NOW AND THEN

Rheumatic disease and the Australian Aborigine

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Abstract

Objective—To document the frequency and disease phenotype of various rheumatic diseases in the Australian Aborigine.

Methods—A comprehensive review was performed of the archaeological, ethnographical, and contemporary literature relating to rheumatic diseases in these indigenous people.

Results—No evidence was found to suggest that rheumatoid arthritis (RA), ankylosing spondylitis (AS), or gout occurred in Aborigines before or during the early stages of white settlement of Australia. Part of the explanation for the absence of these disorders in this indigenous group may relate to the scarcity of predisposing genetic elements, for example, shared rheumatoid epitope for RA, B27 antigen for AS. In contrast, osteoarthritis appeared to be common particularly involving the temporomandibular joint, right elbow and knees and, most probably, was related to excessive joint loading in their hunter-gatherer lifestyle. Since white settlement, high frequency rates for rheumatic fever, systemic lupus erythematosus, and pyogenic arthritis have been observed and there are now scanty reports of the emergence of RA and gout in these original Australians.

Conclusion—The occurrence and phenotype of various rheumatic disorders in Australian Aborigines is distinctive but with recent changes in diet, lifestyle, and continuing genetic admixture may be undergoing change. An examination of rheumatic diseases in Australian Aborigines and its changing phenotype may lead to a greater understanding of the aetipathogenesis of these disorders.

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Ancestors of Australian Aborigines migrated to Sahul (continental land mass of Australia and New Guinea) 40–60 000 years before present and have continued to occupy the land to the present time. From a biological perspective the subsequent cultural and biological evolution of these isolated people in their unique environment can be viewed as an excellent opportunity to examine differences in disease expression in comparison with racial groups in other lands. If differences are observed they may provide clues to the aetipathogenesis of these disorders. However, before examining the prevalence and phenotype of rheumatic diseases in the Australian Aborigines we need to acknowledge recent changes in genetic and environmental influences.

Up until white settlement the Australian Aborigines were considered to be genetically a fairly homogenous race but with some striking and distinctive regional variations particularly involving those HLA class II genes involved in immune responses.1 2 For example, in Aboriginal groups from different geographical regions, alleles with an apparent local origin accounts for 20–30% of the DRB1 gene frequencies.7 Furthermore, these indigenous people lived a hunter/gatherer lifestyle rich in cultural traditions but with little variation in their regional environmental influences. However, since settlement racial intermarriages have occurred with a mixture of white, Asian, Polynesian and Melanesian genes. In addition, there have been dramatic environmental changes including changes in diet and nutrition, lifestyle and exposure to various new infectious, chemical and toxic agents, etc. Thus today, tragically, the Australian Aborigines are characterised with high levels of obesity, diabetes, hypertension, chronic renal disease, hepatitis B, alcoholism, chronic unemployment, and suicide.3 It is important therefore to recognise that changes in disease prevalence or phenotype over the past 200 years may reflect these influences and that contemporary observations may not necessarily reflect the situation before white settlement. It should also be noted that rheumatic disease prevalence and perhaps phenotype may also change over time, for example, the probable increased prevalence of rheumatoid arthritis (RA) in the past 200 years.

Sources of information

The Australian Aborigines while rich in oral tradition have no written language and hence their own historical record from these people with regard to rheumatic diseases is not available. We must therefore seek other sources for this information. These alternative sources can be listed as (1) archaeological, (2) ethnographical/historical, (3) contemporary. Archaeological evidence of rheumatic disease is obtained from the examination of skeletal material obtained from aboriginal burial grounds or elsewhere and this study constitutes
the subject of paleopathology. A recent monograph on the paleopathology of Australian Aborigines has been published using data collected from 4500 subjects from representative parts of the continent. Furthermore, according to Steinbock and Ortner and Putschar the rheumatic diseases that can be diagnosed from skeletal remains include osteoarthritis, vertebral hyperostosis, traumatic arthritis, RA, ankylosing spondylitis (AS), pyogenic arthritis, psoriatic arthritis, Reiter’s syndrome, erosive osteoarthritis, and gout. Ethnographical evidence is obtained from the written historical records of observers who noted disease prevalence in their early contact with the Australian Aborigine. There is a rich source of this material and on many occasions involved informants who were medically trained. Contemporary data referred to the observation of current observers. In many instances the data are more complete but as discussed above, this information does not necessarily reflect the situation before white settlement.

Rheumatic diseases

OSTEOARTHRITIS

The paleopathological record describes extensive osteoarthritic changes in aboriginal skeletal remains where osteoarthritis is defined by the presence of one or more of the characteristic features of articular surface eburnation, erosions or pitting and marginal osteophytes. The predominant joints involved are the temporomandibular joint (TMJ), the spine, the elbow and the knee. Of interest, osteoarthritis of the elbow was particularly common and sometimes found in as many as 20% of the elbow joint examined. It was more common in men than women and involved the right elbow more than the left (20% versus 12% \( \chi^2 = 5.74, p<0.02 \)) and distinct regional variation in frequencies was observed. It has been speculated that the explanation for the high frequency of the involvement of the right elbow in men could be related to the excessive joint loading and stress involved in repetitive motions such as spear throwing, throwing weapons and stones and in their hunter lifestyle.

In general, the frequency of knee arthritis was low for those nomadic people living hunter gatherer lifestyles. Osteoarthritis of both knees were particularly common in Queensland coastal women and were attributable to the high level mechanical stress involved in gathering shellfish where the women dug their feet into the thick mud looking for shellfish, producing a strong twisting mechanical strain on the knee joint.

High frequencies of TMJ arthritis have been recorded in most indigenous populations in Australia with the highest frequency (60%) being found in the central Murray and South Coast region where arthritic changes were observed in subjects of both sexes and at a young age. The cause, presumably, for these accelerated changes relate to a multifactorial process involving jaw structure, dentition and tooth loss, type of bite, quantity of grit and abrasions and other inclusions in the food, food preparation and perhaps the use of teeth in net making, etc.

RHEUMATOID ARTHRITIS

There is no paleopathological or ethnographical evidence to support the existence of RA in indigenous people in Australia before or after white settlement. Furthermore, an analysis of the recent historical and contemporary literature reveals no descriptions of classic or definite RA occurring in these people. We have recently commented on the rarity of RA in Australian Aborigines and after a letter seeking further information, we have now been able to describe the clinical, laboratory, and immunogenetic features of seven Aborigines with probable or definite RA. It is of interest that these patients all derive from Northern Queensland and at least two of them have genetic evidence of prior racial mixture. Since this description we are now aware of a further 15 Aboriginal patients with RA all, however, having evidence of prior racial intermarriage (personal communication).

Why should the occurrence of RA be so rare in the Australian Aborigines? Part of the explanation may arise from the scarcity of the shared rheumatoid epitope in these people. The shared rheumatoid epitope is a motif on the MHC class II molecule involving antigen presentation as coded by a number of DRB1 alleles. This shared epitope is found in approximately 35% of healthy white populations but in 85% of patients with RA and may also be a risk factor for more severe disease outcome. The shared rheumatoid epitope occurs infrequently in the Australian Aborigine but it was of interest to note its presence in four of the seven patients described above. In Australia, however, there is some regional variation where the shared rheumatoid epitope occurs with a higher frequency in the Aborigines of the Kimberley and Cape York (northern Queensland) region. It will be of interest to see if RA occurs more commonly in these populations of indigenous people.

RA also seems rare or non-existent in New Guinea Highlanders who probably have the same ancestral origins as the Australian Aborigine. It will be of interest to determine if these people also lack the shared rheumatoid epitope. RA is also absent in a rural Nigerian population. These people also lacked the shared rheumatoid epitope encoded by chromosome six.

GOUT

No paleopathological description of gouty arthritis exists in the skeletal record and there is no historical evidence for acute gout or chronic tophaceous gout in Aboriginal subjects. In 1969 Emmerson and colleagues measured the serum uric acid concentration in 382 Gulf of Carpenteria Aborigines and significantly higher serum urate concentrations were noted. The men compared with the women had consistently higher concentrations in keeping with all racial groups studied throughout the world and concentrations tended to increase with age in both sexes. Of
interest was the finding that the mean serum urate concentration was significantly higher than those of a control white population by approximately 12%. No factors other than racial were found to explain the difference. Furthermore, Emmerson stated in his discussion that despite the higher mean serum uric acid in Aborigines, clinical attacks of gouty arthritis were extremely rare. Indeed, inquiry from the Royal Flying Doctor Service and from many Aboriginal settlements scattered throughout Australia failed to record a single case of gout in an Australian Aborigine. An updated Medline search has also failed to find any description of acute or chronic tophaceous gout in Australian Aborigines although we are now aware of several Aboriginal patients in the Top End who have crystal confirmed gout in association with chronic renal impairment. The scarcity of gout in Australian Aborigines should be contrasted with the situation in the Maori, other Polynesians and their ancestors where hyperuricaemia and clinical gout is very prevalent and furthermore, in Micronesia, was also frequent in the past. The scarcity of acute gout and gouty arthritis in the Australian Aborigine is intriguing and requires further examination.

SYSTEMIC LUPUS ERYTHEMATOSUS

Three studies have revealed a high prevalence of systemic lupus erythematosus (SLE) in Australian Aborigines, one in Western Australia, one in northern Queensland, and one in the top end of the Northern Territory. In this latter and more complete study the prevalence was estimated (a minimal figure) at 1:1900, at least twice the estimated prevalence of systemic lupus erythematosus in New Zealand Maori, other Polynesians and their ancestors. The aetiology of lupus in the Australian Aborigine is intriguing and requires further examination.

OTHER CONNeCTIVE TISSUE DISORDERS

We have personally observed the presence of scleroderma, dermatomyositis and ANCA related vasculitis in Australian Aborigines whose ancestral origins may involve prior genetic admixture.

ANKYLosing APONDYLIStIS AND OTHER B27 RELATED ARTHROPATHIES

There is no paleopathological, ethnographical, historical or contemporary evidence to support the occurrence of AS in the indigenous Australian Aborigines with pure ancestry. Some initial confusion arose when examination of axial skeletal remains suggested AS but a close examination of the published illustrations indicates that they more probably represented subjects with localised vertebral ankylosis following an infective discitis or diffuse idiopathic skeletal hyperostosis (DISH). Recent studies have revealed that AS is a genetic disorder with approximately 97% of the variance being determined by genetic factors. The HLA B27 and its allotypes is one of the important genes underlying the propensity to AS. In tissue typing on Australian Aborigines B27 appears to be at a very low frequency (compared with 6–8% in white populations); indeed, in a study of 186 subjects from central Australia it was not detected. "This absence could be in part, the explanation for the absence or low frequency of AS and other B27 related arthropathies in Australian Aborigines. As such, this finding in Australian Aborigines is similar to the absence of AS in other racial groups who also lack the B27 antigen.

RHEUMATIC FEVER

The prevalence of rheumatic fever and associated streptococcal reactive polyarthritis is high in central and Northern Australian Aboriginal populations. Indeed, one study in the Top End has shown the highest incidence rates of rheumatic fever worldwide with an annual incidence of acute rheumatic fever (1989–1993) between two to seven/104 children aged 4–14 years and 3% of people having established rheumatic heart disease. The cause of rheumatic fever is thought not to be caused by any major predisposition based on ethnicity but by the high carriage rate of the M5 family (historical rheumatic fever associated M types) group A streptococcus in Aboriginal subjects in tropical Australia, which in turn is related to overcrowding and continuous poor living conditions. It is not known whether acute rheumatic fever occurred in Australian Aborigines before white settlement but case descriptions of acute rheumatism were increasingly recorded after settlement.

JOINT AND BONE INFECTION

Evidence of pyogenic arthritis leading to joint ankylosis and chronic bone infection has been commonly observed in the paleopathological record. Widespread osteitis and periostitis frequently involving the long bones or the skull is thought to represent chronic bone infection. Webb discusses in detail whether this represents endemic treponemal infection (Yaws) or the spirochaete possibly being introduced by the Macassans who fished northern Australian seas for the sea cucumber or from a form of sexually transmitted syphilitic bony infection (although the presence of syphilis before colonisation seems unlikely). These bony infections seem particularly common in the central Murray Aboriginal populations where the frequency is as high as 17% although similar changes are seen in skeletal remains from most parts of Australia and are prominent around arid and tropical regions (but absent in Tasmanian Aborigines).

Pyogenic arthritis is common in contemporary records frequently involving gonococcus reflecting the high prevalence of sexually related diseases in the Australian Aboriginal population.
Table 1  Australian Aborigines. Diseases of the joints, bones, and muscles

<table>
<thead>
<tr>
<th>Indigenous treatments</th>
<th>Fractures</th>
<th>Rheumatism</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>- splinting</td>
<td>- emu oil liniment</td>
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<tr>
<td></td>
<td>- hot ash massage</td>
<td>- steam vapour bath over smouldering fire</td>
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<tr>
<td></td>
<td>- seated in hot ash mount</td>
<td>- animal urine as rubefacient</td>
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<td></td>
<td>- local bleeding with scarification, then suck afflicted part</td>
<td>- dried and powdered shea oil apple</td>
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<td></td>
<td>- fumigation over a smouldering fire on which green leaves had been thrown</td>
<td>- fumigation over a smouldering fire on which green leaves had been thrown (Acacia lysiphloia or pinggi waterweed)</td>
</tr>
</tbody>
</table>

++ = common, + = described, − = not described, blank = no data.

Ethnographic/ Paleopathology ++ ++ − +/− + − ++

Indigenous treatments

Contempory ++ ++ − − + +/− ++ − ++ +

Historic

Rheumatism - emu oil liniment
Fractures - splinting
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relative frequencies of these various rheumatic diseases in Australian Aborigines subdivided according to paleopathological, ethnohistorical or contemporary data. It should be observed that the common inflammatory arthritides of gout, RA, and B27 related arthropathies occur rarely in Australian Aborigines in contrast with their common occurrence in white populations. Part of the explanation for the absence of these rheumatic disorders may be because of the lack of predisposing genetic elements. In contrast, SLE, acute rheumatic fever, osteoarthritis of the elbow and TMJ occurs frequently and this is probably most explicable on genetic and lifestyle environmental factors. However, the total picture is far from complete. Further studies into the prevalence and phenotypes of rheumatic diseases occurring in the Australian Aborigine (particularly during times of environmental and genetic change) may give further insights into the aetiopathogenesis of these potentially disabling disorders.

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