PhenoBlocks: Phenotype Comparison Visualizations

What: Patient Phenotype Comparisons (as defined by an ontology)

What is a phenotype?

What is an ontology?

What is a patient phenotype?

Why: Task Abstraction

How: Encode

How: Manipulate Selection

How: Manipulate

Why: Phenotype Comparison

How: Manipulate

How: Facet

What is an ontology?

● Network dataset type (xml like format)
● Similar to trees and DAGs
● Is-a relationships (hierarchical)
● Multiple inheritance
● And so much more!

What is a phenotype?

● Defined by assigning ontology terms to
  ○ {Present}
  ○ {Absent}
  ○ Unknown

What is a phenotype comparison?

“Query case” (undiagnosed)
“Reference case” (in database)

Why: Phenotype Comparison

● Analyse
  ○ Discover potential diagnoses
  ○ Verify existing diagnoses

● Query
  ○ Identify
    ■ Find similar cases
    ■ Compare
    ■ Examine similarities and differences between comparisons
    ■ Summarize
      ■ Understand case comparison profiles at a glance

How: Encode

● Ontology expanded into a tree
● Sunburst layout used
  ○ Shows all nodes
  ○ Gives more space to informative nodes
● Major branches separated
● Colour encodes overlap

How: Manipulate Selection

● Can change query phenotype in G
  ○ Will change visuals on demand
● Clicking on items in B
  ○ Brings up include/exclude dialog
  ○ Causes PhenoBlocks to freeze
● Edit Phenotype button near F
  ○ Also breaks PhenoBlocks

How: Manipulate

● Partitioned by main branches of phenotype ontology
● Small multiples used to juxtapose multiple cases
How: Reduce
- Clicking on categories collapses
- Not yet functional
- Can filter which nodes to display based on membership

How: Embed
- Details on demand
  - Main view (linked highlighting)
  - Small multiples view

Validation
- Worked with two clinicians during and after development
  - Tailored workflow to their needs during development
  - Validated the system based on user feedback after development
- Idiom validation not provided
  - No quantitative comparison to other methods/tools
- Algorithmic validation not provided
  - Slow and buggy, but technically not yet released

Validation: PhenomeCentral
- Buske et al. (2015)

Validation: GeneYenta
- Gottlieb et al. (2015)
  - DOI: 10.1002/humu.22772

End

Hard Handles
- Informs colour, based on information content and disorder frequency

How: Encode
- Colour used to represent whether terms are in present or absent in both reference and query cases

Ontology States
- Nine States
  - Present
  - Reference
  - Absent
  - Unknown

<table>
<thead>
<tr>
<th>Query</th>
<th>Present</th>
<th>Reference</th>
<th>Absent</th>
<th>Unknown</th>
</tr>
</thead>
<tbody>
<tr>
<td>Present</td>
<td>P/P</td>
<td>P/A</td>
<td>P/U</td>
<td></td>
</tr>
<tr>
<td>Absent</td>
<td>A/P</td>
<td>A/A</td>
<td>A/U</td>
<td></td>
</tr>
<tr>
<td>Unknown</td>
<td>U/P</td>
<td>U/A</td>
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