

Viewpoint

Diagnostic categories in rheumatology

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There is an increasing trend for the development of rheumatological 'diagnostic criteria'. This trend is based on the view that we can get progressively closer to ideal patient classification by refining such criteria. I would like to argue that this approach is futile and must be replaced by a method which allows efficient data handling, both for practical clinical management and for scientific inquiry.

There are two ways to define patient groups. Firstly, patients with the same symptom, sign, or laboratory finding can be grouped together without reference to a causal process. Hypertension and impaired glucose tolerance are examples. These groups are useful because treatment is often aimed at the end organ problem. They are also useful for scientific study because they are defined by necessary criteria and thus represent raw data.

Patients may also be grouped using criteria which are neither necessary nor sufficient but which are considered good indicators of a common causal agent or process which cannot be observed directly. The clinical diagnosis of tuberculosis or left heart failure depends on this sort of grouping. Patients with such problems present with widely varying symptoms and signs. It is inappropriate to group on the basis of a rigid set of clinical features. Individual clinical features may be present or absent for reasons which are irrelevant to the disease concept. The essence of this sort of grouping is that there is general agreement about the nature of the common feature which gives rise to a spectrum of clinical illness, be it a tubercle bacillus or pump failure.

In rheumatology we argue about how to group patients because we are using the wrong method of grouping. We use the second type of grouping, with criteria which are neither necessary nor sufficient.

There is, however, no agreed concept of the common cause or process for all patients with one 'rheumatic disease'. In most cases we simply assume that there is a common feature which we have yet to define precisely. Unfortunately, without defining the common feature we have no way of deciding how to devise criteria to recognise it.

The belief that each 'rheumatic disease' has a common causal feature is based largely on clustering of clinical features into patterns, but these clustered associations overlap. The categories we use are partly accidents of history. Reiter's syndrome covers an ill defined group of patients with synovitis, some of whom have urethritis. Some of these patients have a history of diarrhoea, sacroiliitis, nail dystrophy, or keratoderma and will fit equally well into other subcategories of seronegative arthritis.

Recent developments in immunogenetics, presented at the British Society for Rheumatology spring meeting, suggest that the causal factors contributing to rheumatic disease cut across diagnostic groups and interact on a multifactorial basis.^{1,2} HLA-B27 is a risk factor for spondylitis, whether on a background of psoriasis or inflammatory bowel disease. Dr4 is a risk factor for peripheral synovitis, whether associated with rheumatoid factor or not. There is probably no one common factor in subjects with spondylitis which is not shared with other patterns of disease. Some will share a factor with subjects with psoriasis, some will share a factor with subjects with iritis (e.g., B27), and so on. Patients with sacroiliitis and B27 share a common disease process. Patients with ankylosing spondylitis do not share a single disease process but rather a cluster of processes due to various combinations of synergising causal agents. Similarly, rheumatoid arthritis does not correspond to a single process but to a cluster of processes. The causal factors which give rise to spondylitis do not appear to overlap with those which give rise to rheumatoid arthritis. It is

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conceivable, however, that some cases of peripheral synovitis share causal elements with both a proportion of patients with ankylosing spondylitis and a proportion with rheumatoid arthritis.

If the theoretical reasons for using terms such as rheumatoid arthritis are dubious, continued usage can only be justified if it is useful. Diagnostic labels must provide a better basis for predicting outcome and response to therapy than the raw observations from which these labels are derived. This is quite clearly not so. Little useful distinction can be made in terms of prognosis or response to therapy between a patient described simply as having rheumatoid arthritis and one described as having psoriatic arthropathy. On the other hand, a very useful distinction can be made between a patient with three years' pain and swelling of all peripheral joints and a patient with three weeks' swelling of one knee followed by three years of remission. The raw observations turn out to be much more useful than the diagnostic groups.

The idea of abandoning current diagnostic groups may seem alarming, but my experience is that it makes things much easier and encourages logical clinical practice. Different ways of grouping patients are appropriate to different problems, whether of management or inquiry into pathogenesis. It is important to avoid the Procrustean temptation to unify criteria for the sake of tidiness. Problem oriented grouping is common practice in other branches of medicine. Sometimes it is useful to consider all cases of hypothyroidism and sometimes it is useful to consider thyroid disease of autoimmune origin. Similarly, subacute oligoarthritis involving the knee is a useful grouping for practical purposes and reactive arthritis is a useful theoretical category. They are not rival 'diagnoses'.

Most rheumatologists appear to have an unexpressed concept of chronic autonomous synovitis, in the sense of synovitis which is out of proportion to any recognisable stimuli other than those generated by the inflammatory cells themselves. When such synovitis is widespread and destructive it tends to be treated (however reluctantly) with gold or penicillamine regardless of whether there is a positive Rose-Waaler test, antinuclear factor, or coexistent psoriasis. Because drug trials are hidebound by the criteria for rheumatoid arthritis we do not know whether or not this is justified. Sulphasalazine trials may be the first to address this problem sensibly.

A grouping based on the idea of chronic autonomous synovitis can legitimately be defined by criteria which are not necessary or sufficient. Pain, swelling, stiffness, raised erythrocyte sedimentation rate, positive Rose-Waaler test, absence of crystals in synovial fluid or calcinosis on *x* ray, and many

other things can all be used as criteria as long as it is possible to establish that these are sensitive or specific indicators of the concept we want to define.

One of the implications of the multifactorial origins of connective tissue disease is that patients must be grouped using a series of freely sortable features. Whereas infectious diseases resemble species (because they are due to species of micro-organism) rheumatic diseases resemble strains of pedigree cat, because the underlying risk factors can be interbred. Pedigree cats are described by a set of independent features, as in British short haired silver tabby (representing four separate groups of genes).

It is nice to be able to describe illness in terms of causal processes. The best we can do at present is 'peripheral synovitis associated with HLA-B27 and salmonella infection', or 'cartilage fibrillation secondary to meniscectomy'. More often we need to describe patients in terms of features which are either reproducibly observable or which we think reflect an unknown causal factor. Thus a patient may have 'juvenile onset synovitis with rheumatoid nodules, positive antinuclear factor, and negative rheumatoid factor'. This real but unusual case can then be described accurately and each factor of interest studied independently. Similarly, 'peripheral synovitis with urethritis and mouth ulcers' is more precise than 'Reiter's syndrome'. 'Psoriatic arthropathy' conveys less than psoriasis with spondylitis or psoriasis with distal interphalangeal (DIP) arthritis. Instead of saying psoriatic arthropathy without psoriasis we should agree what is special about the arthropathy or spondylitis seen in association with psoriasis and describe it as such (DIP arthropathy, focal pelvospondylitis, or arthritis mutilans).

One advantage of a classification based on independent variables is that scientific study can be based on precise data arranged in such a way as to maximise the chance of fruitful statistical analysis. With diagnostic categories the alternatives are a few big groups defined by unsatisfactory, imprecise criteria or a large number of subgroups containing very small numbers. If patients are classified by a range of independent variables then there are a large number of ways of dividing the patients into two groups each containing enough numbers for statistical analysis. If it is felt that urethritis is the key to some causal pathway, all patients with urethritis can be compared with those without. New genetic studies, however, might suggest that nail dystrophy was a more sensitive indicator of the pathway, or that both problems reflect a pathway not shared by iritis. If patients are precategorised into groups such as Reiter's syndrome, such studies

cannot be done without rescreening a large number of cases.

The other advantage of this stratified classification is that it allows more precise clinical practice since prognosis and treatment depend on the individual aspects of the patient's problem more than they do on the diagnostic group.

The chief criticism of such a classification is its apparent complexity. In fact it is no more complex than the present system. The same data are just being sorted at a different stage in relation to storage. The complexity of the initial classification is outweighed by easier analysis. The only real problem is one of cross referencing, but now that computers are available at a realistic price this problem has disappeared. As long as the clinician

records his findings in an organised fashion machines will do the rest.

In summary, it is suggested that current 'diagnostic' groupings should be abandoned in favour of description using a set of independent features either defined by necessary criteria or based on an agreed pathophysiological concept. Little, if any, new terminology is needed, we merely need to define currently used terms more clearly.

References

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- 2 Grennan D M. Family studies and genetics in rheumatoid disease research [Abstract]. *Br J Rheumatol* (in press). (Spring meeting abstracts supplement.)