Ankylosing Spondylitis in a Xhosa Father and Daughter

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SUMMARY

A Xhosa father and daughter with ankylosing spondylitis are reported. Review of the literature revealed a low incidence of ankylosing spondylitis in Blacks in general, with only isolated reports from the continent of Africa itself.

To our knowledge, there has been no documentation of ankylosing spondylitis in a Black female, or of ankylosing spondylitis in first-degree Black relatives. The clinical features of ankylosing spondylitis in the female, and the role of human leucocyte antigen (HLA) B27 in the disease are also discussed.


Ankylosing spondylitis is a disease which affects young adults, with a male preponderance of 6-10 to 1. The aetiology is unknown, but a single non-sex-linked autosomal dominant inheritance, which manifests a 70% penetrance in males and a 10% penetrance in females, has been proposed by Hersh et al. and by Stecher and Hersh.

No epidemiological data on the prevalence of ankylosing spondylitis in the South African Black is available, except for a recent study by Solomon et al. who found only 1 male with the disease in a Transvaal rural and urban community. There have been only isolated reports from the rest of the African continent over the last 15 years.

To our knowledge, no Black female with ankylosing spondylitis has been reported from South Africa. Furthermore, ankylosing spondylitis occurring in first-degree Black relatives, both with human leucocyte antigen (HLA) B27 present, has not been described. Two such patients are reported.

CASE REPORTS

Patient 1 (Figs 1 and 2)

A 31-year-old Xhosa woman presented with a 3-year history of pain, stiffness and limitation of movement in the lumbar region. The symptoms were worse in the morning, but were present throughout the day. Two years after the onset of her illness, she noted that the disability had spread to involve her thoracic spine and, more recently, her cervical spine. Shortly after the onset of her illness, she developed painful eyes, but the pain cleared spontaneously after 5 days. No other joints have been involved at any stage, and there was no history of dysentery, skin lesions or urinary symptoms. Her father has similar back symptoms and a sister has backache. She was admitted to hospital in 1970 with a respiratory infection.

Examination revealed a young Black woman with the typical features of ankylosing spondylitis. There was marked limitation of all movements of the lumbar spine, with a loss of the normal lumbar curvature on flexion. There was a marked anterior stoop of the thoracic spine, with chest expansion limited to 2.5 cm, as measured at the fourth intercostal space. Her neck was fixed in a flexed

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Fig. 1. Patient 1. Lateral view in upright position.
position and showed marked limitation of movement in all directions. There was flattening of the anterior chest and a protuberant abdomen. No other joints were involved and the rest of the examination revealed no other abnormalities. There was no evidence of a cardiac murmur.

The patient's haemoglobin concentration and white cell count were normal. Her erythrocyte sedimentation rate was 4 mm in the first hour. Her sheep-cell agglutination, latex and antinuclear factor tests were negative and serum protein concentration and liver function were normal. Tissue typing revealed HLA A28, W30, BW17, 27.

Radiological examination of the sacro-iliac joints showed bilateral sacro-iliitis with early ankylosis on the right. The ischial rami showed whiskering and the lumbar spine showed squaring of the vertebral bodies. Features of a 'bamboo' spine and costovertebral ankylosis were also present.

**Patient 2 (Figs 3 and 4)**

The 74-year-old father of the first patient described had noticed a stoop 14 years previously. Pain and stiffness had never been a feature. At no stage were any of his peripheral joints involved, and he had no history of diarrhoea, urethral discharge or eye problems.

Examination of this patient revealed features of ankylosing spondylitis which involved the lumbar, thoracic and cervical spine and were very similar to those of his daughter. There was severe limitation of all spinal movements and chest expansion was reduced to 1.5 cm. There were no cardiac murmurs, eye or skin lesions, and no neurological abnormalities. Tissue typing revealed HLA A9, W30, B7, 27.

Radiological examination revealed total ankylosis of the lumbar, dorsal and cervical spine with complete bony ankylosis of the sacro-iliac joints. There was also involvement of the symphysis pubis and whiskering of the ischial rami and iliac crests. There was, in addition, ankylosis of the costovertebral joints. The patient refused further investigation.

**DISCUSSION**

The clinical and radiological features of the 2 patients described fulfil the criteria for the diagnosis of ankylosing spondylitis in population studies.\(^9\,10\) Baum and Ziff\(^9\) have drawn attention to the low incidence of ankylosing spondylitis in Blacks. In a study of patients in 4 hospitals, they showed that the disease was 4 times more common in White Americans than in Black Americans. Furthermore, the isolated reports on Black Africans suggest an even lower incidence than in Black Americans.

The reports from Africa have been sporadic, and document 9 patients from widely differing latitudes who have the disease.\(^11\) These 9 patients comprise 8 men and
I woman. The woman was a Nigerian, whose symptoms started at the age of 50, and in whom radiological features developed 3 years later. One of the men had atypical features and the diagnosis of ankylosing spondylitis was in doubt.

In an epidemiological survey of rheumatic disease in urban and rural Blacks from the Republic of South Africa only 1 subject, a man aged 66, was found to have ankylosing spondylitis. In another epidemiological study, no clinical ankylosing spondylitis was found in a rural Xhosa population (unpublished data). Palmer thought that the disease was not rare. He reported no cases, but he recalled having seen in Rhodesia at least 5 Black men and 1 woman with the disease.

Ankylosing Spondylitis in the Female

In addition to a lower incidence in females, there are differences between males and females in the clinical and radiological features of ankylosing spondylitis. In the female, the disease has an earlier onset (occasionally precipitated by pregnancy or delivery), a milder course and an earlier morbidity peak. The disease begins more often in the lumbar region, the symphysis pubis is more commonly involved, and there is more symptomatic but less frequent radiological involvement of the cervical spine. Finally, extensive ankylosis of the vertebral bodies seldom occurs, paravertebral calcification is uncommon and radiological changes in the peripheral joints are present.

The female patient we have described had severe clinical and radiological signs of the disease, which were more typical of the presentation of ankylosing spondylitis in males.

**HLA W27 (HLA B27)* and Ankylosing Spondylitis**

The association of HLA B27 with ankylosing spondylitis is now well established. Brewerton *et al.* who first described this association in 1973, showed that 96% of Whites with the disease and 51.7% of first-degree relatives had B27, while only 4% of the control population had the antigen. Schlosstein *et al.* demonstrated that B27 was present in 80% of American Negroes with ankylosing spondylitis, with a control incidence of 4%. Swezey *et al.* reported 5 Black American females with the disease and found B27 in 2. No studies from South Africa on the incidence of B27 in ankylosing spondylitis have been reported.

Baum and Ziff suggested that the higher incidence of ankylosing spondylitis in American Negroes as compared with Black Africans was owing to racial intermixture, which has produced 25% of a White component in the former. Similarly, in the Blackfeet Indians there are fewer Caucasian genes and a lower incidence of ankylosing spondylitis than in the Haidas and Pimas.

Whether B27 is simply a genetic marker, or whether it, or a related antigen, plays a role in the development of ankylosing spondylitis, is open to speculation.

The association between B27 and ankylosing spondylitis is so marked that it is possible to assume either very close genetic linkage of a specific immunological responsiveness gene to the disease or, perhaps, a strong immunological cross-reaction between B27 and the aetiological agent involved. Because of the low incidence of B27 in Black females with ankylosing spondylitis compared with the high incidence in both Black and Caucasian males, Swezey *et al.* suggested that factors other than B27 are operative in the genesis of ankylosing spondylitis in Black females.

More epidemiological studies incorporating tissue typing are of importance in furthering the understanding of the pattern of rheumatic disease in Blacks.

**REFERENCES**


*New nomenclature as recommended after the 6th Histocompatibility Workshop held in Arhus in 1975 (6th press).*