

Scoring and heuristics

- · Knowledge is represented as profiles of findings that occur in diseases
- There are measures of importance and frequency for each finding in each disease
- · Found to be most "scalable" approach for comprehensive decision support systems
- Examples INTERNIST-1/QMR, DxPlain, Iliad

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History of systems using scoring and heuristics approach

INTERNIST-1

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- Original approach, aimed to develop an expert diagnostician in internal medicine (Miller, 1982)
- System originally designed to mimic the expertise of an expert
- diagnostician at the University of Pittsburgh, Dr. Jack Meyers
 Evolved into Quick Medical Reference (QMR) where goal changed to using knowledge base explicitly (Miller, 1986)

- DxPlain used principles of INTERNIST-1/QMR but developed more disease coverage (Barnett, 1987) Only system still available:
 - http://lcs.mgh.harvard.edu/projects/dxplain.html
- · Iliad attempted to add Bayesian statistics to the approach (Warner, 1989)

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INTERNIST-1/QMR knowledge representation

- Disease profiles findings known to reliably occur in the disease
- Findings from history, exam, and laboratory
- Import each finding has a measure of how important it is to explain (e.g., fever, chest pain)
- Properties e.g., taboos, such as a male cannot get pregnant and a female cannot get prostate cancer

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а.	Dx: MYOCANDIAL INFANCTION ACUTE	
	is associated with 134 Finding(s) arranged. (w/Reterences) 1. In Textbook order: History, symptoms, signs, labs 2. By Frequency	EvEr
Þ	Pest Medicel History	
▼	Symptoms of Current Illness Chest Pain Substemal At Rest Chest Pain Substemal Lasting 20 Minute(s) Or Gtr	24 34
	Chest Pain Substemal Unrelieved By Nitroglycerin Onset Abrupt	34 04 33
	Chect Pain Substemal Cruching Chect Pain Substemal Radiating To Neck And/Or Upper Extremity(ies) Chect Pain Substemal Covers	33 00 20
	Ahrinnen Pain Acitta Abdomen Pain Epigastrium	12
	Abdomen Pain Epigastrium Unrelieved By Antacid Abdomen Pain Exacerbation With Exercise	12
	Abdomen Pain Non Colicky Abdumen Pain Present	12 02
	Abdomen Pain Severe	12



INTERNIST-1/QMR scoring algorithm

- Initial positive and negative findings are entered by user
 A disease hypothesis is created for any disease that has
- one or more of the positive findings entered
- Each disease hypothesis gets a score
- Positive component based on evoking strengths of all findings
 Negative component of score based on frequency from findings expected to occur but which are designated as absent
- A diagnosis is made if the top-ranking diagnosis is >80 points (one pathognomonic finding) above the nexthighest one
 - When diagnosis made, all findings for a disease are removed from the list, and subsequent diagnoses are made
- Performed as well as experts in NEJM clinical cases (Miller, 1982)

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Limitations of INTERNIST-1 and evolution to QMR

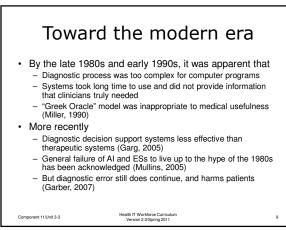
Limitations

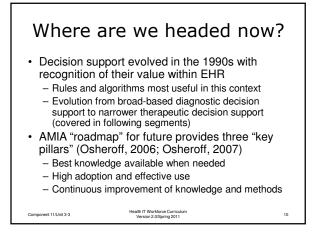
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- Long learning curve
- Data entry time-consuming
- Diagnostic dilemmas not a major proportion of clinician information needs
- Knowledge base incomplete
- Evolution to QMR (Miller, 1986)
 - Less value in "case" mode
 - More value in knowledge exploration mode, e.g.,
 - Rule diseases in and out
 - Obtain differential diagnosesLink to more detailed information
 - Became commercial product but did not succeed in marketplace

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But the quest for diagnostic decision support continues • Isabel (www.isabelhealthcare.com) – "Second generation" approach uses

- Natural language processing to map entered text into findings
 List of differential diagnosis with 30 most likely diagnoses grouped by body system, not probability
- Performance studies

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- Initial development and validation for pediatrics (Ramnarayan, 2006) – reminded of one clinically important case 1 of 8 times
- Subsequently extended and evaluated in emergency department (Ramnarayan, 2007) – displayed correct diagnosis 95% of time and 90% of time showed "must-not-miss" diagnoses
- Now expanded to adult internal medicine (Graber, 2008) pasting in text from NEJM case reports had correct diagnosis suggested in 48 of 50 cases for key text and 37 of 50 cases for all text

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Other continuing approaches – "Googling" for a diagnosis?

- Large quantity of text in Google may hold latent knowledge?
 - Found in a case study to make diagnosis of a rare condition (Greenwald, 2005)
 - When text of NEJM cases entered, 15 of 26 had correct diagnosis in top three suggested (Tang, 2006)

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