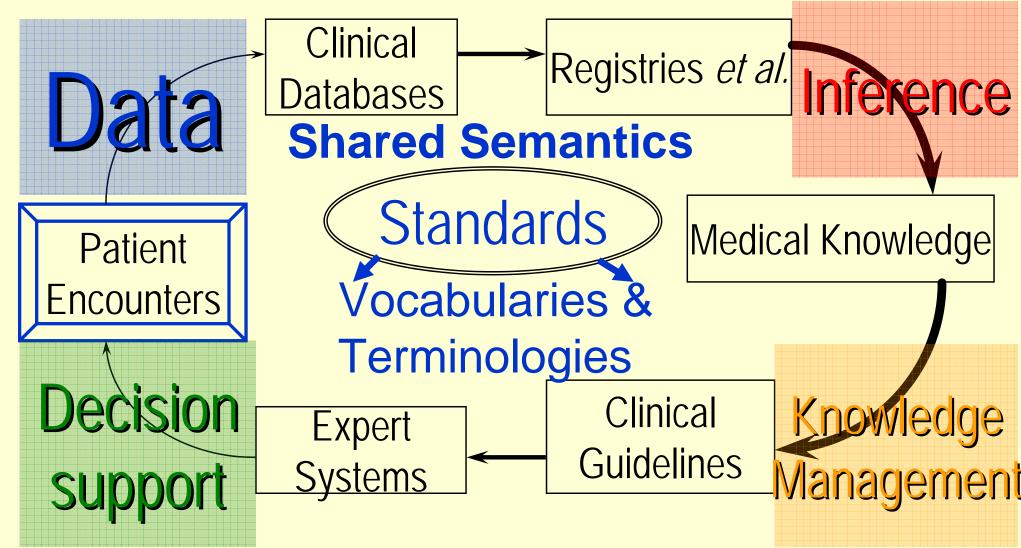


Comments on Building on Data in dbGaP Data Standards and Standard Data

CG Chute MD DrPH Frontiers in Population Genomics Research Meeting December 18, 2007

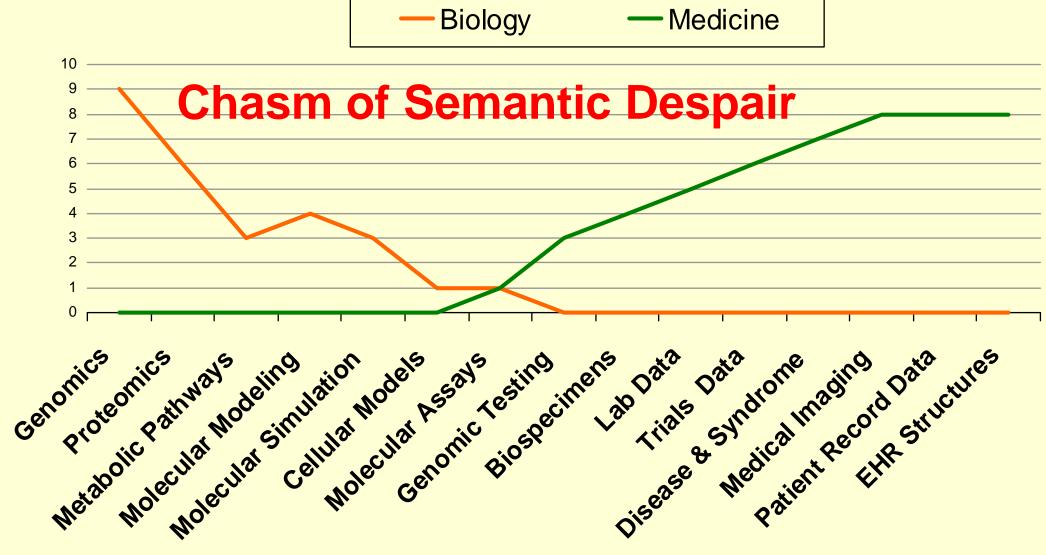


From Practice-based Evidence to Evidence-based Practice





The Continuum Of Biomedical Informatics Bioinformatics meets Medical Informatics





Content vs. Structure Yet Another Chasm

Family History of Breast Cancer *Family History of* Heart Disease *Family History of* Stroke

Terminologic Model

Family History Breast Cancer Heart Disease

Stroke

cer Model Ise

Information Model Equivalent Content

[adapted from Rossi-Mori]



NCBO – A Bridge Across the Chasm

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- It's the Phenotype (observations)... as standard, comparable representations
- Define information routinely collected
- Emphasize computable content rather than relying on textual procedure definitions
- NHGRI role in re-kindling the Roadmap initiative for broadly-based phenotype characterization
 - Align clinical, CTSA, caBIG, population genomics...
 - Coordinate SNOMED, ICD, HITSP, NCBO, HL7,...
- Coordinated aggregation rules/logics [ICD-11]
- Promote "research" as an AHIC use-case