Chapter 1

Introduction

The present chapter discusses the importance of screening in medical diagnosis. It has also elaborated the hurdles related to psychiatric screening, especially in adult psychosis and the tentative ways (conventional and non-conventional ways) of solving them. In this connection, the work of last three decades have been critically analyzed and based on the merits and demerits of the developed tools, the prevailing gaps have been identified as the potential research areas. Special reference has been made on different soft computing tools and their in-built skills, to develop the present day's expert system (ES). Finally, the chapter has defined the aim of the thesis, that is, to develop an ES capable of screening and determining the probability of occurrence of psychotic illnesses in adult population.

1.1 Importance of Screening in Medical Diagnosis

Screening an illness or a group of illnesses, based on a set of sign-symptoms of a patient, is the mainstay towards a successful diagnosis in medical science. Screening primarily leads to tentative identification of a set of possible illnesses, medically termed as differential diagnosis (DD). Here, the pattern of the sign-symptoms are matched with that of the ideal case corresponding to each of the assumed illnesses and based on the closeness, a probability of association is set by the doctors and these are ranked. Later on, based on the ranking, the doctors adopt either preventive and/or curative plan of management. In the next phase, based on the feed-backs of the treatment-response and test-results, the doctors are able to identify the principal disease(s) and this is termed as provisional diagnosis (PD). In nutshell, screening to final diagnosis imposes serial pattern-matching of information with that of the ideal one (that is, target). Thus,

proper screening is the key step to get correct diagnoses. Figure 1.1 has schematically shown how screening leads to a successful diagnosis. Screening of an illness is not always an easy

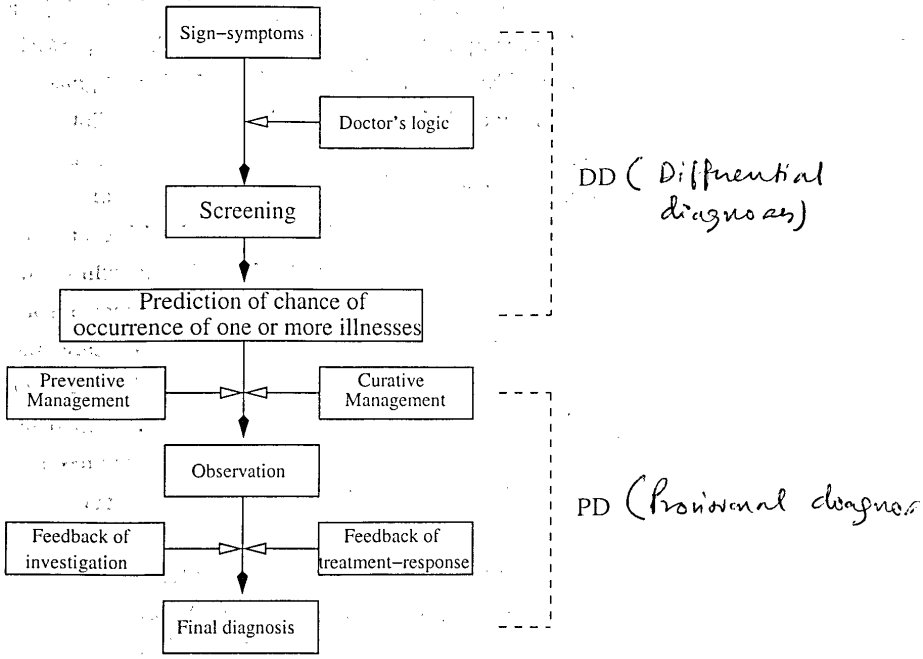

Figure 1.1: A schematic diagram showing the importance of screening in disease-diagnosis.

task. A successful screening requires correct contributions of a doctor's medical skill or logic that should be able to perceive a set of sign-symptoms correctly, as many symptom-constructs are look-alike for many illnesses. As the medical logic is often found to be different among the doctors, the diagnoses tend to vary. Furthermore, the variations of presentation of an illness among the patients make the scenario of disease-identification further Grey and this is one of the key features in psychiatric domain that is extremely complex in nature.

1.1.1 Hurdles in Screening of Adult Psychiatric Population

The present work focuses on screening of adult psychiatric population, as they belong to the worst-affected section of patients and are showing an increasing trend [1]. In psychiatry, the doctors often fail to determine the primary aetiopathology behind a group of symptoms, irre-

spective of age and gender. For example, *depression* can be associated with thyroid diseases or beta-blockers [2]. As already mentioned, correct interpretation of the cause-effect relationships is the key to identify the underlying diseases to be treated. None the less, correctness in identification of the factors, principally lies on how the doctors perceive the patient-information and analyze those using their individual medical logic. In psychiatry, such assertion is often impossible due to the following reasons: (i) too much variations in disease-presentation among the patients, (ii) amalgamation of look-alike diseases behind a symptom-complex (called syndrome), (iii) differences in medical logic of the psychiatrists, (iv) frequent alteration of disease-pattern, (v) absence of an objective tool that can directly measure the morbidity, and (vi) lack of clarity in symptom-illness relationships, as psychiatric symptoms are not always revealed. Thus, a psychiatric screening is usually a long process involving some uncertainties and due to this, treatments are often ill-directed, reflected by frequent drop-outs and adverse drug-effects. Figure 1.2 has pointed out the factors responsible for putting hurdles in psychiatric, especially psychosis screening in adults. The factors like gender biasness, poor patient-psychiatrists ratio, inadequate record-keeping and legal implications have restricted the patients and their relatives to report to the doctors making the scenario further darker. Thus, proper screening of psychotic patients from a population is required. Psychosis is a special type of mental illnesses

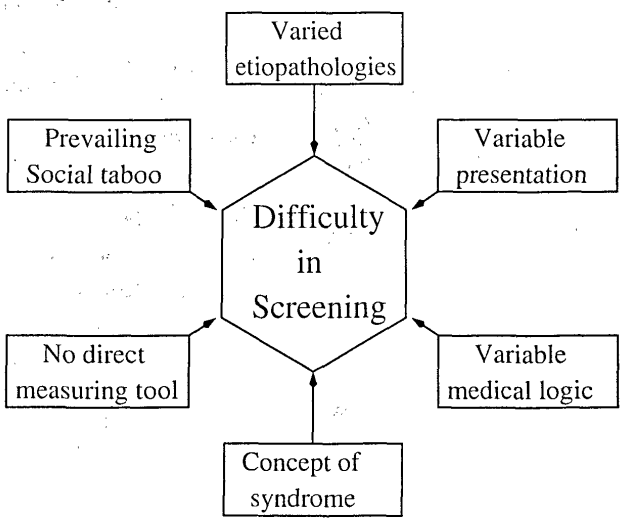

Figure 1.2: A schematic diagram showing the factors responsible for putting hurdles in disease-screening.

falling under psychiatry and it has an immense imprecision that lies in the aetiopathology, presentation (i.e., history, signs and symptoms), treatment-response, follow-up and prognosis. It is prevalent among $2 - 3\%$ of the global population, which is quite high and affecting mostly the young adults [1]. There are several known adult psychotic illnesses and out of those, *Schizophrenia*, *Mania*, *depression with psychotic features*, *delusional disorders*, *schizoaffective disorder*, *organic psychosis*, *catatonia of any cause* are the commonly encountered ones. Regarding the possible aetiopathologies of these psychotic diseases, there are several existing hypotheses, such as the roles of genetic factors [3], birth-complications [4], drug- induced neuropathologies [5], and psychosocial causes [6]. The presentations of these groups of illness vary largely according to age, sex, geography, ethnicity, religion and so on [7, 8, 9]. Despite of several such studies, it is still not defined how much role a factor plays to trigger a psychotic illness. In addition to these constraints, psychiatric history is mostly the *secondary* in nature, as these are collected from the relatives and colleagues of the patient and such information may be erratic and misleading. Moreover, the existing professional biasness on the disease-identification and setting the management plan, prevailing social taboo in the developing country, lack of awareness, unscientific indigenous practices to treat mentally-challenged persons, gender biasness, poor patient-psychiatrists ratio, inadequate record-keeping and legal implications have restricted patients and their relatives to report to the doctors making the scenario more complex. Hence, both screening as well as determining the probability of occurrence of a particular illness among many such need careful analyses.

1.2 Some Tentative Solutions

To solve the above problem of screening adult psychosis, both conventional as well as unconventional approaches have been tried. Conventional approaches include (i) patient-doctor interaction, (ii) use of modern imaging techniques and others. These approaches have their own demerits, for which we take the help of computerization of human experts in the form of an expert system (ES), which is still considered to be the un-conventional approach. In the following subsections, both the conventional as well as un-conventional approaches are discussed.

1.2.1 Conventional Approaches

Present days' clinical practice lies on listening to the history of patients, clinical analysis of their sign-symptoms, grabbing the technical gadgets to screen and grade the illnesses and planning the most useful management. Clinical analysis, done at the very first step, is by far a mixture of intuition, perception, theoretical knowledge and training. These are collectively called as the *medical logic* of a doctor. Due to the effect of individualization, such a logic is variable and the variation will be the maximum, where nothing is directly measurable, as found in psychiatry. The net result may be erroneous screening and planning of management, which is again predominantly seen in psychiatry. To reduce the human-error, various nuclear magnetic resonance imaging techniques, such as functional magnetic resonance imaging (fMRI) [10], positron emission tomography (PET) [11], single photon emission computerized tomography (SPECT) [12] etc. are gaining importance day-by-day in neuropsychiatric research, to find out the underlying pathophysiology of an illness. If the biological reasons are identified, disease-identification automatically becomes easier. However, these tools are costly. Hence, further research may be carried out to develop an expert tool that can reason like the human experts and be cost-effective.

1.2.2 Non-conventional Approaches : Expert Systems

Computerization of human reasoning is, thus, gaining acceleration since the last few decades. There are efforts to make the computers suitable for thinking alike the doctors to solve the diagnostic biasness at the very basic level (that is, screening) using machine intelligence with an intention to benefit the patients at large, in terms of sufferings, productivity and cost of treatment. Such a stupendous attempt is really benevolent in almost all cases, where (i) time does matter and (ii) accuracy is important, for example, neuropsychiatry, cardiology, anesthesiology etc. In this work, we have chosen to deal with the development of an ES, that is able to screen and predict the probability of occurrence of diseases under psychosis. Philosophically, such an attempt is said to be interesting, as the knowledge of medical science and that of information technology are amalgamated with synchrony on a uniquely-shared platform, called Medical Informatics [13]. In the following section, let us see the present depth of research related to development of ES, especially in psychiatry.

To analyze medical data glut, computer scientists and mathematicians always dream to create an artificial or electronic brain that can reason like the doctors [14]. With such a

holistic view, the scientists from both medical as well as engineering fields have proposed a new research area called Artificial Intelligence in Medicine (AIM) [15], which is becoming popular in medical informatics research. The hidden intention behind such a novel philosophy is to develop AI-based ES for medical data analysis, to diagnose, treat and rehabilitate the patients.

ES is a computer program that is written to mimic the logic of human expert through inference methods (called inference engine) to a specific body of knowledge (called the domain). The domain knowledge is represented as the rules in AI-based ES.

To develop an ES, *knowledge extraction* is done by interviewing the human experts. Such knowledge is often heuristic in nature and based on some golden rules of thumb. It is the first step of an ES development. Knowledge can be of various types, such as

1. Knowledge derived from data analysis,
2. Judgmental or subjective knowledge that is based on the valuable experience of human experts,
3. Scientific and theoretical knowledge about the particular domain, and
4. High-level strategic or problem solving knowledge.

Knowledge incorporation is done in the second step, after the rules are generated. The extracted information is refined stepwise by using the feed-back given by both the experts as well as the potential users of ES. Refinement is done by tallying the outputs of the ES (calculated output) with those of the human experts (target) followed by the frequent editing inside the knowledge base (KB) of the ES. Logical incorporation of knowledge is the key to yield a good and reasonable ES. The additional steps of necessary knowledge incorporation into the KB of an ES are as follows.

- Information extractions,
- Code writing,
- Rule generation,
- Establishing a strong reasoning ability, and
- Development of friendly user-interface.