

# Towards a Standard for Anatomy in Health informatics

*Workshop presentation in Balatonfüred  
for the Semantic Mining Summer School*

Dr Gunnar O. Klein, M.D., Ph.D.

*chairman of CEN/TC 251 & eHSCG*

Dept of Medicine, Core facility for Medical Informatics,  
Karolinska Institutet, Stockholm, Sweden

# Anatomy of Standardization

## **Semantic Mining Workpackage 8 Participation in Standardisation Work**

- **To facilitate information exchange between the project and formal standardization in CEN and ISO as well as other relevant groups**

# Workpackage 8

## Participation in Standardization Work

- Promoting the building of information exchange standards that build on research in ontologies and knowledge representation to enable
  - safe integration for the care of the individual
  - efficient analysis of accumulated health information to generate new knowledge  
(Clinical research as well as functional genomics)

# Standard (definition by ISO/IEC)

**Document**, established by **consensus** and approved by a **recognised body**, that provides, for common and repeated use, rules, guidelines or characteristics for activities or their results, aimed at the achievement of the **optimum degree of order in a given context**

# Different types of standards bodies

- **National formal standards bodies**
  - DIN, AFNOR, UNI, BSI, DS, SIS etc
- **ISO (ITU and IEC)**
  - International Organization for Standardization
- **CEN**
  - European Committee for Standardization
- **Informal standards bodies**
  - Often commercial consortias, very important in IT
  - Examples: W3C, Open Group, IETF, Bluetooth, OASIS

# The role of the National Standards Bodies

- **Be a national focal point for interested parties when there is an ongoing international standardization**
  - National experts are nominated to international working groups and the NSB representatives votes for the country in ISO and CEN
- **Publish, distribute and promote standards to users of standards in the country**
  - This may include translation to national language
- **Develop specific national standards for national problems or when no international work exists**
  - Even when international work can be used there is often the need to develop specific national implementation guidelines or profiles restricting the number of optional possibilities of international standards

# ISO

## International Organization for Standardization

**142 countries**  
**2 800 TCs&WGs**  
**12 000 standards**  
**30 000 experts**



# CEN

## European Committee for Standardization

- **Started 1961**
- **28 member countries**
- **6 affiliates**
- **Used by EU in many cases**
- **CEN is mandatory for the countries**



In Europe we decided in 1990 that many of the issues that needed standards for health informatics would best be solved on a European scale rather than national

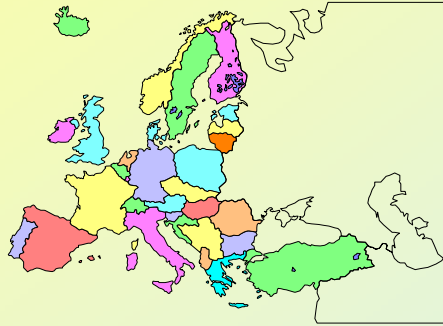


There was no international work and it was felt that there was a need to support **one internal market** for IT products in all of the European Union.

There is also a large number of citizens moving between these countries for work, studies and vacation. **Interoperability** of health information systems was desired



# Standardization of Health Informatics in Europe



***CEN = Comité Européen de Normalisation  
European Committee for Standardization***

**28 EU and EFTA countries are members + 6 affiliate observers**

## **CEN/TC 251**

**Technical Committee on Health Informatics**

**Secretariat is managed by SIS - Swedish standards institute  
[www.centc251.org](http://www.centc251.org)**



# **A political mandate and financial support from EU and EFTA**

- **EU and EFTA policies have given CEN a mandate to produce standards for Healthcare Informatics in Europe**
- **European funding has been extremely important in speeding up the process**

# Scope of CEN/TC 251

**Standardization in the field of Health Information and Communications Technology (ICT) to achieve compatibility and interoperability between independent systems and to enable modularity.**

**This includes requirements on health information structure to support clinical and administrative procedures, technical methods to support interoperable systems as well as requirements regarding safety, security and quality**

# CEN/TC 251

## working groups and convenors

- **I: Information models**
  - Gerard Freriks, Netherlands
- **II: Terminology and knowledge representation**
  - Magnus Fogelberg, Sweden
- **III: Security, Safety and Quality**
  - Gilles Trouessin, France
- **IV: Technology for interoperability**
  - Melvin Reynolds, UK

# New political developments for eHealth

- **The European Commission issued a Communication as a Follow-up on patient mobility**  
(COM(2004) 301, 2004-04-20)
- "EU law gives patients the right to go for treatment in other Member States. But exercising that right can prove difficult. A central aim of our proposal is to explain patients' rights more clearly and ensure they have the information they need to make use of those rights. My final goal is to achieve a European Charter of patients' rights to which everyone can refer." said David Byrne, the European Commissioner for Health and Consumer Protection.

# New political developments

- **The European Commission plan:** (COM(2004) 301, 2004-04-20)
- **Better provision of information to patients on how to obtain treatment in other Member States**
- **Making it easier for national healthcare providers to use spare capacity (such as empty hospital beds) in other Member States;**
- **Networks of health experts and centres of excellence across the EU as well as coordinating assessment of new health technologies;**
- **Systematic exchange of best practice.**
- **A high level group on health services and medical care bringing together Member State representatives and the Commission will be created to drive this process of cooperation. (The open method of co-ordination)**

# e-Health –

**making healthcare better for European citizens:  
An action plan for a European e-Health Area  
Com (356) 2004, 2004-04-30**

## **Interoperability of e-Health systems.**

- Interoperability should enable the seamless integration of heterogeneous systems. This will allow secure and fast access to comparable public health data and to patient information located in different places over a wide variety of wired and wireless devices. However, **this depends on standardisation** of system components and services such as health information systems, health messages, **electronic health record architecture**, and patient identifying services. Work has been launched within European standards organisations to answer this issue partly, **but the take-up of e-Health interoperability standards has been slow and – in addition – to achieve actual interoperability is a separate task.** Interoperable e-Health solutions should also support the technical platform for the implementation of such initiatives as the creation of a European network of centres of reference to promote co-operation across medical institutions across the Community

## Selected targets from COM 356

- By mid 2005, the Commission should produce a summary of European best practices as guidance for Member States.
- By end 2005, each Member State is to develop a national or regional roadmap for e-Health. This should focus on deploying e-Health systems, setting targets for interoperability and the use of electronic health records, and address issues such as the reimbursement of e-Health services.
- By end 2006, Member States, in collaboration with the European Commission, should identify a common approach to patient identifiers. This should take account of best practices and developments in areas such as the European Health Insurance Card and identity management for European citizens.
- By end 2006, Member States, in collaboration with the European Commission, **should identify and outline interoperability standards for health data messages and electronic health records**, taking into account best practices and relevant standardisation efforts.

## Selected targets from COM 356

By end 2005, a European Union public health portal will give access to European level public health information. Health portals shall offer dedicated information on safety at work and health risks in the workplace.

By end 2005, there will be a strengthening of early warning, detection, and surveillance of health threats through enhanced information and communication technologies tools.

Promoting the use of cards in the health care sector. Adoption of implementation of an electronic health insurance card by 2008.

By end 2008, the majority of European health organisations and health regions (communities, counties, districts) should be able to provide online services such as teleconsultation (second medical opinion), e-prescription, e-referral, telemonitoring and telecare.

# CEN/TC 251 achievements-

**A large number of published standards and reports**

- **46 prestandards (ENV or CEN/TS)**
- **8 standards (EN)**
- **9 technical reports**

**A very active work programme  
with both completely new items and  
finalisation to full standards of  
long term work**

- **47 draft standards to become ENs in 2004 or 2005. (10 of them approved awaiting publication)**
- **10 Technical specifications**

# Highlights of achievements

- A new message development methodology based on General Purpose Information Components that are RIM derived
- A new dual model Electronic Health Record architecture that includes archetypes
- An important set of Point of care medical device communication standards jointly with ISO and IEEE
- A basic set of Patient Health Card Standards agreed internationally with ISO

# Highlights of achievements

- **Basic standards for concept based health terminology work including exchange formats in XML**
- **A concept system for continuity of care**
- **Security standards including mechanisms for protection of communication and health specific management**



## **ISO - collaboration with CEN**

**ISO/TC 215 Health informatics was established in 1998 proposed by the US but with strong support from Europe**

# Vienna agreement between CEN and ISO



- There are many examples with thousands of standards processed in collaboration between CEN and ISO.
- The Vienna agreement intends to
  - Avoid duplication of effort and divergence
  - Allow parallel voting process where feasible
- In health informatics a number of European prestandards ENVs have been the starting point for ISO/TC 215 work items
  - Vienna agreement allows the improved ISO documents to be processed in parallel as full European standards.

# European co-operation with HL7

- CEN started a formal co-operation with HL7 under the MoU from the year 2000
- International agreement of the use of the RIM now an ISO work item
- International agreement on Data Types
- CEN experts have learned a lot from HL7
- CEN experts have influenced HL7 in many aspects, major EHRCOM classes now in the RIM

# **CEN/ISSS eHealth Standardization Focus Group [www.CENeHealth.org](http://www.CENeHealth.org)**

**Shall guide the Commission, member states and CEN**

- **What eHealth applications to support ?**
- **What standards exists and should be promoted**
- **What needs to be developed, by whom?**

# Proposed conclusion on the importance of standards

- **Health informatics standards are essential to achieve the goals of eHealth in Europe**
  - **Interoperability between systems and patient information exchange between health care organisations to improve efficiency and quality of care**
  - **Improving patient safety**
  - **Ensuring security meeting legal requirements and protecting privacy of the citizens**
  - **Allowing a growing cross border exchange of health information in Europe for the care of the individual and for collection of population data for public health purposes and biomedical research**

ISO, ITU, IEC, CEN, IEEE, DICOM, HL7, WHO

**Co-operation for better global health**

**The eHealth Standardization  
Co-ordination Group**

# **e-Health Standardization Coordination Group (eHSCG)**

## **Overall objectives**

- **To promote stronger co-operation amongst the key players in the e-Health Standardization area**
- **To promote the use of standards for e-Health**

# Several standards bodies contribute to e-Health

- **E-Health depend on intersector standards**
  - ITU
  - ISO/IEC JTC1
  - IETF
  - W3C
  - ..... (many more also informal consortia do important developments)
- **International bodies with significant work specific for e-Health part of the eHSCG**
  - ISO/TC 215 Health informatics
  - ITU-T/SG 16 Multimedia communication
  - IEC/TC 62 Medical devices (electric aspects)
  - CEN/TC 251 Health informatics
  - IEEE/P1073 Point of Care medical devices
  - HL7 Health care messaging
  - DICOM Medical imaging communication

# World Health Organization

- **WHO eHealth department**
  - with Mr Yunkap Kwankam  
head of the unit  
co-chair the  
eHealth Standardization  
Co-ordination Group
- **The group welcome other  
representatives of the users of  
standards for eHealth**

# e-Health Standardization Coordination Group (eHSCG)

## Terms of Reference

- **Coordination group on all aspects of e-health standardization**
  - Sharing experiences and information on all relevant fields including terminologies
  - Not steering participating parties
- **Strengthen the cooperation amongst the SDOs**
  - Avoiding duplication of efforts
  - Promoting exchange of specifications and experts where relevant
- **Focus on technical aspects taking into consideration regulatory, economic, medical and social issues**

# e-Health Standardization Coordination Group (eHSCG)

## Terms of Reference

- **Consider development paths of new standards required by the user community**
  - Using where appropriate existing standards from different sources
  - Influencing inter-sector standards development with health requirements
- **Guidance for implementations of e-Health applications**
  - Providing information on relevant standards and standards activities
  - Information on products and companies using standards
  - Best practice examples and case studies

# e-Health Standardization Coordination Group (eHSCG)

## Terms of Reference

- Increase awareness of availability of existing standards and the importance of standardization among relevant decision makers (health authorities, health professionals and industries)
  - Conference presentations and reports
  - Website (hosted by ITU)
- Meet regularly co-located with related technical standardization meetings
- Consider the requirements of developing countries

# Many standards exist but are not used enough

- A lot remains to be done
  - There will be a continued need for standards development meeting new application requirements and technical frameworks
- We need global co-operation
  - Respecting pragmatic decisions to use specifications from different bodies (national, european and international)

## **Global standardization of health informatics is welcomed because:**

- **Joint work of the best experts of the world improves the quality of our standards**
- **The market for industrial products (soft and particularly hard) should be more and more global although it must be recognized that for different reasons many systems are developed for national markets only**
- **There is an emerging requirements for cross-border communication of health information which we want to support**
  - **BUT this need is still almost non-existent globally and small even within Europe**

# Does CEN have the expertise for developing an Anatomy Standard ?

- No but ...
- The standard body is an organisational framework that will encourage participation of domain experts for a specific project
- **YOU ARE ALL VERY WELCOME**
- The participants of the CEN working group on Terminology and concept representation are mostly health professionals - physicians

# **CEN/TC 251 work item:**

Health informatics –  
Categorial structure for anatomy

**Preliminary work started in  
2003**

# CatAnat – project leader

Ed Cheetham, M.D. English NHS

**CEN/TC 251 WGII**  
**Terminology and knowledge**  
**representation**  
**Meeting in Berlin June 2004**

# Outline slide

- **Scope and justification**
- **Early model development**
  - Issues
- **Further work**
  - Model refinement
  - Utility of CatAnat
- **Discussion**

# Scope and justification

- **Work item description - scope**
  - to describe a standard syntax that will
    - represent locations and spatial relations between structures in human anatomy, (especially macroscopic)
  - to facilitate
    - harmonisation of systems of concepts in existing anatomical sources
    - development of new tools and knowledge bases

# Scope and justification

- **Work item justification**
  - facilitate systematic use of computer tools for tasks such as documentation, navigation and information retrieval
- **Customers**
  - Cross-referencing groups
  - Developers
  - Quality assurers

# **Categorical structure definition**

**from ISO/DIS 17115 Health informatics –  
Vocabulary for terminological systems**

**domain concept model**

**categorical structure**

**set of formal categories, semantic links (3.1.6) and sanctions (3.2.3) describing potential characteristics (3.2.2) for representing concepts (A.3.2.1) in a domain**

# ISO 1087-1:2000 TERMINOLOGY WORK — VOCABULARY —

## Part 1: Theory and application

**concept**

**unit of knowledge created by a  
unique combination of  
characteristics**

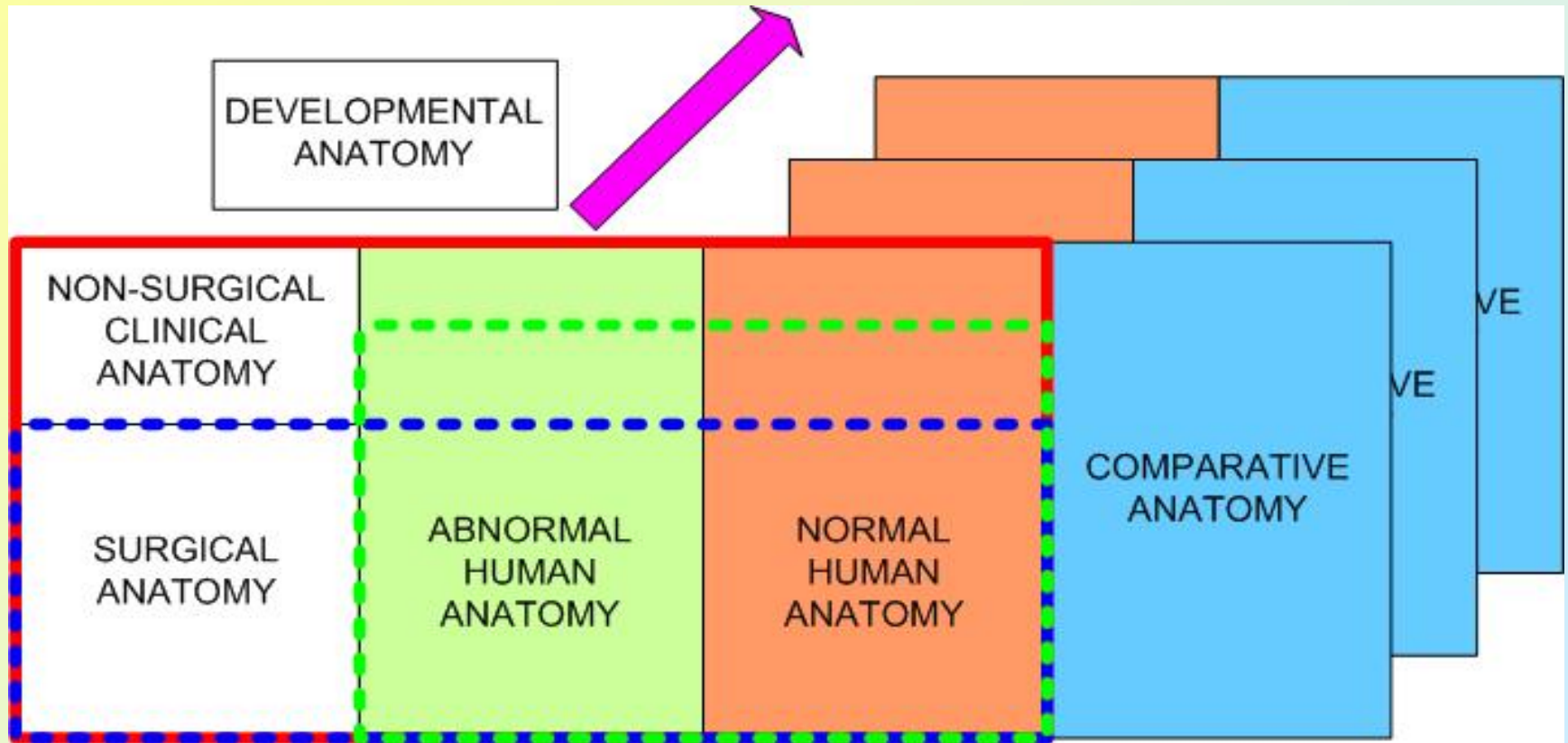
# Scope - current approach

- **Extent of 'model of anatomy':**
  - Human anatomy (static)
  - Developmental anatomy (dynamic)
  - Comparative anatomy
  - ± clinical concept relationships integrating anatomy with fields such as medicine and surgery
  - ? Other components

# Scope - current approach

- Rosse et.al. – Digital Anatomist ‘foundational model of anatomy’
  - *Anatomy taxonomy* (classification of anatomical entities)
  - *Anatomical structural abstraction* (partitive and spatial relationships)
  - *Anatomical transformational abstraction* (time-dependent morphological transformations during the human life-cycle)
  - *Metaknowledge* (principles and rules governing the other three components)
  - ***Not functional***

# Scope – proposed content is the red box



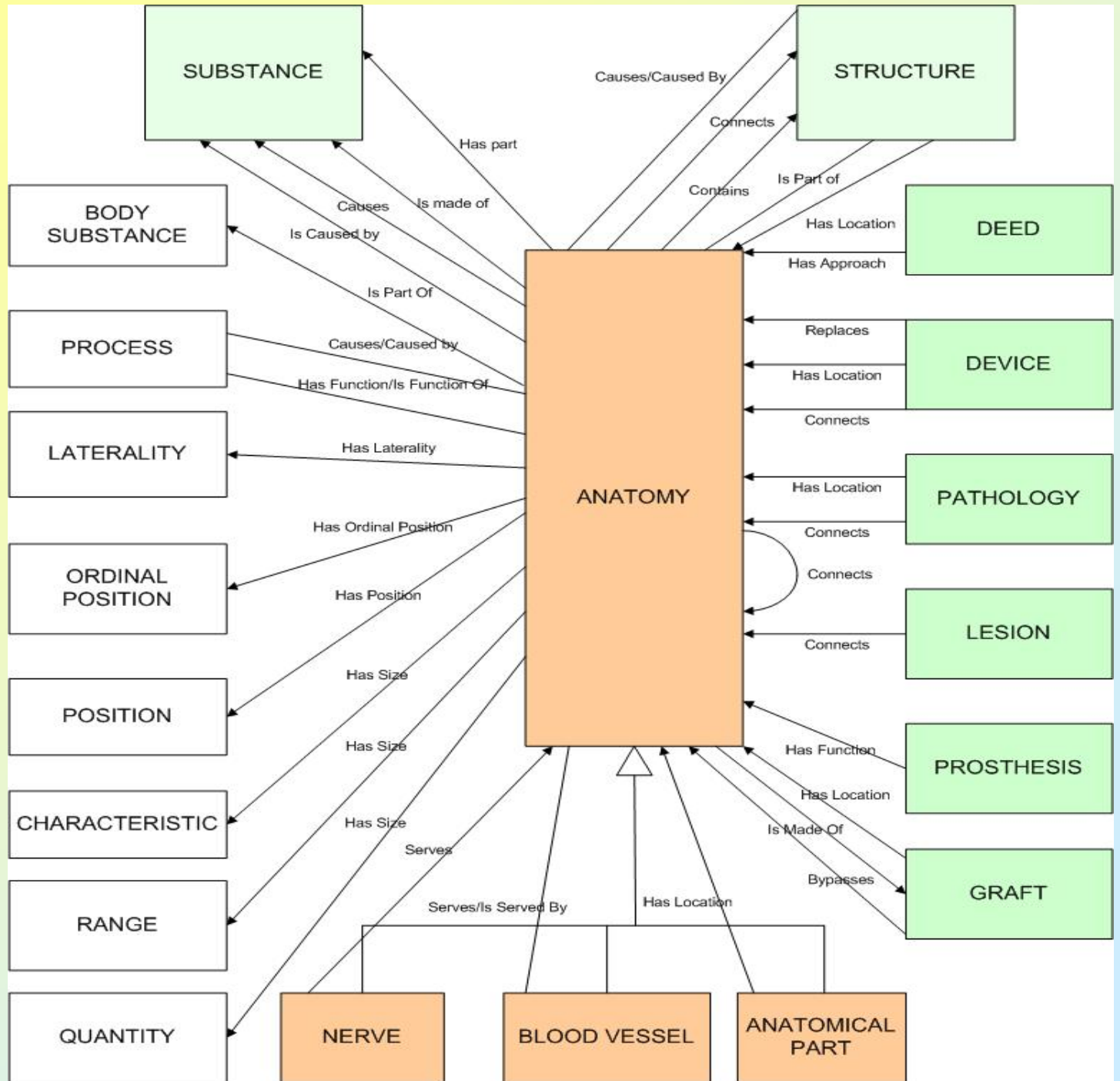
## Scope - proposed model

- **Early problems!**
  - **Overlap between abnormal and developmental anatomy**
  - **E.g. Persistent ductus arteriosus**

# Early model development

- **GALEN-IN-USE. Links and Templates Summary 1996**
  - Link sanctions table
  - Links originating from, or pointing to, GALEN concept 'Anatomy'

# GALEN



# Early model development

- **Findings**

- ‘Pure’ anatomy relationships

- E.g. ANATOMY - HasLaterality – LATERALITY

- Normal/abnormal relationships

- E.g. ANATOMY - HasPart – SUBSTANCE

- ‘Abnormal’ relationships

- E.g. GRAFT – HasLocation – ANATOMY

- **Normal anatomy concepts**

- **Specialisation**

# Early model development

## *Normal anatomy concept – Specialisation*

**Need for categorial structure specialisations:**

- Nerve structure/Blood vessel structure
  - Serves – ANATOMY
  - (nerve, Rosse) Input from/output to
    - NERVE
- Muscle structure
  - Attaches to – ANATOMY
- Cavitated structure v. solid structure
  - Distinct characteristics

# Further work

- Further 'elaboration'
  - Inclusion of other GALEN material
    - E.g Pathology, BodyProcess and BodySubstance
    - Richer set of partitive attributes
  - Inclusion of attributes from WGII/N03-08rev
    - E.g. dimension ('medial/lateral' etc.)
  - Extension to 'pure' anatomy/disorders
- Simplification/rationalisation
  - Informative elements of EN 1828?  
(Health informatics – Categorical structure of surgical procedures)

## Further work

- **Extension to 'pure' anatomy**
  - **Early feedback:**
    - 'It is therefore proposed that CatAnat is tested and extended against textual definitions of anatomy concepts'
      - Simple labels
    - Some preprocessing or paraphrasing prior to dissection
      - More fruitful

## Further work

- **Utility of pursuing CatAnat**
  - Continue Galen-based approach
  - Reasoned adoption of 'digital anatomist'
    - Scale of development
    - Metaclasses
    - Wide intended audience for ontology

# Discussion issues

- The CEN group and project leader is very open for contributions to this work both from Anatomists and Ontology specialists
  - Co-operation with the Digital Anatomists Foundational Model of Anatomy team is encouraged
    - Can we distil a categorial structure ?
  - How can the Semantic mining project channel contributions to a standard to give a formal base using meretopology ?