



Learning Bayesian network parameters from small data sets: application of Noisy-OR gates

Agnieszka Onisko^{a,*}, Marek J. Druzdzel^{b,1},
Hanna Wasyluk^c

^a *Białystok University of Technology, Institute of Computer Science, Wiejska 45A,
Białystok 15-351, Poland*

^b *Decision Systems Laboratory, School of Information Sciences and Intelligent Systems Program,
University of Pittsburgh, Pittsburgh, PA 15260, USA*

^c *The Medical Center of Postgraduate Education and Institute of Biocybernetics and Biomedical
Engineering, Polish Academy of Sciences, Warsaw, Marymoncka 99, Poland*

Received 1 September 2000; accepted 1 March 2001

Abstract

Existing data sets of cases can significantly reduce the knowledge engineering effort required to parameterize Bayesian networks. Unfortunately, when a data set is small, many conditioning cases are represented by too few or no data records and they do not offer sufficient basis for learning conditional probability distributions. We propose a method that uses Noisy-OR gates to reduce the data requirements in learning conditional probabilities. We test our method on HEPAR II, a model for diagnosis of liver disorders, whose parameters are extracted from a real, small set of patient records. Diagnostic accuracy of the multiple-disorder model enhanced with the Noisy-OR parameters was 6.7% better than the accuracy of the plain multiple-disorder model and 14.3% better than a single-disorder diagnosis model. © 2001 Elsevier Science Inc. All rights reserved.

* Corresponding author. Tel.: +48-85-742-8206 extn. 308; fax: +48-85-742-3423.

E-mail addresses: aonisko@ii.pb.bialystok.pl (A. Onisko), mjdruzdzel@reasonedge.com, marek@sis.pitt.edu (M.J. Druzdzel), hwasyluk@cmkp.edu.pl (H. Wasyluk).

¹ Currently with ReasonEdge Technologies, 124 Mt. Auburn Street Suite 200-N, Cambridge, MA 02138-5700, USA.

Keywords: Bayesian network; Noisy-OR gate; Learning conditional probability distributions

1. Introduction

Bayesian networks [19] (also called belief networks) are acyclic directed graphs modeling probabilistic dependencies and independencies among variables. The graphical part of a Bayesian network reflects the structure of a problem, while local interactions among neighboring variables are quantified by conditional probability distributions. Bayesian networks proved to be powerful tools for modeling complex problems involving uncertain knowledge. They have been employed in practice in a variety of fields, including engineering, science, and medicine with some models reaching the size of hundreds of variables.

A major difficulty in applying Bayesian network models to practical problems is the effort that goes in model building, i.e., obtaining the model structure and the numerical parameters that are needed to fully quantify it. The complete conditional probability distribution table (CPT) for a binary variable with n binary predecessors in a Bayesian network requires specification of 2^n independent parameters. For a sufficiently large n , eliciting 2^n numbers from a domain expert may be prohibitively cumbersome. One of the main advantages of Bayesian networks over other schemes for reasoning under uncertainty is that they readily combine existing frequency data with expert judgment within their probabilistic framework. When sufficient amount of data is available, they can be used to learn both the structure and the parameters of a Bayesian network model [3,20,25]. The existing learning methods are theoretically sound and are guaranteed to produce very good results given sufficiently large data sets. However, in case of small data sets, quite typical in practice, learned models can be of lesser quality.

The focus of this paper is learning CPTs in Bayesian network models from small data sets given an existing network structure. Learning CPTs amounts essentially to counting data records for different conditions encoded in the network. Roughly speaking, prior probability distributions are obtained from relative counts of various outcomes for each of the nodes without predecessors. Conditional probability distributions are obtained from relative counts of various outcomes in those data records that fulfill the conditions described by a given combination of the outcomes of the predecessors (we will refer to this combination of parents' outcomes as *conditioning case*). While prior probabilities can be learned reasonably accurately from a database consisting of a few hundred records, learning CPTs is more daunting. In small data sets, many

conditioning cases are represented by too few or no data records and they do not offer sufficient basis for learning conditional probability distributions. In cases where there are several variables directly preceding a variable in question, individual combinations of their values may be very unlikely to the point of being absent from the data file. In such cases, the usual assumption made in learning the parameters is that the distribution is uniform, i.e., the combination is completely uninformative.

A CPT offers a complete specification of a probabilistic interaction that is powerful in the sense of its ability to model any kind of probabilistic dependence between a discrete node Y and its parents X_1, \dots, X_n . However, when learning the conditional probability distribution from data sets, this precision can be illusory. If the size of the data set is small, many of the CPT entries will have been learned from an insufficient number of records, undermining the very purpose of a full specification. In this paper, we propose enhancing the process of learning the CPTs from data by combining the data with structural and numerical information obtained from an expert. Given expert's indication that an interaction in the model can be approximated by a Noisy-OR gate [8,19], we first estimate the Noisy-OR parameters for this gate. Subsequently, in all cases of a small number of records for any given combination of parents of a node, we generate the probabilities for that case as if the interaction was a Noisy-OR gate. Effectively, we obtain a conditional probability distribution that has a higher number of parameters. At the same time, the learned distribution is smoothed out by the fact that in all those places where no data is available to learn it, it is reasonably approximated by a Noisy-OR gate. Noisy-OR distributions approximate CPTs using fewer parameters and learning distributions with fewer parameters is in general more reliable [7]. While applications of the Noisy-OR gates in medical Bayesian models have already been recorded in the past (e.g., [6,13,24]), our method is novel.

We test our approach on HEPAR II, a Bayesian network model for diagnosis of liver disorders consisting of 73 nodes. The parameters of HEPAR II are learned from a data set of 505 patient cases. We show that the proposed method leads to an improvement in the quality of the model as measured by its diagnostic accuracy. While the observed improvement in accuracy is modest (only 6.7% and 14.3% in comparison to a multiple-disorder model and single-disorder model, respectively), it is obtained at a negligible cost, which makes our method attractive in practice.

The remainder of this paper is structured as follows. Section 2 introduces the Noisy-OR gate. Section 3 describes our data set and our model. Section 4 illustrates the structural modifications that we performed on the model in order to apply our method. Section 5 explains the details related to obtaining the Noisy-OR parameters. Finally, Section 6 compares diagnostic accuracy of a model learned using the direct CPT method to models whose parameters are learned using our method.

2. The Noisy-OR gate

Some types of conditional probability distributions can be approximated by canonical interaction models that require fewer parameters. Very often such canonical interactions approximate the true distribution sufficiently well and can reduce the model building effort significantly.

One type of canonical interaction, widely used in Bayesian networks, is known as Noisy-OR gate [5,8,19]. Noisy-OR gates are usually used to describe the interaction between n causes X_1, X_2, \dots, X_n and their common effect Y .² The causes X_i are each assumed to be sufficient to cause Y in absence of other causes and their ability to cause Y is assumed independent of the presence of other causes.

The simplest and most intuitive canonical model is a binary Noisy-OR gate [19], which applies when there are several possible causes X_1, X_2, \dots, X_n of an effect variable Y , where (1) each of the causes X_i has a probability p_i of being sufficient to produce the effect in the absence of all other causes, and (2) the ability of each cause being sufficient is independent of the presence of other causes. The above two assumptions allow us to specify the entire conditional probability distribution with only n parameters p_1, p_2, \dots, p_n . p_i represents the probability that the effect Y will be true if the cause X_i is present and all other causes $X_j, j \neq i$, are absent. In other words,

$$p_i = \Pr(y | \bar{x}_1, \bar{x}_2, \dots, x_i, \dots, \bar{x}_{n-1}, \bar{x}_n). \quad (1)$$

It is easy to verify that the probability of y given a subset \mathbf{X}_p of the X_i s that are present is given by the following formula:

$$\Pr(y | \mathbf{X}_p) = 1 - \prod_{i: X_i \in \mathbf{X}_p} (1 - p_i). \quad (2)$$

This formula is sufficient to derive the complete CPT of Y conditional on its predecessors X_1, X_2, \dots, X_n .

Henrion [8] proposed an extension of the binary Noisy-OR gate for situations where the effect variable can be true even if all its causes are false and called this extended model a *leaky Noisy-OR* gate. Leaky Noisy-OR is applicable to situations in which a model does not capture all possible causes of Y . Arguably, almost all situations encountered in practice belong to this class.

² Throughout this paper, upper case letters (e.g., Y) and indexed upper-case letters (e.g., X_i) will stand for random variables. Lower case letters will denote their outcomes (e.g., x is an outcome of a variable X). In case of binary random variables, the two outcomes will be denoted by lower case and negated lower case (e.g., the two outcomes of a variable X will be denoted by x and \bar{x}). Bold upper case letters (e.g., \mathbf{X}) will denote sets of variables.

This can be modeled by introducing an additional parameter p_0 , called the *leak probability*, the combined effect of all unmodeled causes of Y .

$$p_0 = \Pr(y | \bar{x}_1, \bar{x}_2, \dots, \bar{x}_n). \quad (3)$$

p_0 represents the probability that the effect Y will occur spontaneously, i.e., in absence of any of the causes that are modeled explicitly.

Fig. 1 shows an example of a Noisy-OR gate for the node *Hepatomegaly* (increased liver size). Each of the parents of the node, *Steatosis*, *Toxic hepatitis*, and *Reactive hepatitis* can cause *Hepatomegaly* by itself, although their influence is probabilistic. *Hepatomegaly* can be also caused by some unmodeled factors, which are captured by a leak probability.

In the leaky Noisy-OR gate, p_i ($i \neq 0$) no longer represents the probability that X_i causes Y given that all the other causes are absent, but rather the probability that Y is present when X_i is present and all other explicit causes (all the X_j s such that $j \neq i$) are absent.

Let p'_i be the probability that Y will be true if X_i is present and every other cause of Y , including unmodeled causes, are absent. p'_i is the probability that X_i causes Y . We have

$$1 - p'_i = \frac{1 - p_i}{1 - p_0}. \quad (4)$$

From here, we have

$$p_i = p'_i + (1 - p'_i)p_0. \quad (5)$$

It follows that the probability of Y given a subset \mathbf{X}_p of the x_i which are present is given in the leaky Noisy-OR gate by the following formula:

$$\Pr(Y | \mathbf{X}_p) = 1 - (1 - p_0) \prod_{i: x_i \in \mathbf{X}_p} \frac{1 - p_i}{1 - p_0}.$$

Díez [4] proposed an alternative way of eliciting the parameters of a leaky Noisy-OR gate, which amounts essentially to asking the expert for the parameters p'_i as defined by Eq. (4). The difference between the two proposals has to do with the leak variable. While Henrion's parameters p_i assume that the expert's answer includes a combined influence of the cause in question and the leak, Díez's

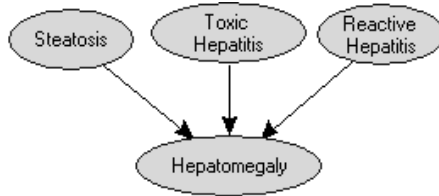


Fig. 1. An example of the Noisy-OR gate.

parameters p'_i explicitly refer to the mechanism between the cause in question and the effect with the leak absent. Conversion between the two parameters is straightforward using Eq. (5). If the Noisy-OR parameters are learned from data, Henrion's definition is more convenient, as the observed frequencies include the leak probability and translate directly into parameters p_i .

Two extensions of the binary Noisy-OR gate to nodes including multiple outcomes have been proposed, the first independently by Díez [4] and Henrion [8] and the second by Srinivas [26]. In our work, we followed the definition of Henrion and Díez. We refer the reader to the original articles and a forthcoming article by Díez and Druzdzel [5] for the details of these extensions.

3. The HEPAR II model and data

Support of a diagnosis in the management of liver disorders has been the focus of a number of research projects in Artificial Intelligence (e.g., [1,2,10–12,22,23,28]). The uniqueness of our approach to this problem is that we are applying decision-theoretic techniques and base our diagnosis on a causal Bayesian network model of the domain of liver disorders.

The starting point of the experiment described in this paper was a single-disorder version of the HEPAR II model, which we describe in the remainder of this section. The HEPAR II project [15,16] aims at applying decision-theoretic techniques to diagnosis of liver disorders. It is a successor of the HEPAR project [2,27], conducted at the Institute of Biocybernetics and Biomedical Engineering of the Polish Academy of Sciences in collaboration with physicians at the Medical Center of Postgraduate Education in Warsaw. The HEPAR system was designed for gathering and processing clinical data of patients with liver disorders and, through its diagnostic capabilities, reducing the need for hepatic biopsy. An integral part of the HEPAR system is its database, created in 1990 and thoroughly maintained since then at the Gastroenterological Clinic of the Institute of Food and Feeding in Warsaw. The current database contains over 800 patient records and its size is still growing. Each hepatological case is described by over 200 different medical findings, including patient self-reported data, results of physical examination, laboratory tests, and, finally, a histopathologically verified diagnosis.

One of the assumptions made in the database that was available to us is that every patient case is ultimately diagnosed with only one liver disorder. In other words, the data set assumed that all disorders were mutually exclusive. This assumption led us to the development of a single-disorder diagnosis model. We elicited the structure of the model (i.e., we selected variables from the data set and established dependencies among them) based on medical literature and conversations with our domain expert, a hepatologist Dr. Hanna Wasyluk (third author) and two American experts, a pathologist, Dr. Daniel Schwartz,

and a specialist in infectious diseases, Dr. John N. Dowling, from the University of Pittsburgh. We estimate that elicitation of the structure took approximately 40 h of the interviews with the experts, of which roughly 30 h were spent with Dr. Wasyluk and roughly 10 h spent with Drs. Schwartz and Dowling. This includes model refinement sessions, where previously elicited structure was reevaluated in a group setting. The model is a causal Bayesian network involving a subset of variables included in the HEPAR database. The most recent single-disorder diagnosis version of the model [17], consists of 66 feature nodes and one disorder node covering, in addition to the hepatologically healthy state, nine mutually exclusive liver disorders: *Toxic hepatitis*, *Reactive hepatitis*, *Functional hyperbilirubinemia*, *Chronic hepatitis (active and persistent)*, *Steatosis hepatis*, *Primary biliary cirrhosis (PBC)*, and *Cirrhosis (compensate and decompensate)*.

The numerical parameters of the model, i.e., the prior and conditional probability distributions, were extracted from the HEPAR database. The data used to extract the numerical parameters contained 505 patient records. All continuous variables were discretized by our expert. One of the assumptions that we used in learning the model parameters was that missing values for discrete finding variables corresponded to state *absent* (e.g., a missing value for *Jaundice* was interpreted as absent). In case of continuous variables, a missing value corresponded to a normal value, elicited from the expert (e.g., a missing value for *Bilirubin* was interpreted as being in the range of 0–1) as the typical value for a healthy patient. We followed here the observation reported by Peot and Shachter [21] that missing values in medical data sets are not missing at random and are either indications of normal or less severe symptoms.

Given a patient case, i.e., values of some of the modeled variables, such as symptoms or test results, the system computes the posterior probability distribution over the possible liver disorders. This probability distribution can be directly used in diagnostic decision support.

4. Structural changes to the HEPAR II model

In order to be able to apply parametric probability distributions, such as Noisy-OR gates, in learning the network parameters, we had to restructure the network in such a way that various nodes express either binary propositions or various grades of intensity of some quantity. The disorder node in the single-disorder diagnosis version of the HEPAR II model is a categorical variable with 10 outcomes that is not suitable for a parametric probability distribution. One way of preparing the structure for these distributions is by breaking the disorder node into separate nodes for each of the disorders. This modification addresses two problems: it relaxes the assumption of mutual exclusivity of disorders and makes the nodes more amenable to parametric quantification.

We have concentrated the structural changes on the disorders. We split the disorder node with its nine mutually exclusive disorders into seven nodes: five binary nodes (*Toxic hepatitis*, *Reactive hepatitis*, *Steatosis*, *Functional hyperbilirubinemia* and *PBC*) and two three-valued nodes (*Chronic hepatitis* and *Cirrhosis*). The feature nodes that we originally modeled as causes/effects of the single-*Liver Disorder* variable were broken down into several groups, specific for each of the nine disorders. As far as the data used in learning the parameters are concerned, we worked with 66 findings and 505 records in the database. The resulting model consisted of 73 nodes (66 feature nodes and seven disorder nodes).

Fig. 2 shows simplified fragments of both models and gives an idea of the structural changes performed in the transition from the single-disorder to the multiple-disorder versions of the model. In particular, the models share each of the four risk factors (*Reported history of viral hepatitis*, *History of alcohol abuse*, *Gallstones*, and *Hepatotoxic medications*) and six symptoms and test results (*Fatigue*, *Jaundice*, *Bilirubin*, *Alkaline phosphatase*, *Ascites*, and *Total proteins*). The single-*Liver disorder* node is replaced by four disorder nodes (*Chronic hepatitis*, *Steatosis*, *Cirrhosis*, and *Toxic hepatitis*).

A consequence of our structural changes was that they decreased the number of numerical parameters required to quantify the model. The main difference between the models is that some of the four new disorder nodes are not connected with some of the risk factors and symptoms. While adding a node might increase the number of parameters, it is compensated by removing an outcome of a variable and removing some arcs. The latter especially leads to a logarithmic decrease in the size of a CPT. Our transformation resulted in a significant reduction of the number of numerical parameters necessary to quantify the network. This, in turn, increased the average number of records for each combination of parents in a CPT. Indeed, the multiple-disorder version of the model required only 1,847 parameters (we counted $\mu = 89.5$ data records per parent combination) compared to the 3714 parameters ($\mu = 16.8$ data records per parent combination) needed for the single-disorder

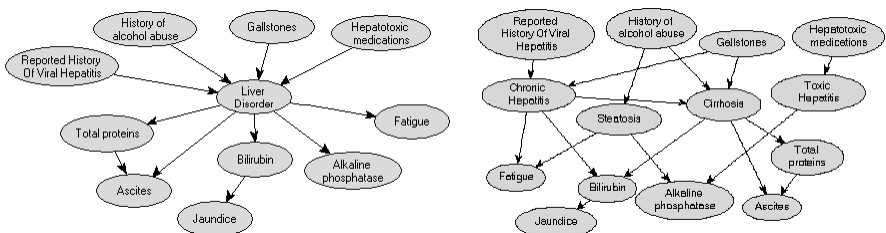


Fig. 2. Simplified fragments of the HEPAR II networks: single-disorder diagnosis (left) and multiple-disorder diagnosis (right) version.

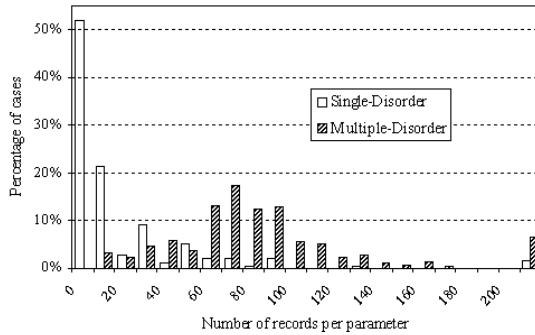


Fig. 3. Distribution over the number of data records per parent combination for the single-disorder and the multiple-disorder models.

version of the model. Fig. 3 shows the distribution of the number of data records per parent combination for the single-disorder and the multiple-disorder models. We can see that over 50% of the parent combinations in the single-disorder model had zero records. In the multiple-disorder model this number is dramatically smaller – only 0.1% of all cases involved zero records and there is quite a high proportion of conditional probability distributions for which tens of records were available. With an increase in the average number of records per parent combination, we can expect the quality of the model parameters to improve.

Unfortunately, structural changes also introduced certain problems. The fact that we used a data set in which each patient record had a single-disorder diagnosis placed us before a difficulty in assessing CPTs of nodes that had several disorder nodes as parents – there were no records in the database for conditions involving combinations of various disorders. We applied a simple solution, in which we included in the calculation all records that described the disorders present in the condition. For example (see Fig. 2), when computing the conditional probability distribution of the node *Fatigue* given presence of both *Chronic hepatitis* and *Steatosis*, we used both: records that were diagnosed as *Chronic hepatitis* and records that were diagnosed as *Steatosis*. This amounted to averaging the effect of various disorders. We also tried taking the maximum effect of all disorders present in the condition, with a very modest improvement in performance. Another limitation of the HEPAR data that had a serious implication on our work is that mutual exclusivity of disorders did not allow us to extract dependencies among disorders. Hepatology often deals with disorders that are consequences of the previous disorders, e.g., a chronic liver disorder implies hepatic fibrosis which can further cause cirrhosis. In the future we plan to model and quantify these dependencies by combining data with expert judgment.

5. Obtaining parameters for Noisy-OR gates

For each combination of a node and its parents (a family) in the multiple-disorder version of the HEPAR II model, we verified with our expert whether the interaction could be approximately modeled by a Noisy-OR gate. The expert identified 25 nodes (from among the total of 62 nodes with parents) that could be reasonably approximated by Noisy-OR gates. Testing the Noisy-OR assumption for each of the gates with the expert was quite straightforward once the expert had understood the concept of independence of causal interaction. When deciding whether an interaction can be approximated by a Noisy-OR gate, we followed the criteria proposed by Díez [6]. An interaction can be approximated by a Noisy-OR gate if it meets the following three assumptions: (1) the child node and all its parents must be variables indicating the degree of presence of an anomaly, (2) each of the parent nodes must represent a cause that can produce the effect (the child variable) in the absence of the other causes, (3) there may be no significant synergy among the causes.

Each of the such identified Noisy-OR gates was subject to the following learning enhancement. Whenever there were sufficiently many records for a given conditioning case, we used these records to learn a corresponding element of the CPT. When there were no or very few data records, we generated the CPT entry from our Noisy-OR parameters (Sections 5.1 and 5.2 describe how we obtained these). Effectively, the complete CPT, once learned, was a general CPT with a fraction of its elements generated using the Noisy-OR assumption. The assumption that we made was that a general conditional probability table will fit the actual distribution better than a Noisy-OR distribution. Noisy-OR will fit better than a uniform distribution in those cases when there was not enough data to learn a distribution. In the following two sections we describe two methods of obtaining the Noisy-OR parameters of the gates in question.

5.1. Obtaining Noisy-OR parameters from data

We learned the Noisy-OR parameters from data for each of the 25 Noisy-OR gates identified by our expert using Eq. (1). We learned the leak parameter using Eq. (3). Obtaining the parameters from records that contain a combination of values of parent outcomes would be less reliable, as there would be certainly fewer such records (a conjunction of two events is at most as likely as each of these events in separation). We tried to obtain better estimates of the Noisy-OR parameters by fitting the Noisy-OR distribution to a larger fragment of a CPT but this simple approach yielded the best performance.

5.2. Expert assessments of Noisy-OR parameters

For each of the 25 Noisy-OR gates identified by the expert, we also obtained all numerical parameters using direct expert elicitation. There was a total of 189 parameters and the assessment took a total of about 4 h of expert time.

Initially, we posed the expert two types of questions, corresponding to the two theoretical formalizations of the Noisy-OR gate proposed in the literature. The first type of questions focused on the parameters p_i (Eq. (1)) and was based on Henrion's [8] definition. For the example network fragment in Fig. 1, it amounted to:

What is the probability that *Toxic hepatitis* results in *Hepatomegaly* when neither *Reactive hepatitis* nor *Steatosis* are present?

The second type of questions focused on parameters p'_i (Eq. (4)) and was based on Díez's [4] definition. For the example network fragment in Fig. 1, it amounted to:

What is the probability that *Toxic hepatitis* results in *Hepatomegaly* when no other cause of *Hepatomegaly* is present?

We stumbled across two interesting empirical questions: (1) which of the two definitions is more intuitive for a human expert, and (2) which leads to better quality assessments. While we have not tested (2), in the course of elicitation our expert clearly developed preference for Díez's definition. Using Eq. (5), we subsequently converted the parameters elicited from the expert in Díez's format into Henrion's format, which is the current native format of our software, GeNIe and SMILE.

While we have no objective basis for comparing the quality of expert assessment to the numbers obtained from the data (please note that we evaluated our model using the data, so the best we can say is whether the expert's judgments matched the data or not), we observed a systematic difference between the two: our expert provided usually higher estimates than those learned from the data ($\mu = 0.32$ versus $\mu = 0.19$ for the expert and data, respectively).

6. Comparison of diagnostic accuracy of the models

We performed a series of empirical tests of diagnostic accuracy of various versions of the model. In order to make the comparison fair, we used the same data set for learning the parameters of each of the models. Our data set contained 505 patient records classified in nine different disorder classes. In each case we used the same measure of accuracy: diagnostic performance using the

leave-one-out method [14]. Essentially, given $n = 505$ data records, we used $n - 1$ of them for learning model parameters and the remaining one record to test the model. This procedure was repeated n times, each time with a different data record. In our tests, we used as observations only those findings that were actually reported in the data (i.e., we did not use the values that were missing, even though we used their assumed values in learning). The diagnosis for each patient case was calculated given the evidence, i.e., a subset of the 66 possible observations such as symptoms, signs and the laboratory tests results. These data did not include the results of a biopsy.

By accuracy we mean the proportion of records that were classified correctly. Whenever we report accuracy within a class, we report the fraction of records within that class that were classified correctly.

6.1. Single- versus multiple-disorder diagnosis model

Our first empirical test focused on a comparison of the diagnostic performance for the single-disorder and the multiple-disorder models. We were interested in overall performance of the models in terms of classification accuracy (each of the disorders was viewed as a separate class that the program predicted based on the values of all the other variables). This test is very conservative against the multiple-disorder model, as this is the task for which the single-disorder version of the model was designed. We were interested in both (1) whether the most probable diagnosis indicated by the model is indeed the correct diagnosis, and (2) whether the set of k most probable diagnoses contains the correct diagnosis for small values of k (we chose a “window” of $k = 1, 2, 3$, and 4). The latter focus is of interest in diagnostic settings, where a decision support system only suggest possible diagnoses to a physician. The physician, who is the ultimate decision maker, may want to see several alternative diagnoses before focusing on one. Results were for the multiple-disorder version of the model approximately 45% (compared to 42% for the single-disorder version), 59% (57%), 70% (68%), and 77% (78%) for $k = 1, 2, 3$, and 4, respectively. In other words, the most likely diagnosis indicated by the model was the correct diagnosis in 45% of the cases. The correct diagnosis was among the four most probable diagnoses as indicated by the model in 77% of the cases. The performance of both versions of the model was similar, with the multiple-disorder version being more accurate. While this performance may not seem spectacular, we would like to point out that the problem that our model addresses is hard. The clinic, in which the data is collected, is a specialist clinic and its patient population consists of typically hard cases that are referred to it by other medical centers. The performance of the ‘naive Bayes’ approach [9] applied to our problem is 41% giving an indication of the difficulty of the problem.

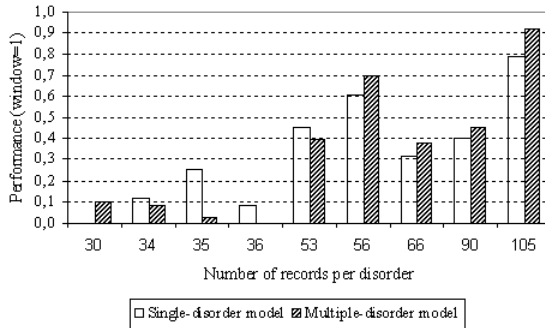


Fig. 4. Diagnostic accuracy as a function of the number of disorder cases in the database (class size) for the single- and multiple-disorder diagnosis models, window = 1.

In order to gain some insight into when multiple-disorder version of the model is better, we looked at the relationship between the number of records in the database for each class and the diagnostic accuracy within that class. Fig. 4 shows this relationship for the window of size 1 (i.e., the most likely disorder). It is clear that accuracy of both models increases significantly with the number of data records. Another interesting trend is that the multiple-disorder model performed often better than the single-disorder model for those disorders that had many records. This promises a higher diagnostic value of our approach when the available data set is sufficiently large, i.e., when the quality of parameters is high.

6.2. Plain CPT model versus CPT smoothed by Noisy-OR parameters

Our second test aimed at comparing the diagnostic accuracy of the plain multiple-disorder model to the models whose probabilities were smoothed out using the Noisy-OR parameters. Here, we focused on three models: (1) the plain multiple-disorder model (i.e., general CPT) and two models enhanced with: (2) Noisy-OR parameters obtained from data, and (3) Noisy-OR parameters assessed by the expert.

As explained in Section 3, our enhancement process replaced those elements of the CPT that had not enough data records to learn a distribution reliably, i.e., when the number of records found in the data set was lower than a *replacement threshold* (we specified this threshold as a percentage of all records in the data set, i.e., a threshold of 10% corresponds roughly to 50 records). Fig. 5 shows the relationship between the replacement threshold and the percentage of all CPT entries that were replaced by the Noisy-OR distributions. The percentage of replaced CPT entries seems to be directly proportional to the replacement threshold.

Fig. 6 shows the results for the three tested models for the window size of 1. It pictures the diagnostic accuracy of the models as a function of the



Fig. 5. Percentage of conditional probability distribution entries replaced by Noisy-OR distributions as a function of the replacement threshold.

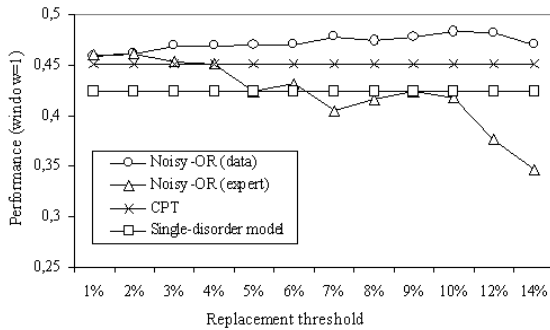


Fig. 6. Diagnostic accuracy as a function of the replacement threshold, window = 1.

replacement threshold. In addition we included the results for the single-disorder model. It appears that the highest accuracy was reached by the model whose CPTs were enhanced with the Noisy-OR parameters learned from data. The highest accuracy achieved by the models was 45%, 48%, and 46% for the CPT model, the data Noisy-OR model, and the expert Noisy-OR model respectively.

Fig. 7 shows the performance within each class for the three models. Again we observed that for almost each of the disorders, the data Noisy-OR model performed better than the other models.

7. Discussion

The transformation of the model performed in order to prepare it for Noisy-OR gates has shown that Bayesian network models readily accommodate

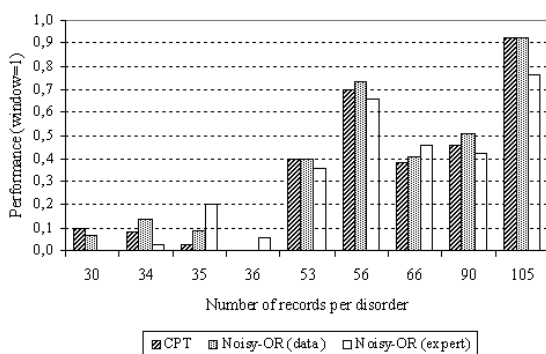


Fig. 7. Diagnostic accuracy as a function of the number of disorder cases in the database (class size) for the CPT and two versions of the model with Noisy-OR parameters.

multiple-disorder diagnoses. It was relatively easy to derive the multiple-disorder version of the model from the existing single-disorder version. We estimate that the total time spent with the expert was less than 10 h, one fourth of the original effort to build the network.

Diagnostic accuracy of the multiple-disorder model enhanced with the Noisy-OR parameters was 6.7% better than the accuracy of the plain multiple-disorder model and 14.3% better than the single-disorder diagnosis model. This increase in accuracy has been obtained with very modest means – in addition to structuring the model so that it is suitable for Noisy-OR nodes, the only knowledge elicited from the expert and entered in the learning process was which interactions can be viewed as approximately Noisy-OR. This knowledge was straightforward to elicit. We have found that whenever combining expert knowledge with data, and whenever working with experts in general, it pays off generously to build models that are causal and reflect reality as much as possible, even if there are no immediate gains in accuracy.

We have also observed that the diagnostic accuracy of the model based on numbers elicited from the expert (as opposed to learned from data) was quite good for diseases with well understood risk factors and symptoms. The accuracy tends to be lower in case of those diseases whose mechanisms are not exactly known, for example Functional hyperbilirubinemia, Reactive hepatitis, or PBC, even if the number of records in the data set was very small.

Our future research plans include expert verification of the probability distributions of those nodes that have several disorder nodes as parents. As we mentioned above, these parameters cannot be learned from our data and the arbitrary assumptions that we made in the learning process may have had a negative effect on the diagnostic performance of the system. We also plan to focus on disorder-to-disorder dependencies. This information is lacking from the database, so here again we will have to rely on expert judgment. A question

that we find worth pursuing is whether there are any properties of individual nodes that influence whether diagnostic accuracy of the model will be served by Noisy-OR or CPT used in individual cases.

Acknowledgements

Support for this research was provided by the Air Force Office of Scientific Research, grant F49620-00-1-0112, by the National Science Foundation under Faculty Early Career Development (CAREER) Program, grant IRI-9624629, by the Polish Committee for Scientific Research, grant 8T11E02917, by the Białystok University of Technology grant W/II/1/00, by the Medical Centre of Postgraduate Education, Poland, grant 501-2-1-02-18/00, and by the Institute of Biocybernetics and Biomedical Engineering, Polish Academy of Sciences, grant 16/ST/00. Our collaboration was enhanced by travel support from Poland to USA by NATO Collaborative Linkage Grant PST.CLG.976167. An earlier version of this paper was presented in the CaNew'2000 Workshop on Bayesian and Causal Networks: From Inference to Data Mining [18]. We would like to thank anonymous reviewers for the CaNew'2000 Workshop, reviewers for this special issue, and most of all Dr. Javier Díez for their helpful comments on the paper. The HEPAR II model was created and tested using SMILE, an inference engine, and GeNIe, a development environment for reasoning in graphical probabilistic models, both developed at the Decision Systems Laboratory, University of Pittsburgh, and available at <http://www2.sis.pitt.edu/~genie>.

References

- [1] K.P. Adlassnig, W. Horak, Development and retrospective evaluation of HEPAXPERT-I: A routinely-used expert system for interpretive analysis of hepatitis and serologic findings, *Artificial Intelligence in Medicine* 7 (1995) 1–24.
- [2] L. Bobrowski, HEPAR: Computer system for diagnosis support and data analysis, Prace IBIB31, Institute of Biocybernetics and Biomedical Engineering, Polish Academy of Sciences, Warsaw, Poland, 1992.
- [3] G.F. Cooper, E. Herskovits, A Bayesian method for the induction of probabilistic networks from data, *Machine Learning* 9 (4) (1992) 309–347.
- [4] F.J. Díez, Parameter adjustment in Bayes networks, The generalized Noisy-OR gate, in: *Proceedings of the Ninth Annual Conference on Uncertainty in Artificial Intelligence*, (UAI-93), Washington, DC, 1993, pp. 99–105.
- [5] F.J. Díez, M.J. Druzdzel, Canonical probabilistic models for knowledge engineering, *Forthcoming*, 2001.
- [6] F.J. Díez, J. Mira, E. Iturralde, S. Zubillaga, DIAVAL, a Bayesian expert system for echocardiography, *Artificial Intelligence in Medicine* 10 (1997) 59–73.

- [7] N. Friedman, M. Goldszmidt, Learning Bayesian networks with local structure, in: M.I. Jordan (Ed.), *Learning and Inference in Graphical Models*, MIT Press, Cambridge, MA, 1999, pp. 421–459.
- [8] M. Henrion, Some practical issues in constructing belief networks, in: L.N. Kanal, T.S. Levitt, J.F. Lemmer (Eds.), *Uncertainty in Artificial Intelligence*, vol. 3, Elsevier, Amsterdam, 1989, pp. 161–173.
- [9] P. Langley, W. Iba, K. Thompson, An analysis of Bayesian classifiers, in: *Proceedings of the Tenth National Conference on Artificial Intelligence*, AAAI Press, Menlo Park, CA, 1992, pp. 223–228.
- [10] P.J.F. Lucas, Refinement of the HEPAR expert system: Tools and techniques, *Artificial Intelligence in Medicine* 6 (1994) 175–188.
- [11] P.J.F. Lucas, A.R. Janssens, Development and validation of HEPAR, an expert system for the diagnosis of disorders of the liver and biliary tract, *Medical Informatics* 16 (3) (1991) 259–270.
- [12] P.J.F. Lucas, R.W. Segaar, A.R. Janssens, HEPAR: an expert system for diagnosis of disorders of the liver and biliary tract, *Liver* 9 (1989) 266–275.
- [13] B. Middleton, M.A. Shwe, D.E. Heckerman, M. Henrion, E.J. Horvitz, H.P. Lehmann, G.F. Cooper, Probabilistic diagnosis using a reformulation of the Evaluation of the INTERNIST-1/QMR knowledge base: II. Evaluation of diagnostic performance, *Methods of Information in Medicine* 30 (4) (1991) 256–267.
- [14] A.W. Moore, M.S. Lee, Efficient algorithms for minimizing cross validation error, in: *Proceedings of the 11th International Conference on Machine Learning*, Morgan Kaufmann, San Francisco, 1994.
- [15] A. Oniško, M.J. Druzdziel, H. Wasyluk, A probabilistic causal model for diagnosis of liver disorders, in: *Proceedings of the Seventh International Symposium on Intelligent Information Systems (IIS-98)*, Malbork, Poland, June 15–19, 1998, pp. 379–387.
- [16] A. Oniško, M.J. Druzdziel, H. Wasyluk, A Bayesian network model for diagnosis of liver disorders, in: *Proceedings of the Eleventh Conference on Biocybernetics and Biomedical Engineering*, vol. 2, Warsaw, Poland, December 2–4, 1999, pp. 842–846.
- [17] A. Oniško, M.J. Druzdziel, H. Wasyluk, Extension of the Hepar II model to multiple-disorder diagnosis, in: S.T. Wierzbchoń, M. Kłopotek, M. Michalewicz (Eds.), *Intelligent Information Systems, Advances in Soft Computing Series*, Physica-Verlag, Heidelberg, 2000, pp. 303–313.
- [18] A. Oniško, M.J. Druzdziel, H. Wasyluk, Learning Bayesian network parameters from small data sets: Application of Noisy-OR gates, in: *Working notes on the European Conference on Artificial Intelligence (ECAI) Workshop Bayesian and Causal Networks: From Inference to Data Mining*, Berlin, Germany, August 22, 2000.
- [19] J. Pearl, *Probabilistic Reasoning in Intelligent Systems: Networks of Plausible Inference*, Morgan Kaufmann, San Mateo, CA, 1988.
- [20] J. Pearl, T.S. Verma, A theory of inferred causation. in: J.A. Allen, R. Fikes, E. Sandewall (Eds.), *KR-91, Principles of Knowledge Representation and Reasoning: Proceedings of the Second International Conference*, Morgan Kaufmann, San Mateo, CA, Cambridge, MA, 1991, pp. 441–452.
- [21] M. Peot, R. Shachter, Learning from what you don't observe, in: *Proceedings of the Fourteenth Annual Conference on Uncertainty in Artificial Intelligence (UAI-98)*, Morgan Kaufmann, San Francisco, CA, 1998, pp. 439–446.
- [22] R.J. Richards, J.K. Hammitt, J. Tsevat, Finding the optimal multiple-test strategy using a method analogous to logistic regression. The diagnosis of hepatolenticular degeneration (Wilson's disease), *Medical Decision Making* 16 (1996) 367–375.
- [23] S. Shiomi, T. Kuroki, H. Jomura, T. Ueda, N. Ikeoka, K. Kobayashi, H. Ikeda, H. Ochi, Diagnosis of chronic liver disease from liver scintiscans by fuzzy reasoning, *Journal of Nuclear Medicine* 36 (1995) 593–598.

- [24] M.A. Shwe, B. Middleton, D.E. Heckerman, M. Henrion, E.J. Horvitz, H.P. Lehmann, G.F. Cooper, Probabilistic diagnosis using a reformulation of the INTERNIST-1/QMR knowledge base: I. The probabilistic model and inference algorithms, *Methods of Information in Medicine* 30 (4) (1991) 241–255.
- [25] P. Spirtes, C. Glymour, R. Scheines, *Causation, Prediction, and Search*, Springer, New York, 1993.
- [26] S. Srinivas, A generalization of the Noisy-OR model, in: *Proceedings of the Ninth Annual Conference on Uncertainty in Artificial Intelligence (UAI-93)*, Washington, DC, 1993, pp. 208–215.
- [27] H. Wasyluk, The four year's experience with HEPAR-computer assisted diagnostic program, in: *Proceedings of the Eighth World Congress on Medical Informatics (MEDINFO-95)*, Vancouver, BC, July 23–27, 1995, pp. 1033–1034.
- [28] Y.K. Zhao, T. Tsutsui, A. Endo, K. Minato, T. Takahashi, Design and development of an expert system to assist diagnosis and treatment of chronic hepatitis using traditional Chinese medicine, *Medical Informatics* 19 (1994) 37–45.