UPDATE ON CADIAG-2: A FUZZY MEDICAL EXPERT SYSTEM FOR GENERAL INTERNAL MEDICINE

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ABSTRACT. A survey on CADIAG-2 a diagnostic consultation system for general internal medicine is presented. The knowledge representation and the inference engine of CADIAG-2 is based on fuzzy set theory and fuzzy logic. CADIAG-2 is integrated into the medical information system of a large hospital. Results obtained by applying the system in the areas of rheumatology, pancreatic diseases, and gall bladder and biliary tract diseases are briefly discussed.

1. Introduction

The central goal of the CADIAG-2[1)] project is the development of a medical consultation system for general internal medicine.

Its underlying clinical issues are to assist in the differential diagnostic process:

(a) by indicating all possible diseases which might be the cause of patient's pathological findings, with special emphasis on rare diseases;
(b) by offering further useful examinations to confirm or to exclude gained diagnostic hypotheses or to find stronger support for them; and
(c) by indicating patients' pathological findings not yet accounted for by the expert system's proposed diagnoses.

[1)] CADIAG stands for *C*omputer-*A*ssisted *DIAG*nosis.

W. H. Janko et al. (eds.), Progress in Fuzzy Sets and Systems, 1–6.

After gaining experience with the medical expert system CADIAG-1 which was formally based on first-order predicate logic and pattern matching (Adlassnig et al. (1985)), a successor system CADIAG-2 was developed and implemented (Adlassnig (1980), Adlassnig (1986)). This system applies fuzzy set theory to model inherent vagueness of medical concepts and fuzzy logic to infer diagnostic conclusions.

At present, CADIAG-2's knowledge base contains disease profiles and complex rules for about 295 diseases, among them 185 rheumatic diseases (69 joint diseases, 12 diseases of the spinal column, 38 diseases of soft tissue and connective tissue system, 45 diseases of cartilage and bone, 21 regional pain syndromes) (Kolarz & Adlassnig (1986)) and 110 gastro-enterological diseases (35 gall bladder and bile duct diseases (Akhavan-Heidari & Adlassnig (1988)), 10 pancreatic diseases (Adlassnig et al. (1984)), 37 colon diseases, 28 disease of the peritoneum).

2. The Medical Consultation System CADIAG-2

2.1. Integration of CADIAG-2 into WAMIS

The CADIAG-2 system is integrated into the medical information system WAMIS[2)] of the Vienna General Hospital (Adlassnig et al. (1986)). This integration allows the collection of patient's findings for CADIAG-2 via the routine medical documentation and laboratory system of WAMIS.

Through a data abstraction and aggregation process (Adlassnig (1988)), patient data are made available to the CADIAG-2 system which tries to infer diagnoses from these abstracted findings in a data-driven manner.

In addition, patient data not routinely collected in WAMIS can be added to CADIAG-2 through a man-machine interface which processes medical terms given in natural language. A word segmentation algorithm allows usage of medical synonyms and abbreviations; moreover, it accepts various orthographic variants and takes different medical suffixes into account (Adlassnig & Grabner (1985)).

The CADIAG-2 system was designed in such a way that three modes of application in our hospital are possible:

(a) the screening and monitoring mode applied at a very early stage of the diagnostic process;

(b) the consultation mode applied after complete data collection; and

(c) the textbook mode without connection to the central patient data base.

[2)] WAMIS is the German acronym for *W*iener *A*llgemeines *M*edizinisches *I*nformations-*S*ystem (Vienna General Medical Information System).

2.2. Knowledge representation and inference engine

CADIAG-2's diagnostic process is based on both stored disease profiles and rules (usually very complex ones such as the ARA criteria for rheumatic diseases (Arnett et al. (1988)).

Two relationships define the association between findings and diseases in these disease profiles:

(a) the necessity of occurrence of a certain finding with a disease (frequency of occurrence degree); and

(b) its sufficiency to infer the disease (strength of confirmation degree).

The same relationships are applicable to define the associations between the antecedents and consequents of rules.

The inference process of CADIAG-2 aims at generating one or more differential diagnoses and—at the same time—at excluding some or all remaining diagnoses. A diagnosis is either established as definitely confirmed or proposed as a diagnostic hypothesis to be confirmed or excluded after additional examinations are performed.

Diagnoses are indicated as definitely confirmed if pathognomonic findings were found in the patient or confirming rules were triggered by patient's findings. Because of the hierarchical relationships among diseases in CADIAG-2, diagnoses at a higher level in the disease hierarchy are confirmed as well if sub-diagnoses are indicated as being confirmed.

Excluded diagnoses are established by either present excluding criteria or absent obligatory criteria. Excluding criteria may be single excluding findings, excluding rules or other, already established diagnoses which exclude other diagnoses. Findings and rule criteria defined to be obligatory present in the patient to establish a certain diagnosis but are definitely absent consequently exclude the respective diagnosis. Definitely excluded disease categories in the disease hierarchy cause also the exclusion of the entire set of the respective sub-diagnoses, if any.

Diagnoses being confirmed and excluded at the same time—which might happen due to contradictory patient data and/or knowledge base errors—are termed diagnostic contradictions. They are displayed separately stating the reason of being established.

Diagnostic hypotheses are generated if a diagnosis is:

(a) neither confirmed, nor excluded, nor a contradictory result; and

(b) the strength of confirmation of at least one present finding, one triggered rule, or one already established sub-diagnosis is equal or higher than a given threshold ϵ $(0 < \epsilon < 1)$.

Since the application of fuzzy set theory allows for mathematical modeling of borderline findings, the degree of presence of a finding (degree of membership in a fuzzy set) is combined with its strength of confirmation. If the resulting value, which is a measure of certainty of the concluded disease, lies between the threshold ϵ and unity (unity means full confirmation), the respective disease has to be taken into consideration as a diagnostic hypothesis.

In addition, diagnostic hypotheses are ranked according to a score of support. This score is calculated on the basis of:

(a) the number of single findings present or present to a certain degree and having a relationship to the disease under consideration;

(b) the degree of presence of these findings; and

(c) the degrees for frequency of occurrence and strength of confirmation between these findings and the respective disease.

Diagnoses which are neither confirmed, nor excluded, nor diagnostic hypotheses, nor contradictory results are put into a category denoted by 'not generated diagnoses'. This allows the physician to obtain a complete survey of all diseases included into CADIAG-2's knowledge base.

In CADIAG-2, two forms of knowledge acquisition have been applied:

(a) acquisition of knowledge from medical experts; and

(b) semiautomatic acquisition of medical knowledge from a patient data base.

Medical experts provide definitional and judgmental knowledge from textbooks and their own practical experience. The estimation of appropriate values for the frequency of occurrence and strength of confirmation degrees is assisted by an automatic procedure which calculates the respective values from stored patient records with known diagnoses (Adlassnig & Kolarz (1986)).

Due to the large number of medical relationships contained in CADIAG-1 and CADIAG-2, intense efforts have been made to verify consistency and completeness of the respective knowledge bases.

For CADIAG-1, a program was developed that verifies the internal consistency of the stored medical knowledge and—in case of inconsistencies—provides the line of reasoning for subsequent correction (Barachini & Adlassnig (1987)). Because of the possible homomorphic mapping of CADIAG-2's finding-to-disease relationships into the finding-to-disease relationship categories of CADIAG-1, this program can partially be applied to CADIAG-2's knowledge base as well (Adlassnig & Kolarz (1986)).

3. Results

At present, extended clinical tests of the accuracy and acceptance of the CADIAG-2 system are being in process (Kolarz & Adlassnig (1986), Adlassnig (1987), Akhavan-Heidari & Adlassnig (1988)).

Results were obtained by applying the system to 544 clinical cases (426 rheumatic cases, 47 pancreatic cases, 71 gallbladder and bile duct cases). Among them, there were 38 multiproblem cases with two discharge diagnoses. For each of these cases, between 200 and 800 findings were available, which were either present, present to a certain degree, or definitely absent. This large number of findings is the result of the complete data collection in the associated medical departments where the tests are being carried out.

The analysis of the CADIAG-2 diagnoses compared with confirmed clinical or, if available, surgery or anatomic-pathological diagnoses yielded an accuracy of about 92%, where the respective evaluation criterion was whether the gold standard diagnosis was either confirmed or among the first three hypotheses in the ranked list of hypotheses. In the multiproblem cases, each discharge diagnosis was evaluated separately.

Very good results could be reached in cases with acute problems and in cases where specific investigations such as X-ray and ultrasonography provided sufficient medical evidence to confirm or to hypothesize a present disease. Unsatisfactory outcome was obtained in some cases with a history of therapy that had let to improved clinical patterns and normalized laboratory test results.

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