Knowledge Representation and Acquisition in Internal Medicine: HEPexpert as an Example

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Background

The acquisition of medical knowledge and the knowledge representation framework of a medical expert system are closely related to each other. We have experienced in the past, that the crucial point of knowledge base development is to provide an intuitive formalism of how to transfer human expertise into a systematized and structured form as required by computer expert systems.

Objective

The aim of this project was to explore new ways of knowledge engineering that are exemplified on liver diseases (hepatology) [1]. Special emphasis was laid on the determination of the strength of confirmation of a present symptom for establishing a disease as diagnostic hypothesis. Variants of the positive predictive value are calculated for this purpose. Other topics of interest were disease hierarchies, cooperating knowledge bases, and systemic interrelationships among symptoms, signs, test results, and diagnoses.

Material and Methods

Based on statistical (Bayesian), frequentistic, and fuzzy logical models of knowledge representation and reasoning this project aimed at the development of a methodological and mathematical framework that could be used for future medical expert system development. The project was performed in close cooperation between an experienced clinician and computer scientists. Some major problems that were dealt with specifically were

- the unsatisfactory precision of the definitions of diseases and findings,
- the vague boundaries between normal and pathological parameter values,
- the influence (alteration) of findings by therapeutic procedures,
- the redundancy of findings,
- the interdependency between findings, and
- patient-specific factors.

Several mathematical models were developed to facilitate the knowledge acquisition process. As an example, a formalism was developed that allows an interpretation of the strength of confirmation value as a frequentistic interpretation of probability with and without considering the prevalence of the diseases [2,3]. Based on this formalism a semi-automatic computation of the strength of confirmation values from the inverse frequency of occurrence values is feasible.

Results

At present the knowledge base covers 125 classical hepatological diseases and about 950 symptoms, signs, and test results. A pool of typical hepatological disease patterns (35 hypothetical cases) was established to allow a pre-clinical evaluation of the knowledge bases.

Technical Specification

Most of the pre-clinical testing was performed by Microsoft Excel 97 Macros. An early prototype, named HEPexpert, was realized in Java (Java Development Kit (JDK) 1.1.5) to allow a comprehensive evaluation in a clinical environment (Fig. 1).

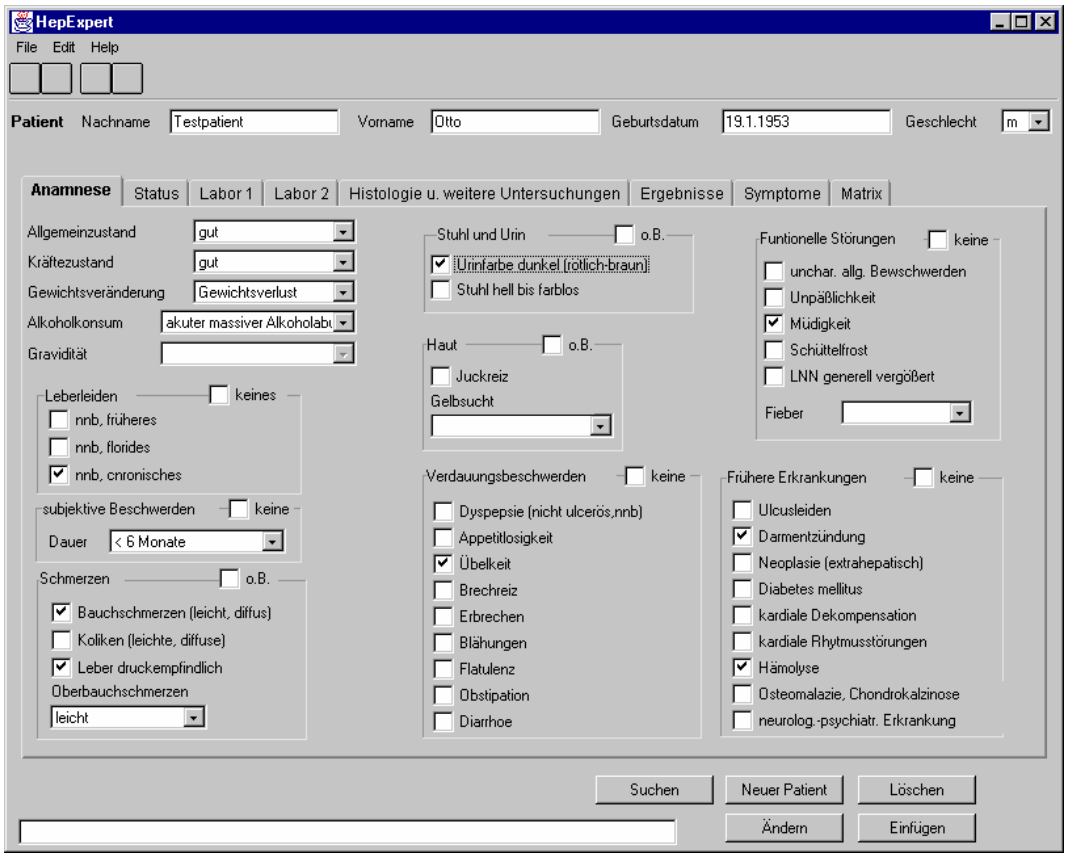

Fig. 1: HEPexpert Java prototype (in German)

Conclusion

Our results showed that the development of large knowledge bases, as required for comprehensive clinical domains such as hepatology, can be essentially facilitated by employing intuitive knowledge representation formalisms. The use of fuzzy set theory extends the scope of the formalisms to vague and uncertain knowledge.

References

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[3] Shortliffe, E.H. & Buchanan, B.G. (1984). A Model of Inexact Reasoning in Medicine. In Shortliffe, E.H. & Buchanan, B.G. (eds.) *Rule-Based Expert Systems*, Addison-Wesley, Massachusetts, 233-262.