Artificial Intelligence in Medicine: an Introduction

Acknowledgement

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Introduction

From the very earliest moments in the modern history of the computer, scientists have dreamed of creating an 'electronic brain'. Of all the modern technological quests, this search to create artificially intelligent (AI) computer systems has been one of the most ambitious and, not surprisingly, controversial.

It also seems that very early on, scientists and doctors alike were captivated by the potential such a technology might have in medicine (e.g. Ledley and Lusted, 1959). With intelligent computers able to store and process vast stores of knowledge, the hope was that they would become perfect 'doctors in a box', assisting or surpassing clinicians with tasks like diagnosis.

With such motivations, a small but talented community of computer scientists and healthcare professionals set about shaping a research program for a new discipline called Artificial Intelligence in Medicine (AIM). These researchers had a bold vision of the way AIM would revolutionise medicine, and push forward the frontiers of technology.

AI in medicine at that time was a largely US-based research community. Work originated out of a number of campuses, including MIT-Tufts, Pittsburgh, Stanford and Rutgers (e.g. Szolovits, 1982; Clancey and Shortliffe, 1984; Miller, 1988). The field attracted many of the best computer scientists and, by any measure, their output in the first decade of the field remains a remarkable achievement.

In reviewing this new field in 1984, Clancey and Shortliffe provided the following definition:

'Medical artificial intelligence is primarily concerned with the construction of AI programs that perform diagnosis and make therapy recommendations. Unlike medical applications based on other programming methods, such as purely statistical and probabilistic methods, medical AI programs are based on symbolic models of disease entities and their relationship to patient factors and clinical manifestations.'

Much has changed since then, and today this definition would be considered narrow in scope and vision. Today, the importance of diagnosis as a task requiring computer support in routine clinical situations receives much less emphasis (J. Durinck, E. Coiera, R. Baud, et al., "The Role of Knowledge Based Systems in Clinical Practice," in: eds Barahona and Christenen, Knowledge and Decisions in Health Telematics - The Next Decade, IOS Press, Amsterdam, pp. 199- 203,
1994), So, despite the focus of much early research on understanding and supporting the clinical encounter, expert systems today are more likely to be found used in clinical laboratories and educational settings, for clinical surveillance, or in data-rich areas like the intensive care setting. For its day, however, the vision captured in this definition of AIM was revolutionary.

After the first euphoria surrounding the promise of artificially intelligent diagnostic programmes, the last decade has seen increasing disillusion amongst many with the potential for such systems. Yet, while there certainly have been ongoing challenges in developing such systems, they actually have proven their reliability and accuracy on repeated occasions (Shortliffe, 1987).

Much of the difficulty has been the poor way in which they have fitted into clinical practice, either solving problems that were not perceived to be an issue, or imposing changes in the way clinicians worked. What is now being realised is that when they fill an appropriately role, intelligent programmes do indeed offer significant benefits. One of the most important tasks now facing developers of AI-based systems is to characterise accurately those aspects of medical practice that are best suited to the introduction of artificial intelligence systems.

In the remainder of this chapter, the initial focus will thus remain on the different roles AIM systems can play in clinical practice, looking particularly to see where clear successes can be identified, as well as looking to the future. The next chapter will take a more technological focus, and look at the way AIM systems are built. A variety of technologies including expert systems and neural networks will be discussed. The final chapter in this section on intelligent decision support will look at the way AIM can support the interpretation of patient signals that come off clinical monitoring devices.

**Box 1 - The Turing test**

How will we know when a computer program has achieved an equivalent intelligence to a human? Is there some set of objective measures that can be assembled against which a computer program can be tested? Alan Turing was one of the founders of modern computer science and AI, whose intellectual achievements to this day remain astonishing in their breadth and importance. When he came to ponder this question, he brilliantly side-stepped the problem almost entirely.

In his opinion, there were no ultimately useful measures of intelligence. It was sufficient that an objective observer could not tell the difference in conversation between a human and a computer for us to conclude that the computer was intelligent. To cancel out any potential observer biases, Turing's test put the observer in a room, equipped with a computer keyboard and screen, and made the observer talk to the test subjects only using these. The observer would engage in a discussion with the test subjects using the printed word, much as one would today by exchanging e-mail with a remote colleague. If a set of observers could not distinguish the computer from another human in over 50% of cases, then Turing felt that one had to accept that the computer was intelligent.
Another consequence of the Turing test is that it says nothing about how one builds an intelligent artefact, thus neatly avoiding discussions about whether the artefact needed to in any way mimic the structure of the human brain or our cognitive processes. It really didn't matter how the system was built in Turing's mind. Its intelligence should only to be assessed based upon its overt behaviour.

There have been attempts to build systems that can pass Turing's test in recent years. Some have managed to convince at least some humans in a panel of judges that they too are human, but none have yet passed the mark set by Turing.

**AI can support both the creation and the use of medical knowledge**

Human cognition is a complex set of phenomena, and AI systems can relate to it in two very different ways. Proponents of so-called 'strong' AI are interested in creating computer systems whose behaviour is at some level indistinguishable from humans (see Box 1). Success in strong AI would result in computer minds that might reside in autonomous physical beings like robots, or perhaps live in 'virtual' worlds like the information space created by something like the Internet.

An alternative approach to strong AI is to look at human cognition and decide how it can be supported in complex or difficult situations. For example, a fighter pilot may need the help of intelligent systems to assist in flying an aircraft that is too complex for a human to operate on their own. These 'weak' AI systems are not intended to have an independent existence, but are a form of 'cognitive prosthesis' that supports a human in a variety of tasks.

AIM systems are by and large intended to support healthcare workers in the normal course of their duties, assisting with tasks that rely on the manipulation of data and knowledge. An AI system could be running within an electronic medical record system, for example, and alert a clinician when it detects a contraindication to a planned treatment. It could also alert the clinician when it detected patterns in clinical data that suggested significant changes in a patient's condition.

Along with tasks that require reasoning with medical knowledge, AI systems also have a very different role to play in the process of scientific research. In particular, AI systems have the capacity to learn, leading to the discovery of new phenomena and the creation of medical knowledge. For example, a computer system can be used to analyse large amounts of data, looking for complex patterns within it that suggest previously unexpected associations. Equally, with enough of a model of existing medical knowledge, an AI system can be used to show how a new set of experimental observations conflict with the existing theories. We shall now examine such capabilities in more detail.

**Reasoning with medical knowledge**

Expert or knowledge-based systems are the commonest type of AIM system in routine clinical use. They contain medical knowledge, usually about a very specifically defined task, and are able to reason with data from individual patients to come up with reasoned conclusions.
Although there are many variations, the knowledge within an expert system is typically represented in the form of a set of rules.

There are many different types of clinical task to which expert systems can be applied.

Generating alerts and reminders. In so-called real-time situations, an expert system attached to a monitor can warn of changes in a patient's condition. In less acute circumstances, it might scan laboratory test results or drug orders and send reminders or warnings through an e-mail system.

Diagnostic assistance. When a patient's case is complex, rare or the person making the diagnosis is simply inexperienced, an expert system can help come up with likely diagnoses based on patient data.

Therapy critiquing and planning. Systems can either look for inconsistencies, errors and omissions in an existing treatment plan, or can be used to formulate a treatment based upon a patient's specific condition and accepted treatment guidelines.

Agents for information retrieval. Software 'agents' can be sent to search for and retrieve information, for example on the Internet, that is considered relevant to a particular problem. The agent contains knowledge about its user's preferences and needs, and may also need to have medical knowledge to be able to assess the importance and utility of what it finds.

Image recognition and interpretation. Many medical images can now be automatically interpreted, from plain X-rays through to more complex images like angiograms, CT and MRI scans. This is of value in mass-screenings, for example, when the system can flag potentially abnormal images for detailed human attention.

There are numerous reasons why more expert systems are not in routine use (Coiera, 1994). Some require the existence of an electronic medical record system to supply their data, and most institutions and practices do not yet have all their working data available electronically. Others suffer from poor human interface design and so do not get used even if they are of benefit.

Much of the reluctance to use systems simply arose because expert systems did not fit naturally into the process of care, and as a result using them required additional effort from already busy individuals. It is also true, but perhaps dangerous, to ascribe some of the reluctance to use early systems upon the technophobia or computer illiteracy of healthcare workers. If a system is perceived by those using it to be beneficial, then it will be used. If not, independent of its true value, it will probably be rejected.

Happily, there are today very many systems that have made it into clinical use. Many of these are small, but nevertheless make positive contributions to care. In the next two sections, we will examine some of the more successful examples of knowledge-based clinical systems, in an effort to understand the reasons behind their success, and the role they can play.
Diagnostic and educational systems

In the first decade of AIM, most research systems were developed to assist clinicians in the process of diagnosis, typically with the intention that it would be used during a clinical encounter with a patient. Most of these early systems did not develop further than the research laboratory, partly because they did not gain sufficient support from clinicians to permit their routine introduction.

It is clear that some of the psychological basis for developing this type of support is now considered less compelling, given that situation assessment seems to be a bigger issue than diagnostic formulation. Some of these systems have continued to develop, however, and have transformed in part into educational systems.

DXplain is an example of one of these clinical decision support systems, developed at the Massachusetts General Hospital (Barnett et al., 1987). It is used to assist in the process of diagnosis, taking a set of clinical findings including signs, symptoms, laboratory data and then produces a ranked list of diagnoses. It provides justification for each of differential diagnosis, and suggests further investigations. The system contains a data base of crude probabilities for over 4,500 clinical manifestations that are associated with over 2,000 different diseases.

DXplain is in routine use at a number of hospitals and medical schools, mostly for clinical education purposes, but is also available for clinical consultation. It also has a role as an electronic medical textbook. It is able to provide a description of over 2,000 different diseases, emphasising the signs and symptoms that occur in each disease and provides recent references appropriate for each specific disease.

Decision support systems need not be 'stand alone' but can be deeply integrated into an electronic medical record system. Indeed, such integration reduces the barriers to using such a system, by crafting them more closely into clinical working processes, rather than expecting workers to create new processes to use them.

The HELP system is an example of this type of knowledge-based hospital information system, which began operation in 1980 (Kuperman et al., 1990; Kuperman et al., 1991). It not only supports the routine applications of a hospital information system (HIS) including management of admissions and discharges and order entry, but also provides a decision support function. The decision support system has been actively incorporated into the functions of the routine HIS applications. Decision support provide clinicians with alerts and reminders, data interpretation and patient diagnosis facilities, patient management suggestions and clinical protocols. Activation of the decision support is provided within the applications but can also be triggered automatically as clinical data is entered into the patient's computerised medical record.

Expert laboratory information systems

One of the most successful areas in which expert systems are applied is in the clinical laboratory. Practitioners may be unaware that while the printed report they receive from a laboratory was checked by a pathologist, the whole report may now have been generated by a
computer system that has automatically interpreted the test results. Examples of such systems include the following.

• The PUFF system for automatic interpretation of pulmonary function tests has been sold in its commercial form to hundreds of sites world-wide (Snow et al., 1988). PUFF went into production at Pacific Presbyterian Medical Centre in San Francisco in 1977, making it one of the very earliest medical expert systems in use. Many thousands of cases later, it is still in routine use.

• GermWatcher checks for hospital-acquired (nosocomial) infections, which represent a significant cause of prolonged inpatient days and additional hospital charges (Kahn et al., 1993). Microbiology culture data from the hospital's laboratory system are monitored by GermWatcher, using a rule-base containing a combination of national criteria and local hospital infection control policy.

• A more general example of this type of system is PEIRS (Pathology Expert Interpretative Reporting System) (Edwards et al., 1993). During its period of operation, PEIRS interpreted about 80-100 reports a day with a diagnostic accuracy of about 95%. It accounted for about which 20% of all the reports generated by the hospital's Chemical Pathology Department. PEIRS reported on thyroid function tests, arterial blood gases, urine and plasma catecholamines, hCG (human chorionic gonadotrophin) and AFP (alpha fetoprotein), glucose tolerance tests, cortisol, gastrin, cholinesterase phenotypes and parathyroid hormone related peptide (PTH-RP).

Laboratory expert systems usually do not intrude into clinical practice. Rather, they are embedded within the process of care, and with the exception of laboratory staff, clinicians working with patients do not need to interact with them. For the ordering clinician, the system prints a report with a diagnostic hypothesis for consideration, but does not remove responsibility for information gathering, examination, assessment and treatment. For the pathologist, the system cuts down the workload of generating reports, without removing the need to check and correct reports.

Machine learning systems can create new medical knowledge

Learning is seen to be the quintessential characteristic of an intelligent being. Consequently, one of the driving ambitions of AI has been to develop computers that can learn from experience. The resulting developments in the AI sub-field of machine learning have resulted in a set of techniques which have the potential to alter the way in which knowledge is created.

All scientists are familiar with the statistical approach to data analysis. Given a particular hypothesis, statistical tests are applied to data to see if any relationships can be found between different parameters. Machine learning systems can go much further. They look at raw data and then attempt to hypothesise relationships within the data, and newer learning systems are able to produce quite complex characterisations of those relationships. In other words they attempt to discover humanly understandable concepts.
Learning techniques include neural networks, but encompass a large variety of other methods as well, each with their own particular characteristic benefits and difficulties. For example, some systems are able to learn decision trees from examples taken from data (Quinlan, 1986). These trees look much like the classification hierarchies discussed in Chapter 10, and can be used to help in diagnosis.

Medicine has formed a rich test-bed for machine learning experiments in the past, allowing scientists to develop complex and powerful learning systems. While there has been much practical use of expert systems in routine clinical settings, at present machine learning systems still seem to be used in a more experimental way. There are, however, many situations in which they can make a significant contribution.

- Machine learning systems can be used to develop the knowledge bases used by expert systems. Given a set of clinical cases that act as examples, a machine learning system can produce a systematic description of those clinical features that uniquely characterise the clinical conditions. This knowledge can be expressed in the form of simple rules, or often as a decision tree. A classic example of this type of system is KARDIO, which was developed to interpret ECGs (Bratko et al., 1989).

- This approach can be extended to explore poorly understood areas of medicine, and people now talk of the process of 'data mining' and of 'knowledge discovery' systems. For example, it is possible, using patient data, to automatically construct pathophysiological models that describe the functional relationships between the various measurements. For example, Hau and Koiera (1997) describe a learning system that takes real-time patient data obtained during cardiac bypass surgery, and then creates models of normal and abnormal cardiac physiology. These models might be used to look for changes in a patient's condition if used at the time they are created. Alternatively, if used in a research setting, these models can serve as initial hypotheses that can drive further experimentation.

- One particularly exciting development has been the use of learning systems to discover new drugs. The learning system is given examples of one or more drugs that weakly exhibit a particular activity, and based upon a description of the chemical structure of those compounds, the learning system suggests which of the chemical attributes are necessary for that pharmacological activity. Based upon the new characterisation of chemical structure produced by the learning system, drug designers can try to design a new compound that has those characteristics. Currently, drug designers synthesis a number of analogues of the drug they wish to improve upon, and experiment with these to determine which exhibits the desired activity. By bootstrapping the process using the machine learning approach, the development of new drugs can be speeded up, and the costs significantly reduced. At present statistical analyses of activity are used to assist with analogue development, and machine learning techniques have been shown to at least equal if not outperform them, as well as having the benefit of generating knowledge in a form that is more easily understood by chemists (King et al., 1992). Since such learning experiments are still in their infancy, significant developments can be expected here in the next few years.
Machine learning has a potential role to play in the development of clinical guidelines. It is often the case that there are several alternate treatments for a given condition, with slightly different outcomes. It may not be clear however, what features of one particular treatment method are responsible for the better results. If databases are kept of the outcomes of competing treatments, then machine learning systems can be used to identify features that are responsible for different outcomes.

references


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"Artificial intelligence in medicine (AIM) has reached a period of adolescence in which interactions with the outside world are not only natural but mandatory. Although the basic research topics in AIM may be those of artificial intelligence, the applied issues touch more generally on the broad field of medical informatics. To the extent that AIM research is driven by performance goals for biomedicine, AIM is simply one component within a wide range of research and development activities. Furthermore, an adequate appraisal of AIM research requires an understanding of the research motivations, the complexity of the problems, and a suitable definition of the criteria for judging the field's success. Effective fielding of AIM systems will be dependent on the development of integrated environments for communication and computing that allow merging of knowledge-based tools with other patient data-management and information-retrieval applications. The creation of this kind of infrastructure will require vision and resources from leaders who realize that the practice of medicine is inherently an information-management task and that biomedicine must make the same kind of coordinated commitment to computing technologies as have other segments of our society in which the importance of information management is well understood."