An Auxiliary System for Medical Diagnosis Based on Bayesian Belief Networks

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Abstract: *In this paper we propose a development tool, based on Bayesian belief networks, specific for medical diagnosis. The main feature of the proposed system is to provide a simple and integrated tool for designing diagnostic applications. With an easy-to-use interface, this tool gives the users (experts in the medical domain) the possibility to design applications without deep background knowledge on Bayesian belief networks theory. Taking advantage of the simple structure of diagnostic models (composed of causal disease-symptom relations), an efficient statistical inference mechanism is implemented, relieving the computational burden. The developed system provides an Internet page-based interface to interact with the expert when using the designed application for diagnosis. An application of this tool in the context of sleep-disorders diagnosis is presented, illustrating the functionality of the system.*

Keywords: *Bayesian belief networks, auxiliary diagnostic system, probabilistic inference, diagnostic system development tools, decision support systems, sleep disorders.*

1. INTRODUCTION

The problem of medical diagnosis can be stated as follows: given a set of *symptoms* (clinical data) and *signals,* or *test results* (tests performed on the patient), assess pathological situations, identifying which *diseases* justify the particular findings.

Computer assisted techniques have been proposed in the past to help solving the diagnostic problem [3,8,18]. Commonly known as *expert systems*, approaches, undertaking distinct formalisms for modeling domain knowledge, include: rule-based or production systems, frame-based systems, semantic networks, neural networks, and Bayesian belief networks (BBNs). Explanatory mechanisms are more easily implemented with rule-based systems and graphical models, such as the BBN paradigm, making these preferred solutions to the diagnostic problem.

Typical schemes for treating uncertainty, often explored in rule-based systems include: probabilistic models (Bayesian approach), certainty factors, approximate reasoning, Dempster-Shafer theory [8,13,18]. Limitations with some of these solutions have been referred: assumption of statistical independence between symptoms; locality of uncertainty modeling, not being able to cope with context changes; non-coherent uncertainty calculus. The probabilistic model for uncertain reasoning, provided by BBNs, offers a flexible alternative to these approaches, overcoming these difficulties.

The principles underlying BBNs [6,14,17] derive from Bayes theory in the 18th century; however due to the heavy calculation load required, only in the mid 1980's this theory gained renewed interest: Bayesian belief networks were introduced to expert systems [17] and efficient inference methods were proposed [14,15]. They have been applied in a variety of domains [6,14], such as agriculture, computer vision, computing, information processing, coding, tutoring systems, knowledge discovery and medicine. Concerning medical applications (see, for instance [14] and the references therein), examples of BBNs-based systems include: *Child* for congenital heart diseases; *MUNIN* as well as *Painulim* for neuromuscular diseases; and *Pathfinder* for lymph-node pathology.

Presently there are powerful and universal tools for developing Bayesian belief networks, such as HUGIN [11], MSBN [16] and JavaBayes [12]. An extended list of software for BBNs can be found in [19]. These tools range from simple software modules [12] to versatile, graphical interfaces that assist the designer of new applications. Offering multiple options in an application independent framework, the use of such tools often requires a deep knowledge of the BBN's formalism and methods.

In this paper we propose a BBN-based development tool, specific for medical diagnosis. The main feature of the proposed system is to provide a simple and integrated tool for designing diagnostic applications. With an easy-to-use interface, this tool gives the user the possibility to design his particular application without knowledge of BBNs background theory. Moreover, the proposed tool produces the diagnosis results in an output layout specially designed for the user-expert analysis. In addition, the user-interface (when diagnosing) is made through an Internet browser. Finally, considering that most of the medical diagnostic models have a simple structure (composed of causal disease-symptom relations), the proposed tool implements efficient statistical inference mechanisms, taking advantage of that simplified model and relieving the computational burden.

Details of the proposed system are presented in the next sections. Bayesian belief networks are briefly introduced in section 2, where a classical diagnostic model is illustrated. The diagnostic tool is described in section 3; details are given about the system interface, functionality and implementation. An example, concerning the application of the system in the context of sleep-disorders is presented in section 4. Finally, conclusions and suggestions for further work are presented in section 5.

2. BBN-BASED DIAGNOSTIC MODEL

Bayesian belief networks provide a probabilistic model for uncertain reasoning, adequate for handling the diagnostic problem. A BBN for a given domain represents the joint probability distribution, $p(\mathbf{x})$, over the set of variables, **X,** of the domain, as a set of local distributions combined with a set of conditional independence assertions; that allows the construction of the global joint probability distribution from the local distributions as

$$p(\mathbf{x}) = \prod_{i=1}^{n} p(x_i \mid pa(x_i)) \qquad (1)$$

where, for each variable x_i, $pa(x_i) \subseteq \{x_1, \ldots, x_{i-1}\}$ is a set of variables that renders x_i and $\{x_1, \ldots, x_{i-1}\}$ conditionally independent. The BBN structure encodes the assertions of conditional independence as a directed acyclic graph such that: (a)- each node corresponds to a variable; (b)- the parents of the node corresponding to x_i are the nodes associated to the variables in $pa(x_i)$. The pair formed by the structure (graph) and the collection of local distributions, $p(x_i \mid pa(x_i))$, for each node in the domain, constitute the Bayesian belief network for that domain.

Concerning the context of medical diagnosis, a Bayesian belief network is a directed acyclic graph in which diseases and symptoms, as well as tests results, are graphically represented as nodes (see Fig. 1), arcs expressing causal relationships between these variables.

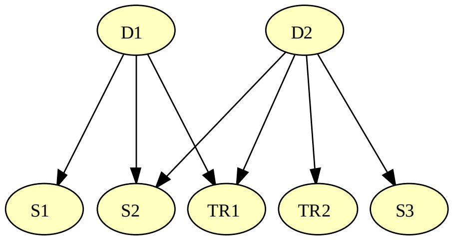


Figure 1. Example of a Bayesian belief network modelling the classical diagnostic problem. The root nodes (*D*-nodes) represent diseases, while the descendants model symptoms and tests results (*S*-nodes and *TR*-nodes respectively). A *D*-node connects to an *S*-node if the first is a possible cause for the symptom in the *S*-node.

Because the model is built in causal form (arrows in the network point from causes to effects), designing the graphical representation of a diagnostic problem is reasonably straightforward, using medical knowledge of the domain.

The basic task for the diagnostic system, under a probabilistic framework, is to compute the posterior probability distribution for a set of query variables, given exact values for some evidence variables. That is, the system computes the probability P(*Query*|*Evidence*). In the example of Fig. 1, *D*-node D_1 is a possible query variable and S_1 and TR_1 could serve as evidence variables – typical diagnostic reasoning. In Bayesian belief networks any node can serve as either a query or an evidence variable. Four distinct kinds of inference can thus be performed:

- Diagnostic inference (from effects to causes). Ex: given a symptom S_1 infer the probability of the pathology D_1, $P(D_1|S_1)$.
- Causal inference (from causes to effects). Ex: given disease D_2 find the most likely symptoms, $P(S_i|D_2)$.
- Intercausal inference (between causes of a common effect). Ex: given S_2 infer $P(D_1| S_2)$, but adding evidence that D_2 is true makes the probability of D_1 go down. Although D_1 and D_2 nodes are independent the presence of one makes the other less likely. This phenomenon has been termed *explaining away* [20];
- Mixed inferences (combining two or more of the above).

3. AUXILIARY DIAGNOSTIC DEVELOPMENT TOOL

The proposed system provides an integrated development tool for designing specific diagnostic applications. With an easy-to-use interface, this tool gives the user the possibility to design his particular application not requiring knowledge of BBNs background theory.

3.1. Interface and Functionality

The diagnostic system essentially provides a graphical interface for domain knowledge introduction and a Web page (input/output) interface to interact with the expert using the system for diagnosing.

Two interaction phases are supported: (1)- *design* of an application in a particular domain; (2)- routine *usage* of the application as an auxiliary diagnostic system.

In the first phase, a graphical interface (see Fig. 2) enables a structured introduction of the domain knowledge, ultimately defining the structure of the BBN (topology and CPTs). The main concepts/components handled by the graphics interface are the diseases, symptoms and test results (corresponding to *D*-nodes and *S*-nodes as well as *TR*-nodes referred previously). Despite of its default boolean-valued (with *absent* and *present* values), the user can choose the variable (node) to be labeled-valued, for example to characterize a test result with possible values: *low*, *normal*, *high* and *too high*. At the same time the user is allowed to add help text to each node, that latter on will be accessed via Internet.

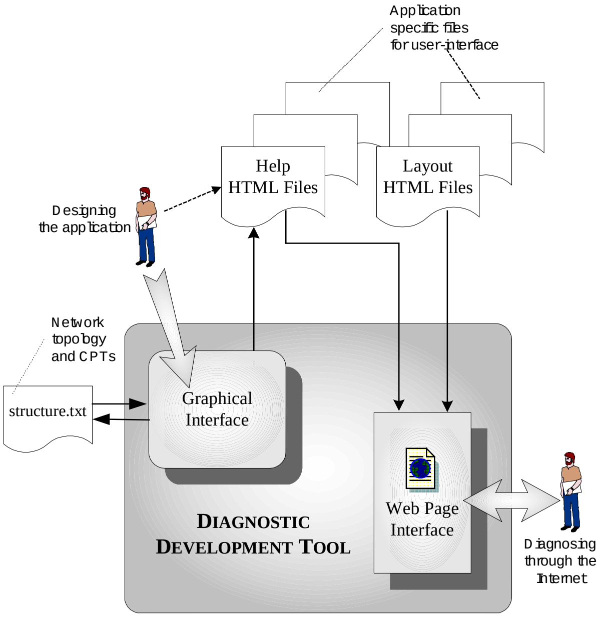


Figure 2. System interface and functionality.

Additionally to designing the network structure (knowledge in the given medical context) the user has the possibility to customize help information by simply writing in HTML *Help* files. Moreover, layout customization of the diagnostic interface is also possible through changes in the respective HTML *Layout* files.

A preliminary version of this system used HUGIN [11,14] environment to do the graphical interface (taking advantage of the HUGIN output file describing the user-defined structure). The complete graphical interface is currently under development.

After the application has been designed, the expert accesses the system through an Internet browser – *usage* phase. By interacting with the Internet page the user can operate for diagnosing, putting in action the diagnostic system with its inference mechanism. Fig. 5 shows an example of the interface, corresponding to the application described in section 4. Any variable node can be set as evidence, all typical queries described in section 2 being possible through this interface (diagnostic, causal, intercausal and mixed inferences).

3.2. Architecture and Implementation

A block diagram representing the architecture and implementation of the diagnostic development tool is presented in Fig. 3. The system consists of three major parts: a) the inference algorithm module; b) a Java interface module; and finally the diagnostic-system (input/output) Internet page.

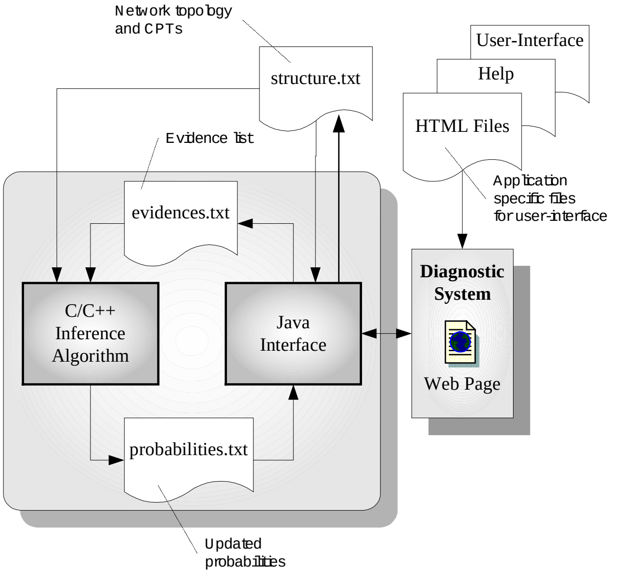


Figure 3. Block diagram representing the diagnostic system architecture and implementation.

The inference algorithm is implemented in C/C++ language and its main function is the computation of the query-variables probabilities: given the evidence nodes (entered by the user in the Internet page and stored in *evidences.txt*) computes the probabilities for the other nodes. An output file is produced with all actual variable probabilities (*probabilities.txt*). Considering that most of the medical diagnostic models have a simple structure (composed of causal disease-symptom relations), this algorithm takes advantage of that simplified model with efficient computation for probabilities inference based on a forward-backward procedure [6,17,18].

The Java interface module supports the information exchange between the Internet diagnostic-system page and the inference algorithm. Hiding particular aspects of the implementation, this module gives the users a friendly interface. The graphical tool currently being developed is also integrated in the Java interface module. Notice (in Fig. 3) that this module outputs information to the external *structure.txt* file.

Forming the core of the system, these two modules interact with the diagnostic-system Internet page; this provides interface with the user for input/output. Its associated HTML files provide the designed output layout and help files.

When designing the application the user indirectly introduces knowledge in *structure.txt* and HTML help files. Later, when opening the diagnostic-system Internet page, the network topology and CPTs are loaded to the inference module, reading data from the *structure.txt* file. After requested by the user, the diagnostic system runs the algorithm for probabilities inference. The Internet page exchanges data with the Java interface (evidences and updated probabilities) each time the user interacts with the system.

This development tool was designed to work on a PC platform requiring a Java runtime interpreter to interact with the file system (load/save files) and a Java-compatible browser to use the associated HTML files. It however can be exported to other Java virtual machines such as workstation, hand-held PC, cellular phone and others.

4. APPLICATION TO SLEEP-DISORDERS DIAGNOSIS

An example of an application of this tool is the *Sleep-Disorders Diagnostic System* (SDDS). It is a medical decision-support system to help in diagnosing sleep disorders, using clinical and polysomnographic data. The construction of this application was based on the experience of previous work on medical diagnosis [4,5].

Part of the domain knowledge is represented in Fig. 4 showing the network topology concerning the following pathologies: *Psycophysiological Insomnia*, *Idiopathic Insomnia*, *Obstructive Sleep Apnea Syndrome* and *Narcolepsy*; the corresponding *D*-nodes are respectively PSIC, IDIO, OSAS and Narco. The network topology and its conditional probabilities were specified from medical literature [2] and from consultation with experts in the field.

The user accesses the SDDS via an Internet browser, which displays an output like the one illustrated in Fig. 5. By propagating the probabilities through the network less experienced experts can use this network as an aid in diagnosis of sleep-disorders.

An example of interaction with the SDDS follows: for a given patient, the initial subset of symptoms observed to be present or absent are entered through a radio button interface; this is shown on the left side of the Internet page illustrated in Fig. 5. The diagnostic action is then selected which triggers the propagation of the probabilities through the network giving a ranking of the current disease probabilities and even a list of the most likely symptoms not yet instantiated. Having established the presence/absence of the next set of observed symptoms a new diagnostic action can be made helping to confirm, or refute, the presence of a certain disease (or more than one). On the right side of Fig. 5 note that the PSIC probability is higher than the other pathologies in the presence of DFW, INS and CAB symptoms. Observe also that DFSD symptom became more likely to be present because of the PSIC increased probability while the other symptoms turned to be less probable since they have no correlation with this disease (PSIC).

Selecting a certain pathology to be present and then observing the most likely associated symptoms is another example of using this system. This mode, referred as causal inference in section 2, is helpful for inexperienced experts to study the disease-symptoms correlation.

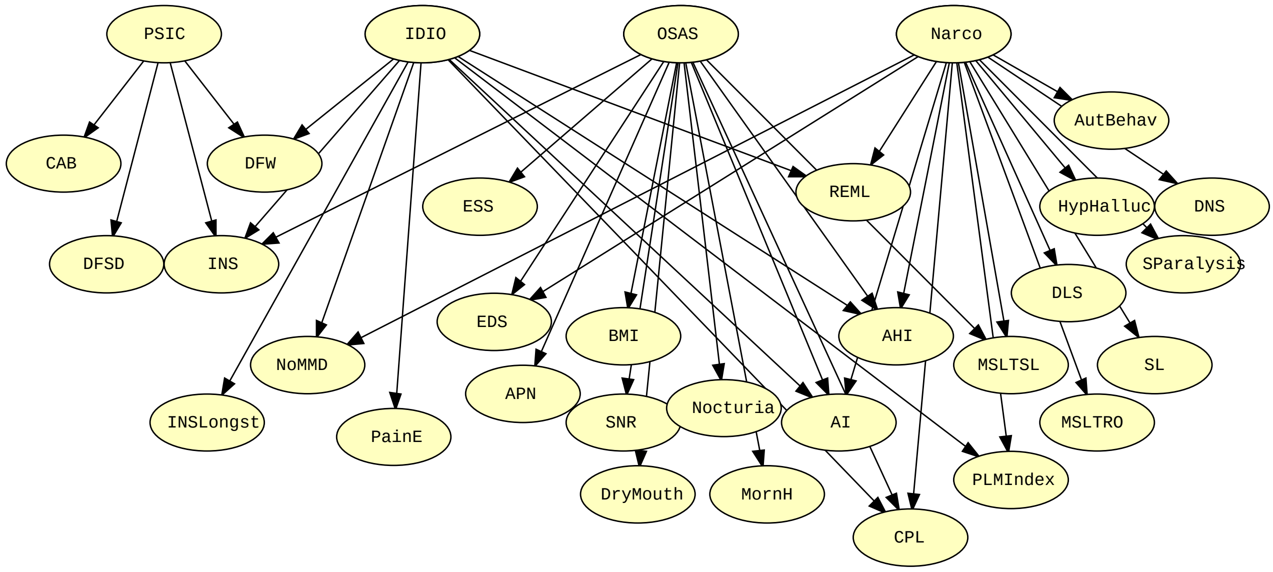
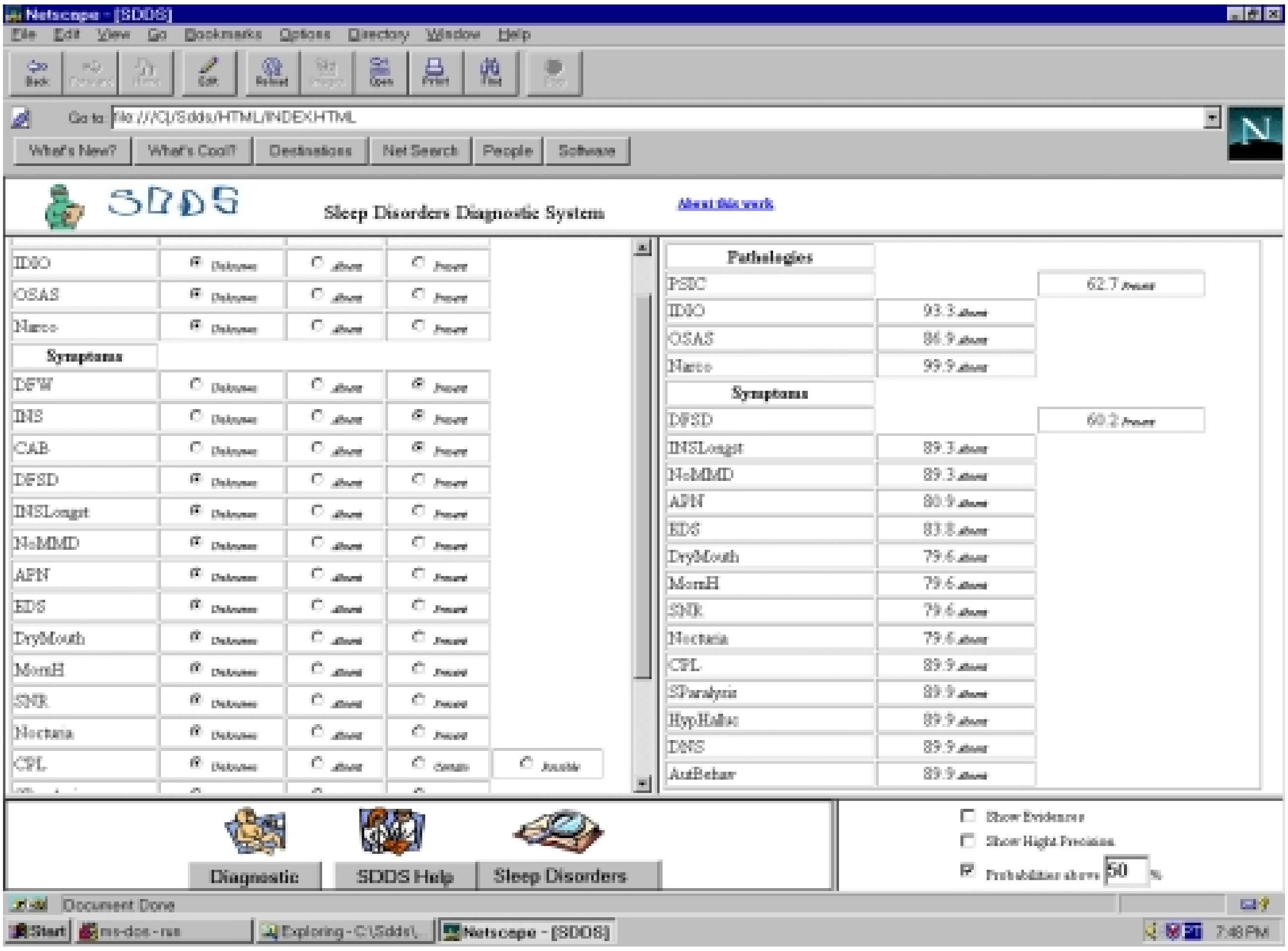


Figure 4. Bayesian belief network topology modeling the sleep-disorders diagnostic problem.



Pathologies		
PSIC		62.7
IDIO	93.3	
OSAS	86.9	
Narco	99.9	
Symptoms		
DFSD		60.2
INSLongst	89.3	
NoMMD	89.3	
APN	80.9	
EDS	83.8	
DryMouth	79.6	
MornH	79.6	
SNR	79.6	
Nocturia	79.6	
CPL	89.9	
SParalysis	89.9	
HypHalluc	89.9	
DNS	89.9	
AutBehav	89.9	

☐ Show Evidences
☐ Show Hight Precision
☑ Probabilities above 50 %

Figure 5. The SDDS Internet page, showing the results of a diagnosis.

The user interface also provides an on-line manual, supported by HTML help files, with the explanations for all the pathologies and symptoms, as well as the polysomnographic data (see the example in Fig. 6).

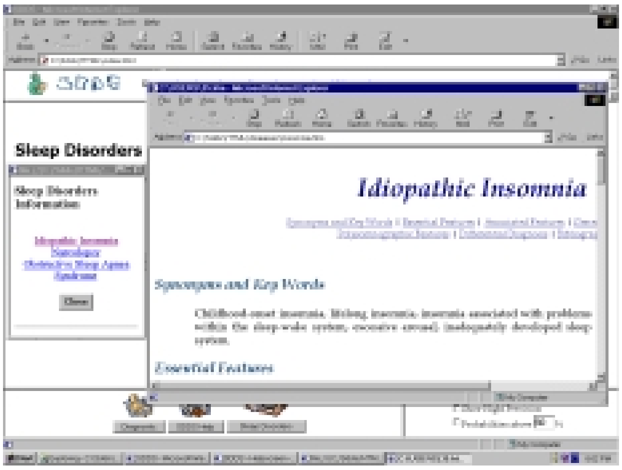


Figure 6. The SDDS Internet page with one of the associated help files.

5. CONCLUSIONS

This paper presented a development tool for designing medical diagnosis applications. With an easy-to-use interface, this tool gives the user-experts the possibility to design their particular applications without BBNs background knowledge. Taking advantage of the simple structure of diagnostic models (composed of causal disease-symptom relations), efficient statistical inference mechanisms were implemented, relieving the computational burden. Moreover the developed system provides an Internet Page (input/output) interface to interact with the expert when using the designed application for diagnosis.

An example concerning the application of this tool in the context of sleep-disorders diagnosis (SDDS) was designed and presented.

Since *Decision Theory and Adaptive Systems Group* at *Microsoft Research* is working on XBN format [21] moving toward to a *Bayesian Network Interchange Format* (BNIF) represented in XML[1], work is currently being made to use this format (XBN) in the diagnostic system. Adding this facility to the interface between the system and the user designing the network (to represent network topology and CPTs) new advantages are presented: possibility of using the MSBN free software, system versatility and future compatibility with other similar systems.

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[1] The *Extensible Markup Language*, representation introduced by the World Wide Web Consortium

Future work includes the embedding of learning algorithms [9,10], validation and comparative studies of the SDDS [5], as well as the application of the tool to other medical domain.

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