

## Two-step Diagnostic Strategy in Rheumatology

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### Abstract

**Introduction:** Reaching a specific diagnosis in any field of medical science is a learning process and diagnosis in rheumatology may have a steep learning curve. An easy solution for arriving at diagnosis in rheumatology is recommended to adopt two-step diagnosis.

**Discussion:** The first step based on symptoms and their duration defines the generic type of pathology in question and basic symptoms are related to anatomic compartment as source of symptom. After this first step, final diagnosis is not too far away. At the end of this first step, patient's condition is described as a rule, needing not more than ten words.

In the second step, detailed investigation to distinguish between conditions causing these symptoms is proceeded and explored leading to a differential diagnosis map, which is based on available rheumatological knowledge. The way it is structured should be on line of reasoning.

Structuring of clinical reasoning process necessarily improves with experience but rheumatic diseases are known to defy any rigid syndromic classification and systemic lupus erythematosus is a classical example presenting with predominant articular symptoms or a variety of extra articular symptoms.

**Conclusion:** Diagnosis of the syndrome and disease itself always requires a thorough well-planned questioning. It is not possible without a careful general examination of all the areas with presenting symptoms and thorough investigations.

### Introduction

How to reach a specific diagnosis in a particular rheumatology patient is a tangible problem warranting an easy solution. This specific diagnosis can be reached via main rheumatological syndromes. It is recommended to adopt "Two-step diagnosis" enroute to diagnostic strategy.

### Priorities

1. Learn how to ask questions
2. Course of questioning
3. Let it not be a random search making it ineffective in reaching a diagnosis

### First Step

First step is to define generic type of pathology in question. It is therefore suggested that pattern of symptoms is to be considered.

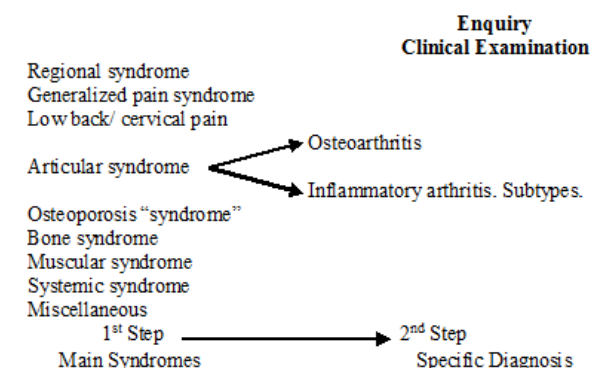
Initial interview and observation should be designed to establish the type of problem being dealt with, to which the case or complaint belongs. Existence of typical features of a given syndrome must be determined and distinguished from others. It is imperative not to delve into details that have no impact on these objectives. It is essential to be as precise as possible in details that are most decisive in identifying or excluding a particular syndrome.

In rheumatology, basic symptoms are related to anatomic compartment as source of symptom: articular/ periarticular/ muscular/ systemic etc.

### Goal

First goal is to get to the right neighborhood, where we won't be too far from our final diagnosis.

In order to know what is being looked for, in the first step of differential diagnosis one must be familiar with main syndromes. (Fig. 1)



**Fig. 1: Differential diagnosis in the rheumatology**  
**First step: main syndromes**

### Caution

- Can get the syndrome – wrong
- Do not misinterpret / assume about uncertain sign
- Diagnosis of rheumatoid arthritis – flawed - assume muscular origin
- Rheumatology – symptoms / signs – subjective & imprecise
  - Usually assess in terms of probability than certainty
  - Avoid making mistakes – must take critical View of leading evidence in one / another direction
- Oedema of foot with local pain
  - Arthritis / venous malformation + local pathology
- Differentiate absolutely certain signs / still doubtful

### Our reasoning based on key words:

Arthritis / Arthralgia – implications of the word  
Minimum 2 signs amongst “pain, warmth, swelling, redness, limitation of function” qualify for the condition to be called arthritis, whereas pain alone will be called as arthralgia

### Magic of Ten Words

After thorough interrogation and clinical examinations describe your patients with as few, accurate keywords as possible-as a rule will not need more than ten words

- Young man with inflammatory knee pain – no systemic signs

- Child with symmetrical, knee swelling and additive, chronic, predominating peripheral poly-arthritis with no axial involvements

- Generalized pain with no objective finding

### Second Step in achieving diagnosis

After arriving at a particular set of symptoms – joint disease / generalized pain / bone pain / back pain - proceed to detailed investigation to distinguish between conditions that can cause these symptoms- this is the second step in differential diagnosis.

Exploration of the symptoms should concentrate on:-

(i) Which features are unique to the condition

(ii) Features which distinguish it from alternatives

For example rheumatoid arthritis and psoriatic arthritis may involve the elbow. Conversely inflammatory involvement of the lumbar spine or the distal interphalangeal joint is common in psoriatic but

not in rheumatoid arthritis. Thus, differentiating between these two conditions existence of spondylitis and the involvement of the distal interphalangeals enable to differentiate whereas the elbow involvement becomes immaterial but for the final management.

It is in this second step of differential diagnosis that investigations play more important role.

A general clinical examination and a complete regional examination of all the affected areas are essential to final diagnosis. Accurate diagnosis always evolves after combination of the questioning outcome and the findings of the regional examination.

Structuring of clinical reasoning process is necessarily simplistic and with experience, this diagnostic strategy can improve.

Rheumatic diseases defy any rigid syndromic classification and presentation of many rheumatic conditions can take many forms with symptom pointing more strongly to one or other syndrome. This concept of clinical spectrum is well illustrated by the disease systemic lupus erythematosus. This can present with predominant articular symptoms or a variety of extra articular symptoms (systemic syndrome). Similarly polymyositis can present with weakness (muscular syndrome) or with arthritis (articular syndrome) or photosensitivity (systemic syndrome).

A differential diagnosis map based on our knowledge and reasoning as per following table's leads to reach a diagnosis:

**Table A. What suggests periarticular pain?**

Local or regional distribution
Most frequent sites: shoulder and elbow
Selectivity of painful movements
Active mobilization much more painful than passive
No range limitations in passive mobilization
Palpation of the structure is painful
Specific distention or resisted movements painful

**Table B. What suggests neurogenic pain?**

Distribution in a dermatome or peripheral nerve territory
Dysesthetic nature of pain
Most common sites: sciatica, carpal tunnel syndrome, ulnar syndrome
Normal local osteoarticular examination
Local alterations in the neurological examination (late onset)
Exacerbation with the Valsava maneuver (in radiculopathies)
Exacerbation with mobilization of the spine (in radiculopathies)
Tinel's sign (in peripheral nerve entrapment)

**Table C. What suggests referred pain?**

Local or regional distribution
Uncharacteristic rhythm
Dysesthetic nature (neurogenic pain)
Associated symptoms (neighboring joints, viscera, neurological changes)
Normal local examination

**Table D. What suggests monoarthropathy?**

Local distribution of pain Typical pattern: inflammatory or mechanical Pain with all movements of the joint Crepitus, swelling, effusion or local heat or redness Pain on palpation along the joint margins Limited active and passive mobility Specific maneuvers for periarticular lesions: negative
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**Table E. What suggests generalized pain syndrome?**

Pain "all over" Diffuse distribution with little focus in the joints Migratory nature Worse after exercise Distribution inconsistent with polyarthropathy Dramatic description Clinical examination with no objective alterations Laboratory tests and imaging normal
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**Table F. Mechanical and inflammatory rhythms of pain**

<b>Inflammatory pattern</b>	<b>Mechanical pattern</b>
Worse in the morning	Worse in the evening
Improves with movement	Worsens with movement
Pain at rest, no pain free position	Eases at rest, pain free positions
Prolonged morning stiffness (>30 minutes)	Short morning stiffness (<10 minutes)
Stiffness after rest > 5 minutes	Stiffness after rest < 2-3 minutes

**Table G. Main features of inflammatory arthritis and osteoarthritis, on clinical examination**

<b>Arthritis</b>	<b>Osteoarthritis</b>
Firm, rubbery swelling	Stony swelling
Spindle shaped swelling	Irregular, nodular swelling
Pain along the joint margins	Focal pain along the joint margin
No crepitus (or fine crepitus)	Rough crepitus
Signs of inflammation	No signs of inflammation*
Extra-articular signs are common	No related systemic signs
Any joint	Predominantly in weight-bearing joints & hands

\*Osteoarthritis may be associated with inflammatory signs during acute flares. Longstanding inflammatory arthritis may result in secondary osteoarthritis: thus features from both conditions may coexist.

**H. When should we think of osteoporosis?**

<b>Risk factors</b>	<b>Manifestations</b>
Post-menopausal women Early menopause Late menarche Low weight & height	• Whenever a patient of any age presents a history of low-impact fractures i.e. fractures caused by falls of less than his/ her own height, whether it manifests itself clinically or radiologically, in a spinal x-ray for example
<b>Family history of osteoporosis</b>	
Prolonged corticosteroid therapy Sedentary lifestyle	
Insufficient intake of dairy products	
Diseases causing osteoporosis: malabsorption, hyperthyroidism, hyperparathyroidism, chronic alcoholism, liver disease ...	• Whenever an x-ray suggests low bone mass (radiological osteopenia)

**I. Main systemic manifestations of rheumatic diseases**

<b>Associated diseases (in descending order of frequency)</b>	
<ul style="list-style-type: none"> <li><b>Constitutional manifestations</b></li> <li>Fever</li> <li>Weight loss</li> <li>Severe fatigue</li> </ul>	Systemic lupus erythematosus Systemic sclerosis Rheumatoid arthritis Mixed connective tissue disease Vasculitis ...
<ul style="list-style-type: none"> <li><b>Skin &amp; mucosal manifestations</b></li> <li>Photosensitivity</li> <li>Skin rash</li> <li>Purpura, ulcers</li> <li>Hair loss</li> <li>Oral &amp; genital aphthae</li> <li>Dry eyes &amp; mouth</li> <li>Red eye</li> <li>Balanitis ...</li> </ul>	Systemic lupus erythematosus Rheumatoid arthritis Psoriatic arthritis (psoriasis) Sjogren's syndrome Systemic sclerosis Reactive arthritis Behcet's disease & other vasculites ...
<ul style="list-style-type: none"> <li><b>Serositis</b></li> <li>Pleurisy/ pleural effusion</li> <li>Pericarditis</li> </ul>	Connective tissue diseases
Raynaud's phenomenon	Idiopathic Raynaud's phenomenon Systemic sclerosis Systemic lupus erythematosus ...
Dysphagia	Systemic sclerosis
Dyspnea	Connective tissue diseases
Lower limb edema, hypertension	Connective tissue diseases
Lower limb edema, hypertension	Connective tissue diseases
Lymphadenopathy	Connective tissue diseases
Muscular weakness	Myositis, overlap syndromes

**J. Most common causes of musculo-skeletal pain referred from visceral origin**

<b>Visceral origin</b>	<b>Location of referred pain</b>
Retroperitoneum	Upper lumbar region
Biliary tree	Right shoulder
Heart	Left shoulder & arm
Urinary tract	Lumbar & inguinal regions
Genital organs	Sacral region
Pleura	Thoracic wall
Esophagus	Dorsal region & sternum

**K. Some relevant questions in systematic enquiry in rheumatology**

<b>General</b>	<b>Known associated diseases Fever? Recent weight loss?</b>
Skin	Skin problems? Rash? Psoriasis? Nodules? Skin sensitivity to light? Hair loss?
Eyes & mucous membranes	Aphthae or mouth ulcers? Genital ulcers? Gritty eyes (as if they had sand in them?) Painful red eyes? Dry mouth? Loss of vision?
Respiratory tract & circulatory system	Shortness of breath? Cough? Chest pain? Lower – limb edema?

	Raynaud's phenomenon?
Digestive tract	Dysphagia? Dyspepsia? History of gastric ulcers? Diarrhoea?
Urinary tract	Change in urine color? Kidney stones?
Genitalia	Discharge? Dyspareunia?
Nervous system (peripheral & central)	Dysesthesia? Weakness? Convulsions? Alterations in behavior?
Global	Any other concerns that we have not talked about?

#### L. Most common diagnoses as per age group & gender

	Women	Men
Children 0-16	Trauma Post – viral arthritis Hip lesions Scoliosis Juvenile idiopathic arthritis	
Young adults 17-35	Fibromyalgia Rheumatoid arthritis	Soft tissue lesions Reactive arthritis Ankylosing spondylitis Rheumatoid arthritis
Middle – aged 36-65	Fibromyalgia Soft tissue lesions Osteoarthritis Rheumatoid arthritis Systemic lupus erythematosus	Soft tissue lesions Gout Osteoarthritis Rheumatoid arthritis
Elderly 65	Osteoarthritis Osteoporosis Rheumatoid arthritis Polimyalgia rheumatica Gout & psudogout	

#### Conclusion

Just as all roads lead to Rome, many routes can be followed to reach exact diagnosis depending on predominant manifestations in a systemic disease. One should not skip any of the stages of the disease without running the risk of making a mistake. Diagnosis of the syndrome and disease itself always requires a thorough well-planned questioning and it may not be possible and it is not possible without a careful and general examination of all the areas with presenting symptoms.