Expert Systems in Government Symposium

Kamal N. Karna, Editor

IEEE COMPUTER SOCIETY

The MITRE Corporation

THE INSTITUTE OF ELECTRICAL AND ELECTRONICS ENGINEERS, INC.

IEEE

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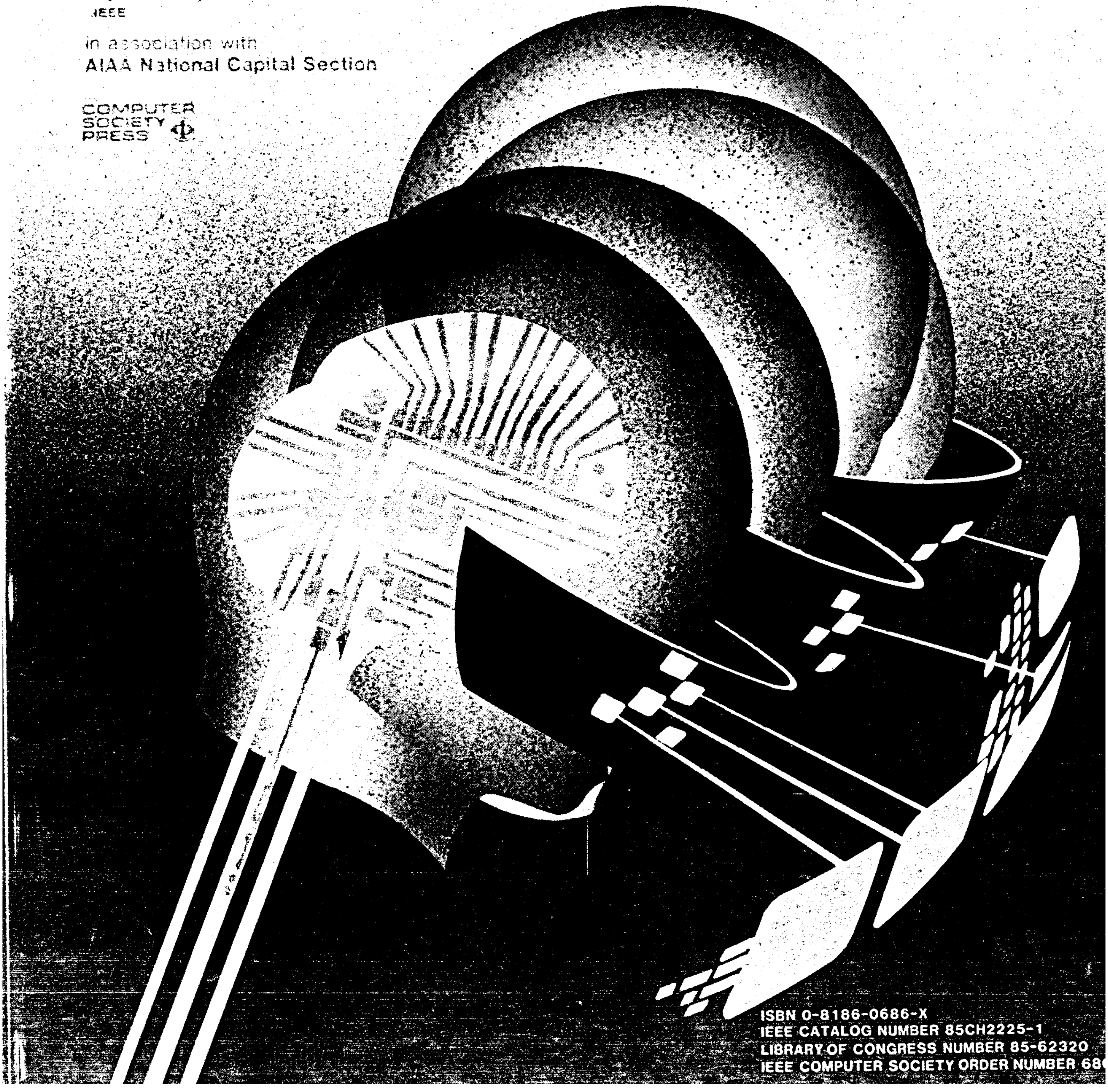
AIAA National Capital Section

COMPUTER SOCIETY PRESS

October 24-25, 1985
McLean, Virginia



ISBN 0-8186-0686-X
IEEE CATALOG NUMBER 85CH2225-1
LIBRARY OF CONGRESS NUMBER 85-62320
IEEE COMPUTER SOCIETY ORDER NUMBER 68

A MODELING SCHEME FOR DIAGNOSIS

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ABSTRACT

Model-based diagnostic expert systems need knowledge representations of spatial structure and function. In this paper, we argue that a semantic network representation is an effective approach to this problem and supports diagnostic reasoning, interactive graphics, image analysis, and natural language interfaces. As a case in point, we describe the neuroanatomic module of an expert system for neurologic diagnosis, NEUREX, currently under development.

1. Introduction

The objective of a *diagnostic expert system* (DES) is to assess the internal status of the *subject* of the diagnostic evaluation, be it a patient or a device, from observed behavior. There are two approaches to the design of such a system. The first maps complaints (symptoms) or behavioral changes (objective findings) to symbolic names for the underlying cause of the internal problem (specific diseases). The mapping is guided by *rules* which proceed through intermediate stages to reach diagnostic conclusions. Such systems are based on *shallow* knowledge, because they do not relate input data and conclusions to the *structure* of the subject and to the *function* associated with components of structure. The second approach makes such associations, using *deep* knowledge in the form of a *model* of the subject[5, 8]. A deep reasoning system is also said to be based on *first principles*[9]. Understanding how the subject is structured and how it functions extends a system's diagnostic capabilities. For instance, a machine repair DES needs to know how the machine *works* in order to analyze complex malfunctions; whereas simple disorders may be handled by mapping symptoms directly to specific types of dysfunction -- by knowing empirically how the machine *fails*.

Representation of *spatial structure* and the *function* associated with the components of that structure is a key element in building a model-based DES. Spatial (physical) structure alone captures the physical characteristics of the components and their interconnections. The functions of the components and their interactions explain the causal relations between the internal status of the subject and its behavior. There are several possible levels of abstraction (or grain sizes) in such representations. The level chosen depends on the needs of the diagnostic task. For example, it can be mapped into an abstraction called *logical* structure. A subject may have more than one logical structure associated with a particular way of decomposing function. Since diagnosis is an analytic process, logical structure follows from physical structure; whereas in a synthesis problem, the reverse is true. Thus we need a representation that allows us to describe spatial structure, function associated with the components of that structure, and the logic of the structural-functional relationships necessary for diagnostic reasoning. In this paper, we outline the constituents of these elements and argue the effectiveness of a semantic network representation. The latter accommodates a large, diverse information base; allows structure to be associated with function; and can support diagnostic reasoning, interactive graphics for data entry and explanatory output, image analysis, and natural language interfaces. To exemplify the use of semantic networks in this manner, we present a proposed representation of functional neuroanatomy from an expert system for neurological consultation under development at our institution.

Section 2 is a brief review and critique of basic approaches to representing spatial knowledge. Section 3 describes the semantic network approach and argues its effectiveness. Section 4 introduces the fundamentals of neurological diagnosis. Section 5 reviews previous works on computerized neurological consultation. Section 6 discusses the representation of spatial structure and function in our system, NEUREX. This system was previously referred to as NEUROLOGIST-II[30].

2. Spatial Structure and Function Representation

The spatial structure of a subject refers to its organization in three-dimensional (3-d) physical space. In special cases it may be two dimensional. The representation of 3-d spatial structure has been studied in computer science for a long time[15, 25]. The methods used are either *analogical* or *propositional* [1].

An analogical representation is a detailed *geometrical* description. This includes mathematical equations and division of 3-d space into volume elements (voxels) where sets of voxels specify the curves, surfaces or objects within the space[25]. They have the following advantages: the spatial structure of the entity represented can be succinctly and unambiguously defined, inference rules for spatial reasoning can be implemented by algorithms from computational geometry, graphics and image processing techniques can be adapted without much difficulty. However, they usually overspecify the real world, since every entity can not or need not be given an exact geometrical description. Furthermore, they sometimes have little relationship to the cognitive approaches which human beings use.

A propositional representation abstracts salient *topological* features in order to describe entities in terms of shape, position, etc. and spatial relationships by adjacency, connectivity, direction, etc. This format favors modeling intelligence and is a promising way to overcome some of the disadvantages inherent in analogical representations. However, it too has limitations: not all structural information can be expressed by propositions. Sometimes, spatial information is better depicted in pictures.

The function associated with components of the structure

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defines the behavior of the subject. For example, the logical relationships between the input and output terminals of such components as "AND gates" and "OR gates" determine how a digital circuit works. Similarly, a muscle is usually innervated by several nerves and provides a certain percentage of the force of a particular movement at a joint. It is important to combine structure and function[18]. However, few investigators have explored this problem in detail. We believe the representational format should accommodate both analogical and propositional information about structure and function, while fulfilling the other requirements noted in Section 1.

3. Semantic Network Representation

Cognitive knowledge is generally organized in the form of *concepts* and their *relations* to each other. A physical entity, each of its physical-spatial properties, and its function are all independent concepts which relate to each other when, in combination, they describe the entity. A complex system can be decomposed in several ways, each corresponding to a different logical structure, into sets of entities which may interweave with each other. The relations between entities such as spatial relation and functional relation are also specific concepts.

A semantic network[12, 20, 21, 27] is a representation in which each concept (including each relationship between concepts) is represented by a specific *node* linked to other nodes by predefined *arcs*. Fig. 1 shows the general organization of such a network

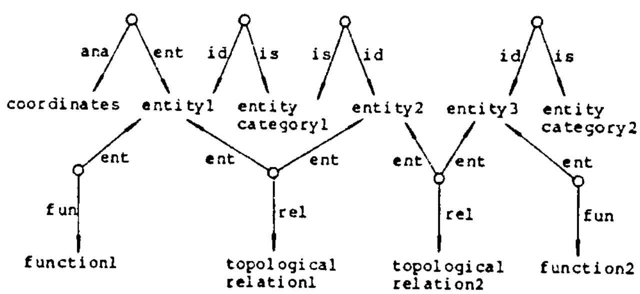

Fig. 1. The general organization of the semantic network representation of spatial structure and function.

representing spatial structure and function. Each conceptually significant entity is represented by a unique node such as "entity1" or "entity2". The node with "ent" arc pointing to "entity1" and "fun" arc pointing to "function1" asserts that "entity1" has function specified by "function1". The analogical information about entity1 (expressed by coordinates, equations, etc.) is asserted by the node with "ana" arc pointing to "coordinates" and "ent" arc pointing to "entity1". Similarly, relations between entities, such as topological connections, are represented by nodes and arcs (see the node with "rel" and "ent" arcs).

Semantic network representations have been used before for implementing expert systems. PROSPECTOR, a geological analysis system[19], used partitioned semantic networks to represent knowledge. The latter consists of production rules and subset and element taxonomic information. The following are the advantages for implementing a DES based on spatial structure and function:

(1) Analogical, propositional and functional knowledge can be integrated into a single network which reflects different levels of abstraction. Each physical entity is "surrounded" by its geometrical and topological descriptions (if any exist), other spatially and/or functionally related entities and its function, i.e., a locally limited search of the network can provide all the information relevant to the entity.

(2) Rule-based inference can be supported. A typical rule consists of two parts, antecedents and consequents both of which may contain variables. To check the satisfaction of the antecedent is to find the existence of certain nodes in the network. The consequents cause new nodes to be built. Complex control strategies can be implemented on top of the basic network processing system[10, 13, 14, 22].

(3) It can be easily expanded and modified. Adding and removing knowledge are in fact adding and removing concepts in the network which are both fundamental operations. The analogical data, e.g., coordinates can be changed independently and there is no need to modify the propositional information as long as the relevant spatial relations still hold.

(4) Procedural knowledge can be represented by the use of function nodes, i.e., nodes representing procedures. For example, spatial reasoning may involve applying algorithms from computational geometry using analogical data in the network. Moreover, probabilistic data[26] such as certainty information of rules can be propagated by procedural calculation.

(5) Interactive graphics for entering symptom data can be well-supported[29]. The analogical data provide the basis to generate relevant drawings which can be used to enter data using locator or pick devices. Explanation capability is also greatly enhanced by generating appropriate drawings and pictures from the analogical data.

(6) New knowledge can be derived from existing knowledge and stored for future use. For example, well-defined topological relations between physical entities can be systematically computed, using analogical information already present, and added to the network.

(7) It has the potential to support computer vision techniques to recognize objects in pictures[1, 25]. The geometric description in the network provides the geometric structure and the topological relation provides the relational structure. The two structures together with other relevant information can be used to guide image segmentation, labeling, and interpretation of the pictures.

(8) It supports the development of a natural language interface between the system and the users since semantic networks are one of the major internal representations used in computational linguistics[23, 24].

In summary, a semantic network is suitable for a model-based DES. The knowledge embedded in the network provides an understanding of how the subject is structured, how it works, and how it may fail. In the following sections, we present an example from the medical domain -- that of neuroanatomic localization, a crucial component of a much large DES which will simulate the behavior of a neurologist.

4. Fundamentals of Neurological Diagnosis

In the first stage of diagnosis -- initialization of the clinical database -- the neurologist collects preliminary data, including qualitative and quantitative descriptions of symptoms (Sxs), the relationships between Sxs, the results of past and present physical examinations (Pxs), adjunctive laboratory data (Lxs), other relevant information, and the overall temporal profile or course of the illness. The clinician documents these in writing on forms and, particularly Pxs, on pictorial drawings[7]. The latter not only indicate the extent of the disability but, when designed appropriately, also provide considerable information about the anatomy underlying the findings. For example, Fig. 2(a) is a line-drawing on a graphics screen of a head and neck. Fig. 2(b) is the same drawing on which is superimposed the distribution of the cutaneous sensory nerves to the area. By drawing sensory loses on Fig. 2(a) and superimposing them on Fig. 2(b), one can readily identify the malfunctioning nerves.

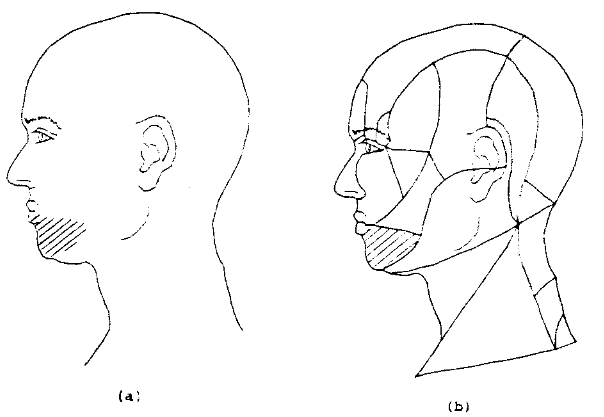

Fig. 2. (a) The view of the head and neck used to indicate a sensory disturbance.
(b) The same view on which is superimposed the distribution of the cutaneous sensory nerves to the area.

In the next stage, the diagnostician uses functional general anatomic and functional neuroanatomic knowledge to infer the presence and site(s) of the cause of the neurologic Sxs, Pxs, and Lxs. This is called *neuroanatomic localization*. It is the scientific foundation upon which the remaining diagnositic analysis is based. It consists of two steps: axial and transverse localization. In the former, clinical data are assigned to their appropriate axial neurosystem(s). The latter is an idiosyncratic phrase conceptualizing a neuroanatomic-physiologic or neuroanatomic-psychologic unit transmitting or processing a specific set of clinically definable functions and roughly paralleling the axial lines of the body and limbs as it extends physically through many of the transverse segments into which the neurologic system is traditionally divided. These neurosystems may or may not be working normally. The "transverse" localization tries to identify the specific transversely-orientated segment(s) of the *central nervous system* (CNS) or *peripheral nervous system* (PNS) where the axial neurosystem(s) are involved, thereby defining the anatomic coordinates of and precisely pinpointing the lesion.

The neurologist (or neurosurgeon) has in mind a functional, 3-d model of the clinically important parts of the neurologic system along with its receptors (the eye, ear, sensors in the skin, etc.) and effectors (skeletal muscle, sweat glands, etc.). He/she uses this in the neuroanatomic localization. Fig. 3 shows a schematic of the spinal cord at three of its 31 levels with the distribution of a few axial neurosystems (tracts) and segment limited (transverse) structures. One half of the cord mirrors the other. The physical

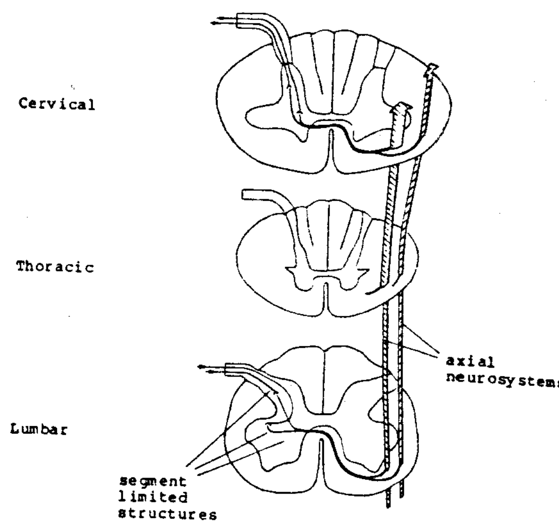

Fig. 3. The anatomic structure of the spinal cord.

positions and, therefore, spatial relations are fixed and vary little from one individual to the next. These pathways, in turn, carry information about homologous areas on each side of the body and the latter are also relatively consistent from one individual to another. Therefore, lesions on one side of the thoracic cord at a given level, regardless of etiology, will cause a predictable pattern of neurologic deficits from one patient to another, so long as they involve the same axial and segment-limited structures. Conversely, a combination of Sxs, Pxs, and Lxs can be traced back to its anatomic source. The latter helps the clinician to decide whether the patient has a single well-circumscribed (focal) lesion, more than one focal lesion (multifocal problem), dysfunction of one or a few entire axial neurosystems (systems-limited disorder), or an uncircumscribed process randomly involving many structures, usually in more than one transverse segment (diffuse disorder). Similar anatomic principles also govern structure, function, and localization in the brain (but with much greater complexity) and the PNS (see Fig. 4 for an example of a simple system connecting with a single transverse segment of the spinal cord).

Having determined the probable site and number of lesions, the clinician combines the neuroanatomic localization with elements of the clinical database (the relationship between Sxs, Pxs, Lxs; and the temporal profile of the illness) to deduce the underlying pathophysiology (ischemia, inflammation, etc.). Anatomy and pathophysiology form patterns suggesting pathogenetic categories of illness (genetically-determined disorders, physical agents, vascular disorders, neoplasia, etc.). The latter plus multiple epidemiologic facts allow the clinician to concentrate attention on a small number of disease-specific etiologies (atherostenotic occlusion of a specific blood vessels, embolic infarction, syphilitic

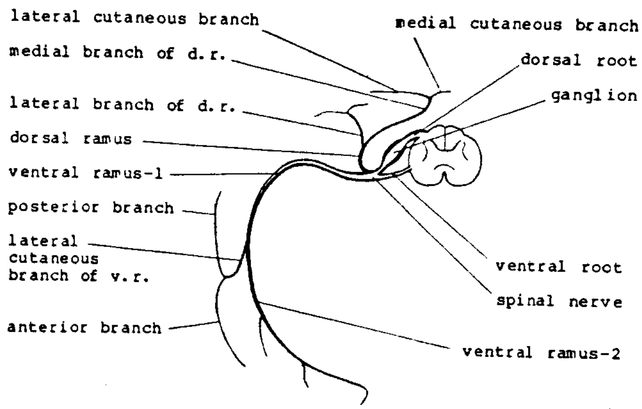

Fig. 4. The origin and distribution of typical spinal nerve.

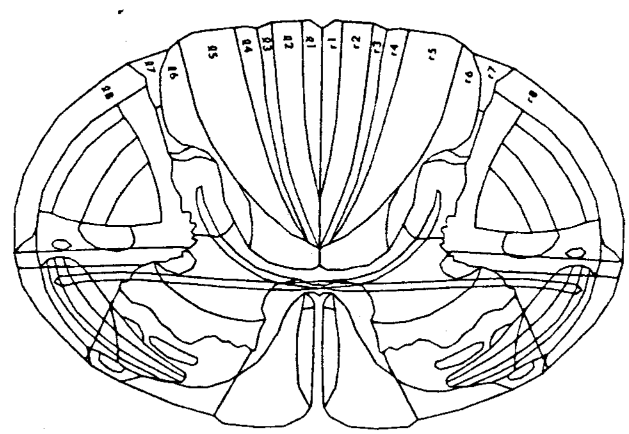

Fig. 5. The fifth cervical segment of the spinal cord (partially labeled).

endarteritis, etc.). Needless to say, hypothesis formation and data generation are on-going, interactive, and mutually correcting. Hypotheses lead to a search for additional data or correction of erroneous information. These, in turn, enhance the statistical probability of one anatomic location, one pathogenetic category, or one specific disease while decreasing the probability of competing alternatives. The diagnostic analysis culminates in rational therapy.

5. Prior Work on Computerized Neurological Consultation

If it is to simulate the analytic behavior of a neurologist, a DES in neurology must have a knowledge representation which effectively integrates both the structure and function of the neurologic system in order to support such tasks as entering clinical data on graphic illustrations of the human body as well as by literal forms, using the neuroanatomic model for diagnostic reasoning, and explaining its anatomic localization to the user on appropriate graphical reconstructions of the neuroanatomy.

A number of researchers studied selected aspects of knowledge representation in neurology and its related fields over the last decade. Some of the implementations are encouraging but clinically impractical. These can be grouped by the representational approach and the portion of neuroanatomy modeled.

5.1. Using Analogical Representation

The parenchymal CNS (brain and spinal cord) is typically represented by line drawings of transverse cross-sections in medical literature. This is a reasonably, idealized, analogical approximation of the 3-d complex without loss of generality. Fig. 5 shows a computerized schematic reconstruction of the major, clinically significant, axial neurosystems and segment-limited structures in the fifth cervical segment of the spinal cord. Each labelled area represents an anatomic region through which fibers of one or more than one axial neurosystems pass as they intersect the "transverse" section. (The right fasciculus gracilis, carrying several types of sensory neurosystem/somatesthesia fibers, intersects the fifth cervical segment in regions r1, r2, and r3.) Since curves can be approximated by lines each cross-section can be represented by a set of polylines and polygons. This approach is used by *Neurologist* of Catanzarite[4] and the earlier version of our system NEUROLOGIST-I[28], where further limitation is imposed that only convex polygons are used to represent nervous tracts and function association is simplified to mapping clinical data to tract (region) status.

Banks and Weimer[2] are using the voxel approach to provide an anatomic knowledge base (SCAN) for neuroanatomic reasoning. The human body is represented as being embedded within a large cube which is then divided into 27 (3*3*3) smaller cubes, each of them is likewise divided into 27, and so on to form a hierarchy of nested cubes. Currently the smallest cubes are each 3mm on a side. The neuroanatomic components are represented by another hierarchy of "objects", each of which is mutually associated with its physical correspondent(s) in the cube hierarchy.

In general, analogical representation defines idealized, structural neuroanatomy succinctly and unambiguously. To a certain extent, function can be associated with structure within the physical model. As will be discussed below, clinical assessable function is assigned to fibers in the PNS. These, in turn, are connected to fibers passing through the CNS areas shown in Fig. 5. It also supports implementation of spatial reasoning using geometric algorithms. For example, two aforementioned systems[4, 28] localize a focal lesion by computing a convex hull[16] which encompasses all the polygons formed by malfunctioning tracts in a given transverse segment (cross-section). If it intersects[3, 19] more than one intact tract, the system will reject the proposition that there is a single focal lesion in that segment. This method allows graphic devices to be used directly -- generating pictures using the geometric information.

However, the analogical approach has a major disadvantage which does not invalidate but clearly limits the conclusions which can de derived from the anatomic analysis. It oversimplifies the real life situation, because it does not provide appropriate levels of abstraction and flexibility. CNS anatomy and physiology, for instance, are very complex. Often, the boundaries of a specific neurosystem are not precise and only approximately predictable. Information is transmitted to a variable extent through several different regions, and functionally different nerve fibers interweave in one area and segregate in another. A circumscribed lesion may affect some but not all fibers passing through it. The convex polygon method reduces the complexity of computation but requires further simplification of these complexities[28]. The voxel approach is uniform and elegant from a mathematical point of view but unnatural from a cognitive perspective. The integrity of objects is not well-preserved: an entity which can be represented by a single cube may be represented by 8 cubes of the same size, each of which involves part of the object merely because it is not aligned against the grid. This will no doubt increase the complexity of reasoning.

5.2. Using Propositional Representation

Propositional representation is a possible way to overcome the above problems. It supports spatial reasoning more effectively[28] and has been used to represent the PNS[6, 11]. As shown in Fig. 4, the connectivity of nerve segments is the most important structural information. Each "transverse section" of the nerve (i.e., root, spinal nerve, dorsal and ventral primary ramus, branch, etc.) is composed of multiple nerve fibers, each with a specific function. The effects of a lesion in any section depends on the type and number of fibers involved in that segment.

Given a group of weak muscles, LOCALIZE[11] traces fibers which supply each affected muscle proximally toward the spinal cord and highlights the pathways. Any set of lesions which includes at least one highlighted segment from each traced pathway will account for all of the deficits. Starting with the set of most distal lesions, the program generates alternative solution sets by replacing set elements with more proximal lesion sites from the highlighted pathways. It tries to reduce the number of hypothesized lesion sites at each convergence point as long as the consistency checks can be satisfied, e.g., all muscles which the program expects to be weak due to a lesion at the convergence point are actually found to be affected. In its present form, LOCALIZE handles weak muscles and does not consider myotatic reflexes or sensory neurosystems.

However, propositional representation has its limits: not all the structural information of the neuroanatomy can be abstracted in the form of propositions, certain geometrical details are lost while topological features and relations are captured; graphics and image processing techniques are not supported, because they rely on geometrical data.

5.3. Rule-Based System Without Modeling

These approaches do not explicitly encode neuroanatomic structure. Rather, neuroanatomic localization depends on the presence or absence of specific symptoms and findings for which the system has rules. Reggia, using a production rule system, concluded that structural knowledge (understanding how the neurologic system is organized) is necessary for complex, spatially-oriented reasoning[17].

5.4. Comment

To be complete, the neuroanatomic model must be an integrated representation of the CNS and the PNS, including the functional distribution of the latter's principal neuroeffectors and clinically testable classes of neuroreceptors. We are not aware of a system which has achieved this goal successfully. Secondly, information about structure should be both analogical and propositional and provide appropriate levels of abstraction to facilitate anatomic localization and graphics techniques. Thirdly, the functional knowledge must support mapping of anatomic dysfunction from Sxs, Pxs, and Lxs. We feel this is the essential knowledge base upon which to define a reasoning mechanism which (1) will simulate a neurologist's clinical approach and (2) will localize a lesion(s) with an accuracy equal to or, preferably, greater than that of an expert.

6. Spatial Structure and Function Representation in NEUREX

The knowledge concerning the spatial structure and function of the neuroanatomy in NEUREX is organized as a semantic network according to the rationale presented in Section 3. The representation is implemented in Franz LISP and SNePS (Semantic Network Processing System[20]) which are completely compatible and mutually callable. The latter allows effective searches of the network knowledge base and the use of both procedural attachments (function nodes) and inference rules.

The anatomic knowledge base consists of neuroanatomy and general anatomy. The CNS is divided into its major "transverse" segments: telencephalon, diencephalon, brainstem (mesenchephalon, pons, and medulla), and spinal cord where material passing along multiple axial neurosystems are analyzed, integrated, relayed, and supplemented or removed. Each major transverse segment is subdivided into smaller segments and regions within segments, corresponding to developmental units and/or facilitating precise localization. Where appropriate, a transverse segment connects on its right and left side with the PNS via the latter's nerve roots or their equivalents. A PNS root is identified by the CNS segment to which it is attached. Each root network (CNS-PNS) innervates (transmits multiple types of sensory information from and/or motor directives to) specific regions or structures of the body. Except for the side of origin and peripheral distribution, homologous right and left CNS-PNSs are identical anatomically and innervate corresponding areas of the body. Each CNS-PNS passes through a system of conduits or peripheral nerves (PNv) to reach its termini. As they extend away from the CNS, PNvs assume a branch-tree structure which segments them "transversely" first into spinal or cranial nerves and, then, into as many additional subunits as needed until the final nerve segments are reached. Thus, the human neurologic system has three general patterns of innervation: one corresponding to CNS pathways; the second, to CNS-PNSs; and the third, to PNvs.

The major functional axial neurosystems transmitting information up or down the neurologic system are made up of smaller units. Each of these "minisystems" is identified uniquely by (1) the specific areas it occupies as it passes through each transverse segment of the CNS or the CNS and PNS; (2) the transverse segment of the CNS in which it originates, terminates, and/or connects with the PNS; (3) the direction it carries data; and (4) a clinically verifiable function. In reality, a functional minisystem represents a very large number of individual nerve fibers relaying the same information in parallel and in series. The point at which one set of its fibers connects with the next is indicated in the model, even though the transferred function does not change.

At present, we are working on the CNS, CNS-PNS and PNv representations of the axial somatic sensory\somatesthetic and somatic motor neurosystems. Both have central and peripheral components. Those minisystems responsible for somesthetic information destined to reach consciousness transmit from the lowest segments of the spinal cord to higher segments of the brain ("upward"). The major volitional motor minisystems transmit directives caudally ("downward"). Regardless of direction, the number of minisystems in these axial pathways decreases as one moves caudally, because they lose a minisystem at each succeeding transverse segment. Therefore, a CNS pattern of innervation -- motor or sensory -- at any given point along the central neuraxis depends on the number of minisystems in the axial pathway and affects the entire field of innervation to the half of the body supplied by these minisystems. A CNS-PNS ("root" or "segmental") pattern corresponds to the peripheral somesthetic or motor innervation of the root. Each CNS-PNS may be purely sensory, entirely motor, or both sensory and motor. If it has a sensory component, a CNS-PNS may carry one or, most commonly, several minisystems; if it has a motor component, it carries two types of minisystems. A PNv sensory or motor pattern may or may not be identical to that of a single CNS-PNS. Those related to the limbs are complex. Several adjacent CNS-PNSs combine in a complicated manner at specific junctional points (plexi) and then partially dissociate at more distal branch points. Therefore, a proximal PNv may involve sensory and/or motor minisystems (and innervation fields) from two or more CNS-PNSs. The composition of its more distal segments will be either the same or less complex, the latter depending on partial dissociation of the minisystems at any succeeding branch point. Conversely, a CNS-PNS may course through many or relatively few PNvs. Despite the complexity of the CNS, CNS-PNS, and PNv patterns, the anatomic pathways involved are unique from origin to final destination and remarkably consistent from one individual to next.

In addition, the body (general anatomy) is divided into its major regions (i.e., head, arms, torso, legs, etc.) and subregions (i.e., upper, middle, and lower third of the right brachium; joints; etc.). The latter serve as anatomic reference points which help to locate the components of the neurologic system, support maps of the cutaneous distribution of sensation, and organize skeletal muscle function.

The structural features of the aforementioned anatomic entities are attached to their corresponding concepts differently according to the features' significance: coordinates of the polyline and polygon representation for cross-sections, regions, and projected views of body parts; connectivity relationships for nerves and branches of nerves.

6.1. Representing the CNS

In the case of the CNS which can be geometrically described by cross-sections through transverse segments, every cross-section and every region in the section is an anatomic concept which among other things has a geometric description (i.e., corresponding coordinates in a common coordinate system) and is represented by an atomic node in the semantic network. More abstract relations such as adjacency or overlap of two regions can be asserted between the corresponding nodes. For example, the transverse segment of the CNS in Fig. 5 is the fifth cervical segment of the spinal cord and is represented by atomic node C5 in Fig. 6. Node C5 has a geometrical description which carries the coordinates of its boundary. Each region in the cross-section is represented by an atomic node, e.g., r1 in Fig. 5 is represented by node r1C5 in Fig. 6. Node r1C5 has a geometrical description which carries the coordinates of its boundary. The spatial relation that r1 and r2 in C5 are adjacent to each other is asserted by the node with "r" arcs pointing to r1C5 and r2C5 and "rel" arc pointing to "adjacent" in Fig. 6. Meanwhile, another set of concepts covers the major axial neurosystems ("tracts" or "pathways") in the CNS. Each nervous tract is represented by an atomic node. This is demonstrated by node 7R in Fig. 6 representing the right fasciculus gracilis. The physical location of the nervous tract in C5 is specified by an assertion indicating the corresponding regions by "path" arcs. Other information such as its specific function, evidence of malfunction, and so on can be further asserted.

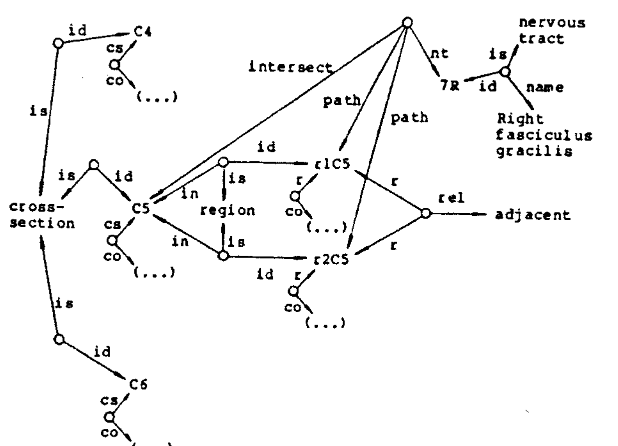

Fig. 6. The semantic network representation of the spinal cord (partially drawn).

6.2. Representing the CNS-PNSs, PNvs and Their Function Distribution

The CNS-PNSs and PNvs are represented topologically as network pathways where each transverse segment is an anatomic concept. Connectivity of segments is asserted between corresponding concepts. When a particular axial minisystem travels through the network, its pathway is specified by assertions relating the minisystem to the segments through which it passes. Each minisystem is identified by its parent neurosystem, its specific function(s), and its peripheral innervation field.

Fig. 7, for instance, illustrates the representation of the simple CNS-PNS system in Fig. 4 (a spinal nerve free of any intervening plexi is a pure CNS-PNS or root system). Anatomically

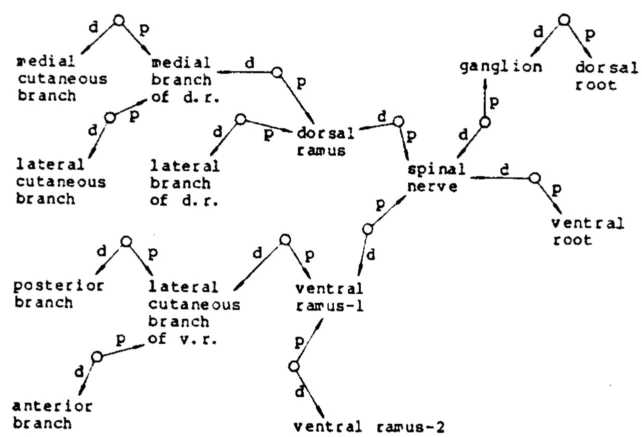

Fig. 7. The representation of the peripheral nervous system (partially drawn). Meaning of the arc labels:

p -- proximal
d -- distal

significant components of the CNS-PNS and transverse nerve segments of the PNv systems are represented by unique atomic nodes and the connectivity relations are specified by nodes with "p" (proximal) and "d" (distal) arcs. The pathway of a particular minisystem can be traced by a sequence of nerve segments and branches linked by "p" and "d" arcs from its origin to its destination.

To represent the CNS-PNS and PNv innervation patterns, we consider each region or structure of the body to be an anatomic concept, each with a geometric description outlining a corresponding region in a display drawing (see Fig. 8). Such information as the transverse nerve segments or CNS-PNS supplying an area are attached by assertions.

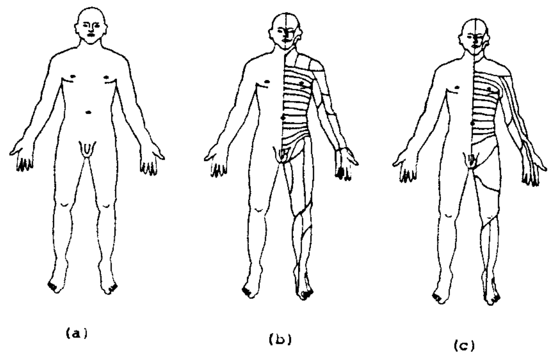

Fig. 8. (a) The outline of the body.
(b) The same drawing on which is superimposed the distribution of peripheral nerves (PNvs).
(c) The same drawing on which is superimposed the distribution of nerve roots (CNS-PNSs).

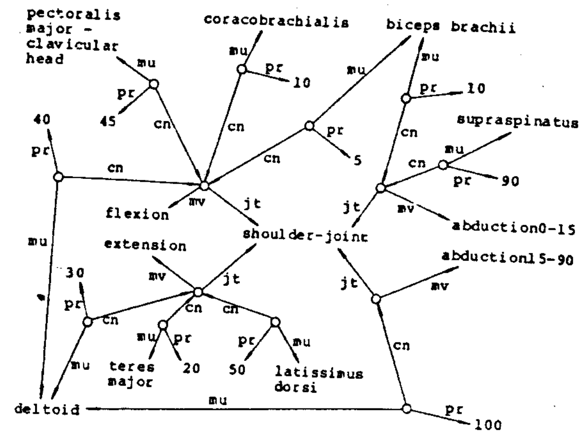

Fig. 9. Four of the shoulder joint movements with muscles involved and their contribution to each relevant movement. Meaning of the arc labels:

jt -- joint
mv -- movement
mu -- muscle
cn -- contribute
pr -- percentage

Additional information concerning anatomy and function are represented as concepts and asserted into the network according to the aforementioned principles. The somatic motor neurosystem, for instance, is responsible for movement which is an attribute of joints. The movement of a joint is controlled by several muscles, and muscles may be supplied by motor minisystems related to one or, as in the case of the limbs, more than one CNS-PNS. In regard to the latter, each motor minisystem contributes a certain percentage to the total innervation of the muscle and, therefore, to regulating the force of the muscle's contraction. Furthermore, a particular movement at a joint may require the synchronous contraction of several muscles. Thus, a muscle may be responsible for all or a fraction of the force of a movement. Muscles, joints, different types of movement, contributions of each muscle to a movement, and contributions of the innervation from each CNS-PNS are all represented by atomic nodes between which their functional relations are asserted. For example, the shoulder joint (movement of the humerus in relation to the scapula) has eight different types of movement: flexion (forward), extension (backward), abduction 0 to 15 degree, abduction 15 to 90 degree, adduction, external (lateral) rotation, internal (medial) rotation and rotator cuff. Flexion is controlled by the following muscles: deltoid (40%), pectoralis major - clavicular head (45%), coracobrachialis (10%) and biceps brachii (5%); extension by deltoid (30%), teres major (20%) and latissimus dorsi (50%); abduction 0 to 15 degree by supraspinatus (90%) and biceps brachii (10%) and abduction 15 to 90 degree by deltoid (100%), etc. The percentage in parentheses is an arbitrary approximation of the contribution which the corresponding muscle makes to the total strength of the movement. The representation of this information is illustrated in Fig. 9. The motor minisystems from each CNS-PNS innervating the muscle are treated in a similar manner. The complete pathway of a minisystem passing through the CNS and PNS is represented by other nodes, specifying its PNv pathway and the anatomic regions of each transverse CNS segment through which it travels.

6.3. Further Details and Examples

Following are further details and examples that demonstrate each of the advantages of the semantic network representation stated in section 3:

(1) Queries about structure and function. A request for the location of a particular minisystem in a cross-section of the CNS will produce a locally limited search of the network; and generate a picture such as Fig. 5 on the screen, highlighting the region, reporting the major axial neurosystem to which it belongs, its origin and termination, the direction it conducts information, and listing all other minisystems passing through the same area along with their function, and so on. Fig. 9 shows the use of the uniform network searching function in SNePS. To find which muscles are involved in the flexion of the shoulder joint, we issue the request (find (mu- cn) (find jt shoulder-joint mv flexion)) which returns a list containing the names of the four muscles involved. To find how much the deltoid contributes to the strength of shoulder flexion, we issue the request (find pr- (find mu deltoid cn (find jt shoulder-joint mv flexion))) and the program returns: (40). To find how paralysis of the deltoid will affect shoulder movement, we issue the request (find mv- (find jt shoulder-joint (cn- mu) deltoid)) and the program returns: (flexion extension abduction15-90).

(2) Rules. A rule might be stated as follows: if a particular minisystem carried by a certain PNv segment is malfunctioning then other minisystems in the same segment must be examined for malfunction. An outline of this rule is described graphically in Fig. 10.

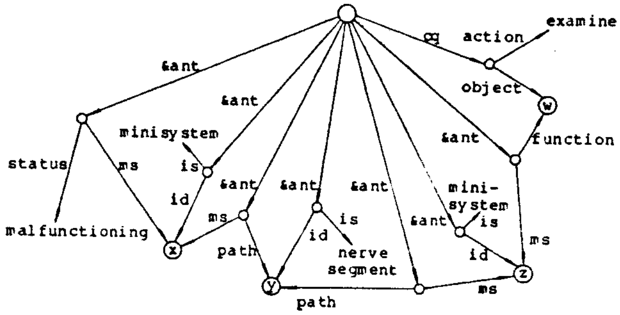

Fig. 10. The graphical description of the following rule in SNePS:

```
IF x is a minisystem &
   x is malfunctioning &
   y is a nerve segment &
   x passes through y &
   z is a minisystem &
   z passes through y &
   z has function w
THEN
   examine function w
```

(3) Revisions of the information base. If, for instance, research discloses that a minisystem passes through a region in a cross-section of the CNS different from that currently believed by the system, one need only build a new node specifying the relation between the new region and the minisystem and remove the old node relating the original region and the minisystem.

(4) Localization. If a lesion defined by a closed curve is located in a given transverse segment of the CNS, then all regions in the segment encompassed by the lesion should be affected and all the minisystems passing through these regions might malfunction. Another example is that since several adjacent or overlapping abnormal regions in the same cross-section tend to be affected by a single lesion, given a set of abnormal regions the task to decide the minimum number of lesions is in fact the task to decide the number of spanning trees in an undirected graph where a vertex represents a region and an edge represents an adjacency or overlap relation.

(5) Graphics support. The geometrical information about CNS-PNS and PNv patterns is used to display pictures such as those in Fig. 2 and 8 on the screen. Using a stylus or similar device, the extent of a sensory Sx or Px can be indicated on the picture. Localization begins by storing the picture in an image array, and comparing the abnormal area(s) to the appropriate underlying CNS-PNS and PNv innervation patterns to establish which of the latter most closely approximates the extent of the lesion. The results are stored as new nodes for further reasoning. For example, the sensory disturbance indicated in Fig. 11(a) partially matches the cutaneous distribution of three PNv segments (Fig. 11(b)) but more closely coincides with a CNS-PNS sensory pattern (Fig. 11(c)), indicating that a nerve root lesion is more likely. If one asks which CNS, CNS-PNS, or PNv pattern

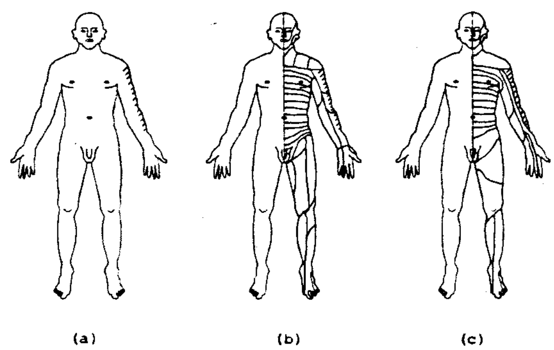

Fig. 11. (a) A sensory disturbance is indicated on the body figure.
(b) It partially matches the cutaneous distribution of three PNv segments.
(c) It closely coincides with a CNS-PNS sensory pattern.

best describes the lesion, the system will return the name of the nerve root, rejecting all other possibilities as unlikely. On the other hand, if the left side of the body is involved from the level of the nipple downward, the system will return the name and a drawing of the most rostral (transverse) segment of the spinal cord (CNS) containing all the somatesthetic-light touch minisystems involved and none of those uninvolved, along with an outline of the lesion. In fact, the geometrical information can be used to construct a 3-d picture on the screen, if the lesion extends through several transverse segments. One can also reverse the directive. Draw a lesion on the display of a particular transverse segment of the CNS and ask the system to outline the expected sensory loss on the appropriate graphic of the body. While we used the somatic sensory neurosystems to demonstrate some features of localization, the same principles can be demonstrated just as well with the somatic motor neurosystems, using weakness of muscles as the input.

(6) We create drawings such as those in Fig. 2, 5 and 8 with a geometric graph editor[29]. Adjacent regions always share a common boundary. This adjacency relationship is inserted into the knowledge base by a system-construction function which asserts an adjacency relation between every pair of regions sharing a common boundary for every cross-section.

(7) Computer-assisted tomography of the CNS. The geometric information in the knowledge based can be increased to provide realistic, age-dependent, geometric structures corrected statistically for normal variation. The system could then be expanded to interface with scanners (X-ray and nuclear

magnetic resonance) to relate tomograpnic output to functional neuroanatomy.

(8) A natural language interface for literal transactions, e.g., patient data entry and diagnosis explanation could be developed.

7. Summary and Conclusions

A model-based diagnostic expert system needs an integrated knowledge representation for spatial structure and function. A properly organized semantic network incorporating analogical and propositional structural information and the associated functional information supports not only diagnostic reasoning but also interactive graphics, image analysis and natural language interfaces. The case of neuroanatomic localization was presented as an example of the capability.

In order to build practical expert systems using all of the ideas presented here an efficient basic semantic network processing system such as SNePS is required in actual implementation. What constitutes spatial knowledge is generally not very well understood. Knowledge engineering in problems requiring spatial knowledge will no doubt reflect this. These problems have to be faced in addition to developing a detailed diagnostic strategy.

ACKNOWLEDGEMENT

This work was supported partially by SUNY/Buffalo Campus RDF grant 150-9867E. The authors wish to thank Dr. Jack Smith of the Ohio State University for his suggestions on improving the paper.

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