

A Systems Model of Diagnosis
Part-1
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A Mathematician looks at his own illness

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Abstract

The problem of diagnosis is common in many disciplines, even though the models, methods and techniques used are very different. Diagnosing problems in complex information systems is a very complex problem similar to diagnosing a particular disease in a patient. In fact, in both disciplines, diagnosis is still considered an art or a skill that a practitioner possesses, and it is even considered an art that can not be taught. However, the approach that information scientists adopted is very different from the approach that medical practitioners adopted towards diagnosis. The medical science is very advanced in the diagnostic tools, laboratory tests, diagnostic imaging and so on. For every corresponding test, there is a list of diseases that are indicative of the test results. However, though such an elaborate diagnostic tests are not available in the information systems (primarily because each information system is different from another), the information scientists use certain mathematical and logical models to arrive at a problem.

In this paper, I presented how a mathematician or an information scientist applies a model driven techniques to diagnosis of a medical problem. I used my own illness as a case study to illustrate the techniques used. The objective of the paper is to explore if there are any ways of bringing the two approaches together. Information Scientists can greatly benefit by developing a statistical approach to diagnosis, developing techniques to conduct simple “blood tests” of their systems to help diagnose the systemic problems, and medical practitioners can benefit by adopting model driven, logical frameworks to minimize their diagnosis time, and use the models to analyze complex cases.

Introduction

This paper is a case study of two different methods of diagnosis. I used my own illness and experience as a case study to illustrate a logical model of diagnosis that is commonly used in information sciences. I suffered from gastro-intestinal problems for about six months, and it took several visits to hospitals, doctors, and several kinds of tests ranging from blood tests to surgery to diagnose the problem. The whole process took about six months, and it cost a lot of time and money before the doctors were able to identify the exact disease and prescribe the course of treatment. As a logician and a mathematician, it was great experience for me to watch the doctors trying to diagnose illness. The method used by most doctors starts by listening to the patient explaining her symptoms, and then they ask a series of questions initially to get a better (technical) understanding of the symptoms, and then they very quickly form in their minds what could be the disease, and using this as the basis, they ask for the presence of other symptoms (for example, do you also have fever in the evening?), and the next step in the diagnostic process is to order certain tests - which basically serve as a confirmation of the diagnosis that the doctor already made. If the tests indeed confirm the initial diagnosis, then all is well, however, if the tests are not positively indicative of any particular disease, or if they fail to confirm the ‘initial’ diagnosis (suspicion is a word used by many doctors, and logicians prefer the word hypothesis), then the doctor orders a few more tests, or goes over the symptoms once more to come up with another diagnosis and so on. In a nutshell this explains the model of diagnosis that is used by many doctors.

However, in my observation as a formal mathematician, it is important to distinguish between the problem being reported by the patient, and the symptoms (or the suffering that the patient undergoes), and the possible disease. Note that from a mathematical point of view, there is a huge difference between the problem and the disease. The disease itself is not the problem, until it is identified as such. The symptoms are also not problems - they are only indications of the manifestation of the problem.

In the rest of the sections, a logical model of diagnosis is presented illustrated by my own case.

The Problem:

For almost last six months, I reported the following problem to all the doctors I visited:

1. Absolutely no hunger - even if I don't eat anything for a number of days, I don't feel hungry even though I become weak.
2. The quantity of stool passed, the quantity of food consumed have no relationship. In other words, the quantity of stool passed is more or less constant, leading to stomach discomfort, bloating and so on.
3. The stomach is always bloated - technically speaking distended and feels full even though ostensibly nothing has been consumed.

An analysis of the problem without taking into account any other information:

A careful analysis of the three problems indicates that there is a fundamental problem with the digestive process of the patient. Not feeling hungry and not being able to flush out after the digestion indicates that the cycle of digestive process (feeling hungry, eating something, digesting the food, and flushing out the post digestive waste material) has either slowed down considerably, and is slowly coming to a grinding halt.

However, the symptoms presented do not indicate any serious illness - the patient only reported discomfort after eating, and discomfort with passing stools, otherwise, the patient is fully functional and looks to be in good health. The innocuousness of the symptoms presented led many doctors to assume that the problem is not serious, probably because they were looking at the symptoms and not connected with the basic problem reported by the patient (that there is a fundamental problem with the digestive process itself).

Logically, all three problems are related together. Let's try and construct a small function to explain the relationship:

1. The quantity of stool and quantity consumed are related variables. After digestion, the residue is what goes out as stool. Assuming that the composition of my diet has always consisted of the same residual factors, we now ask the following question:
 - a. When the quantity consumed is increased, is the stool quantity relatively increasing by the same factor? The answer was no. Therefore, we can conclude that post digestion, there is some solid content sitting in the stomach? Does this lead to "feeling of fullness" and "stomach distention"?

The above conclusion explains Problem 1 and 3 because of the following reasons:

- a. the stomach distends because there is always some solid content in the stomach. (Problem-3)

- b. Assuming that the stomach has no alternative but to keep digesting any material that is put into it, has it been trying to consume the waste material again and again - leading to weight loss, discomfort, distention and the loss of hunger?

This model presented here is very simplistic, because we presented only a static picture and not the dynamics of the complex GI functions. However, there is an important conclusion from a diagnostic point of view which I felt was ignored by many doctors - it is that the distention is because of a solid material in the stomach, and not simply accumulated gas in the stomach. When I repeatedly told the doctors that I don't pass gas - they refused to consider this as important evidence, but instead insisted that I may not know, but I must be passing gas without knowing, and some doctors even started treating me for general constipation and acidity.

With this conclusion (which indicates a serious illness), let's look at the symptoms, and the diagnostic information:

Symptoms, History and Additional Information:

- a. Weight Loss - approximately 10 KG during June'05 - Aug'05. No Weight Loss between Sep - Dec'05.
- b. Abdominal Distention - measured as the difference in the circumference of the belly in the morning and the night. The distention is around 4-5 inches every night.
- c. Irregular bowel movements. The pattern of bowel movements is very difficult to explain. Firstly there was no history of diarrhea. At the beginning stages of the illness, there is used to be an urge to go to toilet, but nothing would happen, or very little stool would come, and there was never a feeling of having completed (emptied) the bowels. But, towards the end, there was not even an urge to go to toilet, and the bowel movements were reduced to once in two days. It is important to note that because of the discomfort, I started eating very little as well. This complicates explaining the pattern of the bowel movements.
- d. Some pain in the lower right and left side of the stomach (only when pressed during the physical examination).
- e. Occasionally there is a needle, prickling type of pain which subsides almost instantly.
- f. Constant, audible bowel sounds. The sounds used to be very high and audible and persistent at the beginning stages of the illness, but again towards the recent times, there are virtually no bowel sounds. The sounds used to be very high during the night time, and my wife - sleeping next to me, could hear them.

During this time of illness, I developed a severe arthritis of the left ankle, and was hospitalized for a week. Initially, the doctors suspected it was some sort of infection, but later after I explained to them several times, my problems with the stomach and general illness, they concluded that it is a reactive arthritis.

Summary of various diagnostic tests

1. Upper GI Endoscope revealed Antral Gastritis
2. Colonoscopic Biopsy Revealed presence of large number of Lymphocytes - *suggesting inflammation*
3. CT-Scan of the abdomen revealed bowel wall thickening, circumferential diffusion and some lymph nodes around the ileo-caecal region, *suggestive of inflammation/infection*
4. Blood tests indicated high CRP and low ESR counts
5. Laparoscopic Examination of the intestines revealed several adhesions around the ileum, and around large and small intestines. It also revealed small bowel congestion

6. Laparoscopic biopsies revealed infiltration of lymphocytes - *indicative of inflammation/infection.*

A logical model of diagnosis

We need to construct a careful model that helps us to diagnose the problem since the symptoms are very generic, non-specific, cannot be represented or demonstrated in front of the doctor. The symptoms indicate a range of problems - therefore, correct diagnosis using common sense approaches and without a scientific model is a very tough task, and even dangerous.

I present below a model for diagnosis. The model basically uses certain divide and conquer techniques. First we choose a range of possibilities and for each possibility - apply a set of heuristics to eliminate or formulate a diagnosis.

Many diseases can be classified into four major categories:- infections, inflammation (auto-immune problems), neurological disorders, and finally functional breakdown of organs involved. The diagnosis, and treatment varies considerably in each of the categories, and each is more difficult to diagnose than the other (in the other order presented).

Let's consider each of these categories to the case study that I presented in the previous sections.

Infection

If we could prove that the problem is indeed infection and pinpoint the infection - it would be the best possible diagnosis from a treatment point of view, mainly because the diagnosis tends to be hundred percent certain, and many infections have a prescribed cure and treatment. However, a diagnostic model cannot be biased towards infection - any bias will influence the choices we make and therefore, the model has to remain strictly non-partisan towards any particular possibility. This is an important consideration and is in fact the basis of any scientific system. However, this basis is ignored by many practitioners - both computer scientists as well as medical practitioners.

1. The symptoms presented indeed point towards several infections.
2. The GI tract is most susceptible for infections
3. The different tests (tests 2-6) only *indicate* infection, but they don't *conclusively prove* the presence of any infection

Therefore, we need some other evidence. Evidence is the logician's best friend and also his worst enemy. This is because, one of the main tenets of formal logic state that *absence of evidence is not evidence of absence*. This is the biggest dilemma - if we cannot find an evidence, we cannot rule out infection - which means that we have an inconclusive diagnosis. At the same time, if we can establish the evidence, then the diagnosis is complete, and we can start a treatment/cure.

In the light of the above observations, let's analyze if there is any evidence of infections in this case. I present the following as evidence of infection:

1. The presence of Gastritis. Gastritis is now known to be a bacterial infection. This is a direct evidence.
2. I had a very severe case of reactive arthritis - which indicates that there is/was some infection in the body. This is an indirect evidence. In order to consider this as an

- evidence, we may have to go into the correctness of the diagnosis of the reactive arthritis.
3. When I was treated for Gastritis with anti-biotics, there was a remarkable improvement in my condition - albeit temporarily. This is neither a direct evidence nor it is an indirect evidence, but it evidence by deduction - since there was improvement, we deduce that there indeed was evidence of infection.
 4. At the same time, by using the same method, we have to suspect the infection theory as well for the following reasons:
 - a. If it were indeed Gastritis, why was there only a temporary improvement in the condition?
 - b. During the treatment period, there was a severe attack of reactive arthritis which indicates that for some reason, the treatment flared up a sleeping giant. It is quite possible that the problem is not at all Gastritis. Until this point, the direct evidence we have is what is identified as Gastritis. We will elaborate on this important evidence in the next section on inflammation.
 5. The symptoms and the problem progressively deteriorated. More importantly *the rate of deterioration* was too rapid. This needs some explanation - even in other categories, the symptoms may worsen over a period of time, but in this particular case, there are two points that are worth considering:
 - a. The rate at which the symptoms worsened is fairly rapid, indicating that the infection is spreading or getting stronger.
 - b. Over time, the 'rate' itself was very rapid - in other words, there was acceleration in the deterioration.
 - c. Does this serve as another evidence of infection?
 6. There is no other evidence contradicting the infection hypothesis. This itself is evidence by rebuttal. If we cannot evidence of any other category of diseases, and there is some evidence in support of infection - then the absence of evidence of other categories has to be treated as evidence in support of infection. A pure logician may not agree with such a heuristic - as there is no such provision in the first order logic, but this is a well established reasoning technique in advanced reasoning systems like non-monotonic reasoning, default reasoning and McCarthy's circumspection logic.

So far, we established the following types of evidence:

1. A direct evidence (Gastritis)
2. Indirect evidence (reactive arthritis)
3. Deductive evidence (temporary improvement in the condition when treated for an infection and later the rate of worsening of the problem)
4. Evidence by rebuttal (there is no other evidence for any other category)

Since there is direct evidence and several other pieces of deductive and indirect evidence in support of infection - we can now reasonably conclude that indeed the problem was some type of infection other than Gastritis.

It now remains to be identified. However, this is something a logician cannot do, it requires the expertise, knowledge and also access to advanced diagnostics of a medical practitioner. However, using several online information sources, I was able to come to the conclusion that the problem must either be Intestinal Tuberculosis or Crohn's Disease.

Inflammation

There is a certain weakness in the infection model that is presented in the previous section. The entire model rests on one piece of direct evidence - it is the endoscopic finding of Antral-Gastritis. The rest of the evidence only supports and builds on the direct evidence.

The physicians came to the *conclusion* of Gastritis by visual examination of the stomach using an endoscope; it was not confirmed by a biopsy. Here, the physician used his experience to conclude by visual examination. Therefore, it is subjective evidence, not objective evidence.

All tests are as indicative of inflammation as they are about infection. There are two types of inflammatory diseases of the GI tract - known together as Inflammatory Bowel Disease - one of them is ulcerative colitis and the other is Crohn's Disease.

The colonoscopy ruled out Ulcerative Colitis. Therefore, only Crohn's Disease remains to be examined. There is strong evidence suggestive of Crohn's and also counter evidence.

One of the major symptoms of Crohns is Diarrhea which is absent in this particular case. This is one reason why many physicians's ruled out Crohns in this case. However, this is based on only what the patient communicated - which may not be very accurate. Since the patient was suffering from a long duration, it is quite possible that he had lose stools and diarrhea, but did not remember it, or he may have thought that it does not quality for diarrhea. We must also consider that the presence of absence of a symptom is not the only way to detect a particular disease. A symptom is usually associated with a disease using a statistical evidence gathered over a period of time. Like any other statistical measure, it is never a hundred percent certainty. Therefore, we cannot consider this as evidence against Crohns. It is an observation, but not scientific evidence.

Another observation against Crohns is that it is mainly a developed country disease, not known to be common in Asian population. Agan, this is a statistical observation, not a scientific evidence.

The evidence in support of Crohns:

1. The affected areas in the GI are multiple. There is inflammation in the small intestines, at the ileo caecal junction, and in the upper abdomen (the Gastritis may not be Gastritis at all, but could be inflammation due to Crohns). The signature of Crohns is that it affects multiple areas of the GI tract.
2. All biopsies indicated inflammation
3. High Level of CRP also indicates inflammation as well.

In order to establish that the disease is Crohns, all we need to do is to establish that what was supposed to be Gastritis is not Gastritis, but it is Crohn's in disguise. If a biopsy sample from the upper abdomen (from the Gastritis area) was taken and checked for a Crohn's profile, then we are on a very firm ground. It establishes two things:

1. It establishes that what is thought to be Gastritis is not Gastritis - therefore, throwing the infection theory out of the window. The entire infection theory rested only one direct evidence - which is Gastritis.
2. It establishes that there is inflammation in multiple places (upper and lower abdomen), which itself is strong evidence in support of Crohns.

Neurological Causes

Even though in this particular case, the evidence is too strong in favor of infection or inflammation, let's assume for the time being that it is not, for the sake of presenting our model completely.

If we had ruled out infection and inflammation completely, then we have to examine the reasons behind the rate of slowness in the function of the GI tract. Is the problem due to the GI tract has considerably slowed down and the patient has not adjusted to the slowness? In such cases, is it due to any neurological problems - for example, motor function problems? Though it seems highly unlikely, it is a possibility.

Functional Failures

In the event of all the above categories of problems are ruled out, we are left with only functional failure of the organs involved as the only alternative. The medical science do not yet understand the causes of functional failures or functional disorders, and there are not much of cures and treatments available for functional failures - though lot of research tries to isolate the functional disorders to one of the three categories mentioned above. In a recent study, it was found that Crohns diseases has a very high statistical correlation with the usage of frozen foods, and refrigerators. Scientists now are trying to establish that Crohns is in fact an infection caused by a certain bacteria that survives in the low temperatures of the frozen foods.

Other logical possibilities

In the event that we find data supporting multiple possibilities or if we find data that is not conclusive, we must consider if there are multiples variables affecting each other inversely or in conjunction. For example, there is a logical possibility that there is small intestinal cancer and intestinal tuberculosis each affecting the other - the cancer preventing the TB from spreading and the TB preventing the cancer from spreading?

Conclusions from the case study

The model of diagnosis presented here - at least in my limited experience with physicians - is very different from the model of diagnosis used frequently.

Most often physicians related symptoms directly to a certain disease, and then try to prove their assumptions. In our case, we don't make any assumptions about diseases - the model is independent of any particular symptoms or diseases.

Our model provides a method to include many observations together about the patient's condition and consider them as diagnostic information. For example, much of the deductive evidence presented in this paper is not obtained from any lab test reports, but logically putting together information. The model also eliminates any subjectivity and preconceived notions as well - for example, questioning the validity of the Gastritis diagnosis without a biopsy.

We make a clear distinction between the patient's problem, the various associated symptoms and different observations and finally even between the problem and the disease. In this particular case, the problem is not abdominal discomfort, but the slowing down of the entire digestive function of the body. This distinction helps us to relate to the problem more intimately and helps us to solve the case holistically. Similarly, there may be more than one disease causing the problem - for example, it might be a case of Irritable Bowel Syndrome, and Inflammatory Bowel Disease and a minor infection all working together. In such cases, we need to isolate the symptoms and diseases and relate all of them to the problem at hand.

The model presented here allows a holistic approach to diagnosis, instead of highly 'test & procedure' driven approaches without a underlying model of diagnosis in place. Reasoning and using heuristics is part of any advanced diagnosis - and therefore, we have to make sure that the heuristic model rests on sound logic and reasoning applied using formal techniques.

In the next part of the paper, we apply the case study to the distributed systems, and draw analogies between the medical diagnosis and the diagnostic difficulties with complex

distributed systems. Finally we present how the model proposed above could be used to enhance the diagnostic capabilities of any systems - be it complex distributed systems, or the most complex system in Mother Nature - the human body.