A Qualitative Review of Differential Diagnosis Generators

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Introduction

Differential diagnosis (DDX) generators have existed for some time, but their use has not been widely adopted in practice. We identified and described the features of a current list of DDX generators.

Methods

We performed a Google search and a literature search using a series of subject headings (MESH) and keywords to identify programs that qualify as differential diagnosis generators. Through consensus, the author group identified four factors critical for a differential diagnosis generator to be useful. First, the program needed to present a list of potential diagnoses rather than text or article references. Second, the program must rank or indicate critical diagnoses that need to be considered or eliminated. Third, the program needed to accept at least two signs, symptoms or disease characteristics. Finally, the program needed to provide the ability to compare the clinical presentation of the different diagnoses presented. The study was limited to programs providing diagnoses in general medicine. Programs focused on one disease process or clinical specialty were excluded. The study was limited to programs developed for the use of healthcare professionals (HCPs), not patients or consumers. Qualitative evaluation criteria were agreed upon by consensus prior to evaluating their use.

Results

Eleven programs were excluded due to specialty specific focus. Another seven programs were excluded after an initial review for reasons that included: inability to compare diagnoses, to enter two symptoms or characteristics, or to rank diagnoses, and generators that were simply a static tree structure with cross linking of internal reference points. Five programs were reviewed with evaluation criteria that are listed in the first column of the results table. When information was not available to the end user, the company producing the software was queried for clarification.

Conclusions:

The programs were useful in presenting and ranking possible diagnoses. Links to both EBM and non-EBM content were plentiful. Our ability to test EHR integration was limited. The DDX generators should prove helpful teaching tools. Use in practice will depend on EHR integration and the number of false alarms generated.

Table 1. Evaluation Criteria Definitions

Criterion	Definition	Diagnosis Pro®	DXPlain®	First Consult©	Isabel©	PEPID
Producer	Publish Name	MedTech USA, Inc 6310 http://www.diagnosispro.com/	Laboratory of Computer Science of the Department of Medicine Massachusetts http://dsplain.org/dxp/dxp.pl	Elsevier Inc./Md Consult http://www.firstconsult.com	Isabel Healthcare Inc., http://www.isabelhealthcare.com/home/default	Pepid Medical Information Services LLC http://www.pepid.com/
Subscription / Licensing Model		Institutional and Individual	Institutional	Institutional and Individual. Available as an add-on to MDConsult.	Institutional and Individual	Institutional and Individual. Available as an add-on to PEPID.
Degree of EHR Integration (Input)	Will the program pull any data from the EHR? What fields? Must findings be pushed into it manually? Does the program incorporate Health Level 7 (HL7) interoperability standards?	No data populated form EHR.	In limited setting (Currently limited to Massachusetts General Hospital EHR), abnormal tests link to a list of associated diseases.	Yes, multiple data fields can be populated from EHR.	Yes, multiple data fields can be populated from EHR.	No data populated from EHR.
HL7 Interoperability Standards		Unknown	Udner development	Yes	Yes	Yes - by default no private patient data transmitted.
Input Method(s)	For example, manual entry, prepopulated from EMR, selected from program list, etc. subquestion: What is the degree of flexibility in entering patient characteristics and symptoms? How user-friendly is the interface?	Manual entry/selection of: signs/symptoms, lab/imaging/diagnostic tests, risk factors. Negative findings not considered.	Manual entry/selection of: signs/symptoms, lab/imaging/diagnostic tests, risk factors. Negative findings not considered.	Manual entry/selection of signs/symptoms. Populated information from EHR. Free text searching of text strings,	Manual entry/selection of signs/symptoms, lab/imaging/diagnostic tests, patient demographics. Populated information from EHR. Negative findings not considered. Numeric data cannot be entered.	Manual entry/selection of signs/symptoms, lab/diagnostic tests, chest xray, patient demographics. Negative findings not considered. Numeric data cannot be entered. Imaging findings other than chest x-ray are not supported.
Mechanism of generating potential diagnoses	What is the ordering of diagnoses based on? (e.g. Baysian, keyword frequency, semantic search, proprietary system etc.) Does the program use natural language processing? Consider any type of weighting that figures into generating the dx.	Results are not rank ordered in any way. Diagnoses are presented in disease categories. Does not rank the suggestions in terms of common versus unusual and offers no advice on how to further refine the suggestions. Underlying logic is not specified.	Rank ordered results from most to least likely; disease prevalence estimated; importance ranked based on criticality of potential diagnosis. Finding assigned two attributes: one relating to the frequency of the finding in the disorder, and one expressing how strongly it suggests that disease. Findings also assigned a disease-independent attribute indicating the importance of the finding. Ranking related to findings that are both important and suggestive of a disorder. Common diseases are given extra weight. Rank of a given disease will be lowered if findings commonly seen in the disease are stated to be absent. The attributes are used to generate an ordered list of diagnoses associated with some or all of a given set of findings.	Diagnoses are presented by age and as a static list for chief complaints based on prevalence. Potentially urgent diagnoses are indicated. No other filtering from within the list is available.	Uses natural language processing search engine to match entered clinical features with similar terms in the diagnostic data set. Each diagnosis has a complete discription of the clinical features with the differential ranked by the strength of the match to the entered clinical features. The differential diagnosis output is displayed in a separate window from the EHR but the clinical feature inputs remain visable. With each clinical feature addition the differential diagnostic output reconfigures the list, taking into acccount all the clinical features entered	Diagnoses presented based on a proprietary scoring system related to the number of selected signs/symptoms consistent with each potential diagnosis plus each sign/symptom is assigned a unique score/weight relative to its importance in differentiating among specific diagnoses. Classic/cardinal disorders in which selections strongly suggest or are pathognomonic are indicated. Critical diagnoses with immediate life or limb threat are indicated.
Lab Values as a Dx Factor	In addition to symptoms, does the program incorporate numeric lab values, positive/negative lab values?	Yes	Yes	No	Yes	Yes
Medications as Dx Factor	Does the program take into account current drugs being given or list possible drugs that can cause the collection of signs or symptoms?	No	No	Yes	Yes	No
Geography as a Dx Factor	Can the program take into account the geographic location of the patient (e.g. for Rocky Mountain Spotted fever or Lyme disease) or the elevation of the patient (e.g. for altitude sickness)?	Yes	Yes	No	Yes	No
Content Source		Textbooks, journal articles and websites.	Proprietary knowledge base.	Proprietary knowledge base. Some of the more obscure topics have very limited information.	Proprietary knowledge base.	Proprietary knoweldge base.
Evidence Based	Is the content provided by the provider/publisher evidence based? Does the program incorporate evidence based guidelines? From what sources? e.g. USPSTF, CDC, etc.	No	Partial. Specific evidence-based recommendations from specialty societies and CDC considered in content development.	Yes. Cochrane Collaboration; BMJ Clinical Evidence; National Guideline Clearinghouse; Evidence graded A-C or "Uncategorized" based on AAFP guidelines	Partial. Specific evidence-based recommendations are considered in content development.	Partial. Specific evidence-based recommendations and analyses which are incorporated contain graded recommendations from FPIN and BEEM
References	Does the program provide references for the diagnoses presented? Can it provide links to full text articles? Is the full text only from vendor sources (e.g. First Consult provides links to MDConsult articles but not other sources). Does the program allow for PubMed linking to allow access to full text of library/institutionally subscribed resources?	No references provided for each disease. Can run preformatted Pubmed search from disease description screen. PubMed links provided do not resolve to the institution's PubMed Linkout to provide full text from institutional/library subscribed content.	References to Medline abstracts and open access guidelines. Can run preformatted a PubMed search and/or a structured Google search of pre-selected medical websites. PubMed links provided but do not resolve to the institution's PubMed Linkout to provide full text from institutional/library subscribed content.	References available in MDConsult will present in full text. PubMed links provided but do not resolve to the institution's PubMed Linkout to provide full text from institutional/library subscribed content.	The "knowledge" choice on the tool bar allows a seach of approximately 90 journals and 7online texts. No link to PubMed.	References to evidence based information from Family Practice Inquiries Network (FPIN) are integrated in PCP module. Other sources are cited throughout. No link to PubMed.
Drug Content Source	What is the sources of any drug information provided? e.g. ASHP, proprietary, etc.	Uncertain. Reference list includes many possible sources for drug information.	No specific drug information provided.	Gold Standard	Martindale and other sources	American Society of Hospital Pharmacists
Minimum level of clinical expertise needed	For what level of HCP is the program suitable? Physician, resident, medical student, nurse, allied.	Resident or higher.	Resident or higher, but good teaching tool at the student level.	Student or higher.	Student or higher.	Student or higher.
Usage Tracking (Institutional Suscription / Licensing)	Is it possible to obtain reports on the level of usage of the program? Possible to determine type of user? If reports are available, what are they based on? e.g. some programs count every click a user makes; others count just entry into the program; others count how many topics were searched. If a program contains several content modules, is it possible to track usage of the DDX module separately from other content?	None mentioned	Yes	Yes	Yes	Yes
Other Features	Does the program contain any additional features? e.g. PEPID has a lab manual, drug interactions checker, etc.	Franch and Spanish interfaces. Side-by-side disease comparison.	Some clinical images. Occupation as a finding. Display of what findings support the disease, the findings known to be part of a disease and other findings, if present, would support the disease.	Since it is integrated with MDConsult, the total program offers textbooks, journals, the Clinics periodicals, 50,000 clinical images, 10,000 patient handouts. Side by side disease comparison.	"Lessons Learned" section where users can share examples of diagnostic errors.	Incorporates lab manual, drug interactions generator, drug database covering 7,500 drugs, approximately 400 interactive clinical calculators, IV compatibility tool, acute care / life support reference section, and 700 evidence based topics (primary care module).

BEEM - Best Evidence in Emergency Medicine
CDC = Centers for Disease Control
CME = Continuing Medical Education
EHR = Electronic Health Record
HL7 = Health Level 7 Interoperability Standards