IHE/HL7 Anatomic Pathology
Joint with Vocabulary

C. Daniel
San Diego
September 13, 2011
Anatomic Pathology Structured Report (APSR)
[APSR] – Anatomic Pathology Structured Report

- 21 CDA templates (Document Content Modules)
  - Generic APSR template
    - All organs & fields of anatomic pathology (inflammatory, vascular, traumatic, metabolic diseases as well as cancer)
  - 20 organ-specific APSR templates
    - Cancer-specific organizers covering 85% of incident cancers
- 490 observations & procedure templates
  - 21 procedure templates
  - 469 observation templates (including 73 TNM observation templates)
Structure: Common hierarchy for all APSR document content modules

- **Header**
  - Reason for AP procedure section [0..1]
  - History of present illness section [0..1]
  - Active problems section [0..1]

- **Structured Body**
  - Clinical information section [0..1]
  - Specimen clinical information entry [0..*]
  - Intraoperative observation section [0..1]
  - Specimen intraoperative observation entry [0..*]
  - Macroscopic observation section [0..1]
  - Specimen macroscopic observation entry [0..*]
  - Microscopic observation section [0..1]
  - Specimen microscopic observation entry [0..*]
  - Report textual summary section [0..1]
  - Diagnosis section [1..1]
  - Specimen diagnosis entry [1..*]
  - Procedure steps section [0..1]
Structure & vocabulary
Element Content Modules (n=11)

- Header (n=5)
- Body (n=5)
  - Describing Anatomic Pathology observations grouped per specimen and per problem
    - Specimen Information Organizer
    - Specimen Collection Procedure (n=21)
      - the characteristics of the specimen (identifiers and type)
      - the procedure that collected it
        - Type of procedure, time interval, performer (person and organization), approach site, target site.
  - Problem Organizer
  - AP Observation (n=469)
    - including TNM observation templates (n=73)
  - Embedded Image
Structure & vocabulary: Example
Anatomic Pathology Observation (data element)

- Breast-In situ neoplasm-Histologic type (template ID 1.3.6.1.4.1.19376.1.8.1.4.446)
  - observation.code : Pathlex code = 436
  - observation.value (Concept Descriptor (CD))
    - Value set OID: 1.3.6.1.4.1.19376.1.8.5.254

<table>
<thead>
<tr>
<th>PathLex code (finding)</th>
<th>Label</th>
</tr>
</thead>
<tbody>
<tr>
<td>2308</td>
<td>Ductal carcinoma in situ with microinvasion</td>
</tr>
<tr>
<td>2309</td>
<td>Lobular carcinoma in situ with microinvasion</td>
</tr>
<tr>
<td>2557</td>
<td>DCIS Comedo</td>
</tr>
<tr>
<td>....</td>
<td>....</td>
</tr>
<tr>
<td>2562</td>
<td>DCIS Solid</td>
</tr>
</tbody>
</table>
Vocabulary constraints

- Available
  IHE_PAT_Suppl_APSR_AppendixValue_Sets - http://www.ihe.net (excel file)

- Scope: Element Content Modules
  - Specimen collection procedure
  - AP observation
Vocabulary constraints

- Also available through Web services
  - STS (Standard Terminology Server)

- About 20 « read only » services are available
- Testables via STS web site

CTS2

[Images of PDF, Adobe, RDF, SKOS, OWL]
PathLex
PathLex, a single lexicon in the Anatomic Pathology domain

- Launched by IHE Anatomic Pathology
  - Collaboration of College of American Pathologists (CAP), ADICAP, French Society of Pathologists (SFP), SEAP (Spanish Society of Pathology).
- Registered as external terminology used by HL7

- Purpose and scope (very similar to those of the RadLex project)
  - “designed to satisfy the needs of Anatomic Pathology information system vendors and users by adopting the best features of existing terminology systems
    - using if possible available concepts defined in reference biomedical terminologies or ontologies like SNOMED-CT or CIM-O (rather than “re-inventing the wheel”)
    - while producing new terms to fill critical gaps.
PathLex

Current status

- **Structure**
  - Permanent identifiers (codes) are meaningless
  - PathLex preferred terms are organized into a is-a hierarchy
    - Histological type
      - Histological type of breast neoplasm
        - Histological type of in situ neoplasm of the breast
  - Multilingual: universal value sets include all possible values available in the local extensions.
    - Common values are therefore available in multiple languages (currently English and French).

- **Open access**
  - “Appendix Value Sets for APSR” as part of the IHE content profile “Anatomic Pathology Structured Report” (APSR) (https://ihe.net)
  - STS (PHAST, France) (CTS2 services)
SNOMED CT in Pathology

Marcial García Rojo
Hospital General Universitario de Ciudad Real, Spain
Arvydas Laurinavicius
National Centre of Pathology, Vilnius, Lithuania
Vytenis Punys
Kaunas University Technology, Lithuania
Stephanie Gehring
School Medical Documentation, Ulm, Germany
Why is it useful? Sharing/exchanging/mining AP data

- Free text reports
  - Coding specimens, anatomy and diagnosis
- Structured reports
  - Coding both the question and the answer
    - Question: Observable entity (Histological grade?)
    - Answer: Diagnosis and qualifiers
Why is it useful? Sharing/exchanging/mining AP images

- Specimen Obtained (types, anatomic location, collection procedure)
- Specimen processing (sampling procedure, preparation procedure, stains, fixatives, embedding)
- Diagnosis...

**Case ID (official): 09B0012219**

**Detailed diagnose:**
BIOPSIA DE PIEL DE REGIÓN DELTOIDEA: PIEL CON HIPERPLASIA FOLLICULAR LINFOIDE REACTIVA. (VER COMENTARIO). COMENTARIO: El cuadro histológico de esta biopsia es superponible al de la biopsia 07B5955. Se envió como caso de consulta al Dr. Pris del CNIO quien confirmó el diagnóstico emitido. INFORME COMPLEMENTARIO 14.12.09: El estudio de PCR realizado en el CNIO muestra un patrón de reordenamiento policional para IgH (CDR2, CDR3) y TCR (G1, G2, B1, B2). No se identifican reordenamientos de BCL2 (MBR y 3'MBR). Estos datos apoyan el diagnóstico de hiperplasia folicular linfoides reactiva.

**Local code:** M01068

**SNOMED CT - FSN Spanish:** hiperplasia linfoides (anomalía morfológica)

<table>
<thead>
<tr>
<th>Case ID (entered)</th>
<th>Digital slide (Intranet)</th>
<th>Digital slide (Internet)</th>
</tr>
</thead>
<tbody>
<tr>
<td>09B-12219-1</td>
<td></td>
<td></td>
</tr>
<tr>
<td>09B-12219-CD3</td>
<td></td>
<td></td>
</tr>
<tr>
<td>09B-12219-L26</td>
<td></td>
<td></td>
</tr>
<tr>
<td>09B-12219-Bcl-2</td>
<td></td>
<td></td>
</tr>
<tr>
<td>09B-12219-kI67</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

**Comment:** It would also be possible to additionally retrieve the corresponding SNOMED CT code per case.
### CAP Anatomic Pathology Subset

<table>
<thead>
<tr>
<th>Hierarchy</th>
<th>Approximate Count</th>
</tr>
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<tbody>
<tr>
<td>Body structure</td>
<td>3,805</td>
</tr>
<tr>
<td>Clinical Finding</td>
<td>2,024</td>
</tr>
<tr>
<td>Procedure</td>
<td>780</td>
</tr>
<tr>
<td>Observable entity</td>
<td>386</td>
</tr>
<tr>
<td>Specimen</td>
<td>264</td>
</tr>
<tr>
<td>Pharmaceutical / Biologic product</td>
<td>83</td>
</tr>
<tr>
<td>Record artifact</td>
<td>67</td>
</tr>
<tr>
<td>Organism</td>
<td>61</td>
</tr>
<tr>
<td>Staging and scales</td>
<td>33</td>
</tr>
<tr>
<td>Substance</td>
<td>24</td>
</tr>
<tr>
<td>Qualifier value</td>
<td>21</td>
</tr>
<tr>
<td>Special concept</td>
<td>12</td>
</tr>
<tr>
<td>Events</td>
<td>9</td>
</tr>
<tr>
<td>Situation with explicit context</td>
<td>9</td>
</tr>
<tr>
<td>Physical object</td>
<td>4</td>
</tr>
<tr>
<td>TOTAL CONCEPTS (Jan 2010)</td>
<td>7,582</td>
</tr>
</tbody>
</table>

In daily practice, this subset was not considered useful by pathologists:

- Mixture of different hierarchies (clinical findings, body structure, observations,..)
- Lack of basic pathology diagnosis
Implementation of SNOMED CT

Gap analysis

- IHE Anatomic Pathology Technical Framework Supplement Anatomic Pathology Structured Reports (APSR) Closed Issues:

- APSR-14 – Gaps in SNOMED CT: It is not straightforward to encode Anatomic Pathology observations and their corresponding value sets described in Volume IV (Value Sets for APSR) using SNOMED CT concepts (missing concepts, issues of postcoordination versus precoordination).

- Therefore these observations and value sets are encoded using a coding system currently being built by the IHE Anatomic Pathology domain (PathLex - OID: 1.3.6.1.4.1.19376.1.8.2.1). PathLex codes are provided with the “Trial Implementation” version of this profile.

- Using SNOMED CT as a reference terminology offers promising perspectives. The terms and expressions of PathLex will be mapped to SNOMED CT concepts in collaboration with IHTSDO. This mapping will be available in the “Trial Implementation” version and completed over time.
Implementation of SNOMED CT in Hospital General de Ciudad Real

- **IHE: Structured Reports Value Sets** (1,840 possible values for observations)
- **HGUCR: 2,320 pathology diagnosis codes mapped to SNOMED CT**
- Search for descriptions that match the local legacy codes was performed using CliniClue Xplorer version 2010 1.243, using SNOMED CT 2011-01-31 International Release and 2010-04-30 Spanish Edition.
Search strategy for AP diagnosis

- Morphologically abnormal structure
  - Searching for a unique ConceptId identifier/SNOMED Clinical Terms Identifier (SCTID) from the body structure (morphologically abnormal structure sub-hierarchy)

- Clinical finding

- Post-coordination
  - Combination of two or more morphologically abnormal structures
  - Combination of qualifiers and a morphologically abnormal structure or a clinical finding
  - Any other combination of terms
Results

- Morphology – Abnormal body structure hierarchy
  - Only 44% of the codes! (1076)
- Disorder hierarchy
  - 48% of the codes
    - In 1104 terms, using “disorder” hierarchy, e.g. Rosacea
    - In 71 terms, using “finding” hierarchy, e.g. World Health Organization (WHO) grade I (central nervous system tumor)
- Postcoordination needed in 19%
  - 3 conceptID needed only rarely
Results

Other hierarchies

- 6% of the local morphological diagnosis that were not well represented using either morphological abnormality or disorder/finding hierarchies

- Usefull SNOMED CT hierarchies
  - Qualifiers (generally combined with clinical or morphological codes) (95 local terms), e.g. Granulomatous
  - Body structures was found useful (68 local terms), e.g. Undescended testis
  - Other: procedures, physical object, substance, organism
Conclusions

- SNOMED CT morphology hierarchy (and other hierarchies of interest in AP) SHOULD be improved
- Pathologists SHOULD become aware of the structure and contents of SNOMED CT
- Pathologists SHOULD become aware of the shift of AP coding process (from Topography, Morphology schema to templates & binding with a polihierarchy and relationship schema)
- Pathologists SHOULD define top level rules for SNOMED CT coding process
- Pathology IS SHOULD allow coding with local terms and mappings to SNOMED CT
Mapping PathLex to SNOMED CT

Preliminary study

Christel Daniel
AP-HP – Paris Descartes University – INSERM

Olivier Bodenreider, Bastien Rance
NLM
Mapping interface terminologies (legacy codes) to SNOMED CT

- CSIRO (Commonwealth Scientific and Industrial Research Organisation) (Hansen, 2011)
- NLM, Bioportal
Mapping PathLex to SNOMED CT (using UMLS)

PathLex Short label
1781

TNM
432

No TNM
1349

UMLS Search
EM/NM
CUI

No CUI

Split label
No new tokens
New tokens

No new tokens
No CUI

New tokens
CUI

Metamap
SNOMED CT

To be explored

Exact match
SNOMED CT
No SNOMED CT

Partial match
SNOMED CT
No SNOMED CT
## Results of the automatic mapping process

<table>
<thead>
<tr>
<th>Matching situations</th>
<th>Number of labels</th>
<th>Percentages of labels</th>
</tr>
</thead>
<tbody>
<tr>
<td>Labels mapped to SNOMED CT through exact match (EM) or normalized match (NM) to UMLS</td>
<td>609</td>
<td>45%</td>
</tr>
<tr>
<td>Labels mapped to another terminology through exact match (EM) or normalized match (NM) to UMLS</td>
<td>79</td>
<td>6%</td>
</tr>
<tr>
<td>Tokens mapped to SNOMED CT through exact match (EM) or normalized match (NM) to UMLS</td>
<td>232</td>
<td>17%</td>
</tr>
<tr>
<td>Tokens mapped to another terminology through exact match (EM) or normalized match (NM) to UMLS</td>
<td>25</td>
<td>2%</td>
</tr>
<tr>
<td>Tokens without any match</td>
<td>80</td>
<td>6%</td>
</tr>
<tr>
<td>Labels without any match and that cannot be split in tokens</td>
<td>324</td>
<td>24%</td>
</tr>
<tr>
<td>TOTAL</td>
<td>1349</td>
<td>100%</td>
</tr>
</tbody>
</table>
# Examples of PathLex labels/expressions with automatic mappings

<table>
<thead>
<tr>
<th>Categories of observations</th>
<th>PathLex label</th>
<th>CUI</th>
<th>SNOMED CT code</th>
</tr>
</thead>
<tbody>
<tr>
<td>Examples of AP macroscopic observation types related to the specimen</td>
<td>Specimen size, largest dimension</td>
<td>C1273739</td>
<td>384627007</td>
</tr>
<tr>
<td></td>
<td>Specimen size, additional dimension</td>
<td>C1273738</td>
<td>384626003</td>
</tr>
<tr>
<td>Examples of AP microscopic observation types related to a lesion related to a lesion</td>
<td>Lesion size, largest dimension</td>
<td>C1275593</td>
<td>396361002</td>
</tr>
<tr>
<td></td>
<td>Lesion site</td>
<td>C0449685</td>
<td>246300000</td>
</tr>
<tr>
<td></td>
<td>Histologic type</td>
<td>C0449574</td>
<td>263541007</td>
</tr>
<tr>
<td></td>
<td>Histologic grade</td>
<td>C0919553</td>
<td>371469007</td>
</tr>
<tr>
<td></td>
<td>Margins involvement</td>
<td>C1269794</td>
<td>371488000</td>
</tr>
<tr>
<td>Types of ancillary techniques</td>
<td>Label</td>
<td></td>
<td></td>
</tr>
<tr>
<td>-------------------------------</td>
<td>-------</td>
<td></td>
<td></td>
</tr>
<tr>
<td>HER2/neu (FISH method)</td>
<td></td>
<td></td>
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</tr>
<tr>
<td>Mismatch Repair Proteins-MLH1 (Immunohistochemistry Study)</td>
<td></td>
<td></td>
<td></td>
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<tr>
<td>Types of histologic grades</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Histologic grade (Clark)</td>
<td></td>
<td></td>
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<tr>
<td>Histologic grade (Gleason-Primary (Predominant) Pattern)</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Histologic grade (Gleason-Total Gleason Score)</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Extension</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Number of lymph nodes with isolated tumor cells ($\leq 0.2$ mm and $\leq 200$ cells)</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Anatomic location</td>
<td>Anterior floor of mouth (qualifier : right, left, medial)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>----------------------------</td>
<td>----------------------------------------------------------</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Distal esophagus</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Histologic types</td>
<td>Atelectasis Extends to the hilar region but does not involve entire lung</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Atypical squamous cells for which a high-grade lesion cannot be excluded (ASC-H)</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Cirrhosis/severe fibrosis (Ishak score 5-6) (F1)</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Combined small cell carcinoma (small cell carcinoma and non-small cell component)</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Complex hyperplasia without cytologic atypia</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>DCIS Comedo</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Ductal carcinoma in situ involving nipple skin (Paget disease) with microinvasion</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Histologic grades</td>
<td>FIGO grade 1</td>
<td></td>
<td></td>
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<tr>
<td>-------------------</td>
<td>-------------</td>
<td></td>
<td></td>
</tr>
<tr>
<td>G1: Nuclei round, uniform, approximately 10 mm; nucleoli inconspicuous or absent</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Low-grade squamous intraepithelial lesion encompassing HPV infection or mild dysplasia (CIN 1)</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Score 2: 10% to 75% of tumor area forming glandular/tubular structures</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Extension</th>
<th>&lt;50% myometrial invasion</th>
</tr>
</thead>
</table>

<table>
<thead>
<tr>
<th>Results of AP ancillary techniques</th>
<th>Amplified (HER2 gene copy &gt;6.0 or ratio &gt;2.2)</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Equivocal (HER2 gene copy 4.0 to 6.0 or ratio 1.8 to 2.2)</td>
</tr>
<tr>
<td></td>
<td>Immunoreactive tumor cells present (&gt; = 1%) (Specify Quantitation)</td>
</tr>
<tr>
<td></td>
<td>Mild to moderate (0-2 per high-power [X400] field) Intratumoral Lymphocytic Response (tumor-infiltrating lymphocytes)</td>
</tr>
<tr>
<td>TNM values</td>
<td>Label</td>
</tr>
<tr>
<td>------------</td>
<td>-------</td>
</tr>
<tr>
<td>pM1c: Metastasis to all other visceral sites or distant metastasis at any site associated with an elevated serum lactic dehydrogenase (LDH)</td>
<td></td>
</tr>
<tr>
<td>pN2: Metastasis in a single ipsilateral lymph node, more than 3 cm but not more than 6 cm in greatest dimension, or in multiple ipsilateral lymph nodes, none more than 6 cm in greatest dimension, or in bilateral or contralateral lymph nodes, none more than 6 cm in greatest dimension</td>
<td></td>
</tr>
<tr>
<td>pT2a: Tumor greater than 3 cm, but 5 cm or less in greatest dimension surrounded by lung or visceral pleura, without bronchoscopic evidence of invasion more proximal than the lobar bronchus (ie, not in the main bronchus); or Tumor 5 cm or less in greatest dimension with any of the following features of extent: involves main bronchus, 2 cm or more distal to the carina; invades the visceral pleura; associated with atelectasis or obstructive pneumonitis that extends to the hilar region but does not involve the entire lung</td>
<td></td>
</tr>
</tbody>
</table>
PathLex as a thematic extension of SNOMED CT? Next steps

- A joint IHE/HL7 Anatomic Pathology – IpaLM initiative
  - Governance & technical issues (tooling) for the management of PathLex.

- IPALM SIG
  - Rajesh Dash, M.D. (r.dash@duke.edu)
  - Andrea Pitkus, CAP (apitkus@cap.org)
  - Technical assistance IHTSDO
    - Yohani Daruis (yda@ihtsdo.org)(support@ihtsdo.org)

- IPALM collaborative site
  - IC0604 members
    - Thomas Schrader (Germany) thomas.schrader@computer.org
    - Bernd Blobel (Germany) bernd.blobel@klinik.uni-regensburg.de
    - Christel Daniel (France) christel.daniel@spim.jussieu.fr
    - Vincenzo Della Mea (Italy) vincenzo.dellamea@uniud.it