

## Case report

# Crohn's spondylitis: a family study

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**SUMMARY** We report a case of HLA B27 negative Crohn's spondylitis with a detailed study of the proband's family, including tissue types of all living relatives. The family included cases of psoriasis, psoriatic spondylitis, and idiopathic ankylosing spondylitis. We believe this to be the first report of these associated diseases occurring in one family. All relatives were B27 negative, including one with psoriatic spondylitis. The aetiological implications of these findings are considered.

The association of Crohn's disease and ankylosing spondylitis is well recognised, as is the familial aggregation of patients with Crohn's disease, with psoriasis, and with psoriatic arthritis. These interrelationships and others in the seronegative spondyarthropathies have been extensively discussed.<sup>1,2</sup> In view of the current interest in these interrelationships we studied the family of a 49-year-old woman who presented with Crohn's spondylitis and whose pedigree included relatives with psoriasis, psoriasis and ankylosing spondylitis, and idiopathic ankylosing spondylitis.

### Case report and pedigree

A 49-year-old Caucasian female developed Crohn's disease at the age of 35. She was treated conservatively with intermittent topical and systemic steroids and sulphasalazine until she was 39, when she underwent a bowel resection and ileostomy. She began to complain of intermittent back pain and stiffness shortly after this and subsequently developed a peripheral arthritis involving her knees, ankles, and right shoulder. In 1980 pelvic and lumbar spine x-rays revealed bilateral sacroiliitis, lumbar spine discitis, and asymmetrical syndesmophyte formation. The diagnosis of Crohn's spondylitis was made and she was commenced on indomethacin 50 mg t.d.s. with good effect. At the age of 34 she had an attack of iritis, but there was no history of psoriasis. A detailed family study, including tissue typing of all 15

living blood relatives, revealed the pedigree shown in Fig. 1.

### Discussion

Pedigrees containing ulcerative colitis, Crohn's disease, and ankylosing spondylitis have been described,<sup>3</sup> as have those containing psoriatic spondylitis, uncomplicated ankylosing spondylitis, and ulcerative colitis.<sup>4</sup> The disease association pattern exhibited by this family is not surprising, therefore, although we believe this is the first report of such a pedigree. The available tissue types of this family are of particular interest. The association of HLA B27 and ankylosing spondylitis is now well recognised. Over 90% of patients carry this tissue type. However, the carriage rate in patients with Crohn's disease and ankylosing spondylitis is probably less than this, being quoted as low as 53% in one study.<sup>5</sup> Similar carriage rates are seen in ankylosing spondylitis associated with psoriasis. The reason for this dilution of B27 is not fully understood, but it has been suggested that the presence of possible genetic determinants for psoriasis and inflammatory bowel disease predispose individuals to develop ankylosing spondylitis in the absence of B27.<sup>6,7</sup> The family reported here would support this hypothesis.

We have been unable to demonstrate B27 in any individual we have tissue typed in the proband's generation or their children. Unfortunately, neither of the proband's parents was available for tissue typing, and only one tissue type was obtained in this generation, but the spondylitic maternal uncle of our proband was B27 negative, and all 4 parental haplotypes were seen in the children and were B27 negative.

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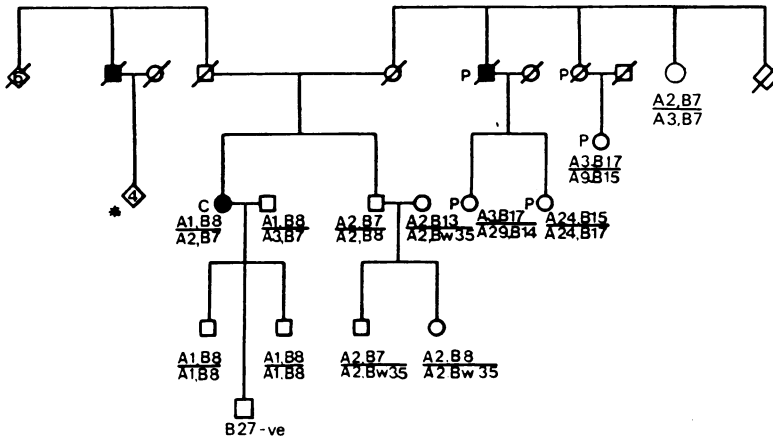


Fig. 1 Family pedigree. P = psoriasis. C = Crohn's disease. ■ = Ankylosing spondylitis. \* = All 4 typed: none are B27, but only 3 haplotypes are exhibited.

It is possible that the occurrence of psoriasis on the maternal side of the family was purely fortuitous. However, it is interesting to note that on this side of the family, in addition to those individuals with psoriasis alone, the spondylitic uncle also had psoriasis.

This family study therefore provides further evidence that the carriage of B27 is not necessary for development of ankylosing spondylitis, particularly in Crohn's disease and psoriasis, and suggests that other genetic and/or environmental factors are important.

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