“Genius diagnosticians make great stories, but they don’t make great health care. The idea is to make accuracy reliable, not heroic.”

Dr. Donald Berwick,
former President and Chief Executive,
Institute of Healthcare Improvement, USA
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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?” 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 Fellow 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 2002, 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 the Society to Improve Diagnosis in Medicine.

Why the 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 effect 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 put 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.

- **Healthcare Reform**
  The changing healthcare payment landscape will force attention on to the quality of diagnosis decision making. The fee for service model is being phased out and performance based or outcomes based payment structures and fixed fee structures like bundled payments and patient centered medical homes will necessitate that mis- or delayed diagnosis frequency be reduced significantly as the costs associated with them will no longer be covered.

- **Medical malpractice**
  Misdiagnosis accounts for 30-40% of all malpractice claims and about 2/3 of all claims in primary care. This recent study looked at 350,706 claims over a 25 year period and found that misdiagnosis was the leading type at 28.6% but, more importantly, accounted for the highest proportion of total payments at 35.2%. “25-Year summary of US malpractice claims for diagnostic errors 1986–2010: an analysis from the National Practitioner Data Bank

- **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.

- **Patient safety**
  In the USA diagnosis error is now in the national spotlight with high visibility events making the national news. A Beckers Hospital Review highlighted diagnosis error to be one of the “10 most pressing patient safety considerations for 2014”. During National Patient Safety Foundation’s Patient Safety Awareness Week a series of three national webinars were presented; over 500 sites signed for the first two webinars and over 900 for the third. All three can be found on the NPSF website at the following link: [http://www.npsf.org/events-forums/patient-safety-awareness-week/diagnostic-error-webinar-series/](http://www.npsf.org/events-forums/patient-safety-awareness-week/diagnostic-error-webinar-series/). In addition an IOM committee will evaluate the existing
knowledge about diagnostic error in medicine from a patient safety perspective. The committee will examine many aspects of diagnosis error including burden and cost and make recommendations to reduce diagnosis error in medicine which may include medical education, reimbursement policies and technology. The report is expected to be out in Summer 2015 and is likely to make a significant impact as per previous IOM reports. The project can be found at: http://www.iom.edu/activities/quality/diagnosticerrormedicine.aspx.

- 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, is almost always preventable and causes 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. There are many intrinsic attributes to us as human beings that contribute to causing diagnosis related errors.

In a 2008 paper in “Medical Teacher,” Robert Trowbridge from Maine Medical Center sets out 12 Tips for teaching avoidance of diagnostic error.²

In 2014 the National Patient Safety Foundation published a very useful checklist entitled “Reducing Diagnostic Error: Ten Things I could do Tomorrow”

Premature Closure

As stated, the more common causes of diagnosis error 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 now 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,” 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” and “Diagnosis of difficult cases in primary care.” In the research, Kostopoulou found that the most significant factor is having a good differential 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 uniformity of the conclusions which are that the clinicians should have broadened their differential or done one in the first place. Examples are this are “Diagnostic Errors in Primary Care: Lessons Learned” by John Ely et al. and “Types and origins of diagnostic errors in primary care settings” 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 in primary care being only 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 a 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.

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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 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, 80% of the time, reveals the diagnosis.

The National Patient Safety Foundation’s Lucien Leape Institute has recently published a major report on patient engagement entitled “SAFETY IS PERSONAL Partnering with Patients and Families for the Safest Care” which pulls the latest evidence and thinking together.

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 so that what they entered forms part of the medical note and a good basis for the start of the consultation.
What diagnosis decision aids are available?

For the healthcare professional….

With the nature of the diagnostic process, technology advances have long been seen 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 and had less need of the tools.

The 1970’s and 80’s 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 record systems.

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 rather than a rules based model.

IBM Watson has more recently entered the medical diagnosis field, seeking to adapt its Jeopardy! - winning system into a tool for diagnosis and treatment. IBM expects to have the first pilot version ready in 2014. Watson aims to use both SNLP and NLP applied to a broad base of 200mn documents from textbooks through blogs.

VisualDx is another system but is based on digital images and allows clinicians to build a visual differential diagnosis based on patient findings which are visible on the skin.

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”. 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 William Bond from the Lehigh Valley Health Network et al, in his paper titled, “Differential Diagnosis Generators:

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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 critical factors when assimilating these types of tools into the daily practice of busy clinicians.

KLAS, a research firm that measures the performance of healthcare technology, conducted a specialty study on Clinical Decision Support (CDS) systems in 2011. The KLAS evaluation process involves validating vendors’ performance at a minimum of 15 unique, acute care provider organizations. The study highlighted the strengths and weaknesses of each system in terms of functionality and vendor performance as reported by healthcare providers.

This report included a category for diagnostic support and included reports on both VisualDx and Isabel. Both scored over 92% and were amongst the highest scores across the entire report. The full report can be purchased from KLAS directly.

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 setting rather than provide real diagnosis help to more sophisticated tools such as the Isabel Symptom Checker.

NHS Direct produces a series of symptom checkers which are currently made available on various health portals such as NHS Choices, Patient.co.uk and NetDoctor. These work for a single symptom or problem and take the user through a series of questions and then conclude with advice as to whether the user should treat themselves at home, see their GP or go to their hospital emergency department.

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.

Everyday Health includes a symptom checker. This works with just one symptom but uses additional questions to come up with a recommendation as to when and where you should go to a doctor.

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

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

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 in 2012 and is closely based on the Isabel diagnosis decision support tool used by healthcare professionals. As a professional grade tool it covers an infinite number of symptoms and several thousand diseases, using free text for symptom input.
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 a clinical adoption issue.

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 reduction in inappropriate 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 their 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 in the EMR. 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, remotely, tablets, 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 actions such as referrals or regular educational forums such as daily
rounds or reports. 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 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.

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 which sets in motion a huge proportion of subsequent health care costs, there is almost no research showing what impact delays and errors of diagnosis have on costs.

About 10 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 in detail which costs changed. The paper was published in 2010 in the International Journal of Medical Informatics.

> “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)”


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 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:

study examining the impact on referrals 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 over 2,000 tests now available, it is nearly impossible for clinicians to remember which tests to order. 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 Workforce 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 showing how the use of Isabel led to a 22% increase in diagnostic accuracy among their year 4th year medical students demonstrates the potential benefit for many clinicians.


   Another study (large PDF file) by the same team compared one group of students using Isabel with another group consulting with a 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 which consulted with their resident became marginally less accurate and inappropriately more confident.

5. **Shorter LOS:** 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.

**The Isabel System – Diagnosis checklist tool**

**How Isabel Works**

The Isabel diagnosis checklist tool 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.

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 at http://en.wikipedia.org/wiki/Natural_language_processing#Statistical_NLP. The most important part of the SNLP application is the database and how the system is trained.

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. For instance, 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 14 years 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 to be understood. Other algorithms are designed 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 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 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.

Mobilizing knowledge. An additional benefit of Isabel is that it serves as a mechanism to organize and mobilize 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, noted below.\(^9\) 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: 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. [http://smdm.confex.com/smdm/2010on/webprogram/Paper5940.html](http://smdm.confex.com/smdm/2010on/webprogram/Paper5940.html)

b. The Rosalind Franklin Medical School in Chicago 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 students who were less skilled prior to beginning Isabel use, increased their accuracy by a greater percentage. The study is cited in Pubmed: “The impact of a diagnostic reminder system on student clinical reasoning during simulated case studies.” Available at: [http://www.ncbi.nlm.nih.gov/pubmed/21330845](http://www.ncbi.nlm.nih.gov/pubmed/21330845).

c. The Rosalind Franklin Medical School also carried out what was effectively the first randomized control trial on Isabel by comparing two groups of 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 was actually marginally less accurate with an inappropriate increase in confidence. This paper was presented at the Diagnostic Error in medicine conference in 2014 and the poster can be accessed [here](http://www.isabelhealthcare.com/home/peer/new1) (large PDF file).

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

\(^9\) Isabel validation process published papers: [http://www.isabelhealthcare.com/home/peer/new1](http://www.isabelhealthcare.com/home/peer/new1)
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, 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 available during late 2014 or 2015.

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

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 three weeks in intensive care and four weeks in a high dependency unit but just survived and, today, is a healthy young teenage 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 10 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, right 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


2. QuantiaMD, the online physician learning collaborative, recently launched a series on diagnostic error (http://www.quantiamd.com/home/diagnosticerrors) featuring recorded short presentations by many of the experts in this area.

3. “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-gcp/QuantiaMD_PreventingDiagnosticErrors_Whitepaper_1.pdf


5. Additional Diagnosis error articles: A collection of important published papers is available online at Isabel Healthcare (http://www.isabelhealthcare.com/home/diagnosis_error/news4)


7. 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.