

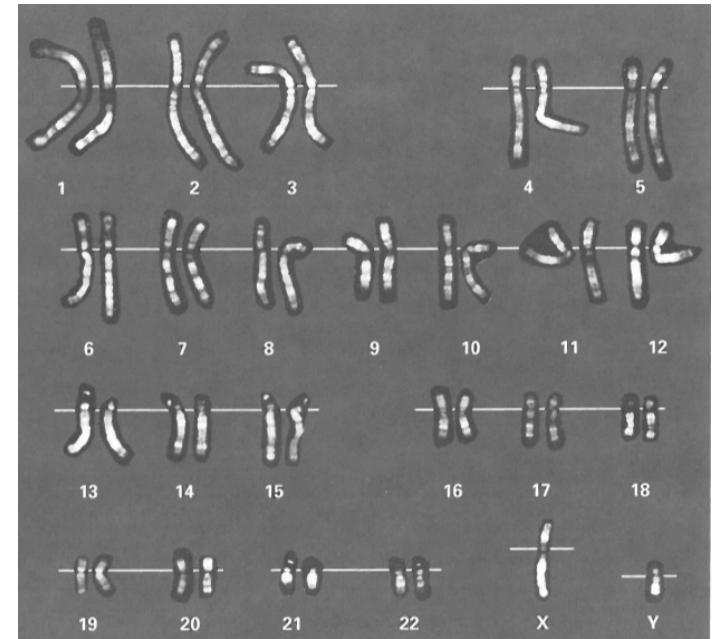
Exploring modelling choices: using non-functional characteristics to test an ontology add-on

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Modelling karyotypes

- What is a karyotype?
- Problem representing karyotypes
 - Can be complicated
 - Not computationally amenable
 - A lot of them
- The Karyotype Ontology (Warrender and Lord, 2013)



Tawny-OWL

- Novel tool – Tawny-OWL (Lord, 2013)
 - Motivated by karyotype work
 - “Textual user interface”
 - Fully programmatic
 - github.com/phillord/tawny-owl
- The Karyotype Ontology
 - Pattern-driven approach
 - Rapidly change and refine ontology
 - Investigate the effect an extension on the reasoning performance



The affects relation

- A deletion event affects a sequence of bands between two breakpoints
 - 46,XX,del(1)(q42) → { 1q42, 1q43, 1qTer }
- “How do we model the affects restriction?”
- *A priori*, it is difficult to determine which representation will work best
 - non-functional characteristics such as reasoning time
- Investigate three different “affects” models



Approach

Three step process:

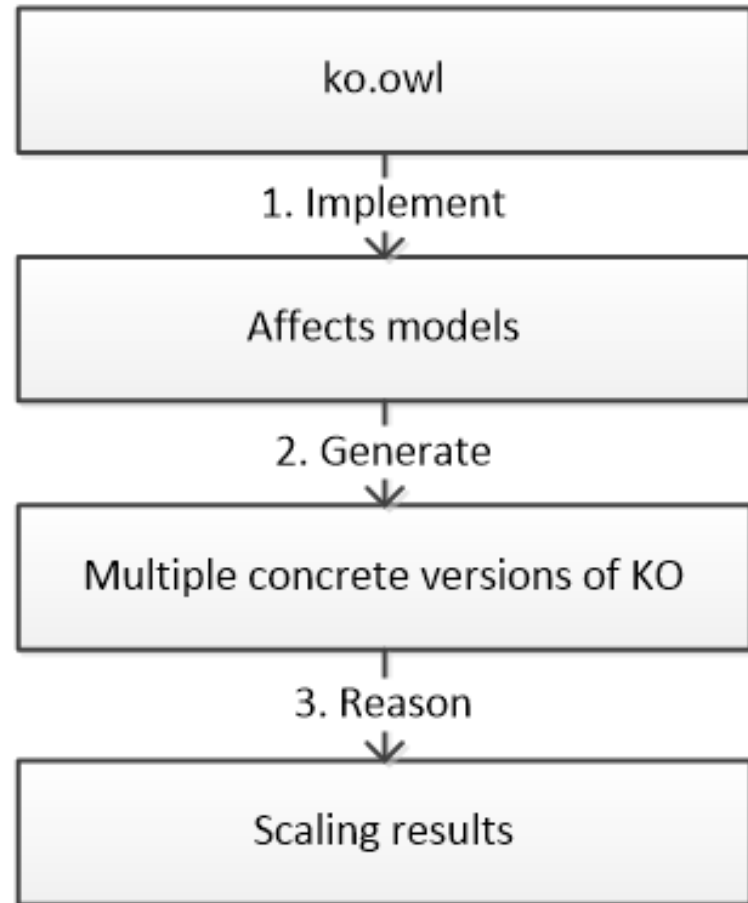
1. Implement

- Closure, Sequence, Data

2. Generate

- 1600 ontologies
- Vary in size

3. Reason

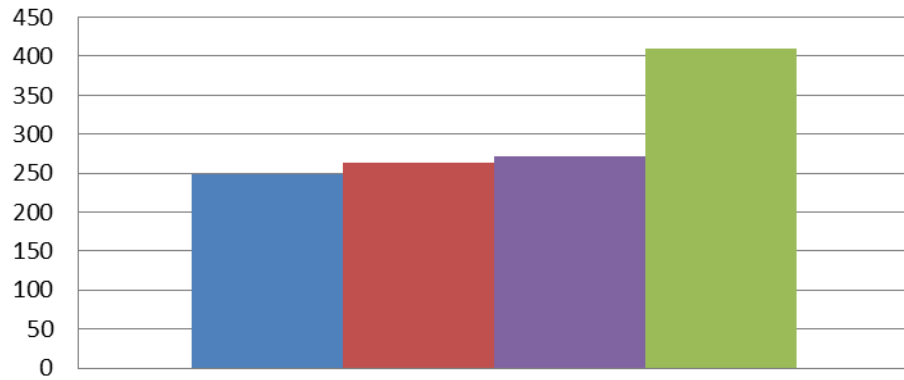


LEGEND

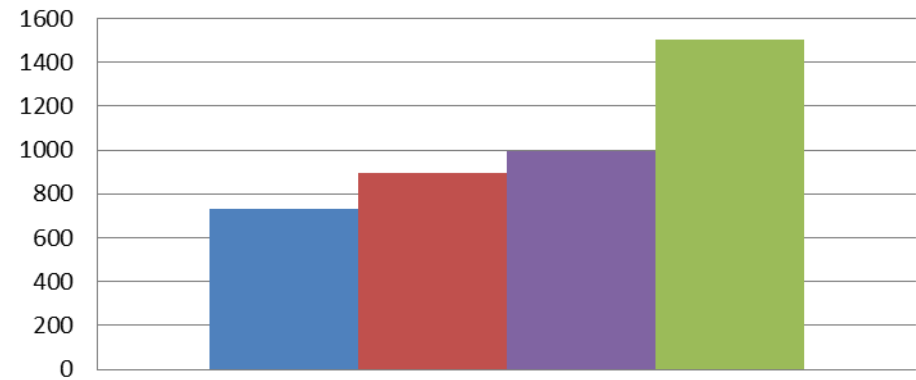
- Null
- Closure
- Sequence
- Data

Results

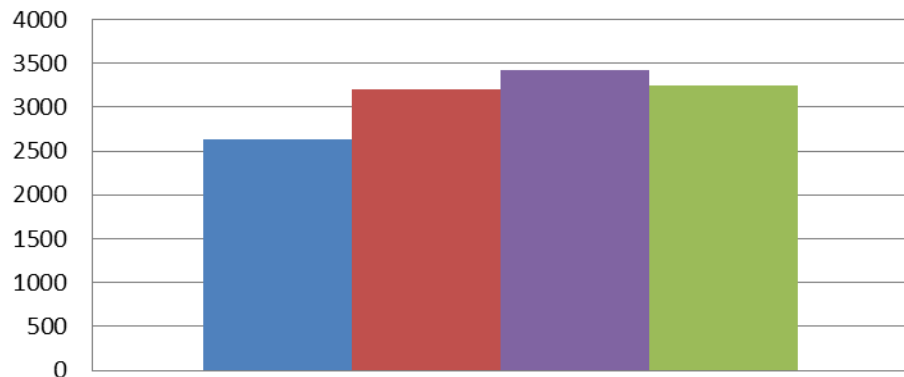
10 karyotypes



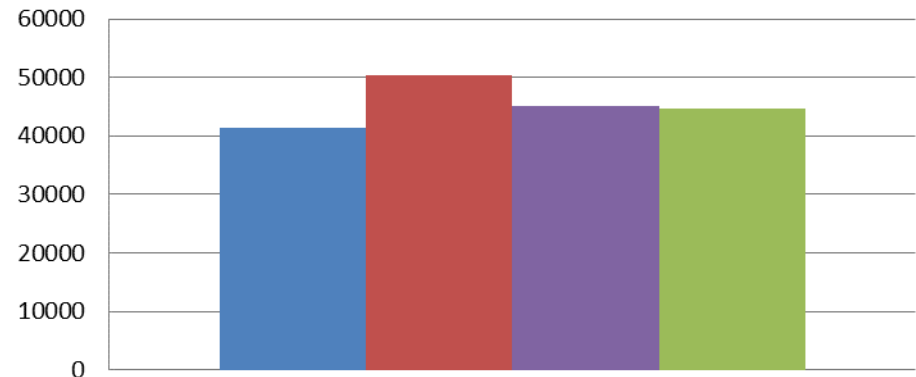
100 karyotypes



1,000 karyotypes



10,000 karyotypes



Summary

- Test performance, scalability and extensibility
- Pattern-driven and programmatic approach
 - Automatically and consistently regenerate ontologies easily and quickly
- Investigation of three distinct representations of the affects relation
 - Number of karyotypes
 - Number of abnormalities
- Use real world data
 - K-parser - github.com/fujimuramasa/k-parser/



Contacts and links

- Contacts:
 - Jennifer D. Warrender - jennifer.warrender@newcastle.ac.uk
 - Phillip Lord - phillip.lord@newcastle.ac.uk
- Thanks:
 - Anthony Moorman
- Links:
 - Tawny-OWL - github.com/phillord/tawny-owl
 - tawny-tutorial - github.com/phillord/tawny-tutorial
 - tawny-karyotype - github.com/jaydchan/tawny-karyotype
 - tawny-karyotype-scaling - github.com/jaydchan/tawny-karyotype-scaling



References

Lord, P. (2013). The Semantic Web takes Wing: Programming Ontologies with Tawny-OWL. OWLED 2013. Available: arxiv.org/abs/1303.0213

Warrender, J. D. and Lord, P. (2013a). The karyotype ontology: a computational representation for human cytogenetic patterns. Bio-Ontologies 2013. Available: arxiv.org/abs/1305.3758



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QUESTIONS?

“Affects” models

Original
No affects relation

```
(defclass 46,XX,del(1)(q42)
  :super
  (exactly 1 hasEvent
    (and Deletion
      (some hasBreakPoint 1q42 1qTer))))
```

Affects 1
ObjectProperty – includes
the use of the closure
axiom

```
(defclass 46,XX,del(1)(q42)
  :super
  (exactly 1 hasEvent
    (and Deletion
      (some hasBreakPoint 1q42 1qTer))))
(some affects 1q42)
(some affects 1q43)
(some affects 1q44)
(only affects
  (or 1q42 1q43 1q44))
```



“Affects” models

Affects 2

ObjectProperty – uses
the sequence ODP

```
(defclass 46,XX,del(1)(q42)
:super
(exactly 1 hasEvent
  (and Deletion
    (some hasBreakPoint 1q42 1qTer)))
(some affects
  (and 1q42
    (some directlyPrecedes 1q43)
    (some directlyPrecedes 1q44))))
```

Affects 3

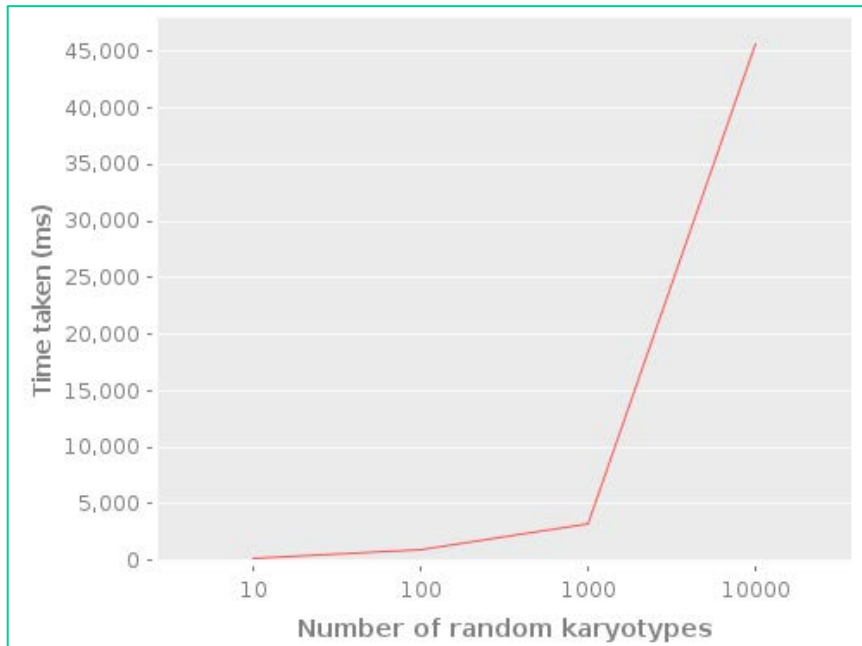
DataProperty – includes
the assignation of ordinal
numbers to the
chromosome bands

```
(defclass 46,XX,del(1)(q42)
:super
(exactly 1 hasEvent
  (and Deletion
    (some hasBreakPoint 1q42 1qTer)))
(only affects (or 11 12 13)))
```

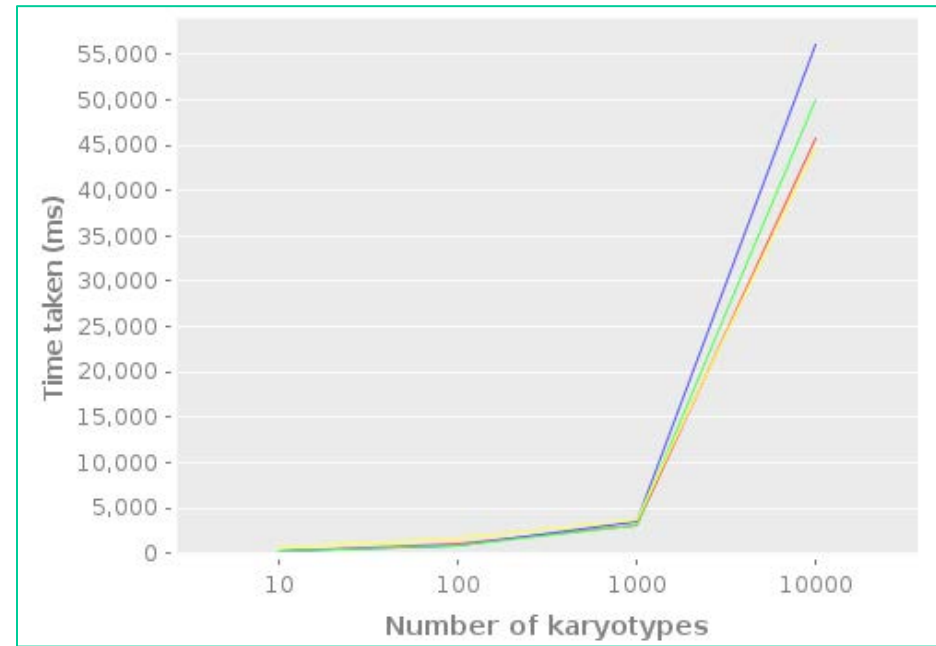


Results

How does the ontology scale as the number of karyotypes increases?



Do the models scale in the same way as the number of karyotypes increases?

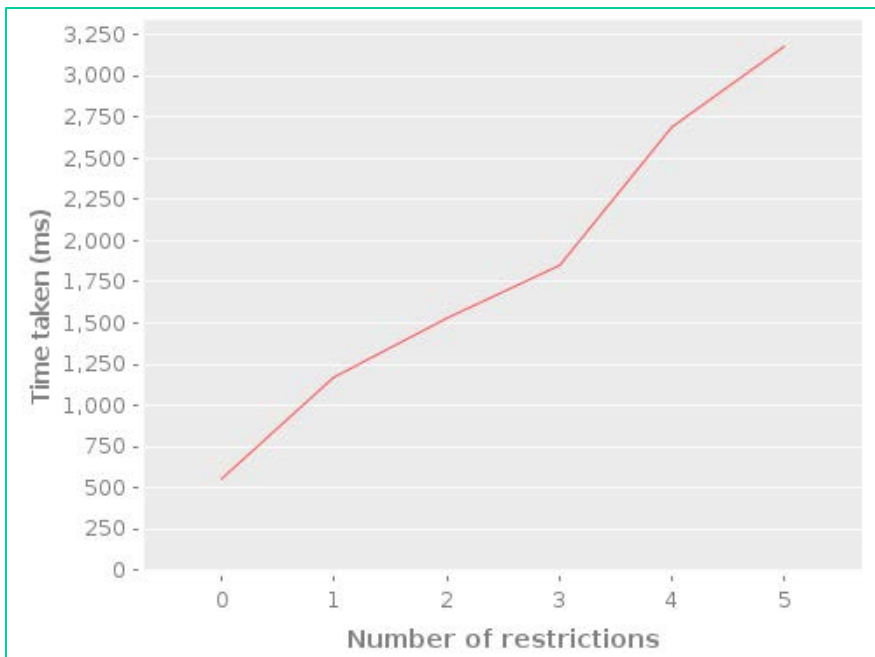


— $a = 0$ — $a = 1$ — $a = 2$ — $a = 3$

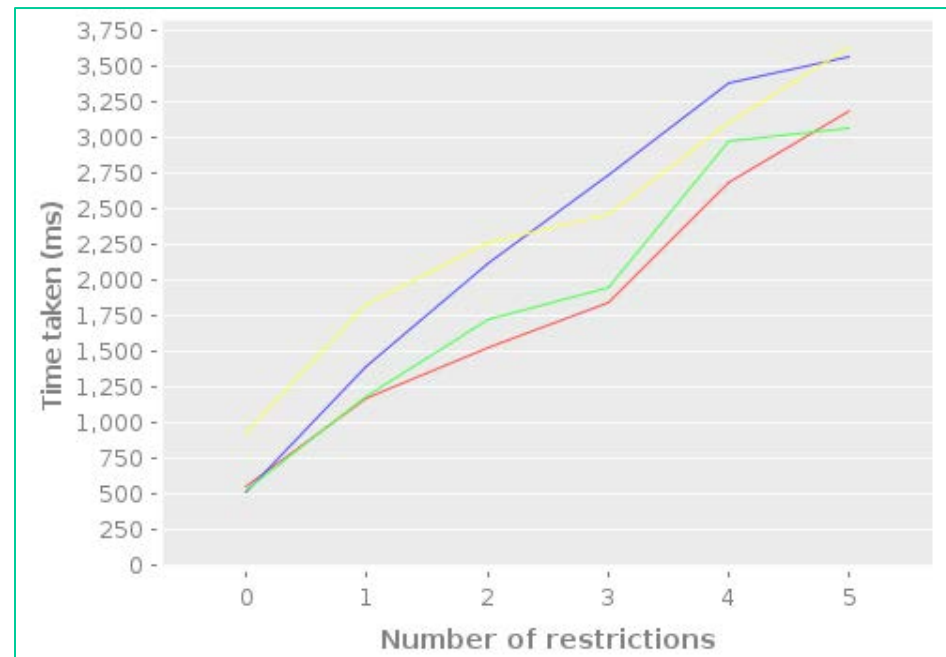


Results

How does the ontology scale as the number of restrictions increases?



Do the models scale in the same way as the number of restrictions increases?



— a = 0 — a = 1 — a = 2 — a = 3

