OEP: Ontology of Evolutionary Process

Practice & Method

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[slide #1]

Introduction

- Main topics today:
  - Ontology—a brief introduction
  - A quick scan of the kinds of phenomena that OEP is meant to illuminate
  - Overview account of the four classes of entities and their relationships according to OEP.
  - Assessment of the state of the discipline of formal ontology for biological sciences
  - Proposed alternative method to solve many problems posed by the dominant school of thought on ontological engineering
  -

[slide #2]

Where are we, and what's next?

[slide #3]

The fact of evolution

- In *The Origin of Species*, Darwin claimed that all organisms share a common ancestor or have an ancestor in a small group of organisms.
- This is the *fact of common descent*.
- Darwin often describes evolution as descent with modification.
- This claim was accepted almost immediately.
- Common descent is used to explain phenomena in every life science discipline.
- The research program: identify relationships of ancestry among the species.
- Establish *what begat what*.

[slide #4]

Mechanisms of evolution

- Darwin made a second claim: natural selection is the central mechanism of evolution.
- If no mechanism of evolution could be identified, species differences and complex adaptation would remain unexplained.
- Hardly anyone accepted Darwin's proposal about natural selection.
- The research program: evaluate Darwin's claim about natural selection and identify alternatives.
- These mechanisms of evolution are used to explain *how and why* dynamics of variation in the organic world occur.

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How and why does . . . occur? Phenomena of evolution

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- Complex adaptation—displace argument from design
- Tempo and mode in macroevolution
- Patterns of molecular evolution
- Embryology, development, morphogenesis
- Differences in major structural elements, *Baupläne*
- Taxonomic relationships
- Phylogenesis
- Origin & nature of species as biological individuals
- Biogeography
- Many, many others . . .

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Where are we, and what's next?

Thomas Gruber's famous definition (1993)

Ontology "Explicit specification of a conceptualization"

Conceptualization "The objects, concepts, and other entities that are assumed to exist in some area of interest and the relationships that hold among them . . . . A conceptualization is an abstract, simplified view of the world that we wish to represent for some purpose."

Gruber's definition explained

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- Gruber's view is that ontologies are a means of information exchange between agents, machine or human.
- If the kind of content differs among data sources, there is a special need for a mechanism for exchange.
- Agents query one another in a standardized formal language.
- The formal language is a model of the general characteristics of the environment of the agents.

An Example

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- The Plant Ontology has the term "plant cell."
- A human researcher queries a robot search agent about specific measurements associated with specific kinds of plant cells.
- The robot searches several databases, each with different data sets about different plant species.
- The researchers curating the databases use the Plant Ontology to organize their data, with a common syntax to encode it.
- Because of this shared description of their specimens and the common encoding format, the robot identifies the correct information in each database.

A naturalistic account of formal ontology

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Ontology The ontology of domain D is a scheme for organizing information about things in D that reflects what those things are and what their relationships are to one another.

Question How do we know what the entities and relationships in D are?

Answer Our best source of information is (are) the science(s) of domain D.

Comment Terms in the ontology represent entities, properties; predicate-like representations of relationships between them.

Example Use Mx for "x has mass."

Example Use Cx for "x is a cell."

Example Use $R(x, y, z)$ for "x is between y and z."

Where are we, and what's next?

The central guiding design principle

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What explains an evolutionary process?

Formulate a general account of those entities and their relationships that explain an evolutionary process.

Desiderata for the ontology

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The ontology must be general enough to apply across:

- Levels of biological hierarchy
- Types of variation
- Types of evolving lineage
- Adaptive and non-adaptive processes
- Scales of time and space

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Daniel Dennett and Richard Dawkins

- Dennett and Dawkins claim that the explanatory aim of evolutionary biology is to displace the argument from design.
- They say that Darwin's central innovation, which has turned out to be a lasting contribution, is the claim that natural selection explains adaptive complexity.
- While acknowledging Darwin's role in naturalizing organic phenomena, it must also be acknowledged that the Dennett-Dawkins view about evolutionary science is not likely to be true.
- The neutralist-selectionist dispute; isolation and subsequent reproductive separation in the origin of animal species; tempo and mode in large-scale evolution.

Sadly, a poverty of resources

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- One strategy of response to Dennett and Dawkins would be to turn to the literature.
- How many papers report the occurrence of natural selection, and how many report the occurrence of (say) drift?
- Unfortunately, there is no way to search bibliographic records or the literature itself to answer this question in a reasonable amount of time.
- PubMed indexing fails miserably at capturing evolutionary topics
  - Not that NLM should be faulted for this—PubMed is, after all, intended for use in medical contexts.
- Free-text searching in abstracts, full article text, titles generates far too many false positives or negatives, depending on the search key.

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- Darwin's marginalia, notes, and published works are continuous with one another—in many cases, his line of thought can be traced from initiation in notes or marginalia to a passage in a published work.
- Growing digital corpus at the Biodiversity Heritage Library presents the opportunity to link a user to a digital copy of a work mentioned by Darwin, even to a particular page or passage.
- Why not add a subject-oriented thread?
- Place Darwin's work in context of historically related works on topics he writes about?
- Example: the role of geographic isolation in the formation of new species.

From Endler's *Natural Selection in the Wild*

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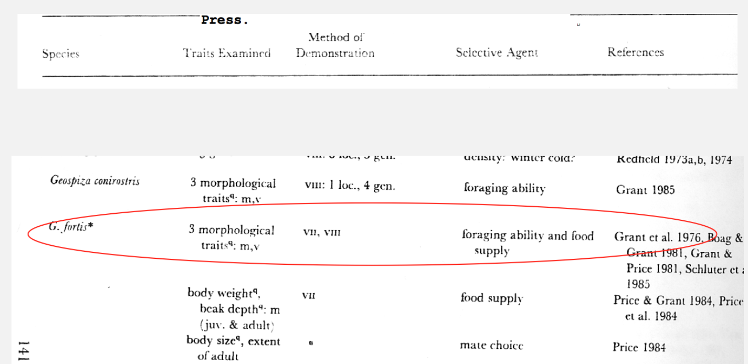

Press.

Species	Traits Examined	Method of Demonstration	Selective Agent	References
			density? winter cold?	Redfield 1973a,b, 1974
Geospiza conirostris	3 morphological traits[q]: m,v	VIII: 1 loc., 4 gen.	foraging ability	Grant 1985
*G. fortis**	3 morphological traits[q]: m,v	VII, VIII	foraging ability and food supply	Grant et al. 1976, Boag & Grant 1981, Grant & Price 1981, Schluter et 1985
	body weight[q], beak depth[q]: m (juv. & adult)	VII	food supply	Price & Grant 1984, Price et al. 1984
	body size[q], extent of adult	*	mate choice	Price 1984

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The essence of an evolutionary process

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Four elements for describing the history of a biological individual:

1. Some lineage or historical individual;
2. Some aspect in which that individual changes;
3. Some "environment," i.e., interaction of the individual as a whole or in parts with its surroundings; and
4. Some pattern or signature of the time-course of the changing aspect of the individual.

Progress?

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Hypothesis: The four-element description can be generalized.

Initial assay: The *human population* changed by *natural selection* from *being primarily lactose intolerant to being tolerant* after *humans learned to domesticate diary animals*.

Assessment: The hypothesis is confirmed.

`EvolvingEntity`

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- A historical individual.
- Is not continuously in the same state with regard to `EvolvableProperty`.

Example: Gene pool
Example: Gene locus
Example: Phylum
Example: Species

Context

- Analogous to environment, i.e., when considering the environment of organisms in their habitats.
- Contains or has an element of `EvolvingEntity` as a part.

Example Habitat of an organism (species or other population-like group of organisms)

Example Physiology of sex cell formation ("genes that violate Mendel's rules".

Example Habitat fragmentation (Influences rates of extinction, i.e., diversity of species in a phylum)

Example Geographic isolation (allopatry)

EvolvableProperty

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- Quality of an element of *EvolvingEntity* that varies in character.
- Analogous to a morphological or physiological trait of an organism.

Example Alleles, interaction of organisms with some one of `Context`

Example Alleles, where `Context` includes genes that violate Mendel's rules

Example Quantitative trait e.g., height

Example Number of species in a phylum

Signature

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- Pattern of change in `EvolvingProperty`; characteristic time-course of `EvolvingProperty`

Example Natural selection—better adapted organisms survive in greater proportion than less-well-adapted organisms

Example Species selection—species that have shorter life cycles have a higher probability of ramifying per unit time

Example Recurring mutations

Example Random drift, e.g., every element of `EvolvingProperty` has an equal probability of producing ancestors

The four classes and their relationships

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Source of *x*	Source of *y*	*Pxy*
`Evolving entities`	`Contexts`	*x* is in context *y*
`Evolving entities`	`Evolvable properties`	*x* has character *y*
`Evolvable properties`	`Signature`	*x* evolves by *y*

Where are we, and what's next?

Kuhn's model of scientific change

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Confusion
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- In *The structure of scientific revolutions*, Thomas Kuhn describes what he claims to be a pattern in the history of science.
- Most of the time, an established science is "normal," that is, there are established theoretical principles, standards of evidence-giving, styles of reasoning.
- Established sciences will eventually reach a point of crisis: there will be some problem that the "normal" resources cannot be used to solve.
- The crisis is not resolved by solving the problem, but by the general acceptance by the scientific community of a new paradigm.

The pre-paradigm and interregnum periods

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- Kuhn identifies characteristics of the period before a science adopts its first paradigm and after a crisis has been reached but before a new paradigm has formed.
  - No discipline-wide agreement on basic issues of method, theory, observation, styles of reasoning
  - Scientists choose one working group over another based on loyalty, social networks, personal interests, happenstance
  - Partly as a result of domination of the field, a new paradigm takes shape around a particular approach
  - In retrospect, the others seem unreasonable, the work of quacks, pseudoscience.

The state of formal ontology in life sciences

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Not looking up

The state of formal ontology in the life sciences most closely resembles the pre-paradigm stage.

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The Open Biological and Biomedical Ontology Group

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- the "OBO" has emerged as the most powerful influence on the community.
- They have become the de facto authority
- There are a set of principles that ontologies must adhere to if they are to be considered compliant
- Because they review ontologies for adherence to these principles, being approved by OBO is regarded similar to the way acceptance of a paper in a peer-reviewed journal is for an article.

Ontological Realism

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- The OBO group promotes a method and interpretation of formal ontology they term "realism"
  - Top-down development of ontologies
  - Ontologies about particular subjects must be formulated in an abstract vocabulary derived from Aristotle, purported to be a general description of reality
  - A central organization enforces compliance, establishes the correct interpretation of the Aristotelian principles.
- The OBO group's aim is to create a set of subject-area ontologies, unified by their adherence to the Aristotelian scheme.

Problems

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- The realist method is poorly suited for many tasks that ontology can be used for.

  For instance Comparing alternative theories; indexing literature; semantic web markup; describing experiments and hypotheses.
- Reformulating the description of one's domain using the Aristotelian vocabulary is difficult, requiring extensive consultation with OBO "gurus."
- Work groups sacrifice a degree of independence and control over how they want to structure their ontologies because the central authority may require changes.

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Ontological naturalism

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- "Bottom-up" development
- Specialist sub-communities in a discipline develop their own ontologies
- Keeping to the vocabulary of the scientific theories of the domain is stressed
- A central organizing body provides resources and expertise to create and maintain links between the sub-community ontologies
- Absent the demand for adherence to Aristotelian principles, a wide range of uses for ontologies are permissible.