



## Spondyloarthropathy, Susceptibility to, 1

### Alternative Names

SPDA1  
Spondyloarthropathy  
Ankylosing Spondylitis, Susceptibility to  
Ankylosing Spondylitis  
AS  
Marie-Strumpell Spondylitis  
Bechterew Syndrome  
Poker Back  
Rheumatoid Spondylitis

### Record Category

Disease phenotype

### WHO-ICD

Diseases of the musculoskeletal system and connective tissue > Dorsopathies

### Incidence per 100,000 Live Births

51-100

### OMIM Number

106300

### Mode of Inheritance

Autosomal dominant with greater penetrance in males

### Gene Map Locus

6p21.3, 6p21.3

### Description

Ankylosing spondylitis (AS) is a chronic, inflammatory, progressive form of seronegative spondylitis, affecting mainly the axial skeleton, particularly the sacroiliac and spinal facet joints, and less commonly extra-articular organs. Chronic spinal inflammation in these joints leads to ankylosis, or complete fusion of the vertebra, resulting in loss of mobility of the spine. AS is a systemic rheumatic disease, and can cause inflammation in joints and in other organs such as eyes, heart, lungs and kidneys, as well. The major symptoms of the disease are pain and morning stiffness of spine and sacral areas. Inflammation in other joints, tendons or organs may also be present.

The disease course is highly variable; some patients showing episodes of transient pain, and some showing chronic severe back pain. Non-skeletal problems include iritis, uveitis, aortitis, pulmonary fibrosis, amyloidosis, and inflammatory bowel disease.

Ankylosing spondylitis is three times more common in men than in women. It typically affects young people, beginning between the ages of 15 and 30. Onset after the age of 40 is uncommon. Incidence rate of the disease has been calculated at 7-10 in 10,000 in Europe.

Diagnosis of the disease is based on clinical examination, X-ray findings and blood tests. The Human Leukocyte Antigen B27 (HLA-B27) has been shown to be strongly associated with AS, and it is used as a genetic marker for the disease. Although ankylosing spondylitis is known to run in families, its pattern of inheritance is unclear. Multiple genetic and environmental factors are likely play a part in determining the risk of developing this disorder. Treatment is mainly in the form of regular exercise and non-steroidal anti-inflammatory drugs. Severe hip involvement may require hip replacement surgery.

### Molecular Genetics

Substantial evidence indicates that AS arises as a consequence of abnormal immune response, which may be triggered by a complex of genetic and environmental factors. Analysis of twin data indicates a predominant role of the genetic component in the occurrence of AS, while the environmental factor has a limited contribution to the etiology of the disease.

A majority of patients with ankylosing spondylitis have been shown to have the tissue type HLA-B27, as compared to low frequencies in the general population. While some studies indicate that only 1% of the people who carry the HLA-B27 develop AS, other studies estimate that this allele accounts for 20-50% of AS genetic risk.



## Epidemiology in the Arab World

### Bahrain

Ebrahim et al. (2000) described a 33-year-old Bahraini male, with a history of localized pain in the lower back. The pain was aggravated on spinal movements, and lifting of heavy loads. MRI studies showed features, typical of discitis, with end plate irregularities in the region of T11-T12. NSAIDs, steroids and methotrexate were administered to the patient, following an initial diagnosis of seronegative rheumatoid arthritis. However, the patient later tested positive for HLA-B27, and a study of his family history showed a 30 year old HLA-B27 positive cousin diagnosed with ankylosing spondylitis. Discitis is a rare occurrence in cases of ankylosing spondylitis. For this, Ebrahim et al. (2000) suggested that the discitis in this patient was probably due to an inflammatory process caused by ankylosing spondylitis.

Kawar and Al-Sayegh (2005) assessed the functional status among patients with ankylosing spondylitis (AS), using the Bath Ankylosing Spondylitis Functional Index (BASFI) to correlate the disease with clinical measures of spinal mobility, back pain, and back morning stiffness. The spinal mobility was evaluated by using three anthropometric measures: Schober's maneuver, chest expansion, and neck rotation. The study included 52 male patients with a mean age of 37.2 years. Disease duration ranged from one year to 18 years. All patients had radiologic evidence of bilateral sacro-iliitis of grade two or higher. Forty-seven patients (90.3%) had axial involvement (neck pain, upper and lower back pain). Peripheral joint involvement (Knee, ankle, elbow or wrist) was observed in 11 patients (21.1%), whereas appendicular joint involvement (hip and shoulder) was detected in 19 patients (36.5%). It was found that 22 patients had back pain >50 on the pain severity scale with mean BASFI 3.21. For back morning stiffness, 42 patients had >50 on the back stiffness severity scale with mean BASFI 4.95. The results showed that patients having severe morning stiffness and severe limitation of lumbar and cervical mobility had higher BASFI scores and, therefore, poorer functional outcome compared with those having severe back pain and limited chest expansion.

### Egypt

[See: Kuwait > Uppal et al. 2006; United Arab Emirates > Al-Attia et al., 1998].

### Jordan

[See: Kuwait > Uppal et al. 2006; United Arab Emirates > Al-Attia et al., 1998].

### Kuwait

Sattar et al. (1988) described a patient with IDDM who, 2-years after diagnosis, went on to develop rheumatoid arthritis, followed by ankylosing spondylitis. HLA typing revealed the presence of HLA A2, A9, B8, B27, DR3, and DR4 antigens. Sattar et al. (1988) suspected that the patient was simultaneously genetically susceptible to all of these diseases.

Uppal et al. (2006) compared the clinical features and HLA-B27 positivity of spondyloarthropathic patients of Middle Eastern Ancestry with those of patients of South Asian ethnicity. Of the 58 patients studied, 29 were Arabs, including 17 Kuwaitis, six Egyptians, three Syrians, two Jordanians, and one Lebanese. A positive family history of the condition in first degree relatives was seen significantly more in the Arab patients. Uppal et al. (2006) attributed these findings to the high prevalence of consanguinity among Arab tribes. About 87% of the Arab patients typed positive for HLA B-27. Of the nine Kuwaiti patients with ankylosing spondylitis and six with undifferentiated spondyloarthritis, seven and three, respectively, were HLA B-27 positive.

[See also: Syria > Sattar et al., 1992].

### Lebanon

[See: Kuwait > Uppal et al. 2006; United Arab Emirates > Al-Attia et al., 1998].

### Oman

[See: United Arab Emirates > Al-Attia et al., 1998].

### Palestine

[See: United Arab Emirates > Al-Attia et al., 1998].

### Saudi Arabia

Al-Arfaj (1996) described the profile of 13 patients with ankylosing spondylitis of Arab origin, treated over a period of four years in Saudi Arabia. HLA-B27 was positive in 67% of these patients. All the patients had symmetric radiographic sacroiliitis and one-third exhibited dominant lower limb oligo- polyarthritis.

### Syria

Sattar (1992) reported a 38 year old Syrian woman with wide spread polyarteritis nodosa following an onset of HLA-B27 positive spondyloarthritis. In 1984, the patient visited the faculty of medicine due to a nine year suffering of low back pain and spinal pain, throughout the same year the patient demonstrated an episode of iritis, mouth ulcers, was diagnosed with seronegative spondyloarthritis, and was found to be positive for the HLA-B27 antigen. The subject underwent a seven year follow-up through which she developed progressive arthritis



of her hips and knees. During 1987, the patient suffered an intermittent fever, frequent headaches, Raynaud's phenomenon, transient blurring of her vision, and occasionally experienced transient weakness of her legs and vascular lesions of a lace pattern (livedo reticularis) on her legs. Throughout 1988, the subject developed a rapid transient bilateral loss of vision and was treated with corticosteroids. Right through the same year, the patient was admitted with an abrupt onset of a cold, blue, and painful right foot which required amputation regardless of the active therapeutic measures that was taken. Six days following the surgery, the patient became dyspnoeic and hypotensive and passed away. Sattar (1992) demonstrated the second case of the coexistence of polyarteritis nodosa and ankylosing spondylitis in a HLA-B27 positive patient anticipating for further research in the association of rheumatic diseases and genetically determined disorders.

Harfouch and Al-Cheikh (2011) assessed the HLA-B27 allele and its subtypes associated with ankylosing spondylitis (AS) in 50 Syrian patients. All patients had chronic inflammatory low back pain and X-ray evidence of bilateral sacroiliitis. None of the patients had symptoms or signs suggesting psoriasis, inflammatory bowel disease, Behcet disease, familial Mediterranean fever, or reactive arthritis. Eight patients demonstrated a family history of spondyloarthritis. HLA-B27 allele was found in 60% of AS patients compared to a frequency of 1.4% in 217 unrelated healthy Syrian controls.

[See also: Kuwait > Uppal et al., 2006; United Arab Emirates > Al-Attia et al., 1998].

#### United Arab Emirates

Al-Attia et al. (1998) undertook a retrospective study, spanning a period of 10 years on 17 Arab patients with ankylosing spondylitis. Patients were from Egypt, Lebanon, Jordan, Oman, Palestine, and Syria. They were all residents of UAE. No single patient was a UAE national. Most of the affected patients were males and the disease onset mostly occurred in adulthood. The very small number of cases suggested that the disease is uncommon in the Arab community. Only 56% of the Arab patients were positive for HLA-B27 as compared to 81% of Asians. Al-Attia et al. (1998) suggested that screening Arabs for B27 along with Bw60 would provide a better demarcation. Extra-articular manifestations were comparatively less numerous, and none of the patients had cardiac involvement.

#### References

Al Attia HM, Sherif AM, Hossain MM, Ahmed YH. The demographic and clinical spectrum of

Arab versus Asian patients with ankylosing spondylitis in the UAE. *Rheumatol Int.* 1998; 17(5):193-6. PMID: 9542780.

Al-Arfaj A. Profile of ankylosing spondylitis in Saudi Arabia. *Clin Rheumatol.* 1996; 15(3):287-9. PMID: 8793262.

Ebrahim RA, Sarwani NI, Kanekar SG. Ankylosing spondylitis presenting with discitis. *Saudi Med J.* 2000; 21(9):884-6. PMID: 11376371

Harfouch EI, Al-Cheikh SA. HLA-B27 and its subtypes in Syrian patients with ankylosing spondylitis. *Saudi Med J.* 2011; 32(4):364-8. PMID: 21483994

Kawar J, Al-Sayegh H. The relationship between clinical activity and function in ankylosing spondylitis patients. *Bahrain Med Bull.* 2005; 27(3):

Sattar MA, Al-Sughyer AA, Siboo R. Coexistence of rheumatoid arthritis, ankylosing spondylitis and dermatomyositis in a patient with diabetes mellitus and the associated linked HLA antigens. *Br J Rheumatol.* 1988; 27(2):146-9. PMID: 3365534

Sattar MA. Coexisting HLA-B27 positive spondyloarthritis and polyarteritis nodosa. *Ann Rheum Dis.* 1992; 51(12): 1338-9. PMID: 1362487

Uppal SS, Abraham M, Chowdhury RI, Kumari R, Pathan EM, Al Rashed A. Ankylosing spondylitis and undifferentiated spondyloarthritis in Kuwait: a comparison between Arabs and South Asians. *Clin Rheumatol.* 2006; 25(2):219-24. PMID: 16240074

#### Related CTGA Records

[Insulin Dependent Diabetes Mellitus](#)

Major Histocompatibility Complex, Class I, B  
[Rheumatoid Arthritis](#)

#### External Links

<http://ghr.nlm.nih.gov/condition=ankylosingspondylitis>

<http://www.emedicine.com/RADIO/topic41.htm>

<http://www.nass.co.uk/>

[http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=GB&Expert=825](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=GB&Expert=825)

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