Biomedical Metamodels

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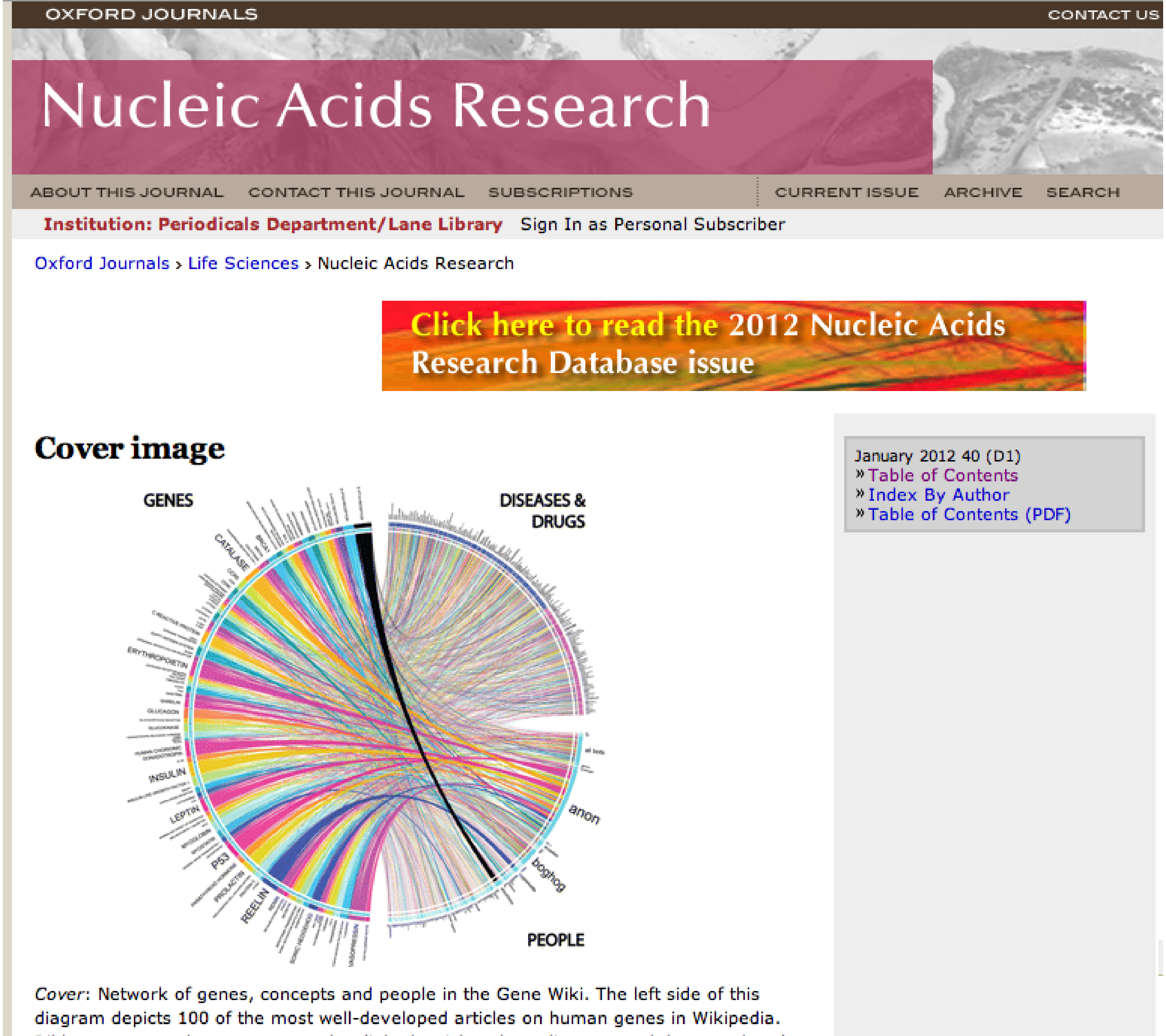
NATIONAL CENTER FOR
BIOMEDICAL ONTOLOGY



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Cover image

Cover: Network of genes, concepts and people in the Gene Wiki. The left side of this diagram depicts 100 of the most well-developed articles on human genes in Wikipedia.

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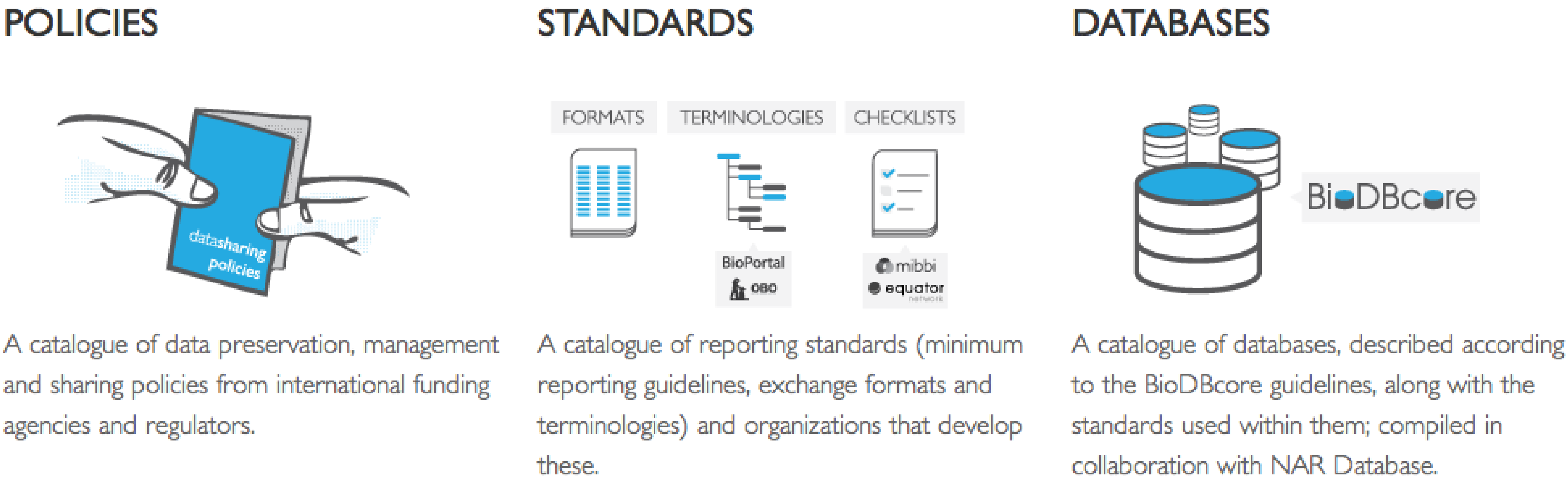
BioSharing Initiative attempts to provide a path through all the data—and all the metadata



POLICIES

A catalogue of data preservation, management and sharing policies from international funding agencies and regulators.

STANDARDS

A catalogue of reporting standards (minimum reporting guidelines, exchange formats and terminologies) and organizations that develop these.

DATABASES

A catalogue of databases, described according to the BioDBcore guidelines, along with the standards used within them; compiled in collaboration with NAR Database.

http://biosharing.org

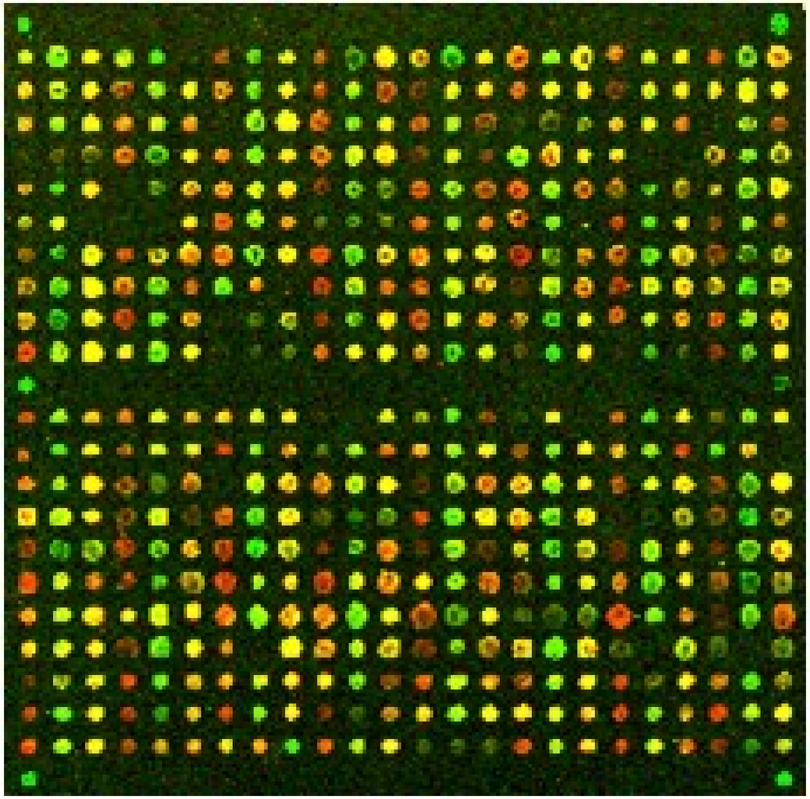
BioDBcore: Uniform descriptors of public biological databases

- Grass-roots effort to develop community-defined descriptions of the core attributes of biomedical databases
- Overseen by the International Society for Biocuration
- Goals are to
  - Overview the landscape of online data resources
  - Encourage consistency and interoperability
  - Promote use of standards

bioDBcore

Microarrays produce tons of gene expression data

- Genes are expressed at different levels in different cells and tissues at different times
- Microarrays bind to specific nucleotide sequences expressed in cells
- Result: Vast amounts of gene-expression data



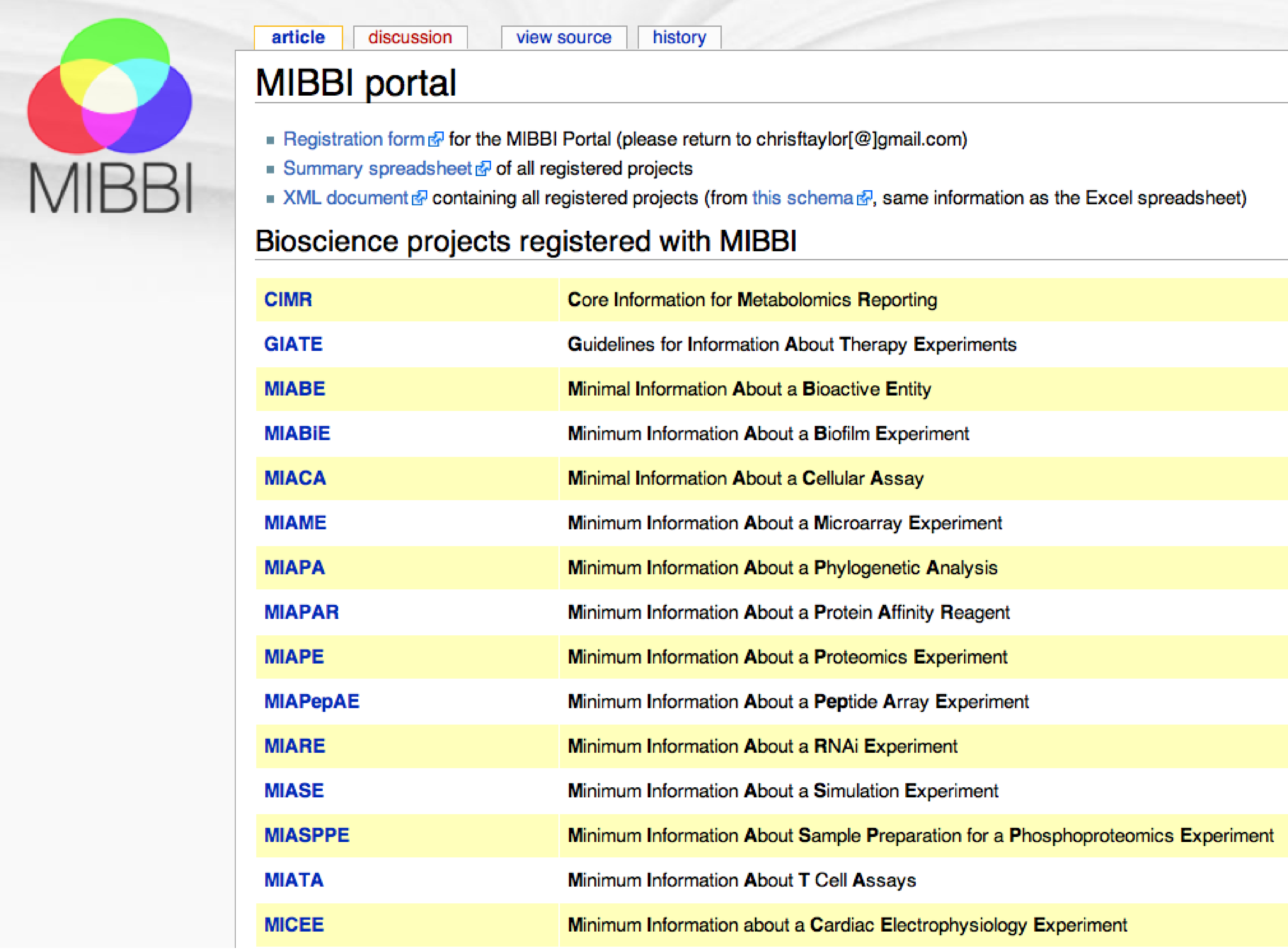
DNA Microarray

Minimum Information About a Microrarray Experiment (MIAME)

- Just a checklist
- At level of detail of what is required in a journal article
- Does not commit to any particular format to enable computational validation or analysis of entries
- Adopted by Gene Expression Omnibus, ArrayExpress and other online databases

Moving beyond checklists

- The Functional Genomics Data Society (formerly the Microarray Gene Expression Data Society; MGED) was founded as the grass-roots effort that created MIAME
- The MicroArray Gene Expression (MAGE) initiative led to
  - **MAGE-ML**: An XML-based markup language
  - **MAGE-TAB**: An Excel-based version of MAGE-ML
  - **MAGE-OM**: A UML-based object language that informs all tags in MAGE-ML
  - **MGED Ontology**: An OWL-based ontology to provide standardized data values of MAGE-OM classes



MIBBI portal

- Registration form for the MIBBI Portal (please return to chrisftaylor[@]gmail.com)
- Summary spreadsheet of all registered projects
- XML document containing all registered projects (from this schema, same information as the Excel spreadsheet)

Bioscience projects registered with MIBBI

CIMR	**C**ore **I**nformation for **M**etabolomics **R**eporting
GIATE	**G**uidelines for **I**nformation **A**bout **T**herapy **E**xperiments
MIABE	**M**inimal **I**nformation **A**bout a **B**ioactive **E**ntity
MIABiE	**M**inimum **I**nformation **A**bout a **Bi**ofilm **E**xperiment
MIACA	**M**inimal **I**nformation **A**bout a **C**ellular **A**ssay
MIAME	**M**inimum **I**nformation **A**bout a **M**icroarray **E**xperiment
MIAPA	**M**inimum **I**nformation **A**bout a **P**hylogenetic **A**nalysis
MIAPAR	**M**inimum **I**nformation **A**bout a **P**rotein **A**ffinity **R**eagent
MIAPE	**M**inimum **I**nformation **A**bout a **P**roteomics **E**xperiment
MIAPepAE	**M**inimum **I**nformation **A**bout a **Pep**tide **A**rray **E**xperiment
MIARE	**M**inimum **I**nformation **A**bout a **R**NAi **E**xperiment
MIASE	**M**inimum **I**nformation **A**bout a **S**imulation **E**xperiment
MIASPPE	**M**inimum **I**nformation **A**bout **S**ample **P**reparation for a **P**hosphoproteomics **E**xperiment
MIATA	**M**inimum **I**nformation **A**bout **T** Cell **A**ssays
MICEE	**M**inimum **I**nformation about a **C**ardiac **E**lectrophysiology **E**xperiment

The HL7 Organization

- ANSI-accredited Standards Development Organization for healthcare applications
- Initial work was applications-layer communication model for data interchange among HIS components
- Current work focuses on
  - Decision support (e.g., Arden Syntax, “Infobuttons”)
  - Enterprise modeling (e.g., Reference Information Model)
  - Clinical Document Architecture, Continuity of Care Documents
  - Clinical terminology

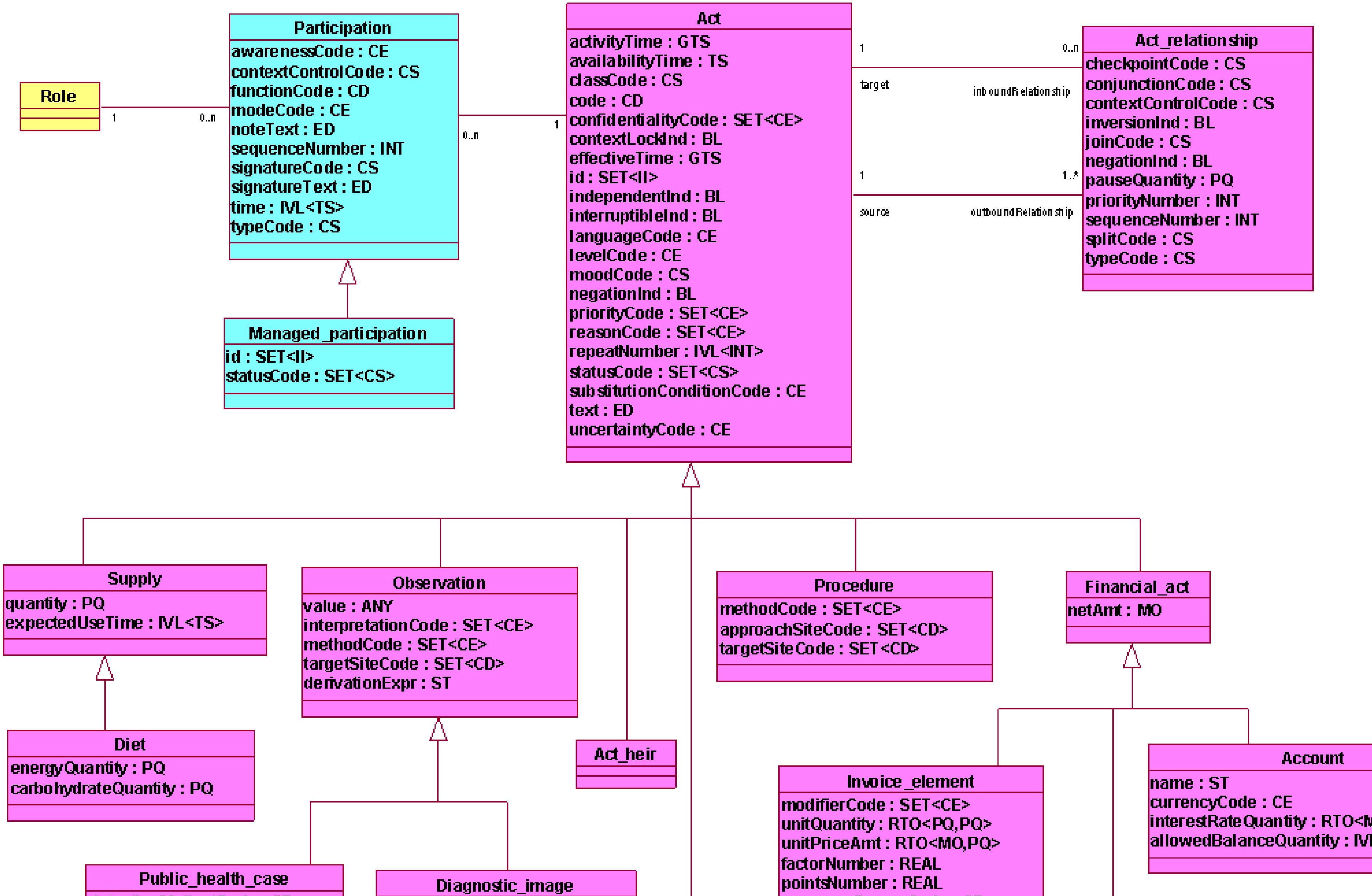
HL7 Version 2 Messaging Standard

```
MSH|^~\&|MegaReg|XYZHospC|SuperOE|XYZImgCtr|20060529090131-
0500||ADT^A01^ADT_A01|01052901|P|2.5  EVN||200605290901||||200605290900  PI
D|||56782445^^^UAReg^PI||KLEINSAMPLE^BARRY^Q^JR||19620910|M||2028-
9^^HL70005^RA99113^^XYZ|260 GOODWIN CREST DRIVE^^BIRMINGHAM^AL^35
209^^M~NICKELL'S PICKLES^10000 W 100TH AVE^BIRMINGHAM^AL^35200^^O
|||||||0105I30001^^^99DEF^AN  PV1||I|W^389^1^UABH^^^^3||||12345^MORGAN^R
EX^J^^^MD^0010^UAMC^L||678
90^GRAINGER^LUCY^X^^^MD^0010^UAMC^L|MED|||||A0||13579^POTTER^SHER
MAN^T^^^MD^0010^UAMC^L|||||||||||||||||||||||||||200605290900  OBX|1|NM|
^Body Height||1.80|m^Meter^ISO+|||||F  OBX|2|NM|^Body
Weight||79|kg^Kilogram^ISO+|||||F  AL1|1||^ASPIRIN  DG1|1||786.50^CHEST PAIN,
UNSPECIFIED^I9|||A
```

HL7 Version 3 Stated Goals

- Apply best practices of software engineering — a model-based methodology
- Adopt three semantic foundations
  1. A reference information model
  2. A robust set of data types
  3. A complete, carefully selected set of terminology domains
- Require all HL7 Version 3 standards to draw from these three common resources

HL-7 Reference Information Model (RIM)



Role

Participation
awarenessCode : CE
contextControlCode : CS
functionCode : CD
modeCode : CE
noteText : ED
sequenceNumber : INT
signatureCode : CS
signatureText : ED
time : IVL<TS>
typeCode : CS

Managed_participation
id : SET<II>
statusCode : SET<CS>

Act
activityTime : GTS
availabilityTime : TS
classCode : CS
code : CD
confidentialityCode : SET<CE>
contextLockInd : BL
effectiveTime : GTS
id : SET<II>
independentInd : BL
interruptibleInd : BL
languageCode : CE
levelCode : CE
moodCode : CS
negationInd : BL
priorityCode : SET<CE>
reasonCode : SET<CE>
repeatNumber : IVL<INT>
statusCode : SET<CS>
substitutionConditionCode : CE
text : ED
uncertaintyCode : CE

Act_relationship
checkpointCode : CS
conjunctionCode : CS
contextControlCode : CS
inversionInd : BL
joinCode : CS
negationInd : BL
pauseQuantity : PQ
priorityNumber : INT
sequenceNumber : INT
splitCode : CS
typeCode : CS

Supply
quantity : PQ
expectedUseTime : IVL<TS>

Diet
energyQuantity : PQ
carbohydrateQuantity : PQ

Observation
value : ANY
interpretationCode : SET<CE>
methodCode : SET<CE>
targetSiteCode : SET<CD>
derivationExpr : ST

Public_health_case

Diagnostic_image

Act_heir

Procedure
methodCode : SET<CE>
approachSiteCode : SET<CD>
targetSiteCode : SET<CD>

Financial_act
netAmt : MO

Invoice_element
modifierCode : SET<CE>
unitQuantity : RTO<PQ,PQ>
unitPriceAmt : RTO<MO,PQ>
factorNumber : REAL
pointsNumber : REAL

Account
name : ST
currencyCode : CE
interestRateQuantity : RTO<M
allowedBalanceQuantity : IV

The HL7 Response to v3 Woes

- Instituted the “Fresh Look” task force in January 2011
- New work on Fast Healthcare Interoperability Resources (FHIR)
  - Still based on Version 3 RIM
  - Adopts “80/20” rule in attempt to simplify things
  - Goal is to make life simpler for implementers, rather than for modelers
  - A modular approach, based on XML

Clinical Document Architecture (CDA)

- First HL7 standard based on the RIM
- XML-based language for marking up narrative clinical reports
- Allows document text to be marked up with terms from the RIM and from controlled terminologies
- Facilitates exchange of clinical documents within and between health-care organizations

Continuity of Care Documents

- CCDs use a refinement of the CDA model that incorporates ideas from yet another standard— ASTM’s Continuity of Care Record (CCR)
- CCD is also an HL7 standard
- Many HIEs use CCDs, as does Microsoft HealthVault
- ONCHIT promotes use of CCD model

REPORT TO THE PRESIDENT
REALIZING THE FULL POTENTIAL OF HEALTH INFORMATION TECHNOLOGY TO IMPROVE HEALTHCARE FOR AMERICANS: THE PATH FORWARD

Executive Office of the President

President's Council of Advisors
on Science and Technology

PCAST Major Conclusions

- "Meaningful use" criteria are necessary first steps
- There must be accelerated progress toward "robust exchange of health information"
- There should be a "universal exchange language" to enable data sharing and an IT infrastructure to support data sharing
- All of this is technologically feasible
- ONC needs to move rapidly in this area

Conclusions

- Metadata initiatives in biomedicine reflect the practical problems of the data deluge
- In the life sciences, there are significant initiatives driven by groups of scientists at the grass roots
- In the clinical area, the principal driving forces have been IT vendors (HL7) and government (CMS, ONC)
- Metadata solutions in biomedicine have at best incomplete penetrance
- Many metadata solutions have been met by open hostility