Why and How to Improve Diagnosis Decision Making

The importance of improving diagnosis decision making and the tools needed to achieve it

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"Improving the diagnostic process is not only possible, but it also represents a moral, professional and public health imperative"

National Academies of Sciences, Engineering and Medicine. 2015. *Improving Diagnosis in Healthcare*. Washington, DC.

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isabel

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Forward – by Mark Graber, MD

In one of the classic papers in our field, Dr. Georges Bordage asked a very simple question: "Why did I miss the diagnosis?"[1] The answer is equally simple: "I just didn't think of it."

Over the past decade we have learned a great deal about why this happens. In short, *it is System 1's fault!* According to the dual process paradigm that describes how doctors think, if we believe we recognize what's going on, we assign the diagnosis automatically and subconsciously, using our "intuition." This is System 1. It works remarkably well, is extremely fast and efficient, and best of all, its almost always correct. The problem we have in medicine is that "almost always" isn't good enough. The mistakes that invariably arise using intuitive thought are the diagnostic errors that lead to inappropriate medical costs, injury, and harm.

System 2 is the counterpart to System 1 and represents the deliberate, conscious consideration of all the diagnostic possibilities. Conscious consideration is the antidote to many of the shortcomings of System 1. Reflection on alternatives would help solve the most common problems that arise when we make cognitive diagnostic errors:

- Being in the wrong context,
- Being influenced by someone else's diagnosis presented to us, and
- Accepting the first diagnosis that explains all the facts (premature closure, satisficing).

Diagnosis decision support systems like **Isabel** are an ideal way for physicians to avoid System 1 errors. The deliberate consideration of alternatives may bring something to mind that just wasn't considered. The process immediately shakes us out of the complacency that is so often encountered in cases of diagnostic error. We can always pause for reflection on our own, but accepting uncertainty, and reviewing a list of alternative diagnoses may be the best antidotes we have for the "I just didn't think of it" problem that underlies much of diagnostic error.

Sincerely,
Dr. Mark Graber, MD

Mark Graber, MD, is a Senior Scientist at RTI's Health Care Quality and Outcomes Program, and Professor Emeritus of Medicine at the State University of New York at Stony Brook. Dr Graber has an extensive background in biomedical and health services research, with over 70 peer-reviewed publications. He is a national leader in the field of patient safety and originated, with Ilene Corina of New York, Patient Safety Awareness Week in 2003, an event now recognized internationally.

Dr. Graber has been a pioneer and national leader of efforts to address diagnostic errors in medicine. He founded and chaired the Diagnostic Error in Medicine conference series and has several landmark publications on this topic. Currently, Dr. Graber directs an AHRQ ACTION study focused on interventions to reduce diagnostic errors in the ambulatory care settings using checklists.

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[1] Bordage G. **Why did I miss the diagnosis**? Some cognitive explanations and educational implications. Acad Med. 1999 Oct;74 (10 Suppl):S138-43. **http://www.ncbi.nlm.nih.gov/pubmed/10536619**

Why is there a need to improve diagnosis decision making?

Diagnosis is the first and most important decision made about the patient - it determines all subsequent treatment and determines the course of each patient encounter. How well this decision is made, therefore, is one of the most significant determinants of healthcare quality and efficiency.

The following are some of the areas where the speed and accuracy of diagnosis have a key impact and where the use of diagnosis decision aids could help affect improvements:

- **Referrals from primary care to specialists** - Research shows that 30-50% of referrals from primary care to specialists are inappropriate, leading to delays in diagnosis, patient dissatisfaction and lengthy waits at specialist clinics.
- **Test ordering -** Surveys and anecdotal evidence place the level of unnecessary and defensive test ordering at 40%. This is extremely costly and subjects patients to unnecessary clinical risk through invasive procedures and radiation exposure.
- **Medical malpractice -** Misdiagnosis accounts for 30-40% of all malpractice claims and about 2/3 of all claims in primary care. Additionally, diagnostic errors are frequently the leading or second leading cause of malpractice claims in the United States, accounting for twice as many alleged and settled claims as medication errors.
- **Patient satisfaction -** Because patient satisfaction will soon account for 30% of Medicare payments, many hospitals are investing in typical customer service initiatives used for years in other industries. However, in many cases these are viewed as gimmicks by patients and will not make up for poor quality of care. A survey of patients' concerns showed that their top concern when visiting their primary care physician is diagnosis, and in hospitals it is their 2nd most important concern.
- **Employee skills -** Healthcare is a knowledge-intensive industry and a key issue underpinning an institution's success is the clinical skills of all its clinicians. One way of boosting skills across the board is to provide tools that increase clinical skills. Although diagnosis is traditionally seen as the preserve of the physicians, it is the nurses who are caring for the patient most of the time and improving their diagnosis skills can lead to an improved level of patient safety and quality of care.

Errors related to missed or delayed diagnoses are a frequent cause of patient injury and, as such, are an underlying cause of patient safety related events. Diagnosis error happens frequently, it is almost always preventable, and it can cause significant harm. There is now a large body of research demonstrating the size of the problem and why it happens. Some of the most useful papers and sources of knowledge are included in the Appendix of this paper.

Diagnosis Errors and Solutions

Literature on diagnosis error abounds (see Appendix to read more), showing that the causes of delays and errors in diagnosis are many, which means that there is no single intervention that can solve the problem.

Some causes are system related, such as test results being misplaced or not received by the physician, and therefore not acted on or communicated to the patient. It is hoped that the introduction of electronic medical records and other technologies, like personal health records and patient based tools, will help reduce the system related causes.

However, the majority of causes are related to how physicians think and the process of working up a patient's diagnosis. In a 2008 paper in "Medical Teacher," Robert Trowbridge from Maine Medical Center sets out **12 Tips for teaching avoidance of diagnostic error**.[2] There are many intrinsic attributes to us as human beings that contribute to causing diagnosis related errors.

Premature Closure and Biases

As stated, the more common causes of diagnosis errors are due to how a doctor thinks. There is now a large body of work describing the many biases that we, as human beings and not just clinicians, are prone to. The research lists over a 100 different biases but the main types that cause the errors in diagnosis are the 'availability' ones. In a time-constrained industry this is to be expected. As Dr. Mark Graber described his landmark paper, "**Diagnostic error in internal medicine**,"[3] a classic cause is "premature closure," where the clinician decides on a diagnosis very quickly, but then fails to consider other reasonable possibilities until it is too late. In any analysis of cases where the diagnosis was delayed or missed, premature closure has been the most common contributing bias.

Cognitive De-biasing

One of the proposed solutions to this cognitive problem is termed "cognitive de-biasing" and involves clinicians being made aware of these issues as part of their medical training. This solution will help, but in order to be sustainable, it needs to be accompanied by the routine use of tools to help at the point of care.

Differential Diagnosis

Another solution commonly proposed is actually very old and is the routine construction of a comprehensive, differential diagnosis. Olga Kostopoulou has carried out a number studies looking at the predictors of diagnostic accuracy, including "**Missing celiac disease in family medicine: the importance of hypothesis generation**"[4] and "**Diagnosis of difficult cases in primary care**."[5] In the research, Kostopulou found that the most significant factor in the process is having a good differential

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[2] "12 Tips for teaching avoidance of diagnostic error," Trowbridge, Robert. **http://informahealthcare.com/doi/abs/10.1080/01421590801965137**

[3] "Diagnostic error in internal medicine." Graber ML, Franklin N, Gordon R., Arch Intern Med. 2005 Jul 11; 165(13):1493-9. **http://www.ncbi.nlm.nih.gov/pubmed/16009864**

[4] "Missing celiac disease in family medicine: the importance of hypothesis generation." Kostopoulou O, Devereaux-Walsh C, Delaney BC., Med Decis Making. 2009 May-Jun;29(3):282-90. Epub 2009 Mar 6. **http://www.ncbi.nlm.nih.gov/pubmed/19270107**

[5] "Diagnosis of difficult cases in primary care." Kostopoulou O., J Health Serv Res Policy. 2010 Jan;15 Suppl 1:71-4., **http://www.ncbi.nlm.nih.gov/pubmed/20075135**

diagnosis that includes what turns out to be the correct diagnosis.

As a growing amount of research is published about misdiagnosis and its causes in various clinical settings, one of the most interesting points to emerge is the consistent conclusion that the clinicians should have broadened their differential or done one in the first place. Examples of this are "**Diagnostic Errors in Primary Care: Lessons Learned**"[6] by John Ely *et al* and "**Types and origins of diagnostic errors in primary care settings**"[7] by Hardeep Singh *et al.*

Although the construction and use of a comprehensive differential diagnosis has been taught for over 100 years, it is not used routinely in medicine. One of the main reasons for this is the time needed to construct one. Due to a lack of time in the ED or primary care, for example, many clinicians rely on their memory to construct a differential. However, with a universe of diagnoses within primary care of 200-300, compared to a total universe of about 12,000 diseases, it is obvious that, on occasions, a clinician will simply not think of a diagnosis either because he did not remember it or never knew it in the first place.

If there is diagnostic doubt, the clinician then typically has to consult with colleagues, read textbooks or research online in order to investigate further. With medical textbooks and online reference resources, it is very difficult to search for something when one does not know what to look for. A search for "toxic shock," for example, will provide huge amounts of information; but, if you are unsure and just know that the patient has ankle pain, ankle edema, diarrhea and fever, then the traditional reference resources are not very helpful in connecting and making sense of all of these signs and symptoms.

In these more unusual or complex clinical presentations, diagnostic decision aids can be particularly helpful, as they are designed to produce a list of likely diagnoses for a given set of signs and symptoms. Their job is to get the clinician thinking about a disease that he had not thought about previously. Instead of taking several hours, days or even years in some cases to suggest the right diagnosis using the traditional methods, the diagnosis decision aids work in seconds. These tools buy the time that the clinician needs to think.

Patient engagement

There is now an increasing body of evidence and growing movement to engage the patient much more in their care and treat them as a partner in the care team rather than as a passive recipient. This is particularly relevant to diagnosis as the patient is clearly the expert on their symptoms and their story, and 80% of the time, the information they can provide reveals a diagnosis. The National Patient Safety Foundation's Lucien Leape Institute has recently published a **major report on patient engagement**.[8]

In order to help patients make sense of their symptoms, they should be encouraged to use properly designed 'symptom checkers' (discussed further in the next section) to help them research their diagnosis and contribute to a differential diagnosis jointly produced by them and their doctor. Patients could even be encouraged to use a symptom checker before a consultation their findings can form part of the medical note and a good basis for the start of the consultation.

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[6] "Diagnostic Errors in Primary Care: Lessons Learned", John W. Ely, MD, Lauris C. Kaldjian, MD, PhD and Donna M. D'Alessandro, MD, **http://www.jabfm.org/content/25/1/87.full**

[7] "Types and origins of diagnostic errors in primary care settings", Hardeep Singh, MD, MPH; Traber Davis Giardina, MA, MSW; Ashley N. D. Meyer, PhD; Samuel N. Forjuoh, MD, MPH, DrPH; Michael D. Reis, MD; Eric J. Thomas, MD, MPH, **http://jamanetwork.com/journals/jamainternalmedicine/fullarticle/1656540**

[8] "SAFETY IS PERSONAL Partnering with Patients and Families for the Safest Care", The National Patient Safety Foundation's Lucian Leape Institute, **https://c.ymcdn.com/sites/npsf.site-ym.com/resource/resmgr/LLI/Safety_Is_Personal.pdf**

What diagnosis decision aids are available?

For the healthcare professional

With the nature of the diagnostic process, technology advances have long been seen as as potentially useful tools to help support the clinician. Initial attempts in the 1960's were focused on the improvement in diagnosis of one specific problem, such as abdominal pain. Although these showed that clinicians did a better job when using them, the tools were time-consuming and proved to be impractical for use in a busy clinical setting, so they were never adopted. Another factor was that the intended users were specialists, who had less need of the tools.

The 1970s and 80s brought the first general diagnostic tools such as **DxPlain**, **QMR,** **Diagnosis Pro** and **Iliad**. These tools were also not widely adopted, primarily due to the time taken to use. Although DxPlain and Diagnosis Pro are still available, QMR and Iliad have all but faded away. These systems were highly developed, but were limited by the technology available when they were launched. The tools are "rules-based systems," which means that each symptom is associated with a particular disease with an assigned probability. These systems work satisfactorily on a small scale, but become difficult to manage on a large scale as each symptom or diagnosis needs to be kept up to date. The rigid nature of a rules-based system also means that the user can only enter a feature that is in the system's database. A by-product of this problem is that it makes it more difficult to fully integrate these systems into electronic medical records.

Isabel marked the new generation of diagnostic tools and was first introduced in 2001. Isabel uses a statistical natural language processing (SNLP) engine applied to a database of disease presentations or illness scripts, rather than a rules based model; it is a much more complex system behind the scenes, but a much simpler system to use.

IBM Watson has more recently entered the medical field, seeking to adapt its Jeopardy! winning system into a tool for diagnosis and treatment. Watson aims to use both SNLP and NLP applied to a broad base of 200 million documents from textbooks through blogs. Latterly, it seems that Watson is more focused on helping with treatment decisions rather than diagnosis.

VisualDx is another system but it is primarily based on digital images and allows clinicians to build a visual differential diagnosis based on actual patient findings.

Google.com is also commonly used as a diagnosis aid. In 2006, the BMJ ran a study entitled "**Googling for a diagnosis – Use of Google as a diagnostic aid: internet based study**".[9] The results showed that Google included the final diagnosis in 58% of cases but only when "statistically improbable phrases" were entered and three possible diagnoses were pre selected from Google's list of documents by 2 specialists. It should be noted that Isabel and DxPlain found the final diagnosis under more realistic test conditions in the 90% range.

Objective Reviews

Few objective reviews of these systems exist, but the most recent review has been published by Nicholas Riches from the University of Manchester in the UK *et al* in their paper "**The Effectiveness of Electronic**

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[9] "Googling for a diagnosis—use of Google as a diagnostic aid: internet based study," Hangwi Tang, Jennifer Hwee Kwoon Ng., BMJ, doi:10.1136/bmj.39003.640567.AE (published 10 November 2006)

Differential Diagnoses (DDX) Generators: A Systematic Review and Meta-Analysis"[10]. This was a systematic review and meta-analysis to investigate the efficacy and utility of DDx generators. The authors concluded that DDx generators have the potential to improve diagnostic practice among clinicians but due to the variability of the studies they were not unequivocal.

An earlier review was published by William Bond from the Lehigh Valley Health Network *et al*, in his paper titled, "**Differential Diagnosis Generators: an Evaluation of Currently Available Computer Programs**."[11]

Overall 4 systems were evaluated on various criteria and scored on a 5-point scale. Isabel and DxPlain scored the highest at 3.45 each. Unfortunately, the study did not take into account two important criteria: first, the ability to integrate into an EMR, and second, the ease of use – both important when assimilating these types of tools into the daily practice of busy clinicians.

You can find more recent studies and objective reviews on the **Isabel Website**, which is updated regularly.

For the patient

Diagnostic tools for the patient are known as 'symptom checkers' and vary enormously in what they attempt to do. They range from triage tools for a single symptom that aim to direct the patient to appropriate care settings rather than provide real diagnosis help, to more sophisticated tools such as the Isabel Symptom Checker.

WebMD is the most visited health portal in the world and offers a symptom checker on its home page. This symptom checker, like many others, is a rules based system which means that the user can only enter the symptoms contained in its database. The tool currently covers around 470 symptoms.

Mayo Symptom Checker is another well known system which covers just the most common symptoms. Only single symptoms can be entered.

AskMD is actually the original Problem Knowledge Coupler system that was designed by Larry Weed which has been developed into a patient tool. Although it only works on a single symptom, it takes the user through a detailed set of questions - often 20 to 25 - about the symptom to try and pin point a cause.

Symcat is a newer symptom checker that uses existing patients records to compare with what a user has entered. At present it covers 474 symptoms, around the same as WebMD.

iTriage is designed to help direct patients to the most appropriate care setting near to them. Currently it covers around 400 symptoms and also works with single symptoms only.

Isabel Symptom Checker was released at the end of 2012 and is closely based on the Isabel DDx Generator used by healthcare professionals. As a professional grade tool it covers an infinite number of symptoms and several thousand diseases. It also includes a triage tool which helps patients decide where to present if they want to see a healthcare professional.

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[10] "The Effectiveness of Electronic Differential Diagnoses (DDX) Generators: A Systematic Review and Meta-Analysis", **journals.plos.org/plosone/article**

[11] "**Differential Diagnosis Generators: an Evaluation of Currently Available Computer Programs**." William F. Bond, Linda M. Schwartz, Kevin R. Weaver, Donald Levick, Michael Giuliano and Mark L. Graber

Implementation of a Diagnosis Decision Support System (DDSS)

After many years of development, diagnosis decision support systems (DDSS) have now come of age and are practical for use in busy clinical settings. The only remaining hurdle is widespread clinical adoption.

This white paper is unable to present information on implementations of other tools, so the following comments are based primarily on the experiences from implementations of the Isabel Healthcare diagnosis tool. In this view, there are 10 key steps for a successful implementation.

Ten Tips for Successful Diagnosis Tool Adoption

1. **Start with Leadership Support -** The administration and clinical leadership must be strongly supportive of the tool's use in the hospital or health system. The institution must have a clear end goal that could be the general improvement in diagnosis quality or a more specific one such a reduction in appropriate referrals or test ordering so there is clear purpose for the use of Isabel. The widespread adoption and regular use of Isabel should always be the means and not the end.

2. **Secure Stakeholder Buy In -** Get buy in from the many other stakeholders in improving diagnosis. As stated at the beginning of this paper, the stakeholders are many and some may not even think that diagnosis is an important element of their business. Examples of key stakeholders are: risk management, education, nurses, mid-level clinicians, utilization management, accountable care champions, patient satisfaction, patient safety and quality etc.

3. **Find a Champion -** Identify a senior clinical champion to drive adoption and awareness. This person needs to be passionate about improving the quality of care and patient safety and not just have diagnosis as one of another 20 projects. Ideally this person also needs to be a practicing clinician or perceived by his peers to be "in the trenches."

4. **Communicate Early -** Before "go live" the key clinicians should be fully aware that the system is coming so that they are expecting it and, ideally, looking forward to it.

5. **Ensure Ease-of-Use -** Access needs to be made very easy with prominent links to the system from various obvious (to the clinicians) places. It's no good if it takes six clicks just to find the system. It can be accessed via the EMR, from a desktop, from favorites pages, library pages and mobile devices, etc.

6. **Plan a Launch Kick Off event -** Have a well-planned launch event to kick start awareness and show that your institution is serious about improving diagnosis. Senior clinicians should be in attendance and be visibly and vocally supportive. It doesn't help with widespread adoption if they take the attitude that using diagnosis decision support systems is beneath them.

7. **Focus on the Right Audience -** The initial focus should be on the right clinical groups such as family practice, hospitalists, internists, pediatrics, residents and nurses. The specialists will generally be less receptive since they will not see the need.

8. **Keep it Top of Mind -** Awareness campaigns should be continual with use of the tool built into or preferably mandated into routine events such as daily rounds, daily reports and other educational forums. Clinical champions should inquire if residents have used the tool and remind them to do so. One champion of an Isabel institution asks his residents whether they "have Isabeled a patient and whether are comfortable that they have thought of everything." Others use triggers built into the EMR that, for example, ask for a list of all patients in their institution that have been there for 2 days or more with no diagnosis and then runs them through Isabel. Another obvious trigger could be patients re-attending for the same complaint.

9. **Promote Successes -** The clinical champion should be on the look out for early wins and then publicize them widely as these make great stories.

10. **Measure the Improvement -** Try to measure the changes and improvements. This task can be a challenge because diagnosis is often not measured at present. However, a picture of success can be drawn by examining changes in referrals, test ordering, patient satisfaction scores, readmissions, and then also surveying clinician opinions.

Return on Investment

Remarkably, although diagnosis is the first and most important decision made about the patient, there is almost no research showing what impact delays and errors of diagnosis have on costs.

About 16 years ago, one study investigated the impact of using a diagnostic decision support system (in this case DxPlain) on the "cost of service for diagnostically challenging cases." The survey showed that costs during the period studied for this type of case fell by 12%. Although the drop is significant, the authors were unable to show which costs changed. Although the study was launched in 2000, **the paper**[12] was not published until 2010 in the International Journal of Medical Informatics.

With the significance and downstream impact of the diagnosis decision, there are many places where its impact can be seen and measured – and this information helps to build a return on investment case:

1. **Avoiding Litigation -** The simplest and starkest impact of diagnosis support is simply an avoided lawsuit. The saving of just one diagnostic malpractice case is likely to pay for the use of the tool for at least a decade.

2. **Appropriate Referrals -** Within an Accountable Care organization, getting referrals to be appropriate will have a key impact on utilization of specialist or secondary care and profitability. About 50% of referrals from primary care to specialists are for diagnosis reasons, therefore the use of a diagnostic decision aid can be effective for improving referral appropriateness, especially when referral management centers have not been shown to be effective. **The Kings Fund published a comprehensive paper on this topic in 2010.** In addition, **an Isabel study examining the impact on referrals**[13] showed a significant benefit with 29% of GP's no longer feeling the need to make a referral after using Isabel.

3. **Appropriate Test Ordering -** With thousands of tests now available, it is nearly impossible for clinicians to remember which tests to order and when they should be used. Over 40% of tests are ordered just for defensive medicine. A DDSS is an effective tool that encourages clinicians to take a good history and consider carefully which tests are necessary to rule in or out the diagnoses they suspect rather than ordering a barrage of tests to hopefully reveal what the diagnosis could be.

4. **Improved work force skills -** Since healthcare is a knowledge-intensive industry the skills level of an institutions' workforce has a direct bearing how well important decisions, such as diagnosis, are made. Logistically, it is practically impossible to sustainably increase the overall sum of knowledge-based skills through additional training. The provision of a DDSS is an alternative and practical way of increasing skills. **The study carried out by Rosalind Franklin Medical School**[14] showing how the use of Isabel led to a 22% increase in diagnostic accuracy among their year 4 medical students

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[12] "The introduction of a diagnostic decision support system (DXplain™) into the workflow of a teaching hospital service can decrease the cost of service for diagnostically challenging Diagnostic Related Groups (DRGs)" Elkin PL, Liebow M, Bauer BA, Chaliki S, Wahner-Roedler D, Bundrick J, Lee M, Brown SH, Froehling D, Bailey K, Famiglietti K, Kim R, Hoffer E, Feldman M, Barnett GO, **www.ncbi.nlm.nih.gov/pubmed/20951080**

[13] "The Impact of a Web Based Diagnosis Checklist System on Specialist Referrals from Primary Care", Isabel Healthcare, **http://smdm.confex.com/smdm/2010on/webprogram/Paper5940.html**

[14] "The impact of a diagnostic reminder system on student clinical reasoning during simulated case studies", Carlson J, Abel M, Bridges D, Tomkowiak J., **http://www.ncbi.nlm.nih.gov/pubmed/21330845**

demonstrates the potential benefit for many clinicians.

Another **study**[15] by the same team compared one group of students using Isabel with another group consulting with the senior colleague, in this case their resident, and found that the group using Isabel became more accurate with their diagnoses and appropriately more confident while the group who consulted with their resident became marginally less accurate and inappropriately more confident.

5. **Shorter LOS -** This means there are less wasted days of care due to delays in diagnosis. Isabel Healthcare has a table that allows an institution to make assumptions about the proportion of patients where there might be diagnostic doubt (10% is a good average rule of thumb) and then the additional bed days or outpatient visits needed if the diagnosis/referral was not made in the optimal time.

6. **Improved Patient Satisfaction -** Many hospitals are investing in customer service techniques from other industries to boost their patient satisfaction scores since these will soon account for 30% of Medicare payments. However, many patients view these as gimmicks and not a tool to improve poor care. Since one of a patient's key concerns when visiting their doctor is getting the right diagnosis, using a DDSS with them has been shown to significantly increase satisfaction, as the patient feels as though they are being listened to and are reassured that a thorough assessment is being done.

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[15] "Does collaboration lead to fewer diagnostic errors?", Jim Carlson, PhD, PA-C; John Tomkowiak, MD MOL; Jeanette Morrison, MD; Wendy Rheault, PhD, DPT, **http://www.isabelhealthcare.com/pdf/collaboration_poster_AAMC_5-28-13.pdf**

The Isabel System – Differential Diagnosis Generator

How Isabel Works

The Isabel Differential Diagnosis (DDx) Generator is designed differently from the rules-based systems started in the 1970's and 1980's and instead uses statistical natural language processing (SNLP) applied to a database of over 10,000 diagnosis presentations or illness scripts.

SNLP software understands the meaning and concepts within natural language. As opposed to NLP that tries to make sense of every word, SNLP works by understanding the key concepts. This ability is similar to a human being who can generally understand what a conversation is about by just hearing a small amount of it rather than having to study every word. Higher success rates occur with applications using SNLP rather than NLP. **A useful explanation and comparison of NLP and SNLP is available here**.[16] The most important part of the SNLP application is the database and how the system is trained.

The Isabel Database

With the excitement about computers and artificial intelligence (AI) of the 1960-1980's, many system developers, in hindsight, were too ambitious about what the computer could do. Trying to build systems that could mimic and then replace clinicians, led to a lack of general acceptance of the technology.

The Isabel engine is designed to leverage what computers do best – quickly comb through massive amounts of medical knowledge and return a short list of possible diagnoses to hand over to the clinician. Isabel uses a database of over 10,000 diagnoses of which 6,000 are diseases and 4,000 are drugs. This database has been manually built and populated over nearly two decades with knowledge about how each disease presents from a multitude of sources. Although for some applications SNLP can automatically categorize documents, the software cannot give sufficient precision for medicine. The key to the performance of the system is not the quantity of knowledge that is indexed but its nature and quality.

Robust Algorithms

Other critical functions of the Isabel system are the many algorithms and files used to improve the quality of the initial query made and to filter the raw output. The main one is the synonym file, which is a functionality that uses a database of similar medical terms. For example, when a query of fever is entered into the system, it also searches for pyrexia and high temperature. This file also allows for common medical abbreviations such as SOB (shortness of breath) to be understood. Other algorithms are in place to filter the raw output so that the results are relevant for the age, gender and region of the patient.

Physician input will always be critical

The computer will never be able to tell the clinician what the patient's diagnosis is, as it never has all the contextual information about the patient such as how sick they look, how many times they have been back, the local population and environment.

Assisting with the Differential

Isabel's job is to take the clinical features that have been extracted by the clinician's careful history taking

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[16] "NLP", Wikipedia, **(http://en.wikipedia.org/wiki/Natural_language_processing#Statistical_NLP)**.

and examination, and produce a short list of possible diagnoses that could be the cause of that combination of signs and symptoms. The clinician can then use this list to form a differential diagnosis for his patient and the relative likelihoods of each diagnosis. Essentially, Isabel enables the clinician to carry out system 2 thinking more quickly (See Mark Graber's Forward in this white paper).

Mobilizing knowledge

An additional benefit of Isabel is that it serves as a mechanism to mobilize and organize knowledge. Traditional reference resources are only useful when you know what you are looking for and have a specific question. Isabel starts from the premise that you don't know what you are looking for, but have a patient with a certain set of clinical features. By providing a list of diagnoses that could be the cause of those clinical features, Isabel provides the clinician with specific questions to ask and effectively then mobilizes knowledge around each diagnosis.

Isabel System – Validation

Isabel has undergone a continual validation process since 2002. The published papers can be accessed from the **Isabel Healthcare website**.[17]

The validation process broadly falls into three categories:

- ✓ **Accuracy studies -** On average these showed that, when given the initial presenting clinical features, Isabel included what turned out to be the correct final diagnosis in 95% of cases.

- ✓ **Utility studies -** In general, these showed that in 10-12% of cases when Isabel was used, it reminded the clinician of an important diagnosis he had not thought of. When done live across three NHS hospitals, it was found that in a quarter of these cases it turned out to be the actual final diagnosis.

- ✓ **Impact studies -** These studies are not homogeneous and can be difficult to carry out since very few institutions currently look at the impact of delays in diagnosis on their own operations. Also, since diagnosis is based on a variety of factors, it is very difficult to attribute a change solely to one factor. The studies that have been done are:

  a) **The effect on referrals from primary care to specialists**[18] - This study was carried out across four practices in the UK. The study looked at the general practitioner's opinion of how the use of Isabel for each usage episode had helped. In 29% of cases the general practitioner said that he no longer felt the need to refer the patient and in 36% of cases where he did have to refer the patient, he said that Isabel had helped him refer more appropriately.

  b) **The Rosalind Franklin Medical School in Chicago**[19] looked at what effect the use of Isabel had on its year 4 medical students' diagnostic skills. They found that, on average, using Isabel improved their diagnostic accuracy by over 20%. It was noteworthy that those

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[17] Isabel validation process published papers: **http://www.isabelhealthcare.com/validation/peer-reviews**

[18] "The Impact of a Web Based Diagnosis Checklist System on Specialist Referrals from Primary Care", Isabel Healthcare, **http://smdm.confex.com/smdm/2010on/webprogram/Paper5940.html**

[19] "The impact of a diagnostic reminder system on student clinical reasoning during simulated case studies", Carlson J, Abel M, Bridges D, Tomkowiak J., **www.ncbi.nlm.nih.gov/pubmed/21330845**

students who were less skilled prior to beginning Isabel use, increased their accuracy by a greater percentage.

C) **The Rosalind Franklin Medical School**[20] also carried out what was effectively the first randomized control trial on Isabel by comparing two groups students and their diagnostic accuracy and confidence. One group used Isabel and the other one consulted their resident. The purpose of this study was to compare the traditional method of consultation with a senior medical colleague with using a decision support tool. The group that used Isabel improved their diagnostic accuracy significantly with an appropriate increase in confidence while the group that consulted their resident actually became marginally less accurate with an inappropriate increase in confidence.

We believe that both of these studies (b & c) have important implications for education and human resources within healthcare.

Isabel System – EMR / HIT Integration

A key advantage of the way Isabel works using free text is that it enables it to integrate easily with other systems and avoid the need for encoded data that has been the great weakness of previous systems.

Isabel can integrate at a 'light level' using the address query string method or be fully built into another system using published Application Programming Interfaces (API).

When integrated into an EMR system Isabel uses the clinical features that have already been entered by the clinician, either by free text or a structured template, to provide back a list of possible diagnoses. Selected diagnoses can then be saved back into the EMR to serve as a record of the differential diagnosis considered for the patient.

- Speech recognition software coupled with NLP can also now be used to parse key information from free text entry and dictated medical notes directly into Isabel.

- The new 'Isabel Active Intelligence' tool combines the power of Isabel and trained Natural Language Processing (NLP) software to enable Isabel to work from a complex free text progress note with no additional data entry. The NLP software automatically extracts the key clinical features from the progress note which allows the clinician to easily select which would be the most appropriate clinical features to pass to Isabel for diagnosis decision support. This new Isabel tool is currently being integrated into two major EMR systems and should be generally available during 2017.

- Current EMR vendors that offer an Isabel interface are: Epic, Cerner, NextGen, Allscripts, SystmOne, T-System, Better Day, and VersaSuite.

About Isabel Healthcare

Isabel was started in 1999 after the founder's daughter, Isabel, suffered a near fatal misdiagnosis. She was then three years of age and had chicken pox. Her local family doctor and emergency department doctors all missed a secondary infection developing which turned out to be necrotising fasciitis. She spent

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[20] "Does collaboration lead to fewer diagnostic errors?", Jim Carlson, PhD, PA-C; John Tomkowiak, MD MOL; Jeanette Morrison, MD; Wendy Rheault, PhD, DPT, **http://www.isabelhealthcare.com/pdf/collaboration_poster_AAMC_5-28-13.pdf**

three weeks in intensive care and four weeks in a high dependency unit but just survived and, today, is a healthy young woman.

Isabel Healthcare develops and markets the world leading diagnosis decision support system called Isabel. The Isabel system was originally started as a charity in 2000 but converted to a for-profit business in 2004 to ensure that it had sufficient funding to continue developing its unique system. The founding charity remains one of the largest shareholders in the company.

Isabel has been a proven diagnosis decision support system used by clinicians for the last 15 years. Over 30 articles have appeared in peer-reviewed articles covering various aspects of the system. The system was selected by the American Medical Association as the diagnosis tool for its portal. More recently the British Medical Journal (BMJ) endorsed Isabel as a new joint product was launched incorporating the BMJ's Best Practice tool.

Today, many high profile health systems, family practices and individual physicians use Isabel to help improve the quality of care they provide.

Conclusion

In the era of changing reimbursement, accountable care organizations, (ACO), bundled payments, pay-for-outcome, it is critical to determine the patient's diagnosis as soon as possible and get them on the appropriate treatment plan or guideline. Minimizing readmissions, unnecessary testing and the costs associated with them is crucial, and these improvements start with getting the diagnosis correct as soon as possible. Today, in 15-30% of the cases there is error or delay in reaching a diagnosis. The future of improved care quality depends on getting the first step of the care continuum, diagnosis, correct as quickly and efficiently as possible.

Although huge sums of money have been invested in EMR systems including CPOE and other patient safety related technology, using technology to assist in mitigating diagnosis error has all too often been put to one side as "too difficult to fix."

The excuse that diagnosis is "too difficult to fix" is now just that: an excuse. This white paper demonstrates that the solutions to diagnostic error are now thoroughly tried and tested, as well as extremely cost effective.

Appendix

Selected Diagnosis Error Related Resources

1. "**Improving Diagnosis in Health Care**" This is the landmark study published by the National Academies of Sciences, Engineering and Medicine in 2015.

2. "**Physician Perspectives on Preventing Diagnosis Errors**." This white paper by QuantiaMD helped launch their new series as it surveyed their users to learn more about the problem. The results represent the views of over 6,000 clinicians, mainly MDs. **http://www.quantiamd.com/q-qcp/QuantiaMD_PreventingDiagnosticErrors_Whitepaper_1.pdf**

3. "**Diagnostic Error: The Hidden Epidemic**," Physician Executive Online, Nov/Dec 2011, Mark Graber and Bob Carlson. This article also includes 53 references to other papers. **http://net.acpe.org/MembersOnly/pejournal/2011/NovemberDecember/Graber.pdf**

4. Additional **Diagnosis error articles**: A collection of important published papers is available online at Isabel Healthcare (http://www.isabelhealthcare.com/diagnostic-errors)

5. **New 'Diagnosis' Journal. 1st issue published in January 2104**

6. **Society to Improve Diagnosis in Medicine**.

Diagnosis Related Reading

A number of important books are also available on the subject of diagnosis:

1. How Doctors Think by Jerome Groopman, MD.
   Groopman presents this landmark book that describes the thought processes a doctor goes through in diagnosis, with examples of how it can go wrong. The book summarizes and makes more accessible the extensive research work carried out on clinical reasoning.

2. Diagnosis. Dispatches from the Frontline of Medical Mysteries by Lisa Sanders. Sanders is the technical adviser to the television show "House." In this book, she uses real stories to illustrate the problems and pitfalls of diagnosis decision making.

3. Over Diagnosed. Making People Sick in the Pursuit of Health by Dr. Gilbert Welch et al. This book is about the pitfalls of over testing in the pursuit of a diagnosis.