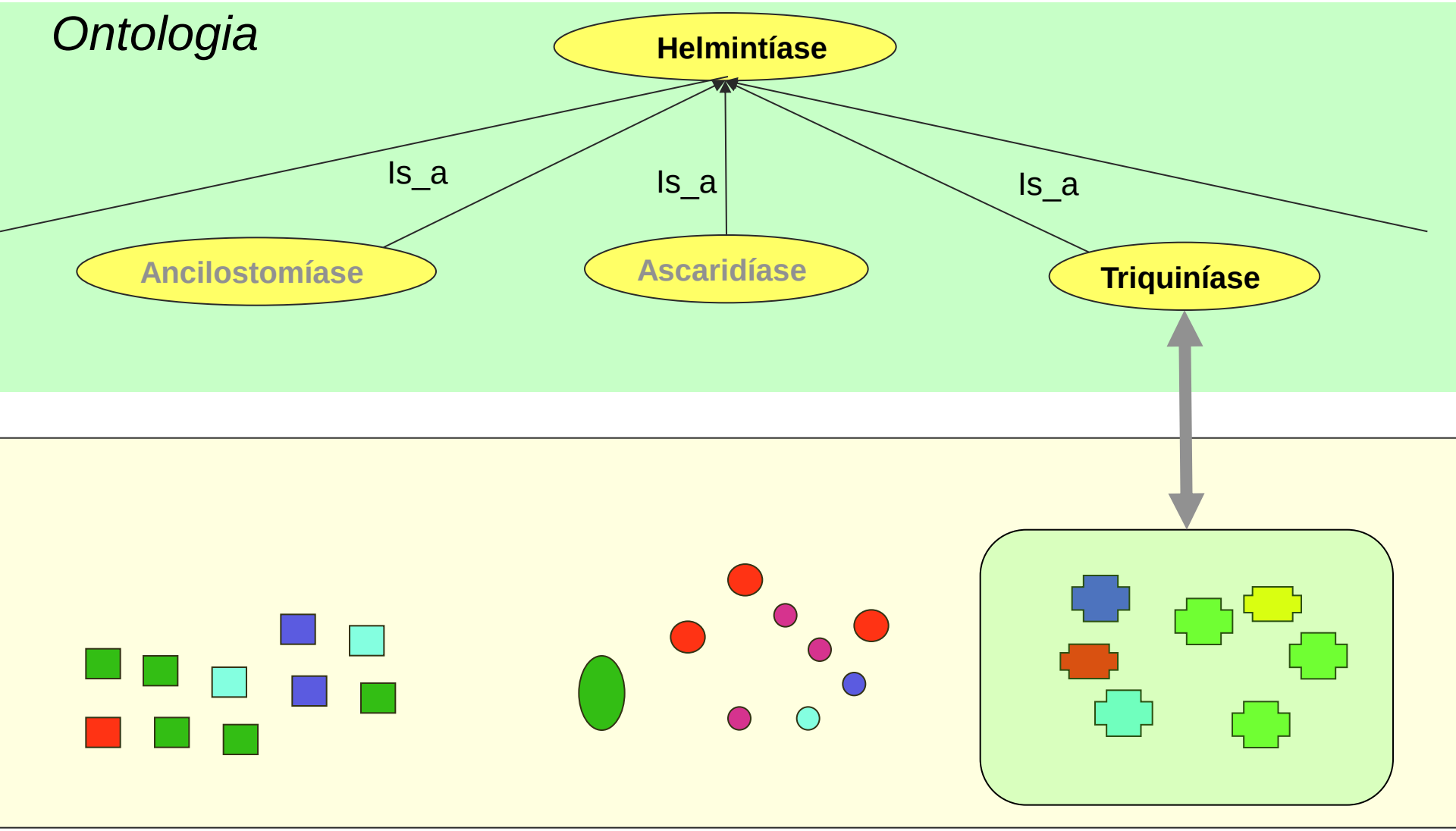
Hierarquias, Tipos, Classes, Indivíduos



Hierarquias, Tipos, Classes, Indivíduos

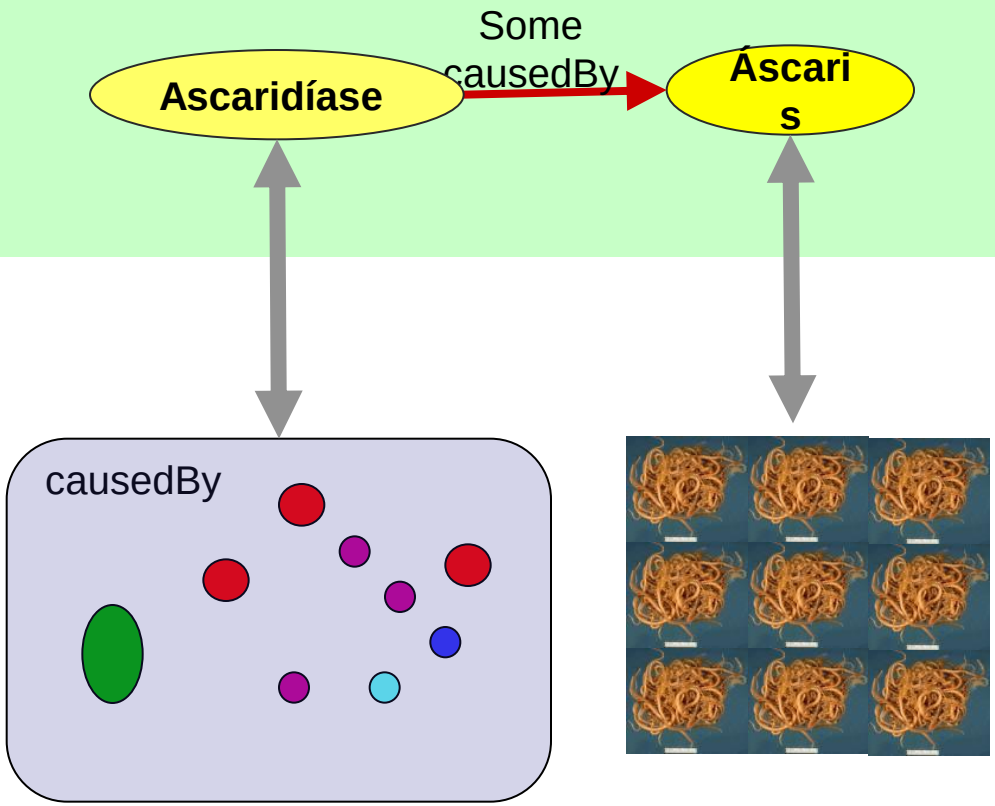


Hierarquias, Tipos, Classes, Indivíduos



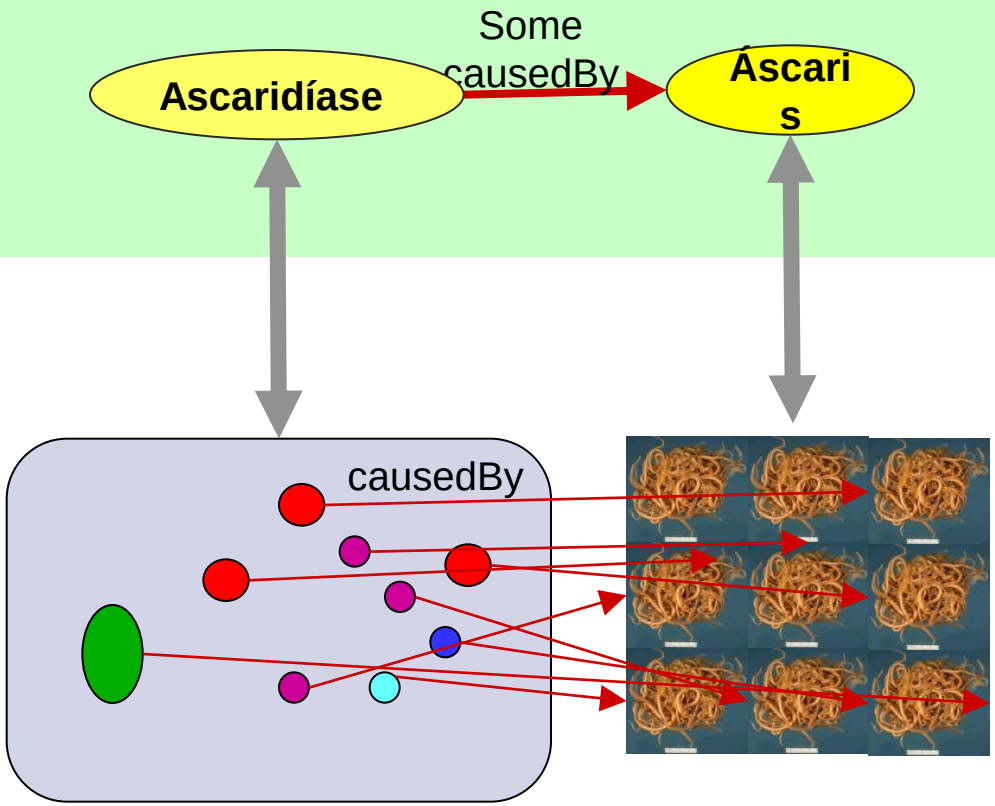
Relações e definições

Ontologia



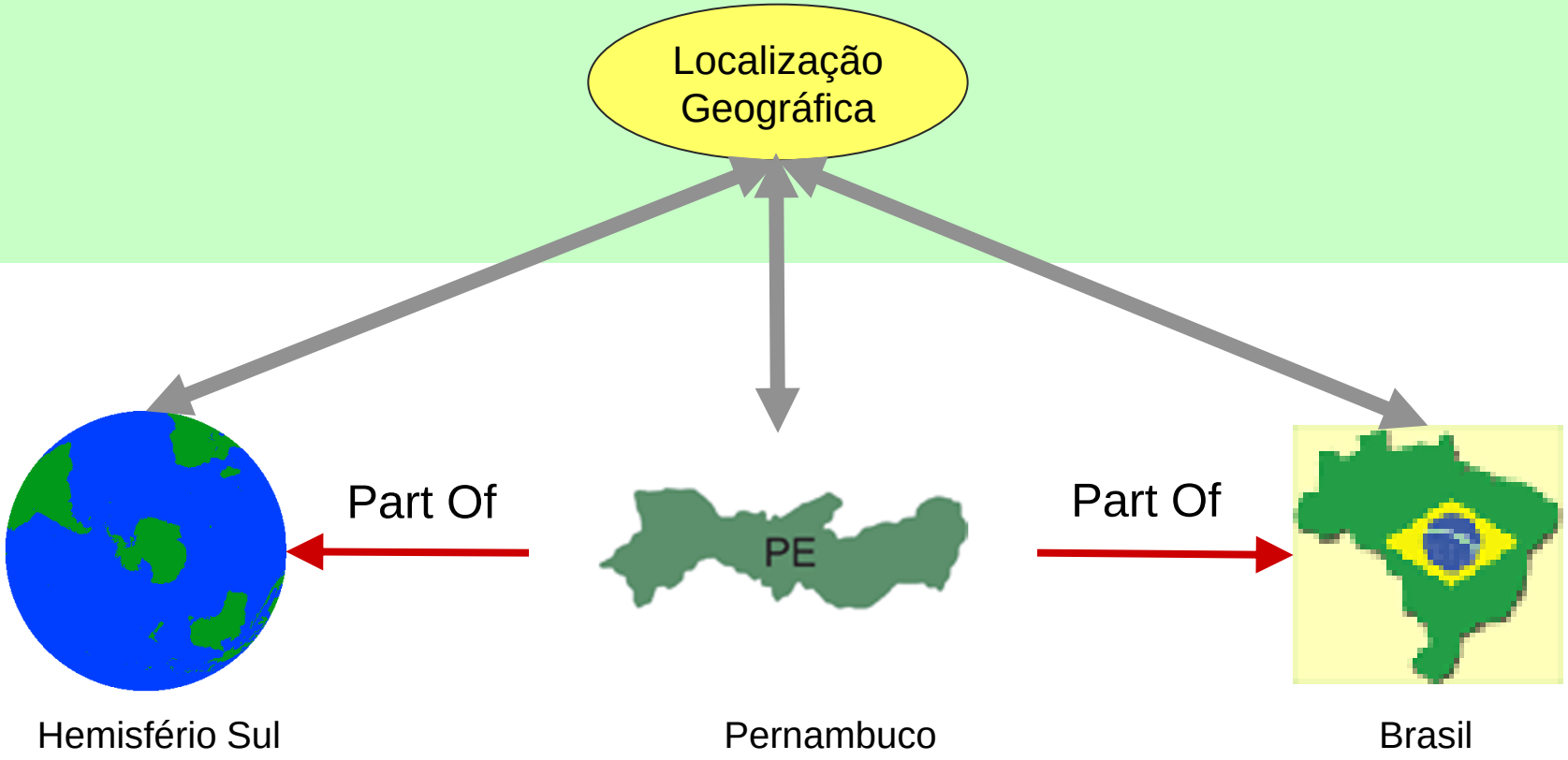
Relações e definições

Ontologia



Relações a nível de indivíduos

Ontologia



Ontologias: características

- Ontologias são hierarquias de tipos
- Esse tipos estendem-se para classes de entidades reais
- Adicionalmente, ontologias provêem relações (e.g. part-of, has-location, caused-by) para descrever e possivelmente definir todas as entidades (individuos, instâncias) de um tipo
- Conforme o domínio, ontologias podem ser estendidos para incluírem indivíduos (e.g. organizações, localizações geográficas

Ontologias em Biologia

- OBO- Open Biological Ontologies: repositório de terminologias desenvolvido para uso compartilhado entre diversos domínios biológicos e médicos.
- Vantagens:
  - Acesso livre
  - Formato padronizado

- anatomy
  - cell type
  - gross anatomy
    - animal gross anatomy
      - C. elegans gross anatomy
      - Drosophila gross anatomy
      - eVoc (Expressed Sequence Annotation
      - fish anatomy
        - Medaka fish anatomy and developmer
        - Zebrafish anatomy and development
      - human developmental anatomy
        - Human developmental anatomy, time
        - Human developmental anatomy, abstı
      - Mosquito gross anatomy
      - mouse anatomy
        - Mouse adult anatomy
        - Mouse anatomy and development
    - microbial anatomy
      - Fungal anatomy
      - Dictyostelium anatomy
    - plant anatomy
      - Arabidopsis anatomy
      - Cereal anatomy
      - Maize anatomy
      - Plant anatomy
  - organ
  - BRENDA tissue / enzyme source
- animal natural history and life history
- chemical
  - chemical entities of biological interest
  - physico-chemical methods and properties
  - physico-chemical process
  - systems biology
- development
  - animal development
    - C. elegans development
    - Drosophila development
    - eVOC (Expressed Sequence Annotation
    - medaka fish anatomy and development
    - Mouse anatomy and development
    - Plasmodium life cycle
    - Zebrafish anatomy and development
  - plant development
    - plant growth and developmental stage
    - Arabidopsis development
    - cereal plant development
- ethology
  - Habronattus courtship
  - Loggerhead nesting
- evidence codes
- experimental conditions
  - biological imaging methods
  - microarray experimental conditions
  - nuclear magnetic resonance experiment co
  - ontology for biomedical investigations
  - physico-chemical methods and properties
  - sample processing and separation techniqu
- genomic and proteomic
  - gene product
    - biological process
    - cellular component
    - event
    - gene product name
    - molecular function
    - molecule role
    - multiple alignment
    - pathway
    - protein
      - protein covalent bond
      - protein domain
      - protein modification
      - protein-protein interaction
    - proteomics data and process provenance
  - sequence types and features
- metabolomics
  - nuclear magnetic resonance experiment condition
- OBO relationship types
- phenotype
  - cereal plant trait
  - eVOC (Expressed Sequence Annotation for Humar
  - human diseases
  - mammalian phenotype
  - medaka fish anatomy and development
  - mouse pathology
  - plant enviromental conditions
  - PATO
- taxonomic classification
  - fly taxonomy
  - NCBI organismal classification
  - SwissProt organismal classification
- vocabularies
  - FlyBase controlled vocabulary
  - MESH
  - NCI Thesaurus

Exemplo: Gene Ontology

- GO:0005575 : cellular_component (80819)
  - GO:0005623 : cell (57332)
    - GO:0005933 : bud (244)
    - GO:0043025 : cell body (3)
    - GO:0000267 : cell fraction (1568)
    - GO:0042995 : cell projection (393)
    - GO:0009986 : cell surface (359)
    - GO:0030312 : external encapsulating structure (361)
    - GO:0042763 : immature spore (17)
    - GO:0005622 : intracellular (46676)
      - GO:0045177 : apical part of cell (78)
      - GO:0005930 : axoneme (59)
      - GO:0045178 : basal part of cell (22)
      - GO:0005938 : cell cortex (379)
      - GO:0046858 : chlorosome (0)
      - GO:0005694 : chromosome (1340)
      - GO:0005929 : cilium (57)
      - GO:0000307 : cyclin-dependent protein kinase holoenzyme complex (40)
      - GO:0005737 : cytoplasm (36347)
        - GO:0009317 : acetyl-CoA carboxylase complex (27)
        - GO:0020022 : acidocalcisome (0)
        - GO:0030929 : ADPG pyrophosphorylase complex (1)
        - GO:0030877 : beta-catenin destruction complex (1)
        - GO:0009504 : cell plate (6)
        - GO:0009346 : citrate lyase complex (9)
        - GO:0000229 : cytoplasmic chromosome (4)
        - GO:0000308 : cytoplasmic cyclin-dependent protein kinase holoenzyme complex (
        - GO:0000177 : cytoplasmic exosome (RNase complex) (29)
        - GO:0000932 : cytoplasmic mRNA proces
        - GO:0000153 : cytoplasmic ubiquitin ligase complex (71)
        - GO:0016023 : cytoplasmic vesicle (4349)
        - GO:0005856 : cytoskeleton (2035)
          - GO:0015629 : actin cytoskeleton (829)
          - GO:0001533 : cornified envelope (24)
          - GO:0030863 : cortical cytoskeleton (149)
          - GO:0045111 : intermediate filament cytoskeleton (132)

Cyclin-dependent protein kinase (CDK) complex found in the cytoplasm.

Ontologias para Endemias

- Organismos
  - Patógenos
  - Vetores
- Estruturas anatômicas
  - Homem, outras espécies
- Doenças
- Procedimentos
  - Terapia, prevenção, controle
  - Investigação científica
- Compostos químicos
  - Biomoléculas
  - Drogas
- Informações geográficas

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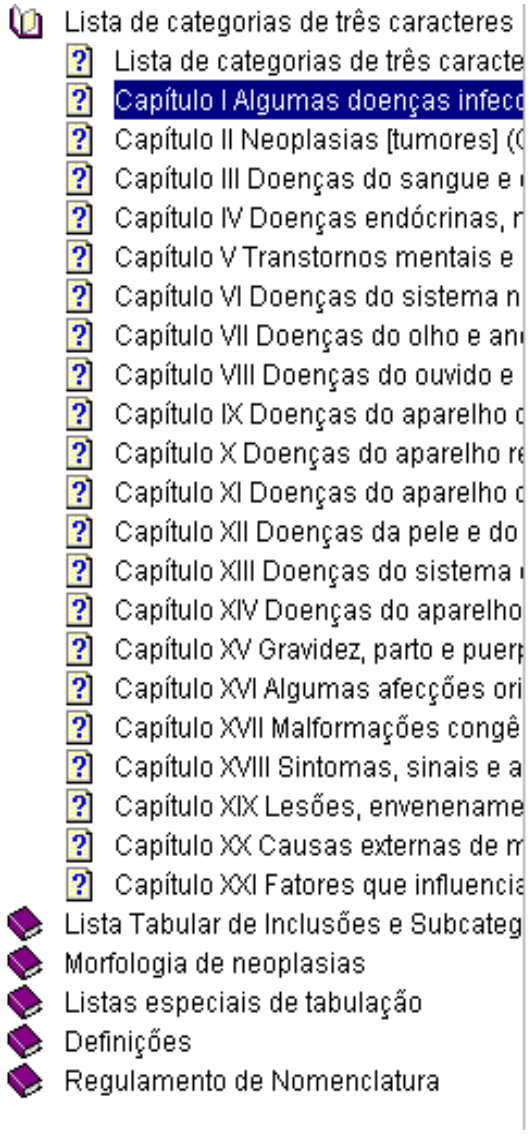
Ontologias em Medicina

- CID – 10 (classificação internacional de doenças)
  - Limitado a doenças
  - Só tem hierarquias
  - Não tem definições e relações

CID (Classificação Internacional de Doenças



B65-B83 Helmintíases
B65 Esquistossomose [bilharziose] [Schistosomíase]
B66 Outras infestações por trematódeos
B67 Equinococose
B68 Infestação por Taenia
B69 Cisticercose
B70 Difilobotríase e esparganose
B71 Outras infestações por cestóides
B72 Dracontíase
B73 Oncocercose
B74 Filariose
B75 Triquinose
B76 Ancilostomíase
B77 Ascaridíase
B78 Estrongiloidíase
B79 Tricuríase
B80 Oxiuríase
B81 Outras helmintíases intestinais, não classificadas em outra parte
B82 Parasitose intestinal não especificada
B83 Outras helmintíases

B85-B89 Pediculose, acaríase e outras infestações
B85 Pediculose e ftiríase
B86 Escabiose [sarna]
B87 Miíase
B88 Outras infestações
B89 Doença parasitária não especificada

B90-B94 Seqüelas de doenças infecciosas e parasitárias
B90 Seqüelas de tuberculose

B77 Ascaridíase

Inclui:

ascaridose
infestação por Ascaris

B77.0 Ascaridíase com complicações intestinais

B77.8 Ascaridíase com outras complicações

B77.9 Ascaridíase não especificada

Ontologias em Medicina

- CID – 10 (classificação internacional de doenças)
  - Limitado a doenças
  - Só tem hierarquias
  - Não tem definições e relações
- SNOMED CT: (*Systematized Nomenclature of Medicine / Clinical Terms*)
  - Todas as disciplinas clínicas (medicina humana e veterinária)
  - 350 000 classes
  - 2007: Propriedade da "*Organização Internacional para o Desenvolvimento de Padrões em Terminologia Clínica (IHTDSO)*". Licenciamento a nível de paises. Membros atuais: US, UK, CA, AU, NL, SW, DK, LT

SNOMED CT

- Hierarquias:

Organism (organism)
Infectious agent (organism)
Infectious agent (organism)
Heterotroph (organism)
Parasite (organism)

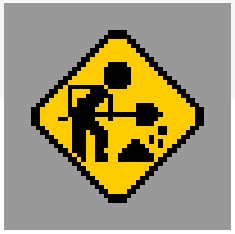
Organism (organism)
Animal (organism)
Invertebrate (organism)
Animal (organism)
Invertebrate (organism)
Helminth (organism)
Nematode (organism)
Parasitic nematode (organism)
Ascaris (organism)

SNOMED CT: Definições

Parent(s):
(Select a parent to make it the "Current Concept".)
Intestinal nematode infection (disorder)

Current Concept:
Ascariasis (disorder)

Child(ren):
(N=7) (Select a child to make it the "Current Concept".)
Adult ascariasis (disorder)
Ascariasis with intestinal complications (disorder)
Ascaris peritoneal granuloma (disorder)
Biliary ascariasis (disorder)
Infection by Ascaris lumbricoides (disorder)
Infection by Ascaris suum (disorder)
Larval ascariasis (disorder)



Current Concept:
FullySpecifiedName: Ascariasis (disorder)
ConceptId: 2435008

Defining Relationships:

Is a	Intestinal nematode infection (disorder)
Causative agent (attribute)	Ascaris (organism)
Causative agent (attribute)	Internal parasite (organism)
Finding site (attribute)	Intestinal structure (body structure)

This concept is fully defined.

Descriptions (Synonyms):

Preferred:	Ascariasis
Synonym:	Round worm infection
Synonym:	Ascariosis
Synonym:	Ascaridiasis
Synonym:	Ascariasis - roundworms
Synonym:	Roundworm infection
FullySpecifiedName:	Ascariasis (disorder)

http://www.ihtsdo.org/

Endemias: Cobertura por SNOMED

- Metodologia:
  Análise de MEDLINE: Amostra de 10 resumos dos últimos 500 indexados com o descritos “*endemic disease*” [MeSH]
- Identificação dos termos técnicos por resumo
- Mapeamento manual de cada termo para SNOMED

49: Mem Inst Oswaldo Cruz. 2007 Jun;102(3):421-4.

Merozoite surface protein 2 allelic variation influences the specific antibody response during acute malaria in individuals from a Brazilian endemic area.

Sallenave-Sales S, Faria CP, Zalis MG, Daniel-Ribeiro CT, Ferreira-da-Cruz Mde F.

Instituto Oswaldo Cruz, Fiocruz, Rio de Janeiro, RJ, 21040-900, Brasil.

The antibody response to Plasmodium falciparum parasites of naturally infected population is critical to elucidate the role of polymorphic alleles in malaria. Thus, we evaluated the impact of antigenic diversity of repetitive and family dimorphic domains of the merozoite surface protein 2 (MSP-2) on immune response of 96 individuals living in Peixoto de Azevedo (MT-Brazil), by ELISA using recombinant MSP-2 proteins. The majority of these individuals were carrying FC27-type infections. IgG antibody responses were predominantly directed to FC27 parasites and were correlated to the extension of polymorphism presented by each MSP-2 region. This finding demonstrated the impact of the genetic polymorphism on antibody response and therefore, its importance on malaria vaccine efficacy.

Domestic dogs are not only reservoir hosts of the American zoonotic visceral leishmaniasis (ZVL) but of the American zoonotic tegumentary leishmaniasis (ATL) as well, for different reasons. However it is still controversial to state that dogs are incriminated as ATL reservoir hosts as there is evidence that humans and dogs are likely to be exposed in the same way to sandfly vector. In Venezuela this issue has not been completely addressed, for this reason we selected a location inside Trujillo city to study eco-epidemiological conditions as well as to survey a significant sample of dogs by Montenegro Skin Test (MST). Antigen was prepared according to standard procedure using Leishmania (V) braziliensis promastigotes (80 microg/ml); response was read 48 hours post-inoculation with an induration size > 5 mm being considered as positive. The study place is an endemic mountainous semi-urban area located at 850-950 masl with an average rainfall of 150 mm/year. We evaluated 61 dogs in 46 houses with 168 human beings. Among the human population 27 cases of ATL were reported (16.1%). With the MST we found 19 positive-reaction dogs (31%) (mean MST size of 9.58 mm, 95% CI: 8.41-10.75) in 13 houses (28%). Multivariate analysis did not reveal significant association between domestic MST positive-dog ownership and human ATL cases (RR = 1.48, p = 0.28). Although some studies have indicated that dog ownership and dog infection rates are associated with an increased risk of human disease in different evaluated places, this question has not been completely answered in Venezuelan studied zones, further research is necessary.

BACKGROUND: Cutaneous larva migrans (CLM) is a common but neglected parasitic skin disease in impoverished communities of the developing world. METHODS: To describe the clinical features and the morbidity associated with CLM, active-case finding was performed during rainy and dry seasons in a rural community in the state of Alagoas, Northeast Brazil. RESULTS: A total of 62 individuals received a diagnosis for CLM, and among them there were a total of 75 larval tracks. The number of tracks per person ranged from 1 to 3. Children <9 years old were affected significantly more often than other age groups (P<.001). Children had their lesions mainly on the buttocks, genitals, and hands, whereas in older patients, the majority of lesions were located on the feet. Twenty-four percent of the 75 lesions were superinfected. Bacterial superinfection was more common among children. All but 1 patient complained about itching, and 84% of the 62 patients complained about sleep disturbances. Itching was classified as being severe by 61% of patients. Approximately 40% of the 62 patients had attempted to treat their creeping eruption previously. Larvae moved forward 2.7 mm per day. There was a significant correlation between the length of the track and the duration of infestation (rho =.53; P<.0001). CONCLUSIONS: CLM is a common parasitic skin disease in this rural community, with different topographic patterns of lesions in children and adults. CLM is associated with considerable morbidity. The length of the track can be used to estimate the duration of infestation.

Resultados

- Por resumo: 71% dos termos têm correspondência no SNOMED
- Termos não representados:

Endemic area
World Health Organization
Peixoto de Azevedo (MT)
Accra
Shandong Province
Northeast Brazil
Developing world
Rainy season
reservoir hosts

Prevalence
Breeding container
Case report
time series study
segmented regression analysis
RPE-Cy5-conjugated antibodies
fluorescein isothiocyanate
Vdelta1
Merozoite surface protein 2
Montenegro Skin Test

Conclusão

- SNOMED CT: ontologia interessante para representar os aspectos clínicos do domínio de endemias
- Insuficiente em conceitos de Saúde Pública, bioestatística, geografia
- Soluções:
  - Extensão local de SNOMED
  - Combinação com outras ontologias
- Para uso na prática: Acordos com IHTSDO a nível de projetos. Mais fácil: Filiação do Brasil à IHTSDO

Freiburg

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XX CONGRESSO BRASILEIRO DE PARASITOLOGIA
Avanços e Desafios

Recife