

REFERRAL AND DIAGNOSIS OF COMMON RHEUMATIC DISEASES BY PRIMARY CARE PHYSICIANS

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SUMMARY

Objective. To describe primary care patterns of referral and diagnoses of patients with rheumatic diseases referred to rheumatologists.

Methods. The medical records of all consecutive patients referred in 1994 by >300 primary care physicians to two rheumatologists at an academic centre were reviewed. The referring physician diagnosis was compared with the rheumatologist's diagnosis. Sensitivity, specificity and predictive values of primary care diagnoses were estimated using the rheumatologist diagnosis as the 'gold standard'.

Setting. University-based rheumatology out-patient clinic.

Results. Over half of the patients referred had a rheumatologist diagnosis of soft-tissue rheumatism or a spinal pain syndrome. Three hundred and forty-seven patients (49%) had a primary care diagnosis of a defined rheumatic disease. Of these, 142 (41%) of the primary care diagnoses were subsequently modified by the rheumatologist. The highest agreement between primary care physician and rheumatologist was observed for crystal-induced arthritis ($\kappa = 0.86$), and the lowest agreement for polymyalgia rheumatica ($\kappa = 0.39$) and systemic lupus ($\kappa = 0.46$). Sensitivity was lowest for a primary care diagnosis of fibromyalgia (48%) and highest for ankylosing spondylitis (94%). Positive predictive values were generally low, in particular for systemic lupus erythematosus (33%) and polymyalgia rheumatica (30%).

Conclusion. Most patients referred to an academic rheumatology centre had soft-tissue rheumatism or other pain syndromes. In general, diagnostic agreement between rheumatologists and primary care physicians was low. Increased emphasis on musculoskeletal disorders should be encouraged in medical education to increase the efficiency of rheumatology referrals.

KEY WORDS: Referral, Diagnosis, Rheumatic diseases, Primary care physicians.

RHEUMATIC diseases comprise >100 different entities with varying clinical characteristics, prognosis and therapy requirements. It has been estimated that rheumatic diseases affect ~10% of the population [1]. Most frequently, the initial contact of a rheumatic patient is with a primary care physician, and >10% of visits to primary care physicians are related to rheumatic diseases [2]. An early diagnosis can facilitate the choice of adequate therapies and rationalize referrals to specialists. Inappropriate diagnosis can result in delays in treatment, inadequate prescription of therapies, 'labelling' of patients with false-positive diagnoses, and inefficient use of resources (e.g. additional testing or unnecessary referrals). The objective of this study was to describe the patterns of primary care referrals to rheumatologists and to evaluate the accuracy in the referral diagnoses of common rheumatic diseases.

METHODS

Seven hundred and eleven consecutive new patients were referred in 1994 by 305 primary care physicians to two rheumatologists at the University of Alberta Hospitals in Edmonton, Alberta, Canada. In 1994, 10 rheumatologists were practising in Edmonton (four at the University of Alberta). Approximately 2500 prim-

ary care and non-specialist physicians were registered with the College of Physicians and Surgeons of Alberta, about half of them in Northern Alberta. The medical records of these patients were reviewed to determine the referring diagnosis by the primary care physician and the rheumatologist, diagnosis both at the initial consultation and during follow-up. Diagnoses were grouped in broad categories including: (a) systemic lupus erythematosus (SLE); (b) rheumatoid arthritis (RA) and related arthritis (juvenile chronic arthritis, palindromic rheumatism); (c) spondyloarthropathies; (d) polymyalgia rheumatica; (e) other connective tissue diseases; (f) localized soft-tissue rheumatism; (g) fibromyalgia; (h) entrapment neuropathies; (i) low back pain and/or cervical pain; (j) osteoarthritis and other localized osteoarticular syndromes; (k) crystal-induced arthritis; (l) miscellaneous (other rheumatic diseases, diseases primarily from other systems). Patients with other miscellaneous diseases were excluded from the analysis since these diagnoses included uncommon or non-musculoskeletal diseases.

The degree of agreement between the rheumatologist and the referring primary care physician for each diagnosis was evaluated using the kappa statistic (a kappa coefficient ≥ 0.7 is considered to indicate substantial agreement). Only patients in whom a diagnosis of musculoskeletal or rheumatic disease had been stated by the primary care physician in the referral were included for this objective. Non-specific symptoms such as arthralgias or myalgias were not considered to be disease diagnoses or defined syndromes, and

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these patients were not included in this analysis. The final rheumatologist diagnosis was considered as the 'gold standard'. Prevalence of a disease was defined as the proportion of individuals who had a rheumatologist diagnosis of a specific disease divided by the total sample included in the analysis. The performance of primary care physicians' diagnoses was evaluated with a Bayesian approach, estimating sensitivity, specificity, and positive and negative predictive values using the rheumatologist diagnosis as the 'gold standard'. In this study, sensitivity can be defined as the probability of the primary care physician detecting a specific rheumatic disease. Specificity can be defined as the probability of the primary care physician excluding a specific rheumatic disease. Positive predictive value is defined as the probability of having a specific rheumatic disease (diagnosed by a rheumatologist) when diagnosed by the primary care physician.

Chi square with Yates correction was used to analyse differences in proportions and two-tailed Student's *t*-tests to evaluate differences in means. The relationship between agreeing diagnosis and primary care physician characteristics was evaluated for physician's age, gender and years since graduation; we also assessed location of practice (urban vs rural) and population size and distance from Edmonton (rheumatologists' practice site).

RESULTS

Table I shows the characteristics of patients included in the study. The most frequent diagnoses by the rheumatologists included localized soft-tissue rheumatism (23%), spinal pain syndromes (16%), fibromyalgia (15%) and osteoarthritis (14%). Fifteen per cent of the referred patients had a connective tissue disease.

TABLE I
Patient characteristics (*n* = 711)

Gender, females	435 (61%)
Mean age (yr, mean \pm s.d.)	49 \pm 16
Patients seen by	
(i) Rheumatologist 1	331 (47%)
(ii) Rheumatologist 2	380 (53%)
Number of consultations by rheumatologist (mean \pm s.d.)	1.5 \pm 1.3
Patients seen only once by rheumatologist	517 (73%)
Rheumatologist diagnosis	
Rheumatoid and related arthritis	45 (6%)
Systemic lupus erythematosus	8 (1%)
Spondyloarthropathies	26 (4%)
Other connective tissue diseases	31 (4%)
Fibromyalgia	109 (15%)
Spinal pain syndromes	112 (16%)
Osteoarthritis	102 (14%)
Localized osteoarticular rheumatism	41 (6%)
Other soft-tissue rheumatism	166 (23%)
Crystal arthropathies	24 (3%)
Other rheumatic diseases	18 (3%)
Diseases primarily from other systems	28 (4%)
Syndromes not yet diagnosed	28 (4%)
No disease	5 (1%)
Total no. of diagnoses	743*
Total no. of patients	711

*32 patients had two diagnoses.

Of the 711 patients referred, 347 (49%) had a diagnosis of a musculoskeletal disease stated by the primary care physician in the referral documentation. In 245 (34%), the reason for referral was not specified. In 113 (16%), the referral diagnosis was non-specific, including only symptoms such as arthralgias or myalgias. Table II shows the agreement between the primary care physicians' and rheumatologists' diagnoses for the 347 patients. Substantial agreement was only observed for crystal-induced arthritis (κ = 0.86). The lowest agreement was observed for SLE and polymyalgia rheumatica (κ = 0.46 and 0.39, respectively).

Table III shows the sensitivity, specificity, and positive and negative predictive values of primary care diagnoses for specific diseases using the rheumatologists' diagnoses as the 'gold standard'. The lowest sensitivity was observed for fibromyalgia (48%) and entrapment neuropathy (50%), and the highest for ankylosing spondylitis (94%) and crystal-induced arthropathies (81%). Specificity was high for all the diseases, but this was expected given the large agreement in negative diagnoses, which included all the patients without the diagnosis of interest (with other diseases). The positive predictive value was high for crystal-induced arthropathies (93%), but low for SLE (33%), polymyalgia rheumatica (30%), RA and other connective tissue diseases (46% for each one).

Figure 1 shows the true-positive, false-positive and false-negative diagnoses by primary care physicians. The total number of patients for each bar (*n*) is the sum of all patients in whom the diagnosis of a particular disease was made, either by the primary care physician (true positive or false positive) or the rheumatologist (true positive or false negative). The purpose of the figure is to compare the proportion of false-positive and false-negative diagnoses. False-positive diagnoses were observed frequently for connective

TABLE II
Agreement between primary care physicians and rheumatologists in patients with specified diagnoses (*n* = 347)

Diagnosis	Diagnosis by primary care physicians (%)	Diagnosis by rheumatologists (%)	κ
Low back pain and/or cervical pain	65 (19)	64 (18)	0.60
Osteoarthritis	51 (15)	39 (11)	0.51
Localized soft-tissue rheumatism	48 (14)	62 (18)	0.63
Rheumatoid arthritis	46 (13)	27 (8)	0.53
Fibromyalgia	43 (12)	60 (17)	0.49
Ankylosing spondylitis	30 (9)	16 (5)	0.63
Other connective tissue diseases	22 (6)	16 (5)	0.50
Crystal-induced arthritis	14 (4)	16 (5)	0.86
Systemic lupus erythematosus	12 (3)	5 (1)	0.46
Polymyalgia rheumatica	10 (3)	5 (1)	0.39
Entrapment neuropathies	6 (2)	6 (2)	0.49
Other	—	16 (5)	
No rheumatic disease	—	15 (4)	
Total	347	347	

TABLE III
Sensitivity, specificity and predictive values of primary care diagnoses using the rheumatologist final diagnosis as the 'gold standard'

Diagnosis	n	Sensitivity (%)	Specificity (%)	+ PV (%)	- PV (%)	Prevalence (%)
Local soft-tissue rheumatism	62	61	96	79	92	17.8
Fibromyalgia	60	48	95	67	90	17.2
Entrapment neuropathy	6	50	99	50	99	1.7
Low back pain/cervical pain	64	67	92	66	93	18.4
Osteoarthritis	39	67	92	51	96	11.2
Crystal-induced arthritis	16	81	99	93	99	4.6
Systemic lupus erythematosus	5	80	98	33	99	1.4
Rheumatoid arthritis	27	78	92	46	98	7.7
Ankylosing spondylitis	16	94	96	50	99	4.6
Polymyalgia rheumatica	5	60	98	30	99	1.4
Other connective tissue diseases	16	63	96	46	98	4.6

n, total number of diagnoses by rheumatologists; +PV, positive predictive value; -PV, negative predictive value.

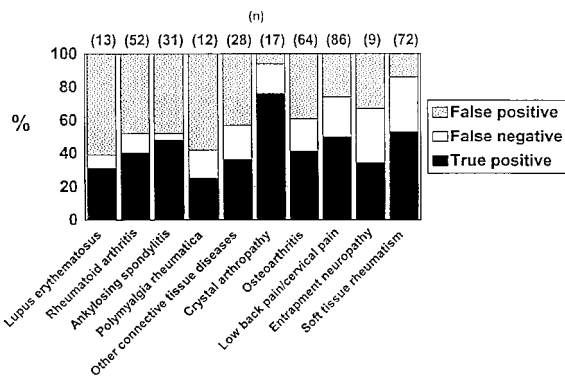


FIG. 1.—True-positive, false-positive and false-negative diagnoses by primary care physicians; (n) represents the sum of all patients in whom a diagnosis of a specific disease was made (by the primary care physician or the rheumatologist).

tissue diseases. False-negative diagnoses were observed most often for soft-tissue rheumatism syndromes.

Overall, 142 (41%) of the referral diagnoses made by a primary care physician were subsequently modified by the rheumatologist. Table IV shows the final rheumatologist diagnosis in those patients with a false-positive diagnosis by the primary care physician. RA

was erroneously diagnosed in 25 out of 46 patients (54%): of these, seven had osteoarthritis and five fibromyalgia. Fifteen of 30 patients (50%) were erroneously diagnosed as having ankylosing spondylitis; seven of these were considered to have non-inflammatory low back pain. Fibromyalgia was often missed in patients diagnosed with low back pain.

No statistical associations were observed between age, gender, years since graduation, practice location of the primary care physician, and agreement with the rheumatologist.

DISCUSSION

The purpose of this study was to examine the patterns of referral and the accuracy in the diagnoses made by primary care physicians when referring patients to a tertiary rheumatology centre. In 1994, 305 different primary care physicians referred 711 new patients to two rheumatologists at the University Hospital in Edmonton. Most of the referred patients had soft-tissue rheumatism or osteoarthritis. Sixty-six per cent of referrals included clinical descriptions or diagnoses in the referral documentation, but only 347 (49%) patients had a definite diagnosis of a rheumatic disease. Others have also observed a reluctance to

TABLE IV
Primary care diagnoses modified by the rheumatologist

Primary care diagnosis	Total	Modified by rheumatologist	True disease (rheumatologist diagnosis)									
			LSTR	FM	LBP	SLE	RA	AS	PR	Other CTD	Misc	Other
			n	n	n	n	n	n	n	n	n	n
Localized soft-tissue rheumatism	48	10 (21%)	—	1	0	0	1	0	1	0	1	6
Fibromyalgia	43	14 (33%)	3	—	5	0	0	0	0	0	2	4
Low back/cervical pain	65	22 (34%)	3	11	—	0	2	0	0	0	3	3
Systemic lupus erythematosus	12	8 (67%)	1	2	0	—	0	0	0	3	0	2
Rheumatoid arthritis	46	25 (54%)	2	5	2	0	—	1	1	2	7	5
Ankylosing spondylitis	30	15 (50%)	3	1	7	0	2	—	0	0	1	1
Polymyalgia rheumatica	10	7 (70%)	2	1	0	0	0	0	—	1	2	1
Other connective tissue diseases	22	12 (55%)	2	4	0	1	0	0	0	—	1	4
Other	71	29 (41%)	8	6	7	0	1	0	0	0	2	5
Total	347	142 (41%)	24	31	21	1	6	1	2	6	19	31

LSTR = localized soft-tissue rheumatism. SLE = systemic lupus erythematosus. Other CTD = other connective tissue diseases. Misc = miscellaneous: crystal arthropathy, osteoarthritis, neuropathy. Other = non-rheumatic diseases, no disease.

include a tentative diagnosis in patients referred by primary care physicians in as many as 80% of the referrals [2–4].

We selected the rheumatologist diagnosis as the 'gold standard' for the diagnosis of rheumatic disease. A concern with our choice of 'gold standard' is that the final rheumatologist diagnosis may have been influenced by the original diagnosis of the primary care physician since the rheumatologists were not blinded to the primary care referrals [5]. In this case, however, if bias occurred, a greater number of inaccurate diagnoses would be expected to occur. Overall, in 41% of the patients, primary care physicians and rheumatologists disagreed on the diagnosis. Rheumatologists used a longer duration of disease to their advantage, and many diseases have an evolving course with worsening or improvement of signs and symptoms over time. Nevertheless, disagreement was common for diseases which normally have clear and specific signs and symptoms at onset (Table II).

The highest diagnostic sensitivity was observed for ankylosing spondylitis (94%) and crystal-induced arthropathies (81%). Low sensitivity was observed for fibromyalgia (48%) and entrapment neuropathy (50%). Specificity was high for all the diseases. This has also been observed by others [4, 5], and is partly due to the way specificity is calculated by 'lumping' all of the other diagnoses together as 'true negatives' for a particular disease, even if they are inaccurate for other diseases. It is obviously desirable to have a diagnosis of rheumatic disease by primary care physicians which is both highly sensitive and specific; nevertheless, for diseases with high morbidity and mortality, such as connective tissue diseases, the most desirable of these two attributes is high sensitivity to diagnose and treat potentially serious diseases promptly. Positive predictive values were low, indicating a tendency to over-diagnose some diseases (false positives), particularly connective tissue diseases. Positive predictive values are important diagnostic attributes from a clinical and economic perspective because they reflect the actual diagnoses (true or false) at the primary care level, which can result in additional testing, referrals or treatment.

Local factors may influence referral of patients and it may be difficult to extrapolate these results to other centres. Nevertheless, a few other studies have also examined diagnostic patterns of rheumatic diseases in primary care. Two Swedish studies published in 1981 and 1983 [2, 3] showed wide variation in the inclusion of tentative diagnoses by primary care physicians: from 20 to 90%. Both studies reported low agreement between rheumatologists and primary care physicians. A Spanish study in 422 patients reported that >50% of primary care diagnoses were modified at the rheumatology clinic [4]. A Canadian study [6] in 149 patients described low sensitivity for many diagnoses. These two last studies found, like ours, a reluctance to establish tentative diagnoses. Another common finding was the frequent confusion of connective tissue diseases with other musculoskeletal disorders, including some

with very different clinical characteristics. We also observed low agreement for connective tissue diseases, with low positive predictive values. It is clear from these studies that primary care physicians in various countries continue to show difficulties in the diagnosis of musculoskeletal disorders. Primary care physicians generally are the initial health care contact for patients with rheumatic diseases. A number of health-related decisions and interventions can arise from their diagnoses: referrals, diagnostic tests, therapies and patient counselling. The economic burden of musculoskeletal disorders is high, among the five most costly groups of diseases in Canada [7]. Costs of inadequate diagnoses and treatment can contribute to inappropriate and inefficient resource utilization. In addition, a delay in the diagnosis may result in a lower response to therapy, disease progression and disability.

At the time of the study, 10 rheumatologists were practising in Edmonton; there were no other rheumatologists in Northern Alberta. Approximately 2500 primary care and non-specialist physicians were registered with the College of Physicians and Surgeons of Alberta, about half of them in Northern Alberta. The 300 different primary care physicians who referred the patients in this study represent ~20–25% of the Northern Alberta primary care physicians, which supports the generalizability of our findings. We realize that patients referred to rheumatologists probably represent the more severe and difficult cases, and that the diagnosis stated by the referring physician may not be definite, and perhaps reflects diagnostic uncertainty more than diagnostic error. However, it is clear that in many cases where primary care physicians suspected connective tissue diseases, the final diagnosis by the rheumatologist was soft-tissue rheumatism, which in most cases should be easily diagnosed by primary care physicians following a thorough history and physical examination. Training of medical students, residents and primary care physicians in musculoskeletal diseases is perceived to be insufficient [8, 9]. The American College of Rheumatology has developed guidelines for the initial evaluation of musculoskeletal diseases [10], but these guidelines need to be tested in clinical settings [11]. The low rate of tentative diagnoses by primary care physicians may relate to low levels of confidence to establish a diagnosis. A survey by Glazier *et al.* [12] in Ontario showed that primary care physicians are less confident in performing a musculoskeletal examination than a cardiovascular examination. This low level of confidence could also potentially impact the initiation of critical interventions such as disease-modifying anti-rheumatic drugs for RA. A previous survey has shown that primary care physicians may be reluctant to prescribe these drugs, despite being aware of studies documenting their potential benefits [13]. Our study did not, however, allow us to evaluate whether changes in therapy occurred as a consequence of changes in diagnosis since we had no access to primary care medical records.

We have shown that primary care physicians misdiagnose rheumatic diseases (false-positive and false-

negative diagnoses), in particular connective tissue diseases. Major concerns from false-positive diagnosis of these diseases could include patient 'labelling' and distress, excessive and inefficient use of diagnostic tests and referrals, and inappropriate therapies with potential for toxicity. False-negative diagnoses could also result in significant therapeutic delays.

Primary care physicians deal with a very large number of patients and conditions, and cannot be expected always to be accurate in their diagnoses. Nevertheless, most primary care referrals to rheumatologists included patients with a final diagnosis of soft-tissue rheumatism or other musculoskeletal pain syndromes, and they often suspected connective tissue diseases in these patients, resulting in a high prevalence of false-positive suspected diagnoses. Rheumatic diseases are among the most common reasons for visits to primary care physicians. Since inaccurate diagnoses in referred patients appear to be frequent, an increased emphasis in medical education on the assessment of musculoskeletal disorders is suggested to improve the efficiency of referrals to rheumatologists.

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