Systematised Nomenclature of Medicine (SNOMED) and the patho-anatomic diagnosis — the basis of the neuro-imaging report

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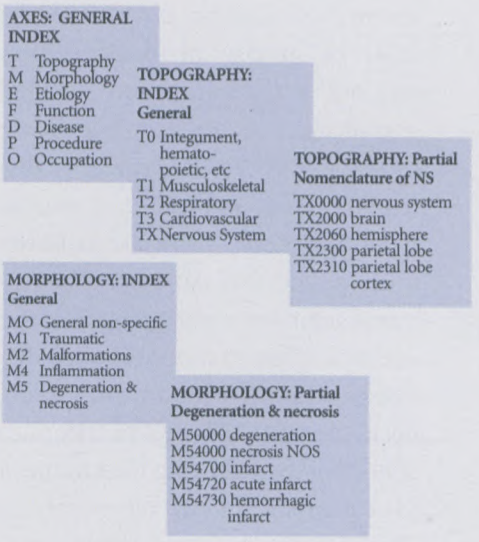
'Diagnosis is the mental act of integrating all the interpretations (of the history and physical findings) and selecting the one explanation most compatible with all the facts of clinical observation. To localise the disease process, i.e. to name the part or parts of the nervous system involved....is called the anatomic diagnosis'. (Adams and Victor, *Principles of Neurology.*)

Introduction

Many clinicians (and most students) are not aware of the abstract nature of the term diagnosis, which is compiled from the method of examination (visual, biochemical, pathological) and the nature of any abnormality within the scope of the examination. Viewed this way, the diagnosis requires a minimum of two components or operators: the region (or organ, system, tissue, cell, organelle, chromosome, or gene locus), and its structure (or morphology, condition, volume, texture, or composition) to which, with imaging, may now be added signal intensity and contrast enhancement.

With the advent of commercial computers in the 1950s, North American pathologists set about codifying the language of pathology in order to facilitate material archiving and retrieval (cases, slides and specimens), a process which had to be totally unambiguous — and therefore ultimately numerical. Systematisation of the basic operators in medicine, i.e **T**opography (anatomic localisation), **M**orphology (structure, both normal and abnormal), **E**tiology, **F**unction (physiologic status) and **D**isease was in the form of 'axes' (probably better regarded as fields) within which the comprehensive components were arranged in a logical numerical hierarchy:



This project, known as the **S**ystematised **N**omenclature **O**f **P**athology (SNOP), was widely adopted in the USA, and became standard usage in the Pathology Departments of UCT and Stellenbosch in the 1960s. Its application in our institutions initially had more to do with data retrieval (despite there then being only one official CPA mainframe), than the intellectual approach to the anatomic diagnosis, as this was never much discussed; certainly it was never emphasised by the consultants in those departments. Nevertheless, the habit of defining the diagnosis in autopsy and surgical pathology in terms of topography and morphology (at the very least) became ingrained among South African pathologists, although the detail and even common sense behind the application varied greatly among individuals.

By the 1980s, the College of American Pathologists had systematised the entire language of medicine (SNOMED), adding **P**rocedure and **O**ccupation fields, the former including every radiologic procedure in use.[1]

No doubt the idea of systematising the pathology nomenclature is simply a continuity of the heritage of European medicine, but as a structural diagnosis is the immediate objective of morbid anatomy, and also radiology, the ability to use topography, morphology, aetiology and function as a syntax in which objective elements are combined to express the causal relationships forming the concept behind the diagnosis, is readily apparent (Fig. 1).

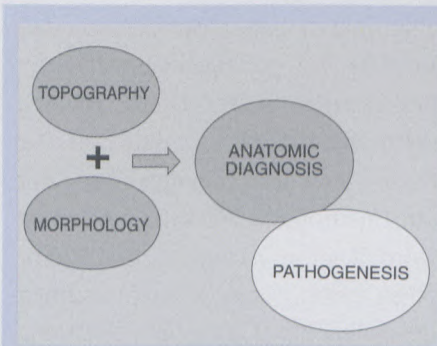


Fig. 1.

Since radiologists have never denied the anatomic basis of their speciality, and since the anatomic diagnosis is identical to both pathology and radiology (Fig. 2), it is hard to understand why such a reliable approach to the visual examination of organs and tissues never caught on among them. It is even harder to see why the RSNA had to invent yet another coding system of its own, some years ago.

The anatomic diagnosis requires two essential fields, i.e Topography (where/which structure) and Morphology (what condition/normal abnormal; if abnormal what is the nature of the abnormality), as depicted in Fig. 3.

However, the addition of other fields, besides allowing for a complete diagnosis, also permits a more detailed expression of causal relationships, referred to as the diagnostic syntax.

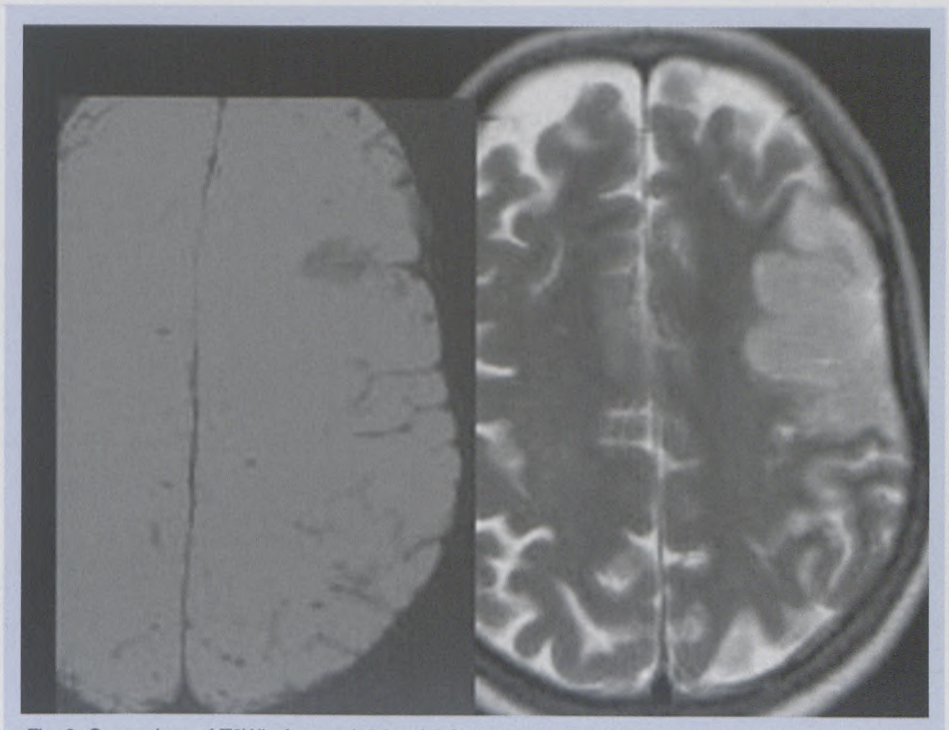


Fig. 2. Comparison of T2WI of acute (48 hour) infarct with the fixed brain specimen to illustrate the topographic and morphologic similarities.

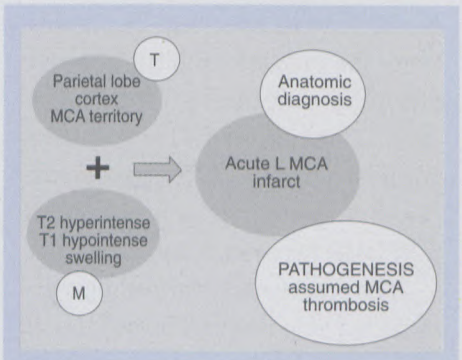


Fig. 3.

Thus, adding the aetiology, functional state of the patient and known disease, clearly defines the medical status of the individual in relation to the acute event, where even the patient occupation may be relevant. For example, using the structural abnormality as depicted in Fig. 2.: (T) parietal lobe : (M) acute infarct / (E) smoker / (F) hypertension / (D) diabetes / (O) radiologist.

If the patient had been subjected to DSA, the diagnosis could be even more informative: (T) terminal L ICA : (M) thrombosis / (Procedure) DSA.

Imaging in general, and MRI in particular, although depriving the examiner of colour and consistency, nevertheless provide other morphologic attributes which are probably more useful and which do not in any way detract from the anatomic diagnosis, namely density/signal intensity and contrast uptake. The problem for both pathologists and radiologists has to do with the relationship between the identified structural abnormality, and its pathogenesis. For example, the focal abnormality shown in Fig. 2 may be just as accurately designated a lesion, not otherwise specified (see below). This is particularly important in CT where a focal hypodense, non-enhancing lesion may have a number of morphologic possibilities.

Problems relating to morphology and pathogenesis

Within radiology, plain films, CT, MR and ultrasound are still primarily concerned with the macroscopic appearance of organs, tissues and spaces, distinctions between normal and abnormal, and the need for accurate, brief, verbal definitions of all of these. So it is surprising to find that quite often a request to an individual to provide the morphologic attributes of a region of interest is met with transient aphasia. This may be partly due to the habit that pathologists have of using morphologic terminology across the macroscopic-microscopic divide.

For example, the word infarct specifically implies ischaemic pannecrosis, and although brain infarct is often diagnosed macroscopically, pathologic confirmation is always microscopic. In radiology, the diagnosis of infarct is routinely made without reference either to its place within the general context of necrosis, or to the cellular reaction. In addition, because the imaging features of ischaemic necrosis may be non-specific, and because ischaemic injury itself may be selective and/or microscopic, there are many instances where the term infarct is either inappropriate or even wrong, and is best avoided altogether. Under these circumstances the only secure way of defining the lesion accurately is by means of the anatomic diagnosis, where the site and morphology can be stated, without any pathogenetic inference.

The hierarchy of diagnostic certainty, which is an essential feature of pathomorphologic description, is optimally expressed using SNOMED. Thus any structural abnormality with non-presumptive morphology is simply a lesion, and to make the total lack of pathogenetic inference quite clear, SNOMED includes a floater NOS — **N**ot **O**therwise **S**pecified. Fig. 4 illustrates the imaging problem of a solitary focal abnormality in the brainstem, exhibiting mild T1 hypointensity, diffuse peripheral contrast uptake (Fig. 4a), volumetric gain, and strong T2/FLAIR hyperintensity (Fig. 4b).

The precise nature or pathogenesis of such a lesion cannot be stated with certainty, so the anatomic diagnosis has to be : Brainstem/pons : lesion, focal, solitary NOS. Simply giving a list of the diagnostic possibilities, which includes infection/inflammation, demyelination, ischaemic injury and neoplasia, could be regarded by the attending physician as unhelpful or even troublesome, so that collateral is always sought, including that most essential item of information, viz. patient age.

In the case of a 40-year old, HIV-negative, Caucasian female, the anatomic diagnosis can be amended to include a pathogenetic inference: brainstem/pons: lesion, focal, solitary, demyelination not excluded; if the history includes a previous episode of optic neuropathy, the modifier can be worded more strongly: brainstem/pons: lesion, focal, solitary, assumed demyelination. Conversely, the same lesion occurring at age 60, would be much more likely to be ischaemic, less likely demyelinative, therefore: brainstem/pons lesion, focal, solitary, assumed infarct. If the clinical presentation included hypertension with focal motor deficit of acute onset, proposing an ischaemic pathogenesis would be reasonable. Conversely, the same lesion in a child, with no useful clinical collateral, would be very unusual, and should remain NOS.

Compared with pathology, imaging has the inestimable advantage of revealing the temporal profile of a structural abnormality. The same solitary lesion (Fig. 4), shown after a month to be T1/FLAIR hypointense and non-enhancing has to be designated a cavity or lacune; and since a cavity is a non-specific structure, to state its pathogenesis would require the same inductive approach. On the other hand, the appearance of a

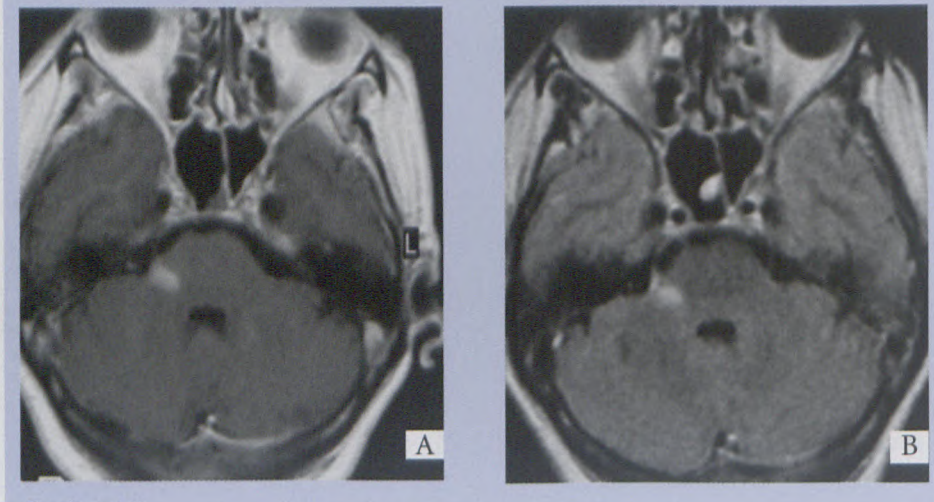


Fig. 4a and 4b.

second focal lesion, adjacent to the ventricle, would confer the strongest presumptive diagnosis of demyelinating disease.

Both aetiology and pathogenesis (aetiopathogenesis) are sometimes inferred from the imaging findings, as in meningitis, where neither the leptomeninges nor the nature of the alteration within them are actually seen. Thus, the uptake of contrast over the surface of the brain including the sulci, identifies an abnormality of the subarachnoid space and therefore the leptomeninges, while the associated findings of focal brain ischaemia and CSF obstruction combine strongly to suggest tuberculous meningitis. This inference is strengthened by collateral findings of CSF pleocytosis, abnormal chest X-ray, childhood state, etc.

Composition of the report

Just as the anatomic diagnosis can be applied directly to imaging, so the report format used by most North American pathologists is entirely appropriate to the needs of radiologists and is comprised of clinical, descriptive, interpretative and diagnostic sections.

Clinical

Essential data include age, race, blood pressure, risk factors, serology/disease status, plain film findings, treatment. Patient occupation may be helpful. Give the reason for the investigation.

Findings

Define the topography in terms of its morphology.

A basic list of structures requiring itemising includes the cerebral hemispheres, CSF spaces, brainstem, cerebellum and craniovertebral junction. Include specific anatomic extras if the clinical data warrant, e.g. temporal lobes in TLE. Dedicated regional studies, e.g. orbit, sella and skull base have their own essential topographic lists.

Define the morphology (normal or abnormal) in purely technical terms, always including symmetry, volume-profile, density, signal intensity and contrast uptake. A focal structural abnormality (i.e. lesion) requires in addition, specification of number, shape, size (in millimetres), conspicuity (also referred to as circumscription), and pattern of contrast uptake.

This component of the report is technically descriptive, and should contain no pathogenetic inferences, e.g. do not identify any structural abnormality or lesion in terms such as infarct, abscess, tumour, etc. Qualifiers (adjectives and adverbs) are often essential to the description, but to be used with care, and then only conventional terms acceptable to morphology, e.g. elongated, lobular, ring-form, elliptical, ventricle-adjacent, diffuse, patchy, etc.

Categorise abnormalities as primary or secondary, e.g. follow the description of a lesion exhibiting volume gain with a statement of mass effect. Omit needless negatives, i.e. if the morphology and signal intensity of the brain are normal, do not list abnormalities which are not present, e.g. midline shift, masses, etc.

Comment

Interpret the findings for the clinician, e.g. lesion attributes should be defined in terms of specificity or lack thereof; if non-specific, the combined imaging and clinical data may suggest the most likely differential morphology such as 'ring-enhancing lesion consistent with' granuloma, primary neoplasia, demyelination, etc.

Avoid specific histologic diagnoses if possible, e.g. in presumed neoplasia, identifying the tumour as primary or metastatic (or equivocal) is often adequate for management. Clinical data can (often should) be used to infer the pathogenesis and causal relationships, e.g. patient known with optic neuritis, pulmonary TB, HIV positivity, etc. If the nature and/or pathogenesis are uncertain, say so (see example below).

Identify the status of the abnormality when possible. Defining morphology in terms of signal and contrast uptake often provides the status of temporal profile, i.e. acute, subacute, etc. particularly in the case of haemorrhage and necrosis. A structural abnormality that is associated with volume loss of parenchyma, and is also unchanged following contrast administration, is likely to represent injury regression. In comparing follow-up studies, lesion status is in fact the main objective of the report.

Anatomic diagnosis

This is the bottom line, should be in telegraphic form, and as has been explained, must possess at least two components, one each from the topography and morphology fields. Search engines work on the association of words, so that the closer the terms are to each other, the more meaningful. However, all the key words, including terms from other fields (Etiology, Function, Disease) should be stated, especially the relevant status (hypertension, HIV, etc.). For example, if the age-status of the individual (childhood, adult, old age) is not specified, the influence of this often critical category on the diagnostic syntax, is lost.

The anatomic diagnosis in an imaging report often requires a statement of its own unique operators, such as the reaction to contrast administration, e.g. Brain/white matter : focal abnormality, non-enhancing/assumed primary neoplasm.

If the density or signal intensity or contrast uptake of the brain parenchyma is/are non-specifically abnormal, use of the terms lesion or encephalopathy is morphologically correct, e.g. brain/white matter : multifocal abnormality/leukoencephalopathy, assumed ischaemia/hypertension.

Examples

In the cases illustrated below selected details from the clinical, technical and interpretive parts of the report are synopsised.

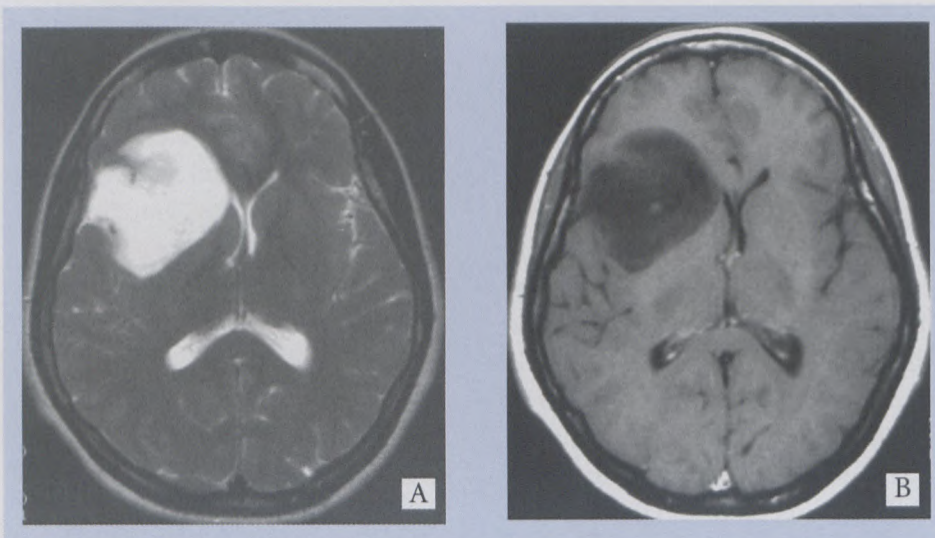


Fig. 5a and 5b.

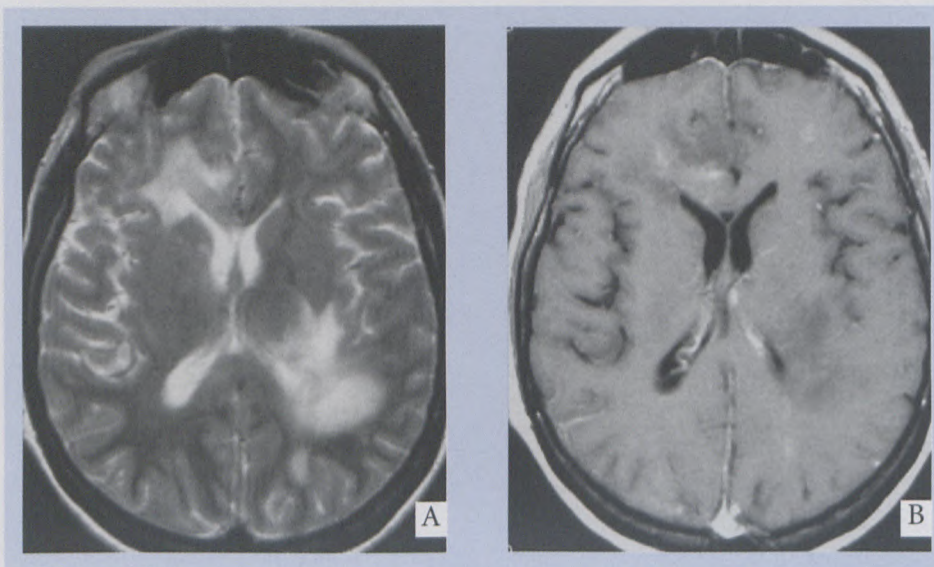


Fig. 6a and 6b.

Fig. 5. Signal (T2 hyper-/T1 hypointensity) structured inhomogeneity, conspicuity, mass effect combine to suggest a primary neoplasm of glial type. Absence of contrast uptake and conspicuity particulary fit astrocytoma.

Anatomic diagnosis: L hemisphere/insula/caudate: mass/primary neoplasm/assumed non-enhancing glioma (Biopsy: protoplasmic astrocytoma, Grade II).

Fig. 6. Subacute dementing process in a 40-year-old HIV-negative Caucasian female; multifocal anbnormality, with volume gain, non-topgraphic, i.e. without selective involvment of grey or white matter or a system; contrast uptake is patchy/linear possibly intravascular, and incongruous, i.e. not always matching the signal alteration. Encephalopathy, nature uncertain, could be infective, vasculitic or neoplastic.

Anatomic diagnosis: Brain/cerebral hemispheres/corpus callosum/thalamus: multifocal encephalopathy, pathogenesis uncertain, consider multifocal microangiopathy, e.g. CNS angiitis/neoplasia or unusual infection not excluded/subacute dementia.

Note: Biopsy undertaken on the strength of the imaging report was inconclusive. Anne Osborn's preferred diagnosis of intravascular lymphoma was confirmed at autopsy.

Fig. 7. Multifocal subacute haemorrhages having inferior frontal polar and lateral temporal lobe topography, with adjacent sudural blood. Picture is almost pathognomonic of coup-contrecoup injury.

Anatomic diagnosis: L occipital-convexity/meninges/subdural space: subacute haematoma. L temporal/inferior frontal lobes: subacute haemorrhage, assumed coup-contrecoup injury/head injury/fall.

Note: Subdural space is inferred from sulcal integrity; topography and morphology of parenchymal haematomas combine very strongly to allow the pathogenetic inference of head

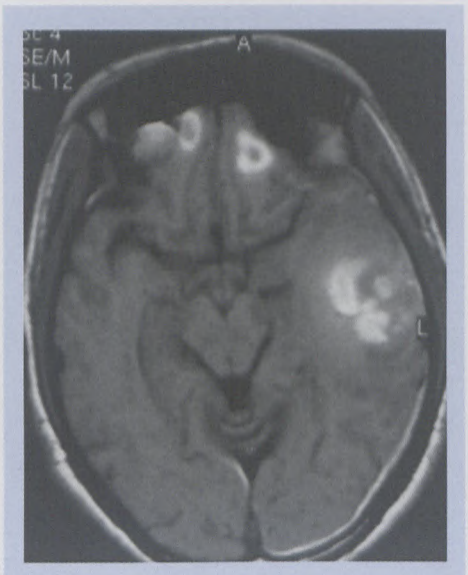


Fig. 7.

injury from a fall — even when no such history is obtained.

Fig. 8. A 56-year-old woman with 2 weeks of headache and limb weakness. Previous history of optic neuropathy. Solitary large mass lesion with rim conspicuity which does not match contrast uptake, including subtle diffusion into the T2 hyperintense core.

Anatomic diagnosis: R centrum semiovale/white matter: ring-enhancing mass/oedema; assumed tumefactive demyelination/dymelinating disease.

Note: original diagnosis was that of glioblastoma; however tidy rim (on all sequences without contrast) and antecedent optic neuropathy allow pathogenetic inference of idiopathic demyelinating disease. Confirmed on biopsy.

Fig. 9. One-year follow-up study of macroprolactinoma treated by decompression and bromocryptine. Imaging features include anatomic disorganisation of the sella-suprasellar region with cystic degeneration of adenoma parenchyma, deformity of the chiasm, etc. consitent with regressive changes, usually stable.

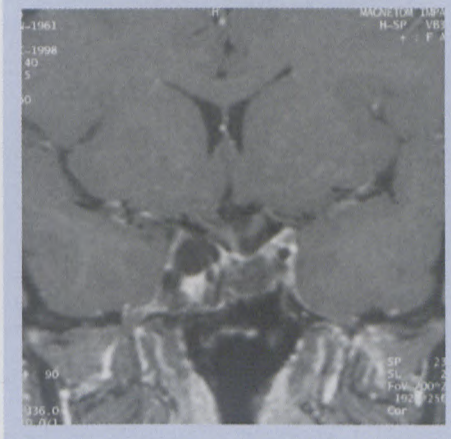


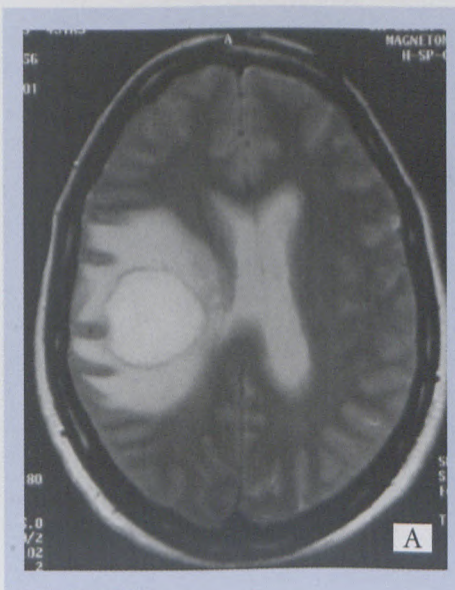
Fig. 9.

Anatomic diagnosis: Sella/suprasellar region: complex abnormality/cystic degeneration/regression/post-treatment status unchanged/macroprolactinoma.

Note: some tissue-remnant enhancement is common, but endocrinologists want to know if the changes are stable. Adding macroprolactinoma will allow search for related terms.

Fig. 10. Infant with severe psychomotor retardation and seizures. Regressive changes comprise bilateral, symmetrical volume loss of the occipitotemporal lobes, with predominant involvement of white matter including diffuse T2 hyperintense signal alteration and cavitation; cortex also atrophic. Topography typical of hypoxic injury.

Anatomic diagnosis: Occipital/temporal lobes: encephalopathy/ multicystic encephalomalacia (Barkovich), assumed perinatal hypotensive hypoxema/infant/cerebral palsy.



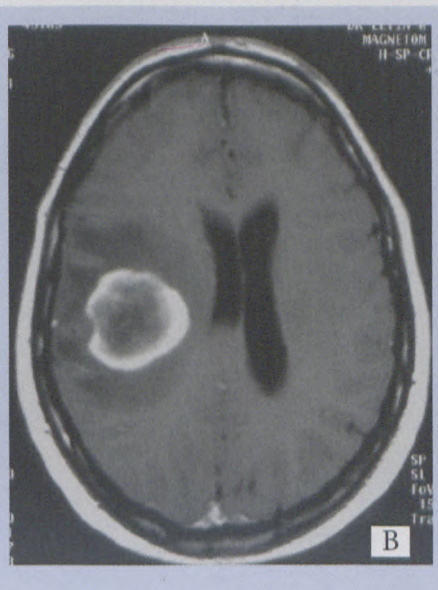


Fig. 8a and 8b.

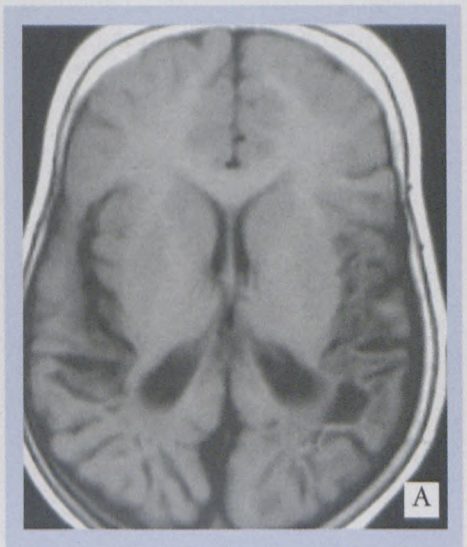


Fig. 10a

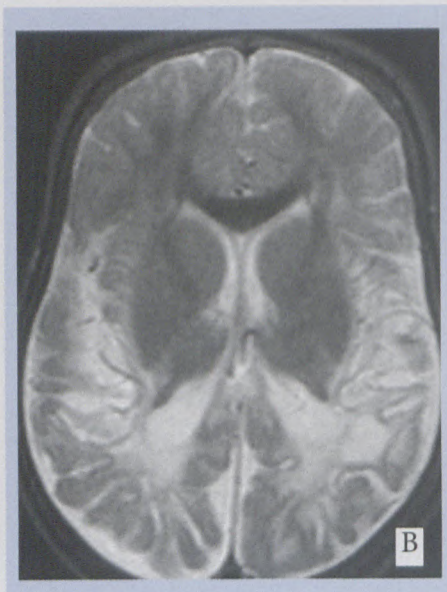


Fig. 10b.

The demise of SNOMED

Key factors contributing to the progressive obsolescence of SNOMED include the adoption of ICD (International Classification of Diseases) by most hospital administrators, and the word-searching power of modern computers. ICD has directory-type access, which appeals to busy individuals confronted with the need to supply key words on forms, but even a cursory examination of some of the ICD hospital diagnoses shows how senseless these often are, and more worryingly, how these diagnoses become hard national statistics. Anatomical pathologists, on the other hand, continue to use SNOMED because of their concern with causal relationships which only a systematised nomenclature can provide, and because of the accuracy which can only be achieved with coding. However, modern search engines, using logical relationships, now make word searches fast and reliable and for practical purposes, departments and practices can operate satisfactorily with the commercial programmes (e.g. Microsoft Jet).

Does there need to be a systematic approach to imaging diagnosis in South Africa?

The lack of a uniform, systematised approach to neuroradiologic diagnosis in South Africa does not seem to worry the fraternity. At the two teaching hospitals in Cape Town, no reporting method in neuroimaging is taught, and this is reflected in the very wide range of reporting styles evident in private practice, where very often the clinical details are omitted, the findings and comment are frequently repetitive, and a final diagnosis is rarely given. Of the many factors operating against systematisation, the most intrinsic have to be deficient knowledge of pathology, and the anti-database mindset. Yet, every radiology department has a museum, and every private practice imaging unit has a film collection, so that defaulting on the database (in the age of IT) is difficult to understand. Apart from the satisfaction of being able to access material quickly and efficiently, there could be important benefits to health care in South Africa if a national imaging database existed, including identification of disease and management trends, optimising studies in relation to clinical categories, and allowing inter-practice consultation. But the anatomic diagnosis would still have be the bottom line, and behind it, a systematic approach to pathogenetic inference.

Reference

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