



Building SNOMED CT Reference Sets for use as Interface Terminologies

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Moving to an e-health environment

- Using information and communications technologies is the only way to maintain Australia's health care system and meet the challenges of an aging population.

Deloitte report to AHMC, 2008



Capturing, Storing and Exchanging Clinical Information

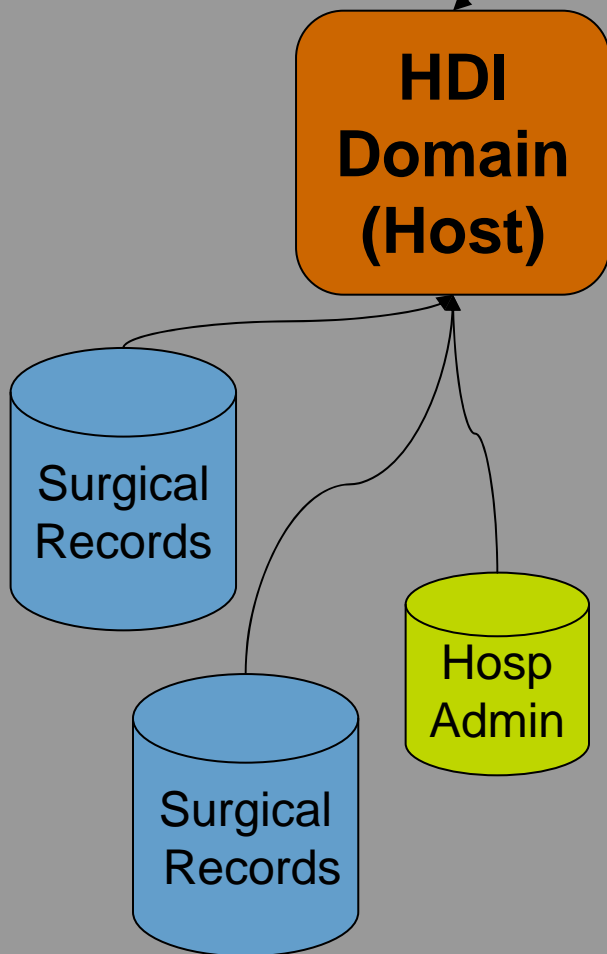
- Capturing and Storing data
 - Information model, such as CEN13606 or openEHR
 - Terminology, such as SNOMED CT
- Exchanging information
 - Messaging standard HL7

Three mapping use-cases for the Snapper platform

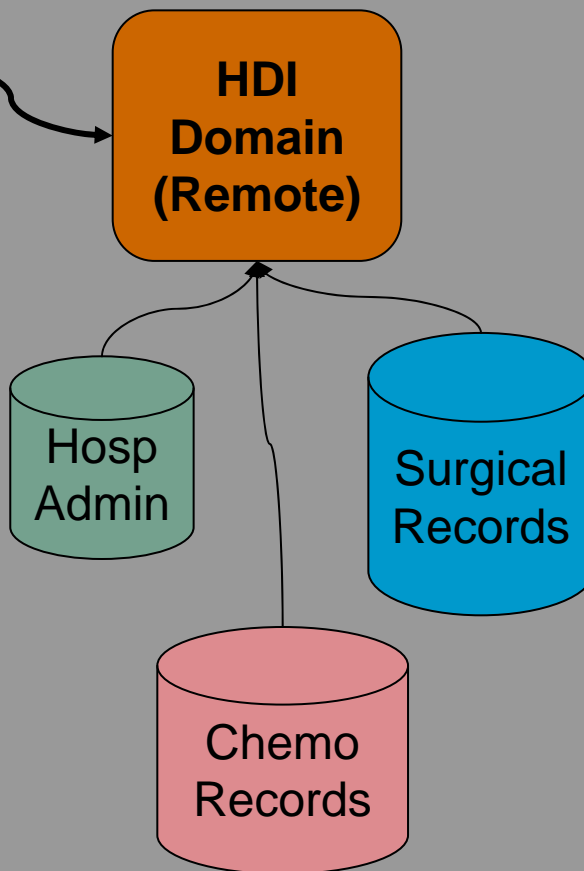
1. To produce a mapping to enable an existing patient data set captured in a different terminology for the purpose of data mining or analytics.
2. To migrate an existing terminology or data dictionary to use SNOMED CT concepts.
3. To develop an extension to SNOMED CT to provide greater depth or fill existing gaps in the ontology.

Why Snapper?

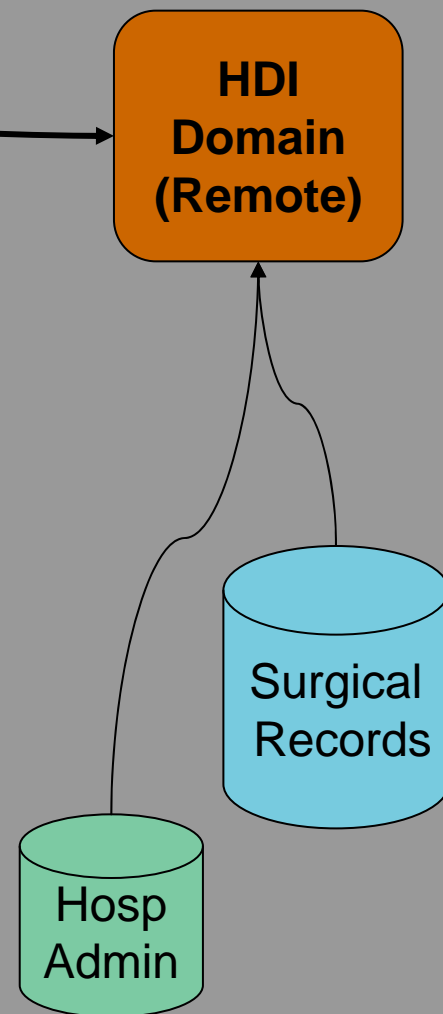
Large teaching hospital



Private hospital

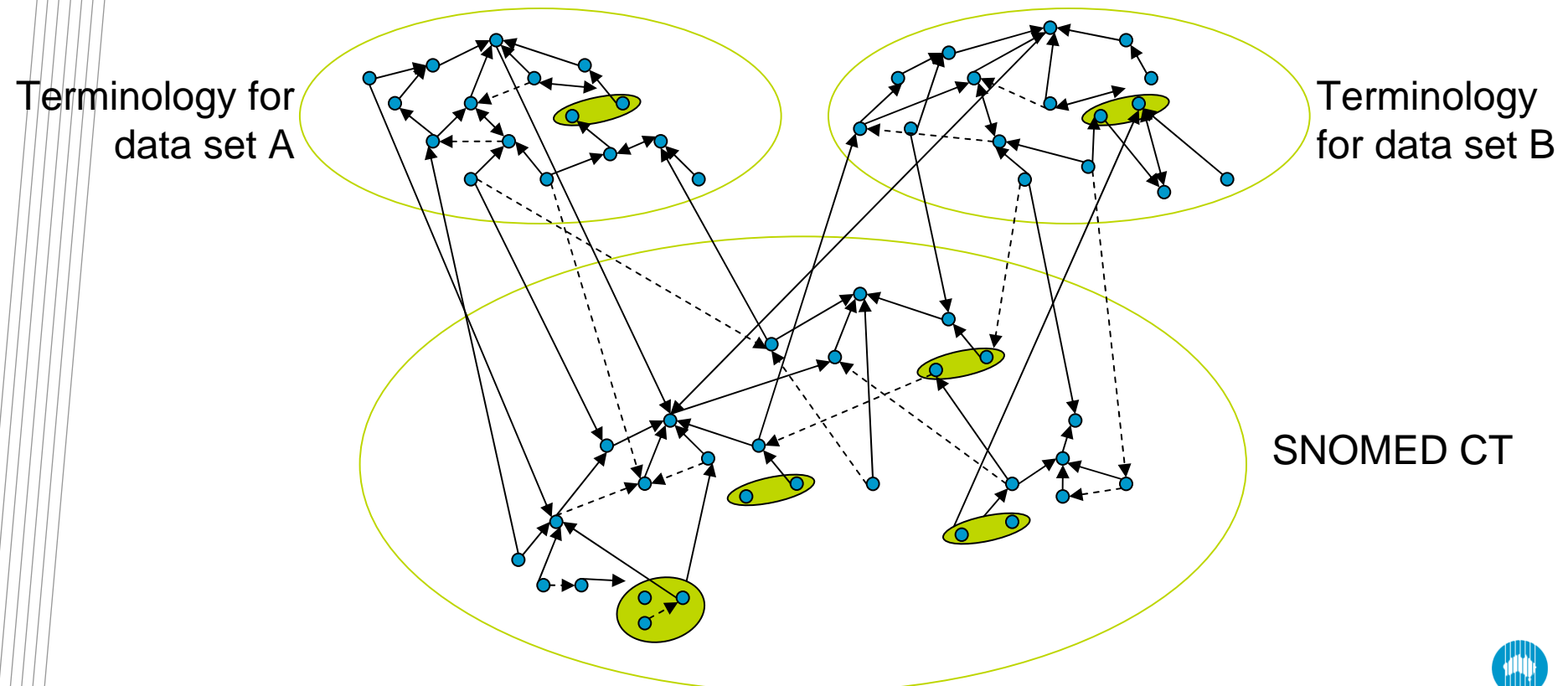


Regional hospital



What to do about adding meaning?

- Hypothesis: Can we use SNOMED CT as a base ontology and add “new bits of knowledge” to fully describe data which is collected using other terminologies?



The ANZICS Mapping

The screenshot displays the Snapper application interface within an Eclipse IDE. The main window shows the 'ANZICS_Mapping_Project/anzics_subcodes.mapping' file. The 'Project Expl' view on the left lists the project structure, including 'ANZICS_Diagnosis', 'ANZICS_Diagnosis', and 'ANZICS_SubCode'. The 'Ontology View' in the center shows a hierarchical structure of concepts, with 'Retinal disorder' and 'Retinal vascular disorder' highlighted. The 'Expression Trace' view at the bottom shows a sequence of concepts: 'Injury of thoracic cavity' → 'Internal injury of chest, abdomen and pelvis' → 'Internal injury of chest, abdomen or pelvis'. The 'Search Ontology' view on the right shows a search for 'chest abdd' with a filter set to 'Clinical finding'. The 'Expression Trace' view also shows a search for 'chest abdd' with a filter set to 'Clinical finding'.

Source Term	Relationship	Snomed Expression	Status
Extremity/ multiple trauma, surgery for	Equivalent	410771003 Surgical procedure for clinical finding	Incomplete
Knee replacement (non-traumatic)	Equivalent	19063003 Arthroplasty of knee (procedure)	Finished

Results from ANZICS mapping

- 396 terms in the ANZICS sub-diagnosis codes
- 164 one-to-one equivalence mappings
- 59 concepts were new child concepts
- 4 concepts were new parent concepts

→ Understanding the source terminology is important

Results from ANZICS mapping

- “Extremity/multiple trauma, surgery for”



- Dilation (without general anaesthesia)

Ontology: tag.aehrc.com,2009-01-31:SnomedCT Empty mappings: 0/396

Source Term	Relationship	Snomed Expression	Status	Comments
Devices for spine fracture/ dislocation	Specialise	303624006 Musculoskeletal device (Finished	
Diabetic hyperglycaemic hyperosmol	Equivalent	371632003 Coma : 363702006 H	Finished	
Diabetic ketoacidosis	Equivalent	420422005 Ketoacidosis in diabetes	Finished	
Dilation (with general anaesthesia)	Equivalent	71025006 Dilation procedure : 363	Finished	
Dilation (without general anaesthesia)	Specialise	71025006 Dilation procedure (proce	Finished	shows need to refer to source to get heart focus
Diverticular disease	Equivalent	397881000 Diverticular disease (dis	Finished	
Diverticular disease, surgery for	Equivalent	397881000 Diverticular disease (dis	Finished	
Drug withdrawal	Equivalent	363101005 Drug withdrawal (disord	Finished	

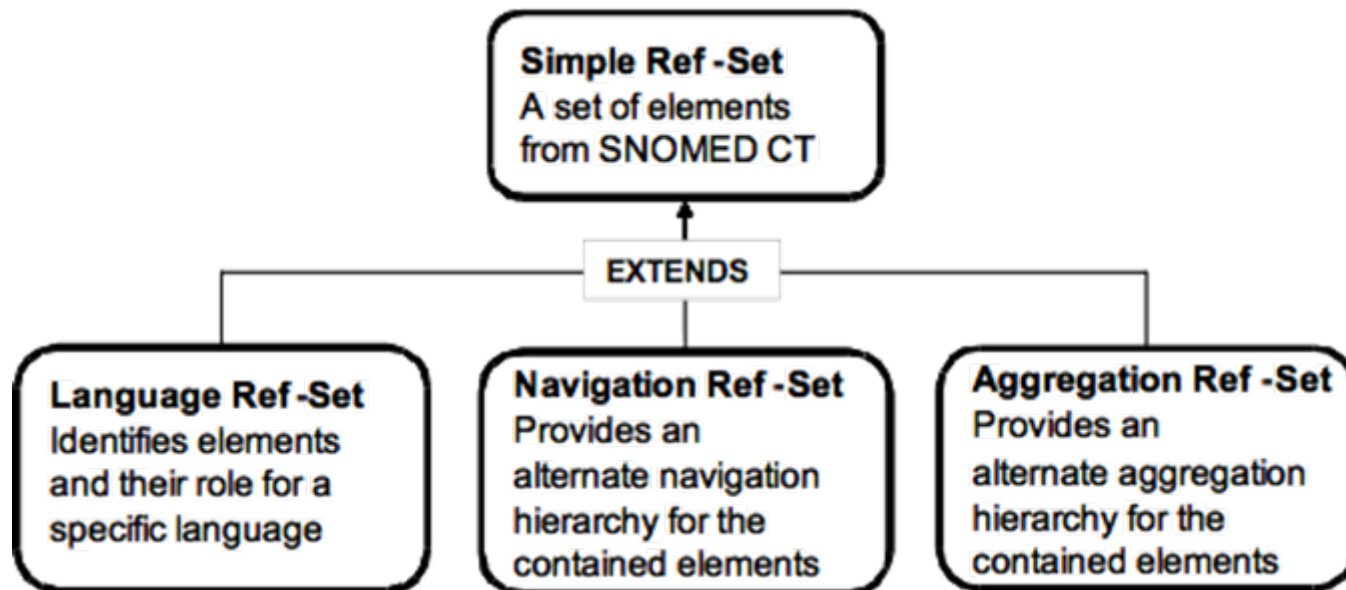
Expression Editor: 71025006 | Dilation procedure (procedure) | : 363702006 | Has focus (attribute) | = 400047006 | Peripheral vascular disease (dis

Graphical Expression: Dilation procedure, Method, Dilation - action, Has focus, Peripheral vascular disease

Other use-cases for Snapper

- Reference Set creation
 - directly from SNOMED
 - based on a previously created mapping
- Text and Image Annotation

Reference Sets for interfaces



Creating a RefSet from a Mapping

Snapper - Demo/Mapping.mapping - Eclipse SDK

Mapping.mapping | GeneOntology.mapping | ANZICS RefSet3.refset

anzics subcodes Simple (Simple)

Id	Component Id	Status	E...e	Annotation
17266470	392031002 Endarterectomy (procedure)	Current		from 'Endarterectomy (
23774816	401314000 Acute non-ST segment elevation myocardial infarction (dis	Current		from 'Infarction, acute
82788067	429595007 Resection of neoplasm of heart (procedure)	Current		from 'Tumour removal,
65958733	236886002 Hysterectomy (procedure)	Current		from 'Hysterectomy for
12838185	108022006 Kidney excision (procedure)	Current		from 'Nephrectomy(oth
77965261	13619001 Thyroidectomy (procedure)	Current		from 'Thyroidectomy a
20180726	65111004 Pelvic exenteration, male (procedure)	Current		from 'Exenteration, pel
26296742	90199006 Transurethral prostatectomy (procedure)	Current		from 'TURP, transureth
08997925	116241004 Pancreaticoduodenectomy (procedure)	Current		from 'Whipple surgery
94155330	38102005 Cholecystectomy (procedure)	Current		from 'Cholecystectomy
25049727	53304009 Parathyroidectomy (procedure)	Current		from 'Thyroidectomy a

Reference Set Group: anzics subcodes Simple (72378870)

Expression Editor | Check Constraints | Graphical Expression

Substance
Situation with explicit context
Organism

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Snapper Reference Set Builder

Snapper - ANZICS_Mapping_Project/ANZICS RefSet3.refset - Eclipse SDK

Project Explorer: ANZICS_Mapping_Project [s]
 Demo
 Demo_1
 Demo2
 DemoOLD
 MappingExamples

Outline: ANZICS
 anzics subcodes Simple
 ANZICS_Diagnosis Simple

anzics subcodes Simple (Simple)

Id	Component Id	Status	Effective Time	Annotation
17266470	392031002 Endarterectomy (procedure)	Current		from 'Endarterectomy
23774816	401314000 Acute non-ST segment elevation myocardia	Current		from 'Infarction, acute
82788067	429595007 Resection of neoplasm of heart (procedure)	Current		from 'Tumour remova
65958733	236886002 Hysterectomy (procedure)	Current		from 'Hysterectomy fo
12838185	108022006 Kidney excision (procedure)	Current		from 'Nephrectomy(o
77965261	13619001 Thyroidectomy (procedure)	Current		from 'Thyroidectomy
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08997925	116241004 Pancreaticoduodenectomy (procedure)	Current		from 'Whipple surgen
94155330	38102005 Cholecystectomy (procedure)	Current		from 'Cholecystectom
25049727	53304009 Parathyroidectomy (procedure)	Current		from 'Thyroidectomy
68205534	34646007 Pericardiectomy (procedure)	Current		from 'Pericardiectom
00893781	22298006 Myocardial infarction (disorder)	Current		from 'MI admitted >
67467590	55853002 Pelvic exenteration, female (procedure)	Current		from 'Exenteration, p
11916790	80146002 Appendectomy (procedure)	Current		from 'Appendectomy

Reference Set Group: anzics subcodes Simple (72... ANZICS_Diagnosis Simple (5... Navigation (64067793)

Search Ontology: tag:aeherc.com,2009-01-31:SnomedCT
 Filter:
 Search: kidn exci
 Kidney excision (procedure)
 Total nephrectomy (procedure)
 Partial nephrectomy (procedure)
 Kidney - local excision (procedure)
 Excision of cyst of kidney (procedure)

ID: 108022006 Defn
 Procedure
 Method
 Excision - action
 Procedure site - Direct
 Kidney structure

Expression Trace Properties Ontology View Console

Selected Object: Ref Set Member 12838185

Diagram illustrating the relationship between various medical procedures and the selected object (Kidney excision):

```

graph LR
    TrunkExcision[Trunk excision] --> KidneyExcision[Kidney excision]
    ProcedureOnUrinarySystem[Procedure on urinary system] --> KidneyExcision
    AbdominalCavityOperation[Abdominal cavity operation] --> KidneyExcision
    RemovalFromUrinaryTract[Removal from urinary tract] --> KidneyExcision
    OperationOnUrinarySystem[Operation on urinary system] --> KidneyExcision
    ExcisionOfPelvis[Excision of pelvis] --> KidneyExcision
    AbdomenExcision[Abdomen excision] --> KidneyExcision
    ProcedureOnKidney[Procedure on kidney] --> KidneyExcision
    KidneyOperation[Kidney operation] --> KidneyExcision
    TotalNephrectomy[Total nephrectomy] --> KidneyExcision
    Nephroureterectomy[Nephroureterectomy] --> KidneyExcision
    IncisionalBiopsyOfKidney[Incisional biopsy of kidney] --> KidneyExcision
    WedgeBiopsyOfKidney[Wedge biopsy of kidney] --> KidneyExcision
    RecipientNephrectomy[Recipient nephrectomy] --> KidneyExcision
    UnilateralNephrectomy[Unilateral nephrectomy] --> KidneyExcision
    DiverticulectomyOfKidney[Diverticulectomy of kidney] --> KidneyExcision
    PartialNephrectomy[Partial nephrectomy] --> KidneyExcision
    ExcisionOfLesionOfKidney[Excision of lesion of kidney] --> KidneyExcision
  
```

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Simple Reference Sets

The screenshot displays the Eclipse IDE with the Snapper application open. The main window shows the 'Colonoscopy Example.refset' project. A table lists concepts with their IDs, component IDs, and annotations. The 'Search Ontology' window is active, showing a search for 'ulc coli' and a list of results including 'Ulcerative colitis (disorder)'. The 'Graphical Expression' window shows a hierarchical diagram of 'Ulcerative colitis' with its associated morphologies and findings. The 'Expression Trace' window at the bottom shows a flow from 'Intestine' to 'Colitis' to 'Chronic colitis' to 'Ulcerative colitis'.

Id	Component Id	Status	Effective Time	Annotation
9551124	34000006	Current		Crohn's disease (disorder)
7318244	64766004	Current		Ulcerative colitis (disorder)
2411618	196990002	Current		Ulcerative proctocolitis NOS
3237177	414156000	Current		Exacerbation of ulcerative colitis
9554049	78712000	Current		Moderate chronic ulcerative colitis
7127426	13470001	Current		Chronic ulcerative ileocolitis
9850739	410484008	Current		Iritis with ulcerative colitis
2081086	78324009	Current		Mild chronic ulcerative colitis

Reference Set Group: Colonoscopy Interface Concepts (25032006)

Search Ontology: tag:aeherc.com,2009-01-31:...

Filter: Clinical finding

Search: ulc coli

Ulcerative colitis (disorder)

- Acute ulcerative colitis (disorder)
- Chronic ulcerative proctitis (disorder)
- Iritis with ulcerative colitis (disorder)
- Eosinophilic ulcerative colitis (disorder)
- Mild chronic ulcerative colitis (disorder)
- Arthropathy in ulcerative colitis (disorder)

ID: 64766004 Defn

- Ulcerative colitis
- Associated morphology
- Chronic
- Finding site
- Colon structure
- Clinical course
- Chronic
- Associated morphology
- Chronic

Expression Trace: Intestine → Colitis → Chronic colitis → Ulcerative colitis

Ulcerative proctocolitis NOS (disorder)

Exacerbation of ulcerative colitis

Moderate chronic ulcerative colitis

Chronic ulcerative ileocolitis

Iritis with ulcerative colitis

Mild chronic ulcerative colitis

Chronic ulcerative rectosigmoiditis

Severe chronic ulcerative colitis

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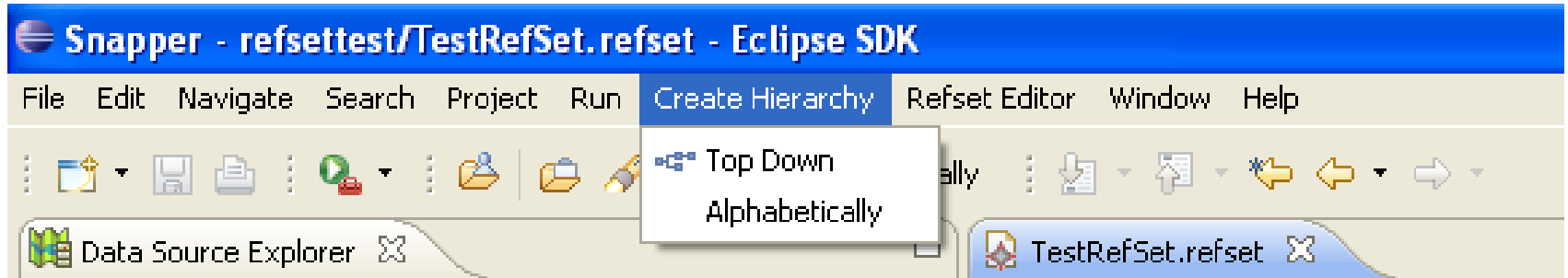
Uses for Simple RefSets

- Limit the terms/concepts which can be used in application
 - Small reference sets for use as pick lists in an application
- Use as filter for searching
 - limit to specific (sub-)hierarchy
 - exclude specific concepts
 - unsuitable for use
 - unnecessarily specific
 - ...
- Standardise concepts for a specific domain

Aggregation Reference Sets

- Ideal for reporting use
- Can help to report as ICD from data captured in SNOMED CT
 - Multiple SNOMED CT codes may
- *"Aggregation Reference Sets would allow the distribution of alternative aggregation hierarchies for SNOMED Concepts."*
 - IHTSDO Reference Set Guide, version 1

Reference Sets with Hierarchies



- 73761001 | Colonoscopy | :
363703001 | Has intent | = 262202000 | Therapeutic intent |
 {
 405813007 | Procedure site - Direct | = 71854001 | Colon structure |,
 424226004 | Using device | = 90412006 | Colonoscope, device |,
 260686004 | Method | =129433002 | Inspection - action |
 }

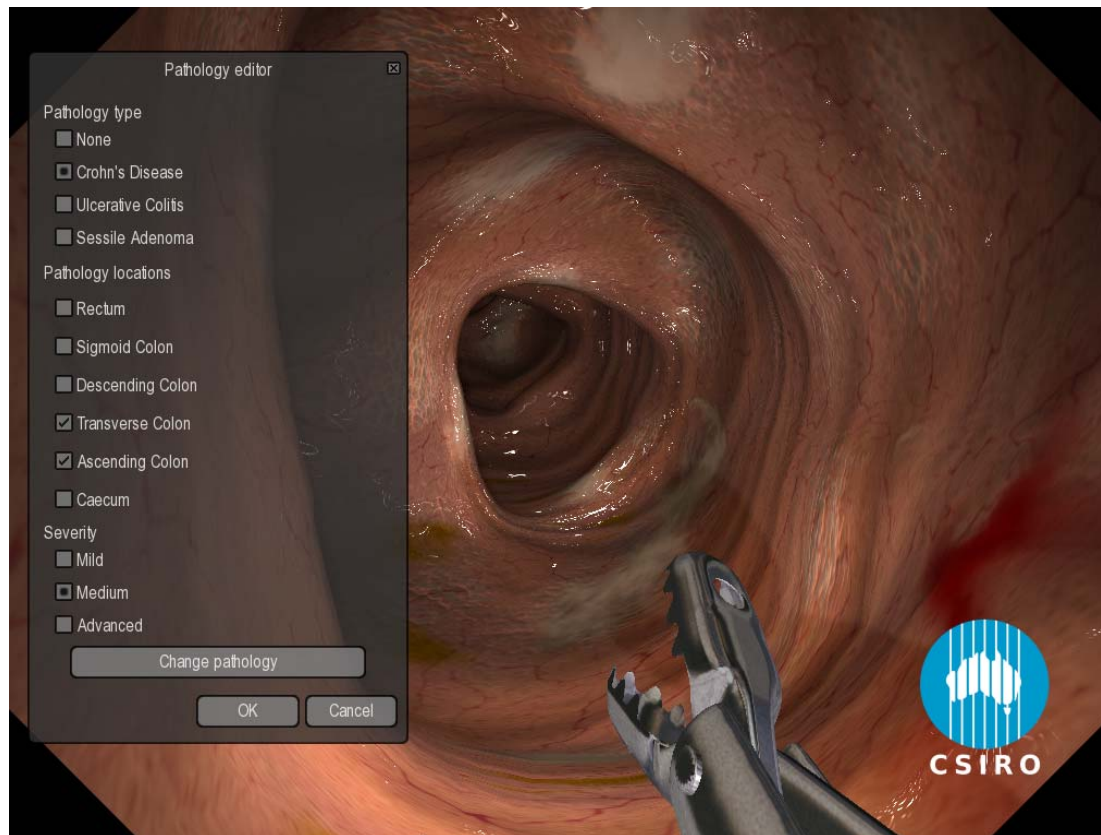
Navigation Reference Sets

- Can be used to drive an application

Using the Hierarchy to drive an applications

- Accessing a Reference Set from ontoserver
 - Retrieve information about a concept
 - <http://ict-ehrcsl:8080/ontoserver/resources/ontology/tag:aehrc.com,2009-07-29:BowelCancerRefSet/concept/2>
 - Retrieve children of a concept:
 - <http://ict-ehrcsl:8080/ontoserver/resources/ontology/tag:aehrc.com,2009-07-29:BowelCancerRefSet/concept/2/children>
 - Retrieve parents of a concept
 - <http://ict-ehrcsl:8080/ontoserver/resources/ontology/tag:aehrc.com,2009-07-29:BowelCancerRefSet/concept/2/parents>

Using the Hierarchy to drive an applications



Proposed Release Format 2.0 Refset Spec

- Much broader scope
- Still work in progress
- Allows for query-based inclusion as well
 - has implications (positive and negative) for version management
- Aggregation and Navigation Refsets no longer explicitly mentioned
 - may (?) be represented using *Association reference set (pattern)*

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